

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
2-aminoadipic 2-oxoadipic aciduria	DHTKD1	614984	1.260,00 €					
2-methylbutyrylglucosuria	ACADSB	600301	796,00 €					
3-beta-hydroxysteroid dehydrogenase deficiency type 2	HSD3B2	613890	468,00 €	437,00 €	755,00 €			x
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL	613898	737,00 €	421,00 €	1.008,00 €			
3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency	HMGCS2	600234	819,00 €	328,00 €	997,00 €			
3-hydroxyisobutryl-CoA hydrolase deficiency	HIBCH	610690	995,00 €					
3MC syndrome type 1	MASP1	600521	1.278,00 €					
3MC syndrome type 2	COLEC11	612502	729,00 €					x
3-methylglutaconic aciduria type 1	AUH	600529	729,00 €	429,00 €	1.008,00 €			x
3-methylglutaconic aciduria type 3	OPA3	606580	468,00 €	343,00 €	661,00 €			x
3-methylglutaconic aciduria type 5	DNAJC19	608977	573,00 €					
3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	SERAC1	614725	1.127,00 €	424,00 €	1.401,00 €			
5-oxoprolinase deficiency	OPLAH	614243	1.127,00 €					
6q24-related transient neonatal diabetes mellitus type 1	UPD chr. 6			328,00 €				
17-beta hydroxysteroid dehydrogenase X deficiency	HSD17B10	300256	573,00 €					
17-hydroxylation activity deficiency	CYP17A1	609300	737,00 €	328,00 €	915,00 €			x
46,XX sex reversal type 1	SRY	480000	374,00 €	328,00 €	552,00 €			
46,XY gonadal dysgenesis, partial, with minifascicular neuropathy	DHH	605423	468,00 €	374,00 €	692,00 €			x
46,XY sex reversal type 8, modifier of	AKR1C4	600451	819,00 €					
Abetalipoproteinemia	MTTP	157147	1.326,00 €					
Accelerated tumor formation, susceptibility to	MDM2	164785	862,00 €	328,00 €	1.040,00 €			
Acetylcholinesterase deficiency	ACHE	100740	573,00 €					
Acetyl-CoA carboxylase deficiency	ACACA	200350	1.955,00 €					
Achalasia addisonianism alacrimia syndrome	AAAS	605378	862,00 €	424,00 €	1.136,00 €			x
Achalasia addisonianism alacrimia syndrome	AAAS	605378	862,00 €	424,00 €	1.136,00 €			x
Achondrogenesis type 1A	TRIP11	604505	1.622,00 €	442,00 €	1.914,00 €			x
Achondrogenesis type 1B	SLC26A2	606718	655,00 €	374,00 €	879,00 €			x
Achondrogenesis type 2	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Achondrogenesis type 2	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Achondroplasia	FGFR3	134934	995,00 €	421,00 €	1.266,00 €			x
Achromatopsia type 2	CNGA3	600053	928,00 €					
Achromatopsia type 3	CNGB3	605080	1.260,00 €	421,00 €	1.531,00 €			x
Achromatopsia type 4	GNAT2	139340	737,00 €					
Achromatopsia type 6	PDE6H	601190	468,00 €					
Acne inversa familial type 3	PSEN1	104311	928,00 €	328,00 €	1.106,00 €			x
Acrocallosal syndrome	KIF7	611254	1.326,00 €	430,00 €	1.606,00 €			x
Acrodermatitis enteropathica	SLC39A4	607059	729,00 €	421,00 €	1.000,00 €			x
Acrodysostosis 2	PDE4D	600129	1.521,00 €	445,00 €	1.816,00 €			x
Acrodysostosis type 1, with or without hormone resistance	PRKAR1A	188830	796,00 €	429,00 €	1.075,00 €			x
Acrofacial dysostosis 1, Nager type	SF3B4	605593	655,00 €	421,00 €	926,00 €			x

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Acromegaly, predisposition to, due to germline GPR101 mutation	GPR101	300393	491,00 €					
Acromelic frontonasal dysostosis	ZSWIM6	615951	1.193,00 €	437,00 €	1.480,00 €			x
Acromesomelic dysplasia, Maroteaux type	NPR2	108961	1.278,00 €	463,00 €	1.591,00 €			x
Acromicric dysplasia	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			x
Acute myeloid leukemia, somatic, DNMT3A related	DNMT3A	602769					1.900,00 €	
Acyl-CoA medium-chain dehydrogenase deficiency	ACADM	607008	862,00 €	426,00 €	1.138,00 €			x
Acyl-CoA multiple dehydrogenase deficiency	ETFA	608053	995,00 €	421,00 €	1.266,00 €			x
Acyl-CoA multiple dehydrogenase deficiency	ETFB	130410	819,00 €	390,00 €	1.059,00 €			x
Acyl-CoA peroxisomal oxidase deficiency	ACOX1	609751	796,00 €					
Acyl-CoA short-chain dehydrogenase deficiency	ACADS	606885	819,00 €	429,00 €	1.098,00 €			x
Acyl-CoA very long-chain dehydrogenase deficiency	ACADVL	609575	1.127,00 €	328,00 €	1.305,00 €			x
Adams-Oliver syndrome type 1	ARHGAP31	610911	1.326,00 €	421,00 €	1.597,00 €			x
Adams-Oliver syndrome type 2	DOCK6	614194	2.028,00 €	824,00 €	2.702,00 €			x
Adams-Oliver syndrome type 3	RBPJ	147183	995,00 €					
Adams-Oliver syndrome type 4	EOGT	614789	1.127,00 €	421,00 €	1.398,00 €			
Adams-Oliver syndrome type 6	DLL4	605185	729,00 €					
Adenine phosphoribosyltransferase deficiency	APRT	102600	491,00 €					
Adenocarcinoma of lung, somatic	BRAF	164757					1.290,00 €	
Adenocarcinoma of lung, somatic	ERBB2	164870					1.900,00 €	
Adenoma, periampullary, somatic	APC	611731					1.900,00 €	
Adenosine triphosphate, elevated, of erythrocytes	PKLR	609712	796,00 €	328,00 €	974,00 €			x
Adenylosuccinase deficiency	ADSL	608222	995,00 €					
Adrenal adenoma, somatic	MEN1	613733					1.900,00 €	
Adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	613815	819,00 €	328,00 €	997,00 €			x
Adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	POR	124015	1.061,00 €	328,00 €	1.239,00 €			x
Adrenal hyperplasia due to steroid 11-beta-hydroxylase deficiency	CYP11B1	610613	819,00 €	421,00 €	1.090,00 €			x
Adrenal hypoplasia	NR0B1	300473	491,00 €	328,00 €	669,00 €			x
Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	CYP11A1	118485	819,00 €					x
Adrenocorticotrophic hormone deficiency	TBX19	604614	737,00 €					
Adrenoleukodystrophy, x-linked	ABCD1	300371	862,00 €	328,00 €	1.040,00 €			x
Adrenoleukodystrophy, x-linked	PLXNB3	300214	2.028,00 €					
Adrenoleukodystrophy/Adrenomyeloneuropathy	ABCD1	300371	862,00 €	328,00 €	1.040,00 €			x
ADULT syndrome, split hand-foot malformation	TP63	603273	1.061,00 €	424,00 €	1.335,00 €			x
Afibrinogenemia, congenital	FGA	134820	655,00 €					
Afibrinogenemia, congenital	FGB	134830	819,00 €	437,00 €	1.106,00 €			
Afibrinogenemia, congenital	FGG	134850	928,00 €					
Agammaglobulinemia and isolated hormone deficiency	BTK	300300	1.338,00 €	328,00 €	1.516,00 €			x
Agammaglobulinemia type 1, autosomal recessive	IGHM	147020	468,00 €					
Agammaglobulinemia type 1, X-linked	BTK	300300	1.338,00 €	328,00 €	1.516,00 €			x
Agammaglobulinemia type 2, autosomal recessive	IGLL1	146770	374,00 €					

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Agammaglobulinemia type 3, autosomal recessive	CD79A	112205	374,00 €					
Agammaglobulinemia type 4, autosomal recessive	BLNK	604515	1.193,00 €					
Agammaglobulinemia type 5, autosomal recessive	LRRC8A	608360	573,00 €					
Agammaglobulinemia type 6, autosomal recessive	CD79B	147245	468,00 €					
Agammaglobulinemia type 7, autosomal recessive	PIK3R1	171833	1.260,00 €					
Agensis of the corpus callosum with peripheral neuropathy	SLC12A6	604878	1.643,00 €	527,00 €	2.020,00 €			x
Aicardi-Goutieres syndrome type 1	TREX1	606609	374,00 €	328,00 €	552,00 €			x
Aicardi-Goutieres syndrome type 2	RNASEH2B	610326	796,00 €	328,00 €	974,00 €			x
Aicardi-Goutieres syndrome type 3	RNASEH2C	610330	374,00 €	328,00 €	552,00 €			x
Aicardi-Goutieres syndrome type 4	RNASEH2A	606034	655,00 €	328,00 €	833,00 €			x
Aicardi-Goutieres syndrome type 5	SAMHD1	606754	1.061,00 €	328,00 €	1.239,00 €			x
Aicardi-Goutieres syndrome type 6	ADAR	146920	1.326,00 €					
Aicardi-Goutieres syndrome type 7	IFIH1	606951	1.127,00 €					
Alacrima, achalasia and mental retardation syndrome	GMPPA	615495	796,00 €					
Alagille syndrome type 1	JAG1	601920	1.643,00 €	328,00 €	1.821,00 €			x
Alagille syndrome type 2	NOTCH2	600275	1.977,00 €	716,00 €	2.543,00 €			x
Aland Island eye disease	CACNA1F	300110	1.802,00 €					
Alazami syndrome	LARP7	612026	862,00 €	445,00 €	1.157,00 €			
Albinism, ocular type I, Nettleship-Falls type	GPR143	300808	729,00 €	328,00 €	907,00 €			x
Albinism, oculocutaneous nonsyndromic	SLC24A5	609802	737,00 €					x
Albinism, oculocutaneous nonsyndromic	SLC24A5	609802	737,00 €					x
Albinism, oculocutaneous type 1A	TYR	606933	655,00 €	328,00 €	833,00 €			x
Albinism, oculocutaneous type 1A	TYR	606933	655,00 €	328,00 €	833,00 €			x
Albinism, oculocutaneous type 1B	TYR	606933	655,00 €	328,00 €	833,00 €			x
Albinism, oculocutaneous type 1B	TYR	606933	655,00 €	328,00 €	833,00 €			x
Albinism, oculocutaneous type 2	OCA2	611409	1.460,00 €	328,00 €	1.638,00 €			x
Albinism, oculocutaneous type 2	OCA2	611409	1.460,00 €	328,00 €	1.638,00 €			x
Albinism, oculocutaneous type 3	TYRP1	115501	655,00 €	437,00 €	942,00 €			x
Albinism, oculocutaneous type 3	TYRP1	115501	655,00 €	437,00 €	942,00 €			x
Albinism, oculocutaneous type 4	SLC45A2	606202	655,00 €	437,00 €	942,00 €			x
Albinism, oculocutaneous type 4	SLC45A2	606202	655,00 €	437,00 €	942,00 €			x
Albinism, oculocutaneous type 5	C10ORF11	614537	819,00 €					x
Albinism, oculocutaneous type 5	C10ORF11	614537	819,00 €					x
Alexander disease	GFAP	137780	796,00 €	437,00 €	1.083,00 €			x
Alkaptonuria	HGD	607474	995,00 €	437,00 €	1.282,00 €			x
Allan-Herndon-Dudley syndrome	SLC16A2	300095	655,00 €	421,00 €	926,00 €			x
Allan-Herndon-Dudley syndrome	SLC16A2	300095	655,00 €	421,00 €	926,00 €			x
Alopecia universalis	HR	602302	1.193,00 €	430,00 €	1.473,00 €			
Alpha-2-macroglobulin deficiency	A2M	103950	1.802,00 €					
Alpha-ketoglutarate dehydrogenase deficiency	OGDH	613022	1.399,00 €					

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Alpha-methylacyl CoA racemase deficiency	AMACR	604489	655,00 €	421,00 €	926,00 €			x
Alpha-thalassemia/mental retardation syndrome	ATRX	300032	1.845,00 €	328,00 €	2.023,00 €			x
Alpha-thalassemia/mental retardation syndrome	ATRX	300032	1.845,00 €	328,00 €	2.023,00 €			x
Alpha-thalassemia/mental retardation syndrome	ATRX	300032	1.845,00 €	328,00 €	2.023,00 €			x
Alport syndrome, autosomal recessive	COL4A3	120070	2.067,00 €	328,00 €	2.245,00 €			x
Alport syndrome, autosomal recessive	COL4A3	120070	2.067,00 €	328,00 €	2.245,00 €			x
Alport syndrome, autosomal recessive	COL4A4	120131	1.973,00 €	328,00 €	2.151,00 €			x
Alport syndrome, autosomal recessive	COL4A4	120131	1.973,00 €	328,00 €	2.151,00 €			x
Alport syndrome, X-Linked	COL4A5	303630	2.067,00 €	655,00 €	2.572,00 €			x
Alport syndrome, X-Linked	COL4A5	303630	2.067,00 €	655,00 €	2.572,00 €			x
Al-Raqad syndrome	DCPS	610534	573,00 €					
Alstrom syndrome	ALMS1	606844	2.028,00 €	484,00 €	2.362,00 €			x
Alternating hemiplegia of childhood type 1	ATP1A2	182340	1.399,00 €	328,00 €	1.577,00 €			x
Alternating hemiplegia of childhood type 2	ATP1A3	182350	1.326,00 €	328,00 €	1.504,00 €			x
Alveolar capillary dysplasia with misalignment of pulmonary veins	FOXF1	601089	491,00 €	328,00 €	669,00 €			x
Alzheimer disease type 1	APP	104760	1.278,00 €	328,00 €	1.456,00 €			x
Alzheimer disease type 2	APOE	107741	655,00 €	437,00 €	942,00 €			
Alzheimer disease type 3	PSEN1	104311	928,00 €	328,00 €	1.106,00 €			x
Alzheimer disease type 4	PSEN2	600759	729,00 €	328,00 €	907,00 €			x
Alzheimers disease, early onset, autosomal dominant	SORL1	602005	2.102,00 €	905,00 €	2.857,00 €			
Alzheimers disease, RTN3 related	RTN3	604249	729,00 €					
Amelogenesis imperfecta type 1A	LAMB3	150310	1.460,00 €	484,00 €	1.794,00 €			x
Amelogenesis imperfecta type 1B	ENAM	606585	1.127,00 €					
Amelogenesis imperfecta type 1C	ENAM	606585	1.127,00 €					
Amelogenesis imperfecta type 1E	AMELX	300391	491,00 €					
Amelogenesis imperfecta type 1F	AMBN	601259	729,00 €					
Amelogenesis imperfecta type 1G	FAM20A	611062	862,00 €					
Amelogenesis imperfecta type 1H	ITGB6	147558	1.127,00 €					
Amelogenesis imperfecta type 2A1	KLK4	603767	491,00 €					
Amelogenesis imperfecta type 2A2	MMP20	604629	729,00 €					
Amelogenesis imperfecta type 2A3	WDR72	613214	1.399,00 €					
Amelogenesis imperfecta type 2A4	C4orf26	614829	468,00 €					
Amelogenesis imperfecta type 2A5	SLC24A4	609840	1.338,00 €					
Amelogenesis imperfecta type 3	FAM83H	611927	796,00 €					
Amelogenesis imperfecta type 4	DLX3	600525	374,00 €	374,00 €	598,00 €			x
Amelotin deficiency	AMTN	610912	737,00 €					
Aminoacylase deficiency	ACY1	104620	796,00 €					
Amish infantile epilepsy syndrome	ST3GAL5	604402	737,00 €					
AMP deaminase deficiency, erythrocytic	AMPD3	102772	1.127,00 €					
Amyloidosis	TTR	176300	491,00 €	437,00 €	778,00 €			

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Amyloidosis, familial visceral	APOA1	107680	491,00 €	437,00 €	778,00 €			
Amyloidosis, finnish type	GSN	137350	1.260,00 €					x
Amyloidosis, primary localized cutaneous, type 1	OSMR	601743	1.326,00 €					
Amyloidosis, primary localized cutaneous, type 2	IL31RA	609510	1.326,00 €					
Amyotrophic lateral sclerosis risk factor	CHGB	118920	729,00 €					
Amyotrophic lateral sclerosis type 1	SOD1	147450	729,00 €	390,00 €	969,00 €			x
Amyotrophic lateral sclerosis type 2, juvenile	ALS2	606352	1.724,00 €	716,00 €	2.290,00 €			x
Amyotrophic lateral sclerosis type 4	SETX	608465	1.825,00 €	328,00 €	2.003,00 €			x
Amyotrophic lateral sclerosis type 6	FUS	137070	995,00 €	445,00 €	1.290,00 €			x
Amyotrophic lateral sclerosis type 8	VAPB	605704	573,00 €					x
Amyotrophic lateral sclerosis type 9	ANG	105850	386,00 €					x
Amyotrophic lateral sclerosis type 10	TARDBP	605078	819,00 €	421,00 €	1.090,00 €			x
Amyotrophic lateral sclerosis type 11	FIG4	609390	1.460,00 €	484,00 €	1.794,00 €			x
Amyotrophic lateral sclerosis type 12	OPTN	602432	928,00 €	424,00 €	1.202,00 €			x
Amyotrophic lateral sclerosis type 14	VCP	601023	1.127,00 €					x
Amyotrophic lateral sclerosis type 16	SIGMAR1	601978	491,00 €					
Amyotrophic lateral sclerosis type 17	CHMP2B	609512	573,00 €					
Amyotrophic lateral sclerosis type 18	PFN1	176610	374,00 €					
Amyotrophic lateral sclerosis type 21	MATR3	164015	1.193,00 €	421,00 €	1.464,00 €			
Amyotrophic lateral sclerosis with frontotemporal dementia	C9orf72	614260				200,00 €		
Amyotrophic lateral sclerosis, CREST related	SS18L1	606472	928,00 €					
Amyotrophic lateral sclerosis, susceptibility to	NEFH	162230	862,00 €	437,00 €	1.149,00 €			x
Amyotrophic lateral sclerosis, VPS54 related	VPS54	614633	1.521,00 €					
Amyotrophic lateral sclerosis, x-linked juvenile and adult-onset ALS	UBQLN2	300264	491,00 €					x
Amyotrophy hereditary neuralgic	SEPT9	604061	1.127,00 €	328,00 €	1.305,00 €			x
Andersen disease	GBE1	607839	1.127,00 €	424,00 €	1.401,00 €			x
Androgen insensitivity	AR	313700	1.061,00 €	328,00 €	1.239,00 €	200,00 €		x
Androgen insensitivity, partial, with or without breast cancer	AR	313700	1.061,00 €	328,00 €	1.239,00 €	200,00 €		x
Androgen-binding protein deficiency	SHBG	182205	573,00 €					
Anemia dyserythropoietic type 1A	CDAN1	607465	1.825,00 €	590,00 €	2.265,00 €			x
Anemia dyserythropoietic type 2	SEC23B	610512	1.278,00 €	437,00 €	1.565,00 €			x
Anemia, neonatal hemolytic, fatal and near-fatal	SPTB	182870	1.927,00 €					
Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive	GLRX5	609588	386,00 €					
Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive	SLC25A38	610819	655,00 €					
Anemia, sideroblastic, with ataxia	ABCB7	300135	1.061,00 €					
Anemia, sideroblastic, X-linked	ALAS2	301300	796,00 €					
Anemia, X-linked	GATA1	305371	655,00 €	421,00 €	926,00 €			x
Angelman syndrome	chr. 15q11			328,00 €				x
Angelman syndrome	UBE3A	601623	1.061,00 €	328,00 €	1.239,00 €			x
Angelman-like syndrome	CDKL5	300203	1.460,00 €	328,00 €	1.638,00 €			x

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Angelman-like syndrome	MECP2	300005	468,00 €	328,00 €	646,00 €			x
Angioedema, hereditary	SERPING1	606860	819,00 €	328,00 €	997,00 €			x
Angiofibroma, somatic	MEN1	613733					1.900,00 €	
Anhaptoglobinemia	HP	140100	737,00 €					
Aniridia	PAX6	607108	1.061,00 €	328,00 €	1.239,00 €			x
Ankyloblepharon-ectodermal defects-cleft lip/palate	TP63	603273	1.061,00 €	424,00 €	1.335,00 €			x
Anterior segment mesenchymal dysgenesis	PITX3	602669	491,00 €					
Anterior segment mesenchymal dysgenesis	PITX3	602669	491,00 €					
Antithrombin III deficiency	SERPINC1	107300	819,00 €	328,00 €	997,00 €			x
Antitrypsin-alpha-1 deficiency	SERPINA1	107400	573,00 €	437,00 €	860,00 €			x
Antley-Bixler syndrome	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Aortic aneurysm, familial thoracic type 3	TGFBR2	190182	862,00 €	328,00 €	1.040,00 €			x
Aortic aneurysm, familial thoracic type 4	MYH11	160745	2.028,00 €	905,00 €	2.783,00 €			
Aortic aneurysm, familial thoracic type 5	TGFBR1	190181	819,00 €	328,00 €	997,00 €			x
Aortic aneurysm, familial thoracic type 6	ACTA2	102620	737,00 €	421,00 €	1.008,00 €			x
Aortic aneurysm, familial thoracic type 7	MYLK	600922	1.724,00 €					
Aortic aneurysm, familial thoracic type 8	PRKG1	176894	1.326,00 €					
Aortic aneurysm, familial thoracic, MAT2A related	MAT2A	601468	819,00 €					
Aortic valve disease type 1	NOTCH1	190198	1.876,00 €	716,00 €	2.442,00 €			x
Aortic valve disease type 2	SMAD6	602931	737,00 €	437,00 €	1.024,00 €			x
Apert syndrome	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Aplastic anemia	PRF1	170280	491,00 €	328,00 €	669,00 €			x
Aplastic anemia	TERC	602322	296,00 €	328,00 €	474,00 €			x
Aplastic anemia, SBDS related	SBDS	607444	491,00 €	390,00 €	731,00 €			x
Apolipoprotein C-II deficiency	APOC2	608083	386,00 €	437,00 €	673,00 €			x
Apparent mineralocorticoid excess	HSD11B2	614232	573,00 €	390,00 €	813,00 €			x
Arginase deficiency	ARG1	608313	655,00 €					
Arginine-glycine amidinotransferase deficiency	GATM	602360	819,00 €	421,00 €	1.090,00 €			x
Argininosuccinic aciduria	ASL	608310	862,00 €	424,00 €	1.136,00 €			x
Aromatase deficiency	CYP19A1	107910	819,00 €	429,00 €	1.098,00 €			
Aromatic L-amino acid decarboxylase deficiency	DDC	107930	1.061,00 €					
Arrhythmogenic right ventricular cardiomyopathy type 1	TGFB3	190230	796,00 €	328,00 €	974,00 €			
Arrhythmogenic right ventricular cardiomyopathy type 5	TMEM43	612048	862,00 €					
Arrhythmogenic right ventricular cardiomyopathy type 8	DSP	125647	1.927,00 €	328,00 €	2.105,00 €			x
Arrhythmogenic right ventricular cardiomyopathy type 9	PKP2	602861	1.127,00 €	328,00 €	1.305,00 €			x
Arrhythmogenic right ventricular cardiomyopathy type 10	DSG2	125671	1.193,00 €	328,00 €	1.371,00 €			x
Arrhythmogenic right ventricular cardiomyopathy type 11	DSC2	125645	1.278,00 €	328,00 €	1.456,00 €			x
Arrhythmogenic right ventricular cardiomyopathy type 12	JUP	173325	1.193,00 €					
Arrhythmogenic right ventricular dysplasia type 2	RYR2	180902	1.955,00 €					x
Arterial calcification type 1, generalized, infantile	ENPP1	173335	1.582,00 €	527,00 €	1.959,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Arterial calcification type 2, generalized, infantile	ABCC6	603234	1.572,00 €	328,00 €	1.750,00 €			x
Arterial Tortuosity Syndrome	SLC2A10	606145	729,00 €	390,00 €	969,00 €			x
Arthrogryposis, distal, type 1A	TPM2	190990	819,00 €	421,00 €	1.090,00 €			x
Arthrogryposis, distal, type 1B	MYBPC1	160794	1.724,00 €	632,00 €	2.206,00 €			
Arthrogryposis, distal, type 2A	MYH3	160720	1.759,00 €	704,00 €	2.313,00 €			x
Arthrogryposis, distal, type 2B	MYH3	160720	1.759,00 €	704,00 €	2.313,00 €			x
Arthrogryposis, distal, type 2B	TNNI2	191043	655,00 €					x
Arthrogryposis, distal, type 2B	TNNT3	600692	1.061,00 €					
Arthrogryposis, distal, type 3	PIEZO2	613629	2.184,00 €	811,00 €	2.845,00 €			
Arthrogryposis, distal, type 5D	ECEL1	605896	1.061,00 €	421,00 €	1.332,00 €			x
Arthrogryposis, distal, type 5	PIEZO2	613629	2.184,00 €	811,00 €	2.845,00 €			
Arthrogryposis, distal, type 7	MYH8	160741	1.977,00 €	842,00 €	2.669,00 €			x
Arthrogryposis, mental retardation, and seizures	SLC35A3	605632	737,00 €					
Arthrogryposis, renal dysfunction, and cholestasis type 1	VPS33B	608552	1.326,00 €					
Arthrogryposis, renal dysfunction, and cholestasis type 2	VIPAS39	613401	1.326,00 €	437,00 €	1.613,00 €			x
Arthrogryposis, renal dysfunction, and cholestasis type 2	VIPAS39	613401	1.326,00 €	437,00 €	1.613,00 €			x
Arthropathy, progressive pseudorheumatoid, of childhood	WISP3	603400	573,00 €					x
Arts syndrome	PRPS1	311850	655,00 €	437,00 €	942,00 €			x
Asparaginesynthetase deficiency	ASNS	108370	796,00 €					
Aspartylglucosaminuria	AGA	613228	819,00 €					
Asperger syndrome susceptibility X-linked type 2	NLGN3	300336	796,00 €	437,00 €	1.083,00 €			
Ataxia and muscle hypotonia	COX20	614698	468,00 €					
Ataxia telangiectasia like disorder	MRE11A	600814	1.326,00 €	437,00 €	1.613,00 €			x
Ataxia, posterior column, with retinitis pigmentosa	FLVCR1	609144	995,00 €	429,00 €	1.274,00 €			x
Ataxia, progressive seizures, mental deterioration, and hearing loss, MT-TV related	MT-TV	590105	296,00 €					
Ataxia-oculomotor apraxia type 1	APTX	606350	819,00 €	328,00 €	997,00 €			x
Ataxia-oculomotor apraxia type 2	SETX	608465	1.825,00 €	328,00 €	2.003,00 €			x
Ataxia-oculomotor apraxia type 3	PIK3R5	611317	1.127,00 €	430,00 €	1.407,00 €			x
Ataxia-oculomotor apraxia type 4	PNKP	605610	796,00 €	424,00 €	1.070,00 €			x
Ataxia-telangiectasia	ATM	607585	1.955,00 €	655,00 €	2.460,00 €			
Atelosteogenesis type 1	FLNB	603381	2.059,00 €	328,00 €	2.237,00 €			x
Atelosteogenesis type 3	FLNB	603381	2.059,00 €	328,00 €	2.237,00 €			x
Athabaskan brainstem dysgenesis syndrome	HOXA1	142955	468,00 €	343,00 €	661,00 €			x
Atherosclerosis, SOAT1 related	SOAT1	102642	1.127,00 €					
Atrial fibrillation type 3	KCNQ1	607542	1.061,00 €	328,00 €	1.239,00 €			x
Atrial fibrillation type 4	KCNE2	603796	386,00 €	328,00 €	564,00 €			x
Atrial fibrillation type 6	NPPA	108780	374,00 €					
Atrial fibrillation type 7	KCNA5	176267	655,00 €					
Atrial fibrillation type 10	SCN5A	600163	1.673,00 €	328,00 €	1.851,00 €			x
Atrial fibrillation type 11	GJA5	121013	729,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Atrial fibrillation type 12	ABCC9	601439	2.028,00 €	905,00 €	2.783,00 €			x
Atrial septal defect type 2	GATA4	600576	729,00 €	328,00 €	907,00 €			x
Atrial septal defect type 3	MYH6	160710	1.673,00 €					
Atrial septal defect type 4	TBX20	606061	737,00 €	437,00 €	1.024,00 €			
Atrial septal defect type 5	ACTC1	102540	737,00 €					x
Atrial septal defect type 8	CITED2	602937	386,00 €					
Atrial septal defect type 9	GATA6	601656	819,00 €	437,00 €	1.106,00 €			x
Atrial septal defect with atrioventricular conduction defects	NKX2-5	600584	655,00 €	328,00 €	833,00 €			x
Atrichia with papular lesions	HR	602302	1.193,00 €	430,00 €	1.473,00 €			
Atrioventricular septal defect type 4	GATA4	600576	729,00 €	328,00 €	907,00 €			x
Atrioventricular septal defect type 5	GATA6	601656	819,00 €	437,00 €	1.106,00 €			x
Atrioventricular septal defect, partial with heterotaxy syndrome	CRELD1	607170	796,00 €					
Attention deficit-hyperactivity disorder	DRD4	126452	491,00 €					
Attention deficit-hyperactivity disorder	DRD5	126452	374,00 €					
Atypical Mycobacterial infection	IFNGR2	147569	655,00 €					x
Atypical Mycobacterial infection	IKBKG	300248	796,00 €					x
Atypical Mycobacterial infection	IL12RB1	601604	1.193,00 €					
Atypical Mycobacterial infection	STAT1	600555	1.399,00 €					
Atypical Mycobacterial infection, IL12RB2 related	IL12RB2	601642	1.127,00 €					
Auditory neuropathy, autosomal dominant	DIAPH3	614567	1.825,00 €					
Auriculocondylar syndrome type 1	GNAI3	139370	737,00 €	421,00 €	1.008,00 €			
Auriculocondylar syndrome type 2	PLCB4	600810	1.876,00 €	758,00 €	2.484,00 €			x
Auriculocondylar syndrome type 2	PLCB4	600810	1.876,00 €	758,00 €	2.484,00 €			x
Autism spectrum disorder	AHNAK2	608570	2.016,00 €					
Autism spectrum disorder	ANKS3		995,00 €					
Autism spectrum disorder	BPIFA3		655,00 €					
Autism spectrum disorder	EN2	131310	491,00 €					
Autism spectrum disorder	RABGGTA	601905	862,00 €					
Autism spectrum disorder, MYO16 related	MYO16	615479	1.876,00 €	905,00 €	2.631,00 €			
Autism spectrum, MXRA5 related	MXRA5		1.399,00 €					
Autism spectrum/ hyperactivity/ bipolar disorder, GRM7 related	GRM7	604101	729,00 €	429,00 €	1.008,00 €			
Autism susceptibility, X-linked type 1	NLGN3	300336	796,00 €	437,00 €	1.083,00 €			
Autism susceptibility, X-linked type 2	NLGN4X	300427	796,00 €	328,00 €	974,00 €			
Autism susceptibility, X-linked type 3	MECP2	300005	468,00 €	328,00 €	646,00 €			x
Autism susceptibility, X-linked type 4	PTCHD1	300828	655,00 €					
Autism susceptibility, X-linked type 5	RPL10	312173	573,00 €					
Autism susceptibility, X-linked type 17	SHANK2	603290	1.521,00 €	328,00 €	1.699,00 €			
Autism, ATP1B4 related	ATP1B4		737,00 €					
Autism, AVPR1A related	AVPR1A	600821	573,00 €					
Autism, C7orf43 related	C7orf43		729,00 €					



Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Autism, CELF6 related	CELF6	612681	862,00 €					
Autism, EFCAB13 related	EFCAB13		1.460,00 €					
Autism, FAAH2 related	FAAH2	300654	862,00 €					
Autism, FCRL6 related	FCRL6	613562	796,00 €					
Autism, GYG2 related	GYG2	300198	796,00 €					
Autism, IQCE related	IQCE		1.338,00 €					
Autism, MBD1 related	MBD1	156535	1.061,00 €					
Autism, NTNG1 related	NTNG1	608818	729,00 €	328,00 €	907,00 €			x
Autism, OR13H1 related	OR13H1		374,00 €					
Autism, OXTR related	OXTR	167055	468,00 €					
Autism, PKHD1L1 related	PKHD1L1	607843	1.955,00 €	905,00 €	2.710,00 €			
Autism, RNF128 related	RNF128	300439	737,00 €					
Autism, RRM1 related	RRM1		1.278,00 €					
Autism, SETD2 related	SETD2	612778	1.876,00 €					
Autism, SLC22A9 related	SLC22A9	607579	819,00 €					
Autism, UNC13B related	UNC13B	605836	1.802,00 €					
Autism, ZNF778 related	ZNF778		796,00 €					
Autism/Mental retardation/Angelman syndrome, susceptibility to, ATP10A related	ATP10A	605855	1.460,00 €					
Autoimmune lymphoproliferative syndrome type 1A	FAS	134637	819,00 €	421,00 €	1.090,00 €			x
Autoimmune lymphoproliferative syndrome type 1B	FASLG	134638	573,00 €	437,00 €	860,00 €			
Autoimmune lymphoproliferative syndrome type 2A	CASP10	601762	862,00 €					
Autoimmune lymphoproliferative syndrome type 2B	CASP8	601763	729,00 €					
Autoimmune lymphoproliferative syndrome type 3	PRKCD	176977	1.260,00 €					
Autoimmune polyendocrinopathy syndrome type 1	AIRE	607358	995,00 €	437,00 €	1.282,00 €			x
Autoinflammation, lipodystroph and dermatosis syndrome	PSMB8	177046	655,00 €	421,00 €	926,00 €			x
Avascular necrosis of the femoral head, primary	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Axenfeld-Rieger syndrome type 1	PITX2	601542	573,00 €	328,00 €	751,00 €			x
Axenfeld-Rieger syndrome type 3	FOXC1	601090	491,00 €	328,00 €	669,00 €			x
Azoospermia induced by Y chromosome microdeletions	AZF region			328,00 €				
Bainbridge-Ropers syndrome	ASXL3	615115	1.572,00 €					
Baller-Gerold syndrome	RECQL4	603780	1.193,00 €	442,00 €	1.485,00 €			
Band-like calcification with simplified gyration and polymicrogyria	OCLN	602876	729,00 €	421,00 €	1.000,00 €			
Baraitser-Winter syndrome type 1	ACTB	102630	573,00 €	421,00 €	844,00 €			x
Baraitser-Winter syndrome type 2	ACTG1	102560	468,00 €	421,00 €	739,00 €			x
Bardet-Biedl syndrome type 1	BBS1	209901	928,00 €					x
Bardet-Biedl syndrome type 2	BBS2	606151	1.127,00 €	424,00 €	1.401,00 €			x
Bardet-Biedl syndrome type 3	ARL6	608845	655,00 €					
Bardet-Biedl syndrome type 4	BBS4	600374	1.127,00 €					
Bardet-Biedl syndrome type 5	BBS5	603650	928,00 €	421,00 €	1.199,00 €			x
Bardet-Biedl syndrome type 6	MKKS	604896	655,00 €	421,00 €	926,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Bardet-Biedl syndrome type 7	BBS7	607590	1.260,00 €					
Bardet-Biedl syndrome type 8	TTC8	608132	1.193,00 €					
Bardet-Biedl syndrome type 9	BBS9	607968	1.338,00 €					
Bardet-Biedl syndrome type 10	BBS10	610148	573,00 €					x
Bardet-Biedl syndrome type 11	TRIM32	602290	819,00 €					x
Bardet-Biedl syndrome type 12	BBS12	610683	819,00 €					
Bardet-Biedl syndrome type 13	MKS1	609883	1.127,00 €	424,00 €	1.401,00 €			
Bardet-Biedl syndrome type 14	CEP290	610142	2.262,00 €	842,00 €	2.954,00 €			x
Bardet-Biedl syndrome type 14	CEP290	610142	2.262,00 €	842,00 €	2.954,00 €			x
Bardet-Biedl syndrome type 15	WDPCP	613580	1.338,00 €					
Bardet-Biedl syndrome, LZTFL1 related	LZTFL1	606568	819,00 €					
Bardet-Biedl syndrome, modifier of, CCDC28B related	CCDC28B	610162	491,00 €					x
Bare lymphocyte syndrome, type 2, complementation group A	CIITA	600005	1.460,00 €					
Bare lymphocyte syndrome, type 2	RFXANK	603200	737,00 €	429,00 €	1.016,00 €			x
Barth syndrome	TAZ	300394	655,00 €	429,00 €	934,00 €			x
Bartter syndrome	SLC12A2	600840	1.775,00 €					x
Bartter syndrome	SLC12A3	600968	1.825,00 €	328,00 €	2.003,00 €			x
Bartter syndrome	SLC12A5	606726	1.704,00 €					
Bartter syndrome	SLC12A7	604879	1.572,00 €					
Bartter syndrome type 1	SLC12A1	600839	1.764,00 €	569,00 €	2.183,00 €			x
Bartter syndrome type 2	KCNJ1	600359	573,00 €	343,00 €	766,00 €			x
Bartter syndrome type 3	CLCNKB	602023	1.193,00 €	328,00 €	1.371,00 €			x
Bartter syndrome type 4a	BSND	606412	468,00 €					
Bartter syndrome type 4b	CLCNKA	602024	1.326,00 €					
Basal cell carcinoma type 7, susceptibility to, somatic	TP53	191170					1.900,00 €	
Basal cell nevus syndrome	PTCH1	601309	1.764,00 €	328,00 €	1.942,00 €			x
Basal cell nevus syndrome	PTCH1	601309	1.764,00 €	328,00 €	1.942,00 €			x
Basal cell nevus syndrome	SUFU	607035	928,00 €	421,00 €	1.199,00 €			x
Basal cell nevus syndrome	SUFU	607035	928,00 €	421,00 €	1.199,00 €			x
Basal cell nevus syndrome due to germline PTCH2 mutation	PTCH2	603673	1.460,00 €					
Basal ganglia calcification type 1, idiopathic	SLC20A2	158378	796,00 €	429,00 €	1.075,00 €			
Basal ganglia calcification type 4	PDGFRB	173410	1.399,00 €	484,00 €	1.733,00 €			x
Basal ganglia calcification type 5, idiopathic	PDGFB	190040	655,00 €	437,00 €	942,00 €			x
Basal ganglia calcification type 6, idiopathic	XPR1	605237	1.061,00 €	445,00 €	1.356,00 €			
B-cell expansion with NFKB and T-cell anergy	CARD11	607210	1.521,00 €					
Beare-Stevenson cutis gyrata syndrome	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Beckwith-Wiedemann syndrome	CDKN1C	600856	374,00 €	328,00 €	552,00 €			x
Beckwith-Wiedemann syndrome	chr. 11p15			328,00 €				x
Beckwith-Wiedemann syndrome	H19	103280	928,00 €	328,00 €	1.106,00 €			x
Beckwith-Wiedemann syndrome	KCNQ1OT1	604115		328,00 €				

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Beckwith-Wiedemann syndrome	NSD1	606681	1.775,00 €	328,00 €	1.953,00 €			x
Bent bone dysplasia syndrome	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Bernard Soulier syndrome type A1	GP1BA	606672	655,00 €					
Bernard Soulier syndrome type A2	GP1BA	606672	655,00 €					
Bernard Soulier syndrome type B	GP1BB	138720	374,00 €					
Bernard Soulier syndrome type C	GP9	173515	386,00 €					
Bestrophinopathy	BEST1	607854	928,00 €	328,00 €	1.106,00 €			x
Beta-Galactosamide alpha-2,6-Sialyltransferase 2 deficiency	ST6GAL2	608472	655,00 €					x
Beta-ureidopropionase deficiency	UPB1	606673	729,00 €	429,00 €	1.008,00 €			
Beta-ureidopropionase deficiency	UPB1	606673	729,00 €	429,00 €	1.008,00 €			
Bethlem myopathy	COL6A1	120220	1.643,00 €	737,00 €	2.230,00 €			x
Bethlem myopathy	COL6A2	120240	1.825,00 €	590,00 €	2.265,00 €			x
Bethlem myopathy type 1	COL6A3	120250	2.106,00 €	755,00 €	2.711,00 €			x
Bethlem myopathy type 2	COL12A1	120320	1.955,00 €	905,00 €	2.710,00 €			x
Bicuspid aortic valve	TIMP1	305370	491,00 €					
Bietti crystalline corneoretinal dystrophy	CYP4V2	608614	729,00 €	328,00 €	907,00 €			
Bifid nose	FREM1	608944	2.028,00 €	800,00 €	2.678,00 €			x
Bile acid malabsorption, primary	SLC10A2	601295	655,00 €					
Bile acid synthesis defect type 2, congenital	AKR1D1	604741	819,00 €					
Bile acid synthesis defect type 3, congenital	CYP7B1	603711	655,00 €	421,00 €	926,00 €			x
Bile acid synthesis defect type 4, congenital	AMACR	604489	655,00 €	421,00 €	926,00 €			x
Biotinidase deficiency	BTD	609019	729,00 €	437,00 €	1.016,00 €			x
Birt-Hogg-Dube syndrome	FLCN	607273	729,00 €	328,00 €	907,00 €			x
Bjornstad syndrome	BCS1L	603647	819,00 €	421,00 €	1.090,00 €			x
Bladder cancer, HRAS related, somatic	HRAS	190020					1.900,00 €	
Bladder cancer, somatic	FGFR3	134934					1.900,00 €	
Bladder cancer, somatic	KRAS	190070					950,00 €	
Bladder cancer, TSC1-related, somatic	TSC1	605284					1.900,00 €	
Blau syndrome	NOD2	605956	1.326,00 €	421,00 €	1.597,00 €			x
Bleeding disorder, platelet-type 8	P2RY12	600515	374,00 €					
Bleeding disorder, platelet-type 15	ACTN1	102575	1.399,00 €					
Bleeding disorder, platelet-type 17	GFI1B	604383	491,00 €					
Blepharophimosis, epicanthus inversus, and ptosis	FOXL2	605597	491,00 €	328,00 €	669,00 €			x
Blepharophimosis-ptosis-intellectual disability syndrome	UBE3B	608047	1.704,00 €					
Bloom syndrome	BLM	604610	1.460,00 €	484,00 €	1.794,00 €			x
Bohring-Opitz syndrome	ASXL1	612990	1.399,00 €	426,00 €	1.675,00 €			
Bone marrow failure syndrome type 1	SRP72	602122	1.260,00 €	430,00 €	1.540,00 €			x
Bone marrow failure syndrome type 2	ERCC6L2	615667	1.399,00 €					
Bone marrow failure syndrome type 2	ERCC6L2	615667	1.399,00 €					
Bone mineral density QTL18, osteoporosis	PLS3	300131	1.193,00 €	328,00 €	1.371,00 €			

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Borjeson-Forssman-Lehmann syndrome	PHF6	300414	819,00 €	429,00 €	1.098,00 €			x
Bothnia retinal dystrophy	RLBP1	180090	573,00 €	421,00 €	844,00 €			
Brachydactyly type A1C	GDF5	601146	573,00 €	343,00 €	766,00 €			x
Brachydactyly type A2	BMP2	112261	468,00 €					
Brachydactyly type A2	BMPR1B	603248	796,00 €					
Brachydactyly type B1	ROR2	602337	862,00 €	328,00 €	1.040,00 €			x
Brachydactyly type E1	HOXD13	142989	374,00 €	328,00 €	552,00 €			x
Brachydactyly-mental retardation syndrome	HDAC4	605314	1.825,00 €					
Brachydactyly-syndactyly syndrome	HOXD13	142989	374,00 €	328,00 €	552,00 €			x
Bradyopsia	RGS9BP	607814	655,00 €					
Bradyopsia	RGS9	604067	1.460,00 €					
BRAF somatic Hotspot: c.1799T>A p.V600E	BRAF	164757						
BRAF, selective sequencing of exon 15	BRAF	164757						
Branched-chain aminotransferase 1 deficiency	BCAT1	113520	862,00 €					
Branched-chain aminotransferase 2 deficiency	BCAT2	113530	796,00 €					
Branched-chain ketoacid dehydrogenase kinase deficiency	BCKDK	614901	737,00 €					
Branchiooculofacial syndrome	TFAP2A	107580	862,00 €					
Branchiootic syndrome type 1	EYA1	601653	1.127,00 €	328,00 €	1.305,00 €			x
Branchiootorenal syndrome type 1	EYA1	601653	1.127,00 €	328,00 €	1.305,00 €			x
Branchiootorenal syndrome type 2	SIX5	600963	737,00 €	374,00 €	961,00 €			x
Breast cancer, male, susceptibility to	BRCA2	600185	525,00 €	330,00 €	705,00 €			
Breast cancer, RINT1 related	RINT1	610089	862,00 €					
Breast cancer, somatic	KRAS	190070					950,00 €	
Breast cancer, susceptibility to	BARD1	601593	928,00 €					
Breast cancer, susceptibility to	PALB2	610355	1.193,00 €	328,00 €	1.371,00 €			x
Breast cancer, susceptibility to	RECQL	600537	928,00 €					
Breast cancer, susceptibility to	XRCC3	600675	729,00 €					
Breast-ovarian cancer	BRCA1	113705	525,00 €	330,00 €	705,00 €			
Breast-ovarian cancer	RAD51C	602774	819,00 €	328,00 €	997,00 €			
Breast-ovarian cancer, familial, susceptibility to, type 4	RAD51D	602954	796,00 €	328,00 €	974,00 €			
Breast-ovarian cancer, familial, type 2	BRCA2	600185	525,00 €	330,00 €	705,00 €			
Brittle cornea syndrome	ZNF469	612078	2.141,00 €					
Brody myopathy	ATP2A1	108730	1.399,00 €	463,00 €	1.712,00 €			x
Bronchiectasis with or without elevated sweat chloride type 2	SCNN1A	600228	862,00 €	421,00 €	1.133,00 €			x
Brown-Vialetto-Van Laere syndrome 1	SLC52A3	613350	573,00 €	390,00 €	813,00 €			x
Brown-Vialetto-Van Laere syndrome type 2	SLC52A2	607882	573,00 €					
Brugada syndrome type 1	SCN5A	600163	1.673,00 €	328,00 €	1.851,00 €			x
Brugada syndrome type 2	GPD1L	611778	737,00 €					
Brugada syndrome type 3	CACNA1C	114205	2.106,00 €	905,00 €	2.861,00 €			
Brugada syndrome type 4	CACNB2	600003	1.338,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Brugada syndrome type 5	SCN1B	600235	573,00 €	374,00 €	797,00 €			x
Brugada syndrome type 6	KCNE3	604433	296,00 €					
Brugada syndrome type 7	SCN3B	608214	491,00 €					
Brugada syndrome type 8	HCN4	605206	729,00 €					
Brugada syndrome type 9	SLMAP	602701	1.460,00 €					
Brunner syndrome	MAOA	309850	1.127,00 €					
Budd-Chiari syndrome	F5	612309	1.673,00 €					x
Buschke-Ollendorff syndrome	LEMD3	607844	1.061,00 €	426,00 €	1.337,00 €			
Butyrylcholinesterase deficiency	BCHE	177400	819,00 €	437,00 €	1.106,00 €			
C1q deficiency	C1QA	120550	374,00 €					
C2 deficiency	C2	613927	1.061,00 €					
C3 deficiency	C3	120700	1.622,00 €	704,00 €	2.176,00 €			x
C5 deficiency	C5	120900	1.845,00 €	905,00 €	2.600,00 €			
C7 deficiency	C7	217070	1.260,00 €					
C syndrome	CD96	606037	1.061,00 €	445,00 €	1.356,00 €			x
CADASIL	NOTCH3	600276	1.876,00 €	328,00 €	2.054,00 €			
CALR, selective sequencing of exon 9	CALR	109091						
Campomelic dysplasia	SOX9	608160	573,00 €	328,00 €	751,00 €			x
Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	PRG4	604283	1.260,00 €	426,00 €	1.536,00 €			
Camurati-Engelmann disease	TGFB1	190180	737,00 €					
Canavan disease	ASPA	608034	573,00 €	328,00 €	751,00 €			x
Cantu syndrome	ABCC9	601439	2.028,00 €	905,00 €	2.783,00 €			x
Capillary malformation-arteriovenous malformation	RASA1	139150	1.643,00 €	328,00 €	1.821,00 €			x
CAPOS syndrome	ATP1A3	182350	1.326,00 €	328,00 €	1.504,00 €			x
CARASIL	HTRA1	602194	729,00 €	421,00 €	1.000,00 €			x
Carbamoylphosphate synthetase I deficiency	CPS1	608307	1.802,00 €	821,00 €	2.473,00 €			x
Carcinoid tumor of lung, somatic	MEN1	613733					1.900,00 €	
Carcinoid tumors, intestinal	SDHD	602690	468,00 €	328,00 €	646,00 €			x
Cardiac defects, CNOT3 related	CNOT3	604910	1.326,00 €					
Cardiac defects, PPP1R8 related	PPP1R8	602636	737,00 €					
Cardiac valvular dysplasia, X-linked	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency	SCO2	604272	468,00 €	343,00 €	661,00 €			
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 2	COX15	603646	796,00 €	421,00 €	1.067,00 €			x
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 3	COA5	613920	374,00 €					
Cardiofaciocutaneous syndrome	BRAF	164757	1.260,00 €					x
Cardiofaciocutaneous syndrome	KRAS	190070	491,00 €	421,00 €	762,00 €			x
Cardiofaciocutaneous syndrome type 3	MAP2K1	176872	796,00 €					
Cardiofaciocutaneous syndrome type 4	MAP2K2	601263	796,00 €	429,00 €	1.075,00 €			
Cardiomyopathy, apical hypertrophic, and neuropathy, MT-ATP8 related	MT-ATP8	516070	296,00 €					
Cardiomyopathy, dilated	MYBPC3	600958	1.825,00 €	328,00 €	2.003,00 €			x

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Cardiomyopathy, dilated type 1A	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Cardiomyopathy, dilated type 1AA	ACTN2	102573	1.460,00 €					
Cardiomyopathy, dilated type 1BB	DSG2	125671	1.193,00 €	328,00 €	1.371,00 €			x
Cardiomyopathy, dilated type 1C	LDB3	605906	1.193,00 €					
Cardiomyopathy, dilated type 1CC	NEXN	613121	1.061,00 €					
Cardiomyopathy, dilated type 1D	TNNT2	191045	1.061,00 €	328,00 €	1.239,00 €			x
Cardiomyopathy, dilated type 1DD	RBM20	613171	1.260,00 €					
Cardiomyopathy, dilated type 1E	SCN5A	600163	1.673,00 €	328,00 €	1.851,00 €			x
Cardiomyopathy, dilated type 1EE	MYH6	160710	1.673,00 €					
Cardiomyopathy, dilated type 1G	TTN	188840	2.225,00 €	905,00 €	2.980,00 €			
Cardiomyopathy, dilated type 1GG	SDHA	600857	1.061,00 €	328,00 €	1.239,00 €			x
Cardiomyopathy, dilated type 1HH	BAG3	603883	737,00 €	328,00 €	915,00 €			x
Cardiomyopathy, dilated type 1I	DES	125660	737,00 €	421,00 €	1.008,00 €			x
Cardiomyopathy, dilated type 1J	EYA4	603550	1.326,00 €	437,00 €	1.613,00 €			
Cardiomyopathy, dilated type 1KK	MYPN	608517	1.278,00 €					
Cardiomyopathy, dilated type 1L	SGCD	601411	737,00 €	328,00 €	915,00 €			x
Cardiomyopathy, dilated type 1LL	PRDM16	605557	1.193,00 €					
Cardiomyopathy, dilated type 1M	CSRP3	600824	491,00 €					
Cardiomyopathy, dilated type 1N	TCAP	604488	386,00 €	343,00 €	579,00 €			x
Cardiomyopathy, dilated type 1O	ABCC9	601439	2.028,00 €	905,00 €	2.783,00 €			x
Cardiomyopathy, dilated type 1P	PLN	172405	296,00 €					x
Cardiomyopathy, dilated type 1R	ACTC1	102540	737,00 €					x
Cardiomyopathy, dilated type 1S	MYH7	160760	1.759,00 €	328,00 €	1.937,00 €			x
Cardiomyopathy, dilated type 1T	TMPO	188380	928,00 €					
Cardiomyopathy, dilated type 1U	PSEN1	104311	928,00 €	328,00 €	1.106,00 €			x
Cardiomyopathy, dilated type 1V	PSEN2	600759	729,00 €	328,00 €	907,00 €			x
Cardiomyopathy, dilated type 1W	VCL	193065	1.582,00 €					
Cardiomyopathy, dilated type 1X	FKTN	607440	928,00 €	328,00 €	1.106,00 €			x
Cardiomyopathy, dilated type 1Y	TPM1	191010	995,00 €					
Cardiomyopathy, dilated type 1Z	TNNC1	191040	491,00 €					
Cardiomyopathy, dilated type 1	CRYAB	123590	468,00 €					x
Cardiomyopathy, dilated type 2A	TNNI3	191044	737,00 €					
Cardiomyopathy, dilated type 2B	GATAD1	614518	491,00 €					
Cardiomyopathy, dilated type 3B	DMD	300377	950,00 €	655,00 €	1.455,00 €			x
Cardiomyopathy, dilated with ataxia	DNAJC19	608977	573,00 €					
Cardiomyopathy, dilated with hypergonadotropic hypogonadism	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Cardiomyopathy, dilated with woolly hair and keratoderma	DSP	125647	1.927,00 €	328,00 €	2.105,00 €			x
Cardiomyopathy, familial hypertrophic	CAV3	601253	386,00 €	328,00 €	564,00 €			x
Cardiomyopathy, familial hypertrophic type 1	MYH7	160760	1.759,00 €	328,00 €	1.937,00 €			x
Cardiomyopathy, familial hypertrophic type 2	TNNT2	191045	1.061,00 €	328,00 €	1.239,00 €			x

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Cardiomyopathy, familial hypertrophic type 3	TPM1	191010	995,00 €					
Cardiomyopathy, familial hypertrophic type 4	MYBPC3	600958	1.825,00 €	328,00 €	2.003,00 €			x
Cardiomyopathy, familial hypertrophic type 6	PRKAG2	602743	1.127,00 €	424,00 €	1.401,00 €			x
Cardiomyopathy, familial hypertrophic type 7	TNNI3	191044	737,00 €					
Cardiomyopathy, familial hypertrophic type 8	MYL3	160790	573,00 €					
Cardiomyopathy, familial hypertrophic type 9	TTN	188840	2.225,00 €	905,00 €	2.980,00 €			
Cardiomyopathy, familial hypertrophic type 10	MYL2	160781	491,00 €					x
Cardiomyopathy, familial hypertrophic type 11	ACTC1	102540	737,00 €					x
Cardiomyopathy, familial hypertrophic type 12	CSRP3	600824	491,00 €					
Cardiomyopathy, familial hypertrophic type 16	MYOZ2	605602	655,00 €					
Cardiomyopathy, familial hypertrophic type 17	JPH2	605267	573,00 €					
Cardiomyopathy, familial hypertrophic type 19	CALR3	611414	796,00 €					
Cardiomyopathy, familial restrictive type 1	TNNI3	191044	737,00 €					
Cardiomyopathy, fatal, MT-TI related	MT-TI	590045	296,00 €					
Cardiomyopathy, hypertrophic, midventricular, digenic	MYLK2	606566	1.127,00 €					
Cardiomyopathy, hypertrophic, MT-TG related	MT-TG	590035	296,00 €					
Cardiomyopathy, hypertrophic, type 18	PLN	172405	296,00 €					x
Cardiomyopathy, hypertrophic, type 24	LDB3	605906	1.193,00 €					
Cardiomyopathy, idiopathic dilated, mitochondrial, MT-TH related	MT-TH	590040	296,00 €					
Cardiomyopathy, infantile hypertrophic, MT-ATP8 related	MT-ATP8	516070	296,00 €					
Cardiomyopathy, left ventricular noncompaction, MYH7B related	MYH7B	609928	1.775,00 €					
Carney complex type 1	PRKAR1A	188830	796,00 €	429,00 €	1.075,00 €			x
Carnitine deficiency	SLC22A5	603377	862,00 €	328,00 €	1.040,00 €			x
Carnitine palmitoyltransferase 1A deficiency	CPT1A	600528	1.278,00 €	430,00 €	1.558,00 €			x
Carnitine palmitoyltransferase 1B deficiency	CPT1B	601987	1.193,00 €					
Carnitine palmitoyltransferase 2 deficiency, infantile	CPT2	600650	819,00 €	390,00 €	1.059,00 €			x
Carnitine palmitoyltransferase 2 deficiency, lethal neonatal	CPT2	600650	819,00 €	390,00 €	1.059,00 €			x
Carnitine-acylcarnitine translocase deficiency	SLC25A20	613698	819,00 €	421,00 €	1.090,00 €			x
Carotid intimal medial thickness type 1	PPARG	601487	655,00 €	328,00 €	833,00 €			
Carpenter syndrome	RAB23	606144	573,00 €	437,00 €	860,00 €			
Carpenter syndrome type 2	MEGF8	604267	1.888,00 €	721,00 €	2.459,00 €			
Cartilage-hair hypoplasia	RMRP	157660	296,00 €	296,00 €	442,00 €			x
Cataract 11, multiple types	PITX3	602669	491,00 €					
Cataract type 17, multiple types	CRYBB1	600929	491,00 €					x
Cataract type 23	CRYBA4	123631	491,00 €					x
Cataract type 41	WFS1	606201	796,00 €	328,00 €	974,00 €			x
Cataract type 43	UNC45B	611220	1.326,00 €					
Cataract, autosomal dominant	GCNT2	600429	995,00 €					
Cataract, autosomal recessive congenital nuclear type 2	CRYBB3	123630	491,00 €					
Cataract, autosomal recessive congenital type 1	CRYAA	123580	374,00 €					x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Cataract, autosomal recessive congenital type 2	FYCO1	607182	1.521,00 €					
Cataract, autosomal recessive congenital type 4	TDRD7	611258	1.338,00 €					
Cataract, autosomal recessive type 38	AGK	610345	1.061,00 €	424,00 €	1.335,00 €			x
Cataract, congenital	SORD	182500	819,00 €					
Cataract, congenital, associated with Marinesco-Sjogren Syndrome	SIL1	608005	819,00 €	429,00 €	1.098,00 €			x
Cataract, cortical pulverulent, late-onset	LIM2	154045	573,00 €					
Cataract, lamellar	HSF4	602438	862,00 €					
Cataract, posterior polar type 2	CRYAB	123590	468,00 €					x
Cataract, pulverulent or cerulean, with or without microcornea	MAF	177075	468,00 €	343,00 €	661,00 €			
Cataract, X-linked	NHS	300457	1.399,00 €	421,00 €	1.670,00 €			x
Cataract-microcornea syndrome	GJA8	600897	468,00 €	343,00 €	661,00 €			x
Cataracts with facial dysmorphism and neuropathy	CTDP1	604927	1.193,00 €	426,00 €	1.469,00 €			x
Catechol-o-methyltransferase deficiency	COMT	116790	468,00 €					
Cell cycle disorder, CDC20 related	CDC20	603618	655,00 €					
Central core disease	RYR1	180901	1.955,00 €	655,00 €	2.460,00 €			x
Central hypoventilation syndrome with or without Hirschsprung disease	PHOX2B	603851	468,00 €	374,00 €	692,00 €	200,00 €		x
Central hypoventilation syndrome with or without Hirschsprung disease	PHOX2B	603851	468,00 €	374,00 €	692,00 €	200,00 €		x
Central hypoventilation syndrome with or without Hirschsprung disease	PHOX2B	603851	468,00 €	374,00 €	692,00 €	200,00 €		x
Central hypoventilation syndrome, congenital	ASCL1	100790	386,00 €	328,00 €	564,00 €			x
Central hypoventilation syndrome, congenital	ASCL1	100790	386,00 €	328,00 €	564,00 €			x
Central hypoventilation syndrome, congenital	ASCL1	100790	386,00 €	328,00 €	564,00 €			x
Central hypoventilation syndrome, congenital	BDNF	113505	573,00 €	328,00 €	751,00 €			
Central hypoventilation syndrome, congenital	ECE1	600423	1.278,00 €					
Central hypoventilation syndrome, congenital	EDN3	131242	573,00 €	328,00 €	751,00 €			x
Central hypoventilation syndrome, congenital	GDNF	600837	491,00 €	328,00 €	669,00 €			x
Central hypoventilation syndrome, congenital	GFRA1	601496	491,00 €	328,00 €	669,00 €			x
Central hypoventilation syndrome, congenital	MECP2	300005	468,00 €	328,00 €	646,00 €			x
Central hypoventilation syndrome, congenital	PHOX2A	602753	491,00 €	374,00 €	715,00 €			x
Central hypoventilation syndrome, congenital	RET	164761	1.338,00 €	328,00 €	1.516,00 €			x
Central hypoventilation syndrome, congenital	ZEB2	605802	862,00 €	328,00 €	1.040,00 €			x
Centronuclear myopathy type 1	MTMR14	611089	1.326,00 €					
Centronuclear myopathy type 3	MYF6	159991	468,00 €					
Centronuclear myopathy type 4	CCDC78	614666	862,00 €					
Cerebellar ataxia	CP	117700	1.326,00 €					x
Cerebellar ataxia and mental retardation with or without quadrupedal locomotion type 3	CA8	114815	819,00 €					
Cerebellar ataxia with deafness and narcolepsy, autosomal recessive	DNMT1	126375	1.802,00 €	704,00 €	2.356,00 €			
Cerebellar ataxia with mental retardation and dysequilibrium syndrome type 2	WDR81	614218	1.338,00 €					
Cerebellar ataxia with spasticity	GBA2	609471	1.193,00 €	424,00 €	1.467,00 €			x
Cerebellar ataxia, mental retardation, and dysequilibrium syndrome type 4	ATP8A2	605870	1.927,00 €	905,00 €	2.682,00 €			
Cerebellar ataxia, nonprogressive, with mental retardation	CAMTA1	611501	1.643,00 €	484,00 €	1.977,00 €			



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Cerebellar ataxia, SNX14 related	SNX14	616105	1.643,00 €					
Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion type 1	VLDLR	192977	1.260,00 €	328,00 €	1.438,00 €			x
Cerebellar-retinal degeneration, infantile	ACO2	100850	1.127,00 €					
Cerebral amyloid angiopathy	CST3	604312	374,00 €	437,00 €	661,00 €			x
Cerebral amyloid angiopathy, APP related	APP	104760	1.278,00 €	328,00 €	1.456,00 €			x
Cerebral cavernous malformations type 1	KRIT1	604214	1.127,00 €	655,00 €	1.632,00 €			x
Cerebral cavernous malformations type 2	CCM2	607929	796,00 €	328,00 €	974,00 €			
Cerebral cavernous malformations type 3	PDCD10	609118	655,00 €	328,00 €	833,00 €			x
Cerebral dysgenesis, neuropathy, ichthyosis, palmoplantar keratoderma syndrome	SNAP29	604202	573,00 €					
Cerebral palsy type 1, spastic quadriplegic	GAD1	605363	1.326,00 €					
Cerebral palsy type 2, spastic quadriplegic	KANK1	607704	1.193,00 €	424,00 €	1.467,00 €			
Cerebrooculofacioskeletal syndrome type 1	ERCC6	609413	1.521,00 €	442,00 €	1.813,00 €			x
Cerebrooculofacioskeletal syndrome type 4	ERCC1	126380	819,00 €					
Cerebrotendinous xanthomatosis	CYP27A1	606530	737,00 €	421,00 €	1.008,00 €			x
Ceroid lipofuscinosis neuronal type 1	PPT1	600722	819,00 €	421,00 €	1.090,00 €			x
Ceroid lipofuscinosis neuronal type 2	TPP1	607998	862,00 €	426,00 €	1.138,00 €			x
Ceroid lipofuscinosis neuronal type 3	CLN3	607042	655,00 €	328,00 €	833,00 €			x
Ceroid lipofuscinosis neuronal type 4	DNAJC5	611203	468,00 €					
Ceroid lipofuscinosis neuronal type 5	CLN5	608102	491,00 €	437,00 €	778,00 €			x
Ceroid lipofuscinosis neuronal type 6	CLN6	606725	655,00 €	437,00 €	942,00 €			x
Ceroid lipofuscinosis neuronal type 7	MFSD8	611124	862,00 €	426,00 €	1.138,00 €			x
Ceroid lipofuscinosis neuronal type 8	CLN8	607837	386,00 €	374,00 €	610,00 €			x
Ceroid lipofuscinosis neuronal type 10	CTSD	116840	737,00 €	421,00 €	1.008,00 €			x
Ceroid lipofuscinosis neuronal type 11	GRN	138945	796,00 €	328,00 €	974,00 €			x
Cervical cancer, somatic	FGFR3	134934					1.900,00 €	
Cervical dystonia	CIZ1	611420	1.326,00 €	421,00 €	1.597,00 €			x
Chanarin-Dorfman syndrome	ABHD5	604780	655,00 €	437,00 €	942,00 €			x
Charcot-Marie-Tooth disease, axonal type 20	DYNC1H1	600112	1.955,00 €					x
CHARGE syndrome	CHD7	608892	2.028,00 €	328,00 €	2.206,00 €			x
Chediak-Higashi syndrome	LYST	606897	1.955,00 €					x
CHILD syndrome	NSDHL	300275	737,00 €	421,00 €	1.008,00 €			x
CHIME syndrome	PIGL	605947	655,00 €					
Chloramphenicol resistance, MT-RNR2 related	MT-RNR2	561010	296,00 €					
Choanal atresia and lymphedema	PTPN14	603155	1.399,00 €					
Cholestasis benign recurrent intrahepatic type 2	ABCB11	603201	1.704,00 €	590,00 €	2.144,00 €			x
Cholestasis intrahepatic, of pregnancy, type 3	ABCB4	171060	1.704,00 €	328,00 €	1.882,00 €			x
Cholestasis progressive intrahepatic type 1	ATP8B1	602397	1.521,00 €	590,00 €	1.961,00 €			x
Cholestasis progressive intrahepatic type 2	ABCB11	603201	1.704,00 €	590,00 €	2.144,00 €			x
Cholestasis progressive intrahepatic type 3	ABCB4	171060	1.704,00 €	328,00 €	1.882,00 €			x
Cholestasis, benign recurrent intrahepatic	ATP8B1	602397	1.521,00 €	590,00 €	1.961,00 €			x

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Cholestasis, infantile, NR1H4 related	NR1H4	603826	796,00 €					
Cholestasis, intrahepatic, of pregnancy, type 1	ATP8B1	602397	1.521,00 €	590,00 €	1.961,00 €			x
Cholesteryl ester storage disease	LIPA	613497	729,00 €	429,00 €	1.008,00 €			x
Chondrocalcinosis type 2	ANKH	605145	796,00 €					
Chondrodysplasia punctata, X-linked dominant	EBP	300205	491,00 €					
Chondrodysplasia punctata, X-linked recessive	ARSE	300180	862,00 €					
Chondrodysplasia with joint dislocations, GPAPP type	IMPAD1	614010	573,00 €					
Chondrodysplasia, Blomstrand type	PTH1R	168468	1.061,00 €	424,00 €	1.335,00 €			x
Chondrosarcoma, familial	EXT1	608177	928,00 €	328,00 €	1.106,00 €			x
Chorea, hereditary benign	NKX2-1	600635	468,00 €	328,00 €	646,00 €			x
Choreoacanthocytosis	VPS13A	605978	1.955,00 €					
Choroidal dystrophy, central areolar type 2	PRPH2	179605	468,00 €	328,00 €	646,00 €			x
Choroideremia	CHM	300390	1.193,00 €	328,00 €	1.371,00 €			x
Chondrodysplasia, acromesomelic, with genital anomalies	BMPR1B	603248	796,00 €					
Chudley-McCullough syndrome	GPSM2	609245	1.260,00 €	445,00 €	1.555,00 €			
Chylomicron retention disease	SAR1B	607690	573,00 €					
Ciliogenesis related disorder	PTPN23	606584	1.521,00 €					
Citrin deficiency	SLC25A13	603859	1.193,00 €	421,00 €	1.464,00 €			x
Citrullinemia	ASS1	603470	995,00 €	424,00 €	1.269,00 €			x
CK syndrome	NSDHL	300275	737,00 €	421,00 €	1.008,00 €			x
Cleidocranial dysplasia	RUNX2	600211	819,00 €	328,00 €	997,00 €			x
Club foot	PITX1	602149	491,00 €	374,00 €	715,00 €			x
CMT1A	PMP22	601097	468,00 €	328,00 €	646,00 €			x
CMT1B	MPZ	159440	655,00 €	328,00 €	833,00 €			x
CMT1C	LITAF	603795	491,00 €					
CMT1D	EGR2	129010	491,00 €	328,00 €	669,00 €			x
CMT1E	PMP22	601097	468,00 €	328,00 €	646,00 €			x
CMT1F	NEFL	162280	573,00 €	328,00 €	751,00 €			x
CMT2A1	KIF1B	605995	2.028,00 €	905,00 €	2.783,00 €			x
CMT2A2	MFN2	608507	1.061,00 €	328,00 €	1.239,00 €			x
CMT2B1	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
CMT2B2	MED25	610197	1.061,00 €	421,00 €	1.332,00 €			x
CMT2B	RAB7A	602298	491,00 €	328,00 €	669,00 €			x
CMT2C	TRPV4	605427	1.127,00 €	424,00 €	1.401,00 €			x
CMT2D	GARS	600287	1.193,00 €	328,00 €	1.371,00 €			x
CMT2E	NEFL	162280	573,00 €	328,00 €	751,00 €			x
CMT2F	HSPB1	602195	374,00 €	328,00 €	552,00 €			x
CMT2I	MPZ	159440	655,00 €	328,00 €	833,00 €			x
CMT2J	MPZ	159440	655,00 €	328,00 €	833,00 €			x
CMT2K	GDAP1	606598	573,00 €	328,00 €	751,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
CMT2L	HSPB8	608014	374,00 €	328,00 €	552,00 €			x
CMT2N	AARS	601065	1.326,00 €					x
CMT2P	LRSAM1	610933	1.582,00 €					
CMT4, CTDP1 related	CTDP1	604927	1.193,00 €	426,00 €	1.469,00 €			x
CMT4A	GDAP1	606598	573,00 €	328,00 €	751,00 €			x
CMT4B1	MTMR2	603557	1.061,00 €	328,00 €	1.239,00 €			x
CMT4B2	SBF2	607697	1.973,00 €	328,00 €	2.151,00 €			x
CMT4C	SH3TC2	608206	1.338,00 €	328,00 €	1.516,00 €			x
CMT4D	NDRG1	605262	1.061,00 €					x
CMT4E	EGR2	129010	491,00 €	328,00 €	669,00 €			x
CMT4E	MPZ	159440	655,00 €	328,00 €	833,00 €			x
CMT4F	PRX	605725	995,00 €	328,00 €	1.173,00 €			x
CMT4H	FGD4	611104	1.326,00 €	424,00 €	1.600,00 €			x
CMT4J	FIG4	609390	1.460,00 €	484,00 €	1.794,00 €			x
CMTDIF	GNB4	610863	819,00 €					
CMTRIB	KARS	601421	862,00 €					
CMTRIB	KARS	601421	862,00 €					
CMTRIB	KARS	601421	862,00 €					
CMTRID	COX6A1	602072	374,00 €					
CMTX1	GJB1	304040	491,00 €	328,00 €	669,00 €			x
CMTX4	AIFM1	300169	1.193,00 €	424,00 €	1.467,00 €			x
CMTX5	PRPS1	311850	655,00 €	437,00 €	942,00 €			x
CoA-2 4-dienoyl reductase 1 deficiency	DECR1	222745	737,00 €					
CoA-3-hydroxyacyl dehydrogenase deficiency	HADH	601609	729,00 €					
CoA-3-methylcrotonyl carboxylase 1 deficiency	MCCC1	609010	1.260,00 €					
CoA-3-methylcrotonyl carboxylase 2 deficiency	MCCC2	609014	1.127,00 €					
COACH syndrome	CC2D2A	612013	1.977,00 €	800,00 €	2.627,00 €			x
COACH syndrome	RPGRIP1L	610937	1.704,00 €					x
COACH syndrome	TMEM67	609884	1.704,00 €					
Coarctation of the aorta	MCTP2		1.460,00 €					
Coat plus syndrome	CTC1	613129	1.338,00 €	484,00 €	1.672,00 €			x
Cockayne syndrome type A	ERCC8		1.127,00 €	421,00 €	1.398,00 €			x
Cockayne syndrome type B	ERCC6	609413	1.521,00 €	442,00 €	1.813,00 €			x
CODAS syndrome	LONP1	605490	1.326,00 €	421,00 €	1.597,00 €			x
Coenzyme Q10 deficiency type 1	COQ2	609825	655,00 €					
Coenzyme Q10 deficiency type 2	PDSS1	607429	729,00 €	421,00 €	1.000,00 €			x
Coenzyme Q10 deficiency type 3	PDSS2	610564	737,00 €	437,00 €	1.024,00 €			x
Coenzyme Q10 deficiency type 5	COQ9	612837	819,00 €	421,00 €	1.090,00 €			x
Coffin-Lowry syndrome	RPS6KA3	300075	1.521,00 €	328,00 €	1.699,00 €			x
Coffin-Siris syndrome, SMARCE1 related	SMARCE1	603111	729,00 €	429,00 €	1.008,00 €			

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Cohen syndrome	VPS13B	607817	1.955,00 €	655,00 €	2.460,00 €			
Colchicine resistance	ABCB1	171050	1.572,00 €					
Cold autoinflammatory syndrome type 2	NLRP12	609648	928,00 €					
Cold-induced sweating syndrome	CRLF1	604237	819,00 €					
Cold-induced sweating syndrome type 2	CLCF1	607672	468,00 €					
Cole disease	ENPP1	173335	1.582,00 €	527,00 €	1.959,00 €			x
Coloboma of optic nerve	PAX6	607108	1.061,00 €	328,00 €	1.239,00 €			x
Coloboma, ocular, autosomal dominant	PAX6	607108	1.061,00 €	328,00 €	1.239,00 €			x
Colobomatous microphthalmia	TENM1	610083	1.802,00 €	674,00 €	2.326,00 €			
Colon cancer, PPARG related, somatic	PPARG	601487					1.900,00 €	
Colorectal cancer, hereditary	NRAS	164790	468,00 €					
Colorectal cancer, hereditary nonpolyposis type 1	MSH2	609309	1.193,00 €	328,00 €	1.371,00 €			
Colorectal cancer, hereditary nonpolyposis type 2	MLH1	120436	1.260,00 €	328,00 €	1.438,00 €			
Colorectal cancer, hereditary nonpolyposis type 4	PMS2	600259	995,00 €	328,00 €	1.173,00 €			
Colorectal cancer, hereditary nonpolyposis type 5	MSH6	600678	1.127,00 €	328,00 €	1.305,00 €			
Colorectal cancer, hereditary nonpolyposis type 6	TGFBR2	190182	862,00 €	328,00 €	1.040,00 €			x
Colorectal cancer, hereditary nonpolyposis type 7	MLH3	604395	1.278,00 €					
Colorectal cancer, hereditary nonpolyposis type 8	EPCAM	185535	819,00 €	328,00 €	997,00 €			
Colorectal cancer, hereditary, susceptibility to	CCND1	168461	491,00 €	328,00 €	669,00 €			
Colorectal Cancer, resistance to cetuximab, EGFR related, somatic	EGFR	131550					1.900,00 €	
Colorectal cancer, somatic	APC	611731					1.900,00 €	
Colorectal cancer, somatic	BRAF	164757					1.290,00 €	
Colorectal cancer, somatic	CTNNB1	116806					1.900,00 €	
Colorectal cancer, somatic	DCC	120470					1.900,00 €	
Colorectal cancer, somatic	EP300	602700					1.900,00 €	
Colorectal cancer, somatic	FGFR3	134934					1.900,00 €	
Colorectal cancer, somatic	FLCN	607273					1.900,00 €	
Colorectal cancer, somatic	NRAS	164790					950,00 €	
Combined cellular and humoral immune defects with granulomas	RAG2	179616	491,00 €	328,00 €	669,00 €			x
Combined D-2- and L-2-hydroxyglutaric aciduria	SLC25A1	190315	737,00 €	437,00 €	1.024,00 €			
Combined immunodeficiency, B cell-negative, T cell-negative, NK cell positive	RAG2	179616	491,00 €	328,00 €	669,00 €			x
Combined immunodeficiency, X-linked, moderate	IL2RG	308380	819,00 €					
Combined malonic and methylmalonic aciduria	ACSF3	614245	729,00 €					
Combined oxidative phosphorylation deficiency type 1	GFM1	606639	1.278,00 €					
Combined oxidative phosphorylation deficiency type 2	MRPS16	609204	468,00 €					
Combined oxidative phosphorylation deficiency type 3	TSFM	604723	729,00 €					
Combined oxidative phosphorylation deficiency type 4	TUFM	602389	862,00 €					
Combined oxidative phosphorylation deficiency type 5	MRPS22	605810	737,00 €					
Combined oxidative phosphorylation deficiency type 6	AIFM1	300169	1.193,00 €	424,00 €	1.467,00 €			x
Combined oxidative phosphorylation deficiency type 7	C12ORF65	613541	386,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Combined oxidative phosphorylation deficiency type 8	AARS2	612035	1.127,00 €					
Combined oxidative phosphorylation deficiency type 9	MRPL3	607118	729,00 €					
Combined oxidative phosphorylation deficiency type 10	MTO1	614667	1.061,00 €	426,00 €	1.337,00 €			x
Combined oxidative phosphorylation deficiency type 11	RMND1	614917	862,00 €					
Combined oxidative phosphorylation deficiency type 12	EARS2	612799	729,00 €	437,00 €	1.016,00 €			
Combined oxidative phosphorylation deficiency type 13	PNPT1	610316	1.521,00 €					
Combined oxidative phosphorylation deficiency type 14	FARS2	611592	573,00 €					
Combined oxidative phosphorylation deficiency type 15	MTFMT	611766	819,00 €	421,00 €	1.090,00 €			
Combined oxidative phosphorylation deficiency type 16	MRPL44	611849	468,00 €					
Combined oxidative phosphorylation deficiency type 17	ELAC2	605367	1.278,00 €					
Combined oxidative phosphorylation deficiency type 18	SFXN4	615564	995,00 €					
Combined oxidative phosphorylation deficiency type 19	LYRM4	613311	491,00 €					
Combined oxidative phosphorylation deficiency type 20	VARS2	612802	1.582,00 €					
Combined oxidative phosphorylation deficiency type 21	TARS2	612805	1.260,00 €					
Combined oxidative phosphorylation deficiency type 22	ATP5A1	164360	729,00 €					
Combined oxidative phosphorylation deficiency type 23	GTPBP3	608536	655,00 €					
Combined oxidative phosphorylation deficiency type 24	NARS2	612803	928,00 €	437,00 €	1.215,00 €			
Combined oxidative phosphorylation deficiency type 25	MARS2	609728	491,00 €					
Combined oxidative phosphorylation deficiency type 26	TRMT5	611023	655,00 €					
Compton-North congenital myopathy	CNTN1	600016	1.521,00 €					
Cone-rod dystrophy	AIPL1	604392	655,00 €	328,00 €	833,00 €			x
Cone-rod dystrophy	UNC119	604011	573,00 €					
Cone-rod dystrophy type 2	CRX	602225	491,00 €	328,00 €	669,00 €			x
Cone-rod dystrophy type 3	ABCA4	601691	2.301,00 €	655,00 €	2.806,00 €			x
Cone-rod dystrophy type 4	PDE6C	600827	1.399,00 €					
Cone-rod dystrophy type 5	PITPNM3	608921	1.399,00 €					
Cone-rod dystrophy type 7	RIMS1	606629	2.028,00 €	905,00 €	2.783,00 €			
Cone-rod dystrophy type 9	ADAM9	602713	1.460,00 €					
Cone-rod dystrophy type 11	RAX2	610362	374,00 €					
Cone-rod dystrophy type 12	PROM1	604365	1.704,00 €	569,00 €	2.123,00 €			x
Cone-rod dystrophy type 13	RPGRIP1	605446	1.582,00 €	328,00 €	1.760,00 €			x
Cone-rod dystrophy type 14	GUCA1A	600364	573,00 €					
Cone-rod dystrophy type 15	CDHR1	609502	1.260,00 €					
Cone-rod dystrophy type 17	CD3G	186740	573,00 €					
Cone-rod dystrophy type 20	POC1B	614784	862,00 €					
Cone-rod dystrophy, C21orf2 related	C21orf2	603191	737,00 €					
Cone-rod dystrophy, X-linked type 3	CACNA1F	300110	1.802,00 €					
Congenital bilateral absence of vas deferens	CFTR	602421	950,00 €	328,00 €	1.128,00 €			x
Congenital disorder of glycosylation, type lp	ALG11	613666	573,00 €					
Congenital disorder of glycosylation, type lq	SRD5A3	611715	491,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Congenital disorder of glycosylation, type Iw	STT3A	601134	1.127,00 €					
Congenital heart defects multiple types	TAB2	605101	729,00 €					
Congenital heart disease and transposition of the great arteries	FOXH1	603621	573,00 €					
Congenital muscular dystrophy and hypoglycosylation of $\alpha$ -dystroglycan	B3GALNT2	610194	1.061,00 €					
Congenital short-bowel syndrome	CLMP	611693	655,00 €					
Contractural arachnodactyly, congenital	FBN2	612570	1.955,00 €	905,00 €	2.710,00 €			
Convulsions, benign familial infantile, 3	SCN2A	182390	1.673,00 €	569,00 €	2.092,00 €			x
Convulsions, familial infantile, with paroxysmal choreoathetosis	PRRT2	614386	468,00 €	374,00 €	692,00 €			x
Coproporphyrinuria	CPOX	612732	737,00 €	328,00 €	915,00 €			
Corneal dystrophy, epithelial basement membrane	TGFBI	601692	1.127,00 €					
Corneal dystrophy, posterior polymorphous, type 1	VSX1	605020	655,00 €					
Corneal endothelial dystrophy type 2	SLC4A11	610206	1.193,00 €					
Corneal intraepithelial dyskeratosis and ectodermal dysplasia	NLRP1	606636	1.460,00 €	424,00 €	1.734,00 €			x
Corneal opacification and other ocular anomalies	PXDN	605158	1.643,00 €					
Cornelia de Lange syndrome type 1	NIPBL	608667	2.145,00 €	655,00 €	2.650,00 €			x
Cornelia de Lange syndrome type 2	SMC1A	300040	1.460,00 €	527,00 €	1.837,00 €			x
Cornelia de Lange syndrome type 3	SMC3	606062	1.764,00 €	611,00 €	2.225,00 €			x
Cornelia de Lange syndrome type 4	RAD21	606462	1.061,00 €	437,00 €	1.348,00 €			
Cornelia de Lange syndrome type 5	HDAC8	300269	928,00 €	429,00 €	1.207,00 €			x
Coronary artery disease in familial hypercholesterolemia, protection against	ABCA1	600046	2.028,00 €					x
Coronary heart disease, susceptibility to, type 6	MMP3	185250	819,00 €					
Corpus callosum, agenesis of, with abnormal genitalia	ARX	300382	737,00 €	328,00 €	915,00 €			x
Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia	IGBP1	300139	737,00 €					
Cortical dysplasia, complex, with other brain malformations, type 1	TUBB3	602661	573,00 €					
Cortical dysplasia, complex, with other brain malformations, type 5	TUBB2A	615101	655,00 €	437,00 €	942,00 €			
Cortical dysplasia-focal epilepsy syndrome	CNTNAP2	604569	1.582,00 €	505,00 €	1.937,00 €			
Cortical malformations, occipital	LAMC3	604349	1.825,00 €					
Corticobasal Degeneration, CFL1 related	CFL1	601442	468,00 €					
Costello syndrome	HRAS	190020	491,00 €	390,00 €	731,00 €			x
Coumarin resistance	VKORC1	608547	573,00 €					
Coumarin/Warfarin resistance due to CYP2C9 variants	CYP2C9	601130	796,00 €	328,00 €	974,00 €			
Cousin syndrome	TBX15	604127	819,00 €					
Cowden syndrome type 1	PTEN	601728	796,00 €	328,00 €	974,00 €			x
Cowden syndrome type 3	SDHD	602690	468,00 €	328,00 €	646,00 €			x
Cowden syndrome type 5	PIK3CA	171834	1.326,00 €	442,00 €	1.618,00 €			
Cowden syndrome type 6	AKT1	164730	995,00 €	445,00 €	1.290,00 €			x
Craniodiaphyseal dysplasia, autosomal dominant	SOST	605740	386,00 €					
Cranioectodermal dysplasia type 1	IFT122	606045	1.673,00 €					
Cranioectodermal dysplasia type 2	WDR35	613602	1.825,00 €					
Cranioectodermal dysplasia type 3	IFT43	614068	737,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Cranioectodermal dysplasia type 4	WDR19	608151	1.927,00 €					
Craniofacial and neuro-developmental abnormalities	DISP1	607502	1.061,00 €					
Craniofacial and neuro-developmental abnormalities, JAG2 related	JAG2	602570	1.643,00 €	548,00 €	2.041,00 €			x
Craniofacial anomalies and anterior segment dysgenesis syndrome	VSX1	605020	655,00 €					
Craniofacial-skeletal-dermatologic dysplasia	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Craniofrontonasal syndrome	EFNB1	300035	491,00 €	328,00 €	669,00 €			x
Craniometaphyseal dysplasia	ANKH	605145	796,00 €					
Craniosynostosis and dental anomalies	IL11RA	600939	729,00 €					
Craniosynostosis type 1	TWIST1	601622	386,00 €	328,00 €	564,00 €			x
Craniosynostosis type 2	MSX2	123101	491,00 €	343,00 €	684,00 €			x
Craniosynostosis type 3	TCF12	600480	1.278,00 €					
Craniosynostosis type 4	ERF	611888	655,00 €					
Craniosynostosis type 6	ZIC1	600470	573,00 €	328,00 €	751,00 €			x
Craniosynostosis, FGFR1 related	FGFR1	136350	1.193,00 €	328,00 €	1.371,00 €			x
Craniosynostosis, nonspecific	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Creatine deficiency syndrome X-linked	SLC6A8	300036	819,00 €	328,00 €	997,00 €			x
Creatine deficiency syndrome X-linked	SLC6A8	300036	819,00 €	328,00 €	997,00 €			x
Creatine phosphokinase, elevated serum	CAV3	601253	386,00 €	328,00 €	564,00 €			x
Creutzfeldt-Jakob disease	PRNP	176640	386,00 €	343,00 €	579,00 €	200,00 €		
Crigler-Najjar syndrome, type 1	UGT1A1	191740	737,00 €	390,00 €	977,00 €			x
Crigler-Najjar syndrome, type 2	UGT1A1	191740	737,00 €	390,00 €	977,00 €			x
Crouzon syndrome	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Crouzon syndrome with acanthosis nigricans	FGFR3	134934	995,00 €	421,00 €	1.266,00 €			x
Cryptorchidism	RXFP2	606655	1.278,00 €					
Cryptorchidism	RXFP2	606655	1.278,00 €					
Currarino syndrome	MNX1	142994	737,00 €	374,00 €	961,00 €			x
Cutaneous telangiectasia and cancer syndrome, familial	ATR	601215	1.955,00 €					x
Cutaneous telangiectasia and cancer syndrome, familial	ATR	601215	1.955,00 €					x
Cutis laxa type 1A, autosomal recessive	FBLN5	604580	796,00 €					
Cutis laxa type 1B, autosomal recessive	EFEMP2	604633	729,00 €	429,00 €	1.008,00 €			x
Cutis laxa type 1C, autosomal recessive	LTBP4	604710	1.673,00 €					
Cutis laxa type 2, autosomal dominant	FBLN5	604580	796,00 €					
Cutis laxa type 2A, autosomal recessive	ATP6V0A2	611716	1.278,00 €					
Cutis laxa type 2B, autosomal recessive	PYCR1	179035	737,00 €					
Cutis laxa type 3A, autosomal recessive	ALDH18A1	138250	1.127,00 €					
Cutis laxa type 3B, autosomal recessive	PYCR1	179035	737,00 €					
Cutis laxa, autosomal dominant	ELN	130160	1.775,00 €	328,00 €	1.953,00 €			
Cyanosis, transient neonatal	HBG2	142250	468,00 €	328,00 €	646,00 €			x
Cylindromatosis, familial	CYLD	605018	1.326,00 €	437,00 €	1.613,00 €			
CYP2C19 related poor drug metabolism	CYP2C19	124020	796,00 €	328,00 €	974,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Cystathioninuria	CTH	219500	796,00 €					
Cystic fibrosis	CFTR	602421	950,00 €	328,00 €	1.128,00 €			x
Cystic fibrosis, SLC6A14 related	SLC6A14	300444	995,00 €	437,00 €	1.282,00 €			
Cystinosis, nephropathic	CTNS	606272	729,00 €	426,00 €	1.005,00 €			x
Cystinosis, nephropathic	CTNS	606272	729,00 €	426,00 €	1.005,00 €			x
Cystinuria	PREPL	609557	1.127,00 €	328,00 €	1.305,00 €			x
Cystinuria	SLC3A1	104614	862,00 €	328,00 €	1.040,00 €			x
Cystinuria	SLC7A9	604144	862,00 €	328,00 €	1.040,00 €			x
Cytochrome c oxidase 1 deficiency	MT-CO1	516030	296,00 €					x
Cytochrome c oxidase 2 deficiency	MT-CO2	516040	296,00 €					
Cytochrome c oxidase 3 deficiency	MT-CO3	516050	296,00 €					x
Cytochrome P450 deficiency	CYP1A2	124060	737,00 €	328,00 €	915,00 €			
Czech dysplasia	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Czech dysplasia	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
D-2-hydroxyglutaric aciduria type 1	D2HGDH	609186	819,00 €	328,00 €	997,00 €			x
D-2-hydroxyglutaric aciduria type 2	IDH2	147650	796,00 €	429,00 €	1.075,00 €			x
Dandy-Walker malformation and occipital cephaloceles, LAMC1 related	LAMC1	150290	1.704,00 €					
Danon disease	LAMP2	309060	928,00 €	421,00 €	1.199,00 €			x
Danon disease	LAMP2	309060	928,00 €	421,00 €	1.199,00 €			x
D-bifunctional protein deficiency	HSD17B4	601860	1.521,00 €	527,00 €	1.898,00 €			x
D-bifunctional protein deficiency	HSD17B4	601860	1.521,00 €	527,00 €	1.898,00 €			x
De Sanctis-Cacchione syndrome	ERCC6	609413	1.521,00 €	442,00 €	1.813,00 €			x
Deafness and male infertility	STRC	606440	2.028,00 €	611,00 €	2.489,00 €			x
Deafness and male infertility, CATSPER2 related	CATSPER2	607249	862,00 €					
Deafness and male infertility, CATSPER2 related	CATSPER2	607249	862,00 €					
Deafness with keratopachydermia and constrictions of fingers and toes	GJB2	121011	374,00 €	328,00 €	552,00 €			x
Deafness, autosomal dominant type 1	DIAPH1	602121	1.673,00 €					
Deafness, autosomal dominant type 2A	KCNQ4	603537	1.061,00 €	437,00 €	1.348,00 €			x
Deafness, autosomal dominant type 2B	GJB3	603324	374,00 €	328,00 €	552,00 €			x
Deafness, autosomal dominant type 3A	GJB2	121011	374,00 €	328,00 €	552,00 €			x
Deafness, autosomal dominant type 3B	GJB6	604418	374,00 €	328,00 €	552,00 €			x
Deafness, autosomal dominant type 4B	CEACAM16	614591	655,00 €					
Deafness, autosomal dominant type 4	MYH14	608568	1.845,00 €					
Deafness, autosomal dominant type 5	DFNA5	608798	737,00 €					
Deafness, autosomal dominant type 6	WFS1	606201	796,00 €	328,00 €	974,00 €			x
Deafness, autosomal dominant type 9	COCH	603196	819,00 €	429,00 €	1.098,00 €			x
Deafness, autosomal dominant type 10	EYA4	603550	1.326,00 €	437,00 €	1.613,00 €			
Deafness, autosomal dominant type 11	MYO7A	276903	2.016,00 €	841,00 €	2.707,00 €			x
Deafness, autosomal dominant type 12	TECTA	602574	1.572,00 €					x
Deafness, autosomal dominant type 13	COL11A2	120290	1.802,00 €					



Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Deafness, autosomal dominant type 15	POU4F3	602460	468,00 €					
Deafness, autosomal dominant type 17	MYH9	160775	2.028,00 €	328,00 €	2.206,00 €			x
Deafness, autosomal dominant type 20	ACTG1	102560	468,00 €	421,00 €	739,00 €			x
Deafness, autosomal dominant type 22	MYO6	600970	1.775,00 €					
Deafness, autosomal dominant type 23	SIX1	601205	374,00 €	343,00 €	567,00 €			x
Deafness, autosomal dominant type 25	SLC17A8	607557	995,00 €					
Deafness, autosomal dominant type 28	GRHL2	608576	1.326,00 €					
Deafness, autosomal dominant type 36	TMC1	606706	1.399,00 €	505,00 €	1.754,00 €			x
Deafness, autosomal dominant type 39, with dentinogenesis type 1	DSPP	125485	491,00 €					
Deafness, autosomal dominant type 40	CRYM	123740	862,00 €					
Deafness, autosomal dominant type 44	CCDC50	611051	862,00 €					x
Deafness, autosomal dominant type 48	MYO1A	601478	1.326,00 €					
Deafness, autosomal dominant type 50	MIR96	611606	156,00 €					
Deafness, autosomal dominant type 52	POU4F3	602460	468,00 €					
Deafness, autosomal dominant type 64	DIABLO	605219	737,00 €					
Deafness, autosomal dominant type 65	TBC1D24	613577	819,00 €	437,00 €	1.106,00 €			x
Deafness, autosomal recessive	GJB3	603324	374,00 €	328,00 €	552,00 €			x
Deafness, autosomal recessive	SUN1	607723	1.338,00 €					
Deafness, autosomal recessive type 1A	GJB2	121011	374,00 €	328,00 €	552,00 €			x
Deafness, autosomal recessive type 1B	GJB6	604418	374,00 €	328,00 €	552,00 €			x
Deafness, autosomal recessive type 2	MYO7A	276903	2.016,00 €	841,00 €	2.707,00 €			x
Deafness, autosomal recessive type 3	MYO15A	602666	2.282,00 €	905,00 €	3.037,00 €			x
Deafness, autosomal recessive type 4	FOXI1	601093	468,00 €	343,00 €	661,00 €			x
Deafness, autosomal recessive type 6	TMIE	607237	468,00 €					
Deafness, autosomal recessive type 7	TMC1	606706	1.399,00 €	505,00 €	1.754,00 €			x
Deafness, autosomal recessive type 8/10	TMPRSS3	605511	1.127,00 €					
Deafness, autosomal recessive type 9	OTOF	603681	1.931,00 €	807,00 €	2.588,00 €			x
Deafness, autosomal recessive type 15	GIPC3	608792	573,00 €					
Deafness, autosomal recessive type 16	STRC	606440	2.028,00 €	611,00 €	2.489,00 €			x
Deafness, autosomal recessive type 18	USH1C	605242	1.643,00 €	569,00 €	2.062,00 €			x
Deafness, autosomal recessive type 22	OTOA	607038	1.825,00 €					
Deafness, autosomal recessive type 23	PCDH15	605514	2.059,00 €	328,00 €	2.237,00 €			
Deafness, autosomal recessive type 24	RDX	179410	1.193,00 €					
Deafness, autosomal recessive type 25	GRXCR1	613283	468,00 €					
Deafness, autosomal recessive type 28	TRIOBP	609761	1.673,00 €					
Deafness, autosomal recessive type 29	CLDN14	605608	386,00 €					
Deafness, autosomal recessive type 30	MYO3A	606808	1.775,00 €	737,00 €	2.362,00 €			x
Deafness, autosomal recessive type 31	DFNB31	607928	995,00 €					
Deafness, autosomal recessive type 35	ESRRB	602167	819,00 €	429,00 €	1.098,00 €			
Deafness, autosomal recessive type 36	ESPN	606351	1.061,00 €					

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Deafness, autosomal recessive type 39	HGF	142409	1.326,00 €					
Deafness, autosomal recessive type 42	ILDR1	609739	819,00 €					
Deafness, autosomal recessive type 48	CIB2	605564	573,00 €					
Deafness, autosomal recessive type 49	MARVELD2	610572	737,00 €					
Deafness, autosomal recessive type 53	COL11A2	120290	1.802,00 €					
Deafness, autosomal recessive type 59	DFNB59	610219	737,00 €					
Deafness, autosomal recessive type 61	SLC26A5	604943	1.582,00 €					
Deafness, autosomal recessive type 63	LRTOMT	612414	729,00 €					
Deafness, autosomal recessive type 66	DCDC2	605755	862,00 €					
Deafness, autosomal recessive type 67	LHFPL5	609427	468,00 €					
Deafness, autosomal recessive type 70	PNPT1	610316	1.521,00 €					
Deafness, autosomal recessive type 74	MSRB3	613719	655,00 €					
Deafness, autosomal recessive type 76	SYNE4	615535	819,00 €					
Deafness, autosomal recessive type 77	LOXHD1	613072	1.931,00 €					
Deafness, autosomal recessive type 79	TPRN	613354	491,00 €					
Deafness, autosomal recessive type 84	PTPRQ	603317	2.145,00 €	905,00 €	2.900,00 €			x
Deafness, autosomal recessive type 86	TBC1D24	613577	819,00 €	437,00 €	1.106,00 €			x
Deafness, autosomal recessive type 89	KARS	601421	862,00 €					
Deafness, autosomal recessive type 91	SERPINB6	173321	819,00 €					
Deafness, autosomal recessive type 93	CABP2	614899	655,00 €					
Deafness, autosomal recessive, type 12	CDH23	605516	1.955,00 €					x
Deafness, congenital with inner ear agenesis, microtia, and microdontia	FGF3	602292	468,00 €	374,00 €	692,00 €			x
Deafness, dystonia, and cerebral hypomyelination, X-linked	BCAP31	300398	737,00 €					
Deafness, nonsyndromic, sensorineural, mitochondrial	MT-RNR1	561000	296,00 €					x
Deafness, X-linked type 1	PRPS1	311850	655,00 €	437,00 €	942,00 €			x
Deafness, X-linked type 2	POU3F4	300039	468,00 €	296,00 €	614,00 €			x
Deafness, X-linked type 4	SMPX	300226	374,00 €					
Deafness, X-linked type 5	AIFM1	300169	1.193,00 €	424,00 €	1.467,00 €			x
Deafness, X-linked type 6	COL4A6	303631	2.016,00 €	905,00 €	2.771,00 €			
Dehydrated hereditary stomatocytosis	PIEZO1	611184	2.106,00 €	796,00 €	2.752,00 €			
Dejerine-Sottas disease	EGR2	129010	491,00 €	328,00 €	669,00 €			x
Dejerine-Sottas disease	GJB1	304040	491,00 €	328,00 €	669,00 €			x
Dejerine-Sottas disease	MPZ	159440	655,00 €	328,00 €	833,00 €			x
Dejerine-Sottas disease	PMP22	601097	468,00 €	328,00 €	646,00 €			x
Dejerine-Sottas disease	PRX	605725	995,00 €	328,00 €	1.173,00 €			x
Delta-beta thalassemia	HBB	141900	491,00 €	328,00 €	669,00 €			x
Dementia, familial, British type	ITM2B	603904	573,00 €					
Dementia, familial, Danish type	ITM2B	603904	573,00 €					
Dementia, frontotemporal	GRN	138945	796,00 €	328,00 €	974,00 €			x
Dementia, frontotemporal	MAPT	157140	1.193,00 €	328,00 €	1.371,00 €			x

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Dementia, frontotemporal	PSEN1	104311	928,00 €	328,00 €	1.106,00 €			x
Dementia, frontotemporal	TARDBP	605078	819,00 €	421,00 €	1.090,00 €			x
Dementia, Lewy body	SNCA	163890	573,00 €	328,00 €	751,00 €			
Dent disease	CLCN5	300008	1.061,00 €	445,00 €	1.356,00 €			x
Dent disease type 2	OCRL	300535	1.460,00 €	505,00 €	1.815,00 €			x
Dentatorubral-pallidoluysian atrophy	ATN1	607462	1.193,00 €			200,00 €		x
Dentin dysplasia, type 2	DSPP	125485	491,00 €					
Dentinogenesis imperfecta, Shields type 2	DSPP	125485	491,00 €					
Dentinogenesis imperfecta, Shields type 3	DSPP	125485	491,00 €					
Dermatitis, atopic type 2	FLG	135940	1.622,00 €	374,00 €	1.846,00 €			x
Dermatopathia pigmentosa reticularis	KRT14	148066	862,00 €	437,00 €	1.149,00 €			x
Desbuquois dysplasia type 1	CANT1	613165	491,00 €					
Desbuquois dysplasia type 2	XYLT1	608124	862,00 €					
Desmoid disease, hereditary	APC	611731	1.927,00 €	328,00 €	2.105,00 €			x
Desmosterolosis	DHCR24	606418	819,00 €					
Developmental delay and microcephaly, SLC1A4 related	SLC1A4	600229	819,00 €					
Developmental delay, GNAQ related	GNAQ	600998	655,00 €	437,00 €	942,00 €			
Developmental delay, KMT2C related	KMT2C	606833	1.955,00 €	905,00 €	2.710,00 €			
D-glyceric aciduria	GLYCTK	610516	573,00 €					
Diabetes insipidus, nephrogenic, autosomal	AQP2	107777	468,00 €	437,00 €	755,00 €			x
Diabetes insipidus, nephrogenic, X-linked	AVPR2	300538	491,00 €	374,00 €	715,00 €			x
Diabetes insipidus, neurohypophyseal	AVP	192340	386,00 €	374,00 €	610,00 €			
Diabetes mellitus type 1	INS	176730	491,00 €	343,00 €	684,00 €			x
Diabetes mellitus, insulin-dependent type 20	HNF1A	142410	729,00 €	328,00 €	907,00 €			x
Diabetes mellitus, insulin-resistant with acanthosis nigricans	INSR	147670	1.399,00 €	463,00 €	1.712,00 €			x
Diabetes mellitus, neonatal	GLIS3	610192	995,00 €					x
Diabetes mellitus, noninsulin-dependent	ABCC8	600509	2.028,00 €	328,00 €	2.206,00 €			x
Diabetes mellitus, noninsulin-dependent	AKT2	164731	796,00 €					
Diabetes mellitus, noninsulin-dependent	KCNJ11	600937	468,00 €	296,00 €	614,00 €			x
Diabetes mellitus, permanent neonatal	ABCC8	600509	2.028,00 €	328,00 €	2.206,00 €			x
Diabetes mellitus, transient neonatal type 2	ABCC8	600509	2.028,00 €	328,00 €	2.206,00 €			x
Diabetes, IGF2 related	IGF2	147470	468,00 €	328,00 €	646,00 €			x
Diamond Blackfan anemia type 15 with mandibulofacial dysostosis	RPS28	603685	374,00 €	437,00 €	661,00 €			x
Diamond Blackfan anemia type 15 with mandibulofacial dysostosis	RPS28	603685	374,00 €	437,00 €	661,00 €			x
Diamond-Blackfan anemia type 1	RPS19	603474	737,00 €	328,00 €	915,00 €			x
Diamond-blackfan anemia type 3	RPS24	602412	737,00 €					x
Diamond-Blackfan anemia type 4	RPS17	180472	573,00 €	328,00 €	751,00 €			x
Diamond-Blackfan anemia type 5	RPL35A	180468	468,00 €	328,00 €	646,00 €			x
Diamond-Blackfan anemia type 6	RPL5	603634	819,00 €	328,00 €	997,00 €			x
Diamond-Blackfan anemia type 7	RPL11	604175	655,00 €	328,00 €	833,00 €			x

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Diamond-Blackfan anemia type 8	RPS7	603658	737,00 €					
Diamond-Blackfan anemia type 9	RPS10	603632	491,00 €					
Diamond-Blackfan anemia type 10	RPS26	603701	468,00 €	328,00 €	646,00 €			x
Diamond-Blackfan anemia type 11	RPL26	603704	374,00 €	437,00 €	661,00 €			x
Diamond-Blackfan anemia type 12	RPL15	604174	468,00 €	437,00 €	755,00 €			x
Diamond-Blackfan anemia type 13	RPS29	603633	374,00 €	374,00 €	598,00 €			x
Diamond-Blackfan anemia type 14 with mandibulofacial dysostosis	TSR2	300945	468,00 €	390,00 €	708,00 €			x
Diamond-Blackfan anemia type 14 with mandibulofacial dysostosis	TSR2	300945	468,00 €	390,00 €	708,00 €			x
Diaphragmatic hernia type 3	ZFPM2	603693	928,00 €					
Diaphyseal medullary stenosis with malignant fibrous histiocytoma	MTAP	156540	737,00 €	328,00 €	915,00 €			x
Diarrhea type 1, secretory chloride, congenital	SLC26A3	126650	1.326,00 €	328,00 €	1.504,00 €			x
Diarrhea type 2 with microvillus atrophy	MYO5B	606540	1.759,00 €	328,00 €	1.937,00 €			x
Diarrhea type 4, malabsorptive, congenital	NEUROG3	604882	386,00 €	343,00 €	579,00 €			x
Diarrhea type 6	GUCY2C	601330	1.704,00 €					
DI-CMTB	DNM2	602378	1.399,00 €	442,00 €	1.691,00 €			x
DI-CMTC	YARS	603623	928,00 €	426,00 €	1.204,00 €			x
DI-CMTD	MPZ	159440	655,00 €	328,00 €	833,00 €			x
DiGeorge syndrome	TBX1	602054	995,00 €	655,00 €	1.500,00 €			x
Dihydropyrimidine dehydrogenase deficiency	DPYD	612779	1.582,00 €	328,00 €	1.760,00 €			
Dihydropyrimidinuria	DPYS	613326	819,00 €					
Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis	DSP	125647	1.927,00 €	328,00 €	2.105,00 €			x
Dimethylglycine dehydrogenase deficiency	DMGDH	605849	1.061,00 €					
Disorders of sex development with cleft palate	FOXF2	603250	573,00 €	328,00 €	751,00 €			
Donnai-Barrow syndrome	LRP2	600073	1.955,00 €	905,00 €	2.710,00 €			
DOOR syndrome	TBC1D24	613577	819,00 €	437,00 €	1.106,00 €			x
Dopamine beta-hydroxylase (DBH) deficiency	DBH	609312	928,00 €					
Doyme honeycob retinal dystrophy	EFEMP1	601548	796,00 €	421,00 €	1.067,00 €			x
Dravet syndrome	GABRG2	137164	796,00 €	429,00 €	1.075,00 €			x
Dravet syndrome	SCN2A	182390	1.673,00 €	569,00 €	2.092,00 €			x
Dravet syndrome, modifier of	SCN9A	603415	1.572,00 €	569,00 €	1.991,00 €			x
Duane Retraction syndrome	CHN1	118423	928,00 €					
Duane Retraction syndrome	SALL4	607343	729,00 €	437,00 €	1.016,00 €			x
Duane Retraction syndrome	SALL4	607343	729,00 €	437,00 €	1.016,00 €			x
Dubin-Johnson syndrome	ABCC2	601107	1.673,00 €	674,00 €	2.197,00 €			x
Dyggve-Melchior-Clausen disease	DYM	607461	1.193,00 €					
Dysautonomia, FRRS1L-related	FRRS1L	604574	491,00 €					
Dyschromatosis symmetrica hereditaria	ADAR	146920	1.326,00 €					
Dyschromatosis universalis hereditaria type 3	ABCB6	605452	1.260,00 €					
Dyserythropoietic anemia	COX4I2	607976	468,00 €					
Dyserythropoietic anemia, congenital, type 1B	C15orf41	615626	862,00 €	421,00 €	1.133,00 €			x

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Dyserythropoietic anemia, congenital, type 3	KIF23	605064	1.278,00 €	484,00 €	1.612,00 €			x
Dyserythropoietic anemia, congenital, type 4	KLF1	600599	468,00 €	374,00 €	692,00 €			x
Dyskeratosis congenita, autosomal dominant type 1	TERC	602322	296,00 €	328,00 €	474,00 €			x
Dyskeratosis congenita, autosomal recessive type 1	NOP10	606471	386,00 €					
Dyskeratosis congenita, autosomal recessive type 2	NHP2	606470	374,00 €					x
Dyskeratosis congenita, autosomal recessive type 4/ autosomal dominant type 2	TERT	187270	1.260,00 €	328,00 €	1.438,00 €			x
Dyskeratosis congenita, autosomal recessive type 5	RTEL1	608833	1.775,00 €					x
Dyskeratosis congenita, autosomal recessive type 6	PARN	604212	1.460,00 €					
Dyskeratosis congenita, autosomal recessive type 7	ACD	609377	995,00 €					
Dyskeratosis congenita, X-linked	DKC1	300126	1.061,00 €	328,00 €	1.239,00 €			x
Dyskinesia, familial, with facial myokymia	ADCY5	600293	1.521,00 €	442,00 €	1.813,00 €			x
Dyslexia	PCDH11X	300246	1.193,00 €					
Dysmorphism, HMG20B related	HMG20B	605535	819,00 €					
Dysprothrombinemia	F2	176930	1.061,00 €					
Dyssegmental dysplasia, Silverman-Handmaker type	HSPG2	142461	1.955,00 €					x
Dyssegmental dysplasia, Silverman-Handmaker type	HSPG2	142461	1.955,00 €					x
Dystonia juvenile-onset	ACTB	102630	573,00 €	421,00 €	844,00 €			x
Dystonia, DOPA-responsive, autosomal recessive	SPR	182125	374,00 €	374,00 €	598,00 €			x
Dystonia-deafness syndrome	TIMM8A	300356	374,00 €	343,00 €	567,00 €			x
DYT1	TOR1A	605204	491,00 €	328,00 €	669,00 €			x
DYT2	HPCA	142622	468,00 €					
DYT3	TAF1	313650	1.927,00 €	821,00 €	2.598,00 €			
DYT4	TUBB4A	602662	655,00 €	390,00 €	895,00 €			x
DYT5A	GCH1	600225	737,00 €	328,00 €	915,00 €			x
DYT6	THAP1	609520	374,00 €	328,00 €	552,00 €			x
DYT8	SLC2A1	138140	819,00 €	328,00 €	997,00 €			x
DYT10	PRRT2	614386	468,00 €	374,00 €	692,00 €			x
DYT11, DRD2 related	DRD2	126450	737,00 €	437,00 €	1.024,00 €			x
DYT11	SGCE	604149	928,00 €	328,00 €	1.106,00 €			x
DYT12	ATP1A3	182350	1.326,00 €	328,00 €	1.504,00 €			x
DYT16	PRKRA	603424	655,00 €	328,00 €	833,00 €			x
DYT18	SLC2A1	138140	819,00 €	328,00 €	997,00 €			x
DYT23	CACNA1B	601012	1.989,00 €					
DYT24	ANO3	610110	1.764,00 €	590,00 €	2.204,00 €			
DYT25	GNAL	139312	928,00 €	421,00 €	1.199,00 €			x
DYT26, myoclonic	KCTD17	616386	819,00 €					
DYT27	COL6A3	120250	2.106,00 €	755,00 €	2.711,00 €			x
Early infantile epileptic encephalopathy type 1	ARX	300382	737,00 €	328,00 €	915,00 €			x
Early infantile epileptic encephalopathy type 2	CDKL5	300203	1.460,00 €	328,00 €	1.638,00 €			x
Early infantile epileptic encephalopathy type 3	SLC25A22	609302	655,00 €					

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Early infantile epileptic encephalopathy type 4	STXBP1	602926	1.278,00 €	437,00 €	1.565,00 €			x
Early infantile epileptic encephalopathy type 5	SPTAN1	182810	2.067,00 €					
Early infantile epileptic encephalopathy type 6	SCN1A	182389	1.572,00 €	328,00 €	1.750,00 €			x
Early infantile epileptic encephalopathy type 7	KCNQ2	602235	1.260,00 €	328,00 €	1.438,00 €			x
Early infantile epileptic encephalopathy type 8	ARHGEF9	300429	796,00 €					
Early infantile epileptic encephalopathy type 9	PCDH19	300460	862,00 €	328,00 €	1.040,00 €			x
Early infantile epileptic encephalopathy type 10	PNKP	605610	796,00 €	424,00 €	1.070,00 €			x
Early infantile epileptic encephalopathy type 11	SCN2A	182390	1.673,00 €	569,00 €	2.092,00 €			x
Early infantile epileptic encephalopathy type 12	PLCB1	607120	1.673,00 €	674,00 €	2.197,00 €			x
Early infantile epileptic encephalopathy type 13	SCN8A	600702	1.825,00 €					
Early infantile epileptic encephalopathy type 14	KCNT1	608167	1.775,00 €	653,00 €	2.278,00 €			x
Early infantile epileptic encephalopathy type 15	ST3GAL3	606494	862,00 €					
Early infantile epileptic encephalopathy type 16	TBC1D24	613577	819,00 €	437,00 €	1.106,00 €			x
Early infantile epileptic encephalopathy type 17	GNAO1	139311	729,00 €					
Early infantile epileptic encephalopathy type 19	GABRA1	137160	796,00 €					
Early infantile epileptic encephalopathy type 20	PIGA	311770	655,00 €	421,00 €	926,00 €			
Early infantile epileptic encephalopathy type 21	NECAP1	611623	737,00 €					
Early infantile epileptic encephalopathy type 23	DOCK7	615730	1.973,00 €	905,00 €	2.728,00 €			
Early infantile epileptic encephalopathy type 24	HCN1	602780	819,00 €					
Early infantile epileptic encephalopathy type 25	SLC13A5	608305	862,00 €	421,00 €	1.133,00 €			
Early infantile epileptic encephalopathy type 26	KCNB1	600397	655,00 €					
Early infantile epileptic encephalopathy type 27	GRIN2B	138252	1.278,00 €	328,00 €	1.456,00 €			x
Early infantile epileptic encephalopathy type 28	WWOX	605131	729,00 €	328,00 €	907,00 €			x
Early infantile epileptic encephalopathy type 29	AARS	601065	1.326,00 €					x
Early infantile epileptic encephalopathy type 30	SIK1	605705	995,00 €					
Early infantile epileptic encephalopathy type 31	DNM1	602377	1.521,00 €					
Early infantile epileptic encephalopathy type 32	KCNA2	176262	468,00 €					
Early infantile epileptic encephalopathy type 33	EEF1A2	602959	737,00 €					
Early infantile epileptic encephalopathy type 40	GUF1	617064	1.193,00 €					x
Early infantile epileptic encephalopathy type 45	GABRB1	137190	729,00 €					
Early infantile epileptic encephalopathy type 47	FGF12	601513	655,00 €					
Early onset glaucoma, phenotype modifier of, COL15A1 related	COL15A1	120325	1.802,00 €	905,00 €	2.557,00 €			
Ectodactyly, ectodermal dysplasia, and cleft lip/palate syndrome type 3	TP63	603273	1.061,00 €	424,00 €	1.335,00 €			x
Ectodermal dysplasia type 4, hair/nail type	KRT85	602767	729,00 €					
Ectodermal dysplasia, ectrodactyly, and macular dystrophy	CDH3	114021	1.193,00 €					
Ectodermal dysplasia, hidrotic	GJB6	604418	374,00 €	328,00 €	552,00 €			x
Ectodermal dysplasia, hypohidrotic, autosomal recessive	EDAR	604095	729,00 €	328,00 €	907,00 €			x
Ectodermal dysplasia, hypohidrotic, autosomal recessive	EDARADD	606603	655,00 €	328,00 €	833,00 €			x
Ectodermal dysplasia, hypohidrotic, with immune deficiency	IKBKG	300248	796,00 €					x
Ectodermal dysplasia, hypohidrotic, X-linked	EDA	300451	729,00 €	328,00 €	907,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Ectodermal dysplasia/skin fragility syndrome	PKP1	601975	995,00 €					
Ectopia lentis et pupillae	ADAMTSL4	610113	1.326,00 €	437,00 €	1.613,00 €			x
Ectopia lentis, familial	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			
Ectopia lentis, isolated, autosomal recessive	ADAMTSL4	610113	1.326,00 €	437,00 €	1.613,00 €			x
Efavirenz, poor metabolism of	CYP2B6	123930	796,00 €	328,00 €	974,00 €			
EGFR somatic Hotspot: c.2573T>G, p.L858R	EGFR	131550						
EGFR, selective sequencing of exons 18-21	EGFR	131550						
Ehlers-Danlos syndrome type 1/2	COL5A1	120215	2.141,00 €	655,00 €	2.646,00 €			x
Ehlers-Danlos syndrome type 1/2	COL5A2	120190	1.989,00 €	842,00 €	2.681,00 €			x
Ehlers-Danlos syndrome type 3	COL3A1	120180	1.989,00 €	328,00 €	2.167,00 €			x
Ehlers-Danlos syndrome type 3	TNXB	600985	2.016,00 €	328,00 €	2.194,00 €			x
Ehlers-Danlos syndrome type 4	COL3A1	120180	1.989,00 €	328,00 €	2.167,00 €			x
Ehlers-Danlos syndrome type 4	COL5A1	120215	2.141,00 €	655,00 €	2.646,00 €			x
Ehlers-Danlos syndrome type 6	PLOD1	153454	1.326,00 €	328,00 €	1.504,00 €			x
Ehlers-Danlos syndrome type 7A	COL1A1	120150	1.825,00 €	328,00 €	2.003,00 €			x
Ehlers-Danlos syndrome type 7B	COL1A2	120160	2.016,00 €	328,00 €	2.194,00 €			x
Ehlers-Danlos syndrome type 7C	ADAMTS2	604539	1.521,00 €	463,00 €	1.834,00 €			x
Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss	FKBP14	614505	468,00 €					
Ehlers-Danlos syndrome, musculocontractural type 1	CHST14	608429	468,00 €	296,00 €	614,00 €			x
Ehlers-Danlos syndrome, musculocontractural type 2	DSE	605942	729,00 €					
Ehlers-Danlos syndrome, progeroid type 1	B4GALT7	604327	491,00 €	421,00 €	762,00 €			x
Ehlers-Danlos syndrome, progeroid type, type 2	B3GALT6	615291	468,00 €					
Ellis-van Creveld syndrome	EVC2	607261	1.460,00 €	463,00 €	1.773,00 €			x
Ellis-van Creveld syndrome	EVC	604831	1.460,00 €	442,00 €	1.752,00 €			
Emberger syndrome	GATA2	137295	655,00 €	328,00 €	833,00 €			
Emery-Dreifuss muscular dystrophy type 1	EMD	300384	573,00 €	421,00 €	844,00 €			x
Emery-Dreifuss muscular dystrophy type 2	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Emery-Dreifuss muscular dystrophy type 4	SYNE1	608441	1.955,00 €	905,00 €				x
Emery-Dreifuss muscular dystrophy type 5	SYNE2	608442	1.955,00 €	905,00 €	2.710,00 €			x
Emery-Dreifuss muscular dystrophy type 6	FHL1	300163	737,00 €	437,00 €	1.024,00 €			x
Encephalomyopathy, mitochondrial, MT-TL2 related	MT-TL2	590055	296,00 €					
Encephalomyopathy, mitochondrial, MT-TR related	MT-TR	590005	296,00 €					
Encephalopathy acute necrotizing type 1	RANBP2	601181	1.931,00 €	611,00 €	2.392,00 €			x
Encephalopathy lethal, due to defective mitochondrial peroxisomal fission	DNM1L	603850	1.338,00 €					
Encephalopathy mitochondrial	VDAC1	604492	655,00 €					x
Encephalopathy mitochondrial with proximal renal tubulopathy due to cytochrome c oxidase deficiency	COX10	602125	737,00 €	328,00 €	915,00 €			
Encephalopathy neonatal severe	MECP2	300005	468,00 €	328,00 €	646,00 €			x
Encephalopathy thiamine-responsive	SLC19A3	606152	655,00 €	421,00 €	926,00 €			x
Encephalopathy, familial, with neuroserpin inclusion bodies	SERPINI1	602445	737,00 €					x
Encephalopathy, mitochondrial, MT-TW related	MT-TW	590095	296,00 €					

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Encephalopathy, progressive, with or without lipodystrophy	BSC12	606158	819,00 €	429,00 €	1.098,00 €			x
Endometrial cancer, familial, MSH6 related	MSH6	600678	1.127,00 €	328,00 €	1.305,00 €			
Endometrial carcinoma, somatic	CDH1	192090					1.900,00 €	
Endometrioid carcinoma, ARID1A related, somatic	ARID1A	603024					1.900,00 €	
Endplate acetylcholinesterase deficiency	COLQ	603033	1.260,00 €					
Enterokinase deficiency	TMPRSS15	606635	1.582,00 €					
Epidermal nevus, somatic	NRAS	164790					950,00 €	
Epidermolysis bullosa dystrophica	COL7A1	120120	2.176,00 €	328,00 €	2.354,00 €			x
Epidermolysis bullosa dystrophica, autosomal recessive, modifier of	MMP1	120353	729,00 €					
Epidermolysis bullosa junctionalis with pyloric atresia	ITGA6	147556	1.521,00 €	527,00 €	1.898,00 €			
Epidermolysis bullosa junctionalis with pyloric atresia	ITGB4	147557	1.977,00 €	842,00 €	2.669,00 €			x
Epidermolysis bullosa simplex	KRT5	148040	819,00 €	328,00 €	997,00 €			x
Epidermolysis bullosa simplex with muscular dystrophy	PLEC	601282	1.955,00 €					x
Epidermolysis bullosa simplex with pyloric atresia	PLEC	601282	1.955,00 €					x
Epidermolysis bullosa simplex, autosomal recessive type 1	KRT14	148066	862,00 €	437,00 €	1.149,00 €			x
Epidermolysis bullosa simplex, autosomal recessive type 2	DST	113810	2.225,00 €					x
Epidermolysis bullosa simplex, autosomal recessive type 2	DST	113810	2.225,00 €					x
Epidermolysis bullosa simplex, Dowling-Meara type	KRT14	148066	862,00 €	437,00 €	1.149,00 €			x
Epidermolysis bullosa simplex, Koebner type	KRT14	148066	862,00 €	437,00 €	1.149,00 €			x
Epidermolysis bullosa simplex, Ogna type	PLEC	601282	1.955,00 €					x
Epidermolysis bullosa simplex, Weber-Cockayne type	KRT14	148066	862,00 €	437,00 €	1.149,00 €			x
Epidermolysis bullosa, generalized atrophic benign	LAMA3	600805	1.955,00 €	905,00 €	2.710,00 €			
Epidermolysis bullosa, junctional	COL17A1	113811	2.028,00 €	874,00 €	2.752,00 €			x
Epidermolysis bullosa, junctional	LAMC2	150292	1.399,00 €					x
Epidermolysis bullosa, junctional, Herlitz type	LAMA3	600805	1.955,00 €	905,00 €	2.710,00 €			
Epidermolysis bullosa, junctional, Herlitz type	LAMB3	150310	1.460,00 €	484,00 €	1.794,00 €			x
Epidermolysis bullosa, junctional, non-Herlitz type	LAMB3	150310	1.460,00 €	484,00 €	1.794,00 €			x
Epidermolysis bullosa, lethal acantholytic	DSP	125647	1.927,00 €	328,00 €	2.105,00 €			x
Epidermolysis bullosa, nonspecific, autosomal recessive	EXPH5	612878	1.399,00 €					
Epidermolytic hyperkeratosis	KRT1	139350	862,00 €					x
Epidermolytic hyperkeratosis	KRT10	148080	796,00 €	437,00 €	1.083,00 €			
Epidermolytic palmoplantar keratoderma	KRT9	607606	819,00 €	437,00 €	1.106,00 €			x
Epilepsy with neurodevelopmental defects	GRIN2A	138253	1.326,00 €	328,00 €	1.504,00 €			x
Epilepsy, childhood absence type 2	GABRG2	137164	796,00 €	429,00 €	1.075,00 €			x
Epilepsy, childhood absence type 4, susceptibility to	GABRA1	137160	796,00 €					
Epilepsy, childhood absence type 5	GABRB3	137192	862,00 €	328,00 €	1.040,00 €			x
Epilepsy, childhood absence type 6, susceptibility to	CACNA1H	607904	1.977,00 €					
Epilepsy, childhood absence, JRK related	JRK	603210	573,00 €					x
Epilepsy, familial focal with variable foci	DEPDC5	614191	1.931,00 €	738,00 €	2.519,00 €			
Epilepsy, familial temporal lobe type 1	LGI1	604619	819,00 €	328,00 €	997,00 €			x



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Epilepsy, familial temporal lobe type 5	CPA6	609562	796,00 €					
Epilepsy, familial temporal lobe type 7	RELN	600514	1.955,00 €					x
Epilepsy, focal, SCN3A related	SCN3A	182391	1.825,00 €					
Epilepsy, HCN2 related	HCN2	602781	729,00 €					
Epilepsy, hearing loss, and mental retardation syndrome	SPATA5	613940	1.260,00 €					
Epilepsy, idiopathic generalized type 10	GABRD	137163	737,00 €	421,00 €	1.008,00 €			x
Epilepsy, idiopathic generalized type 11	CLCN2	600570	1.326,00 €	505,00 €	1.681,00 €			x
Epilepsy, idiopathic generalized type 12	SLC2A1	138140	819,00 €	328,00 €	997,00 €			x
Epilepsy, juvenile absence type 1	EFHC1	608815	928,00 €	429,00 €	1.207,00 €			x
Epilepsy, nocturnal frontal lobe	KCNT1	608167	1.775,00 €	653,00 €	2.278,00 €			x
Epilepsy, nocturnal frontal lobe type 1	CHRNA4	118504	819,00 €	328,00 €	997,00 €			x
Epilepsy, nocturnal frontal lobe type 3	CHRN2	118507	655,00 €	328,00 €	833,00 €			x
Epilepsy, nocturnal frontal lobe type 4	CHRNA2	118502	737,00 €	437,00 €	1.024,00 €			x
Epilepsy, progressive myoclonic 4, with or without renal failure	SCARB2	602257	862,00 €	421,00 €	1.133,00 €			x
Epilepsy, progressive myoclonic type 5	PRICKLE2	608501	819,00 €					
Epilepsy, X-linked, with learning disabilities and behavior disorders	SYN1	313440	995,00 €	426,00 €	1.271,00 €			x
Epileptic encephalopathy, childhood-onset	CHD2	602119	1.802,00 €	905,00 €	2.557,00 €			
Epileptic encephalopathy, Lennox-Gastaut type	MAPK10	602897	1.193,00 €					
Epiphyseal dysplasia, multiple, type 1	COMP	600310	995,00 €	430,00 €	1.275,00 €			x
Epiphyseal dysplasia, multiple, type 3	COL9A3	120270	1.673,00 €					
Epiphyseal dysplasia, multiple, type 5	MATN3	602109	819,00 €					
Epiphyseal dysplasia, multiple, with myopia and deafness	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Episodic ataxia type 1	KCNA1	176260	468,00 €	328,00 €	646,00 €			x
Episodic ataxia type 2	CACNA1A	601011	2.067,00 €	328,00 €	2.245,00 €	200,00 €		x
Episodic ataxia type 5	CACNB4	601949	1.127,00 €	437,00 €	1.414,00 €			
Episodic ataxia type 6	SLC1A3	600111	819,00 €	429,00 €	1.098,00 €			x
Episodic pain syndrome type 2, familial	SCN10A	604427	1.622,00 €					
Episodic pain syndrome type 3, familial	SCN11A	604385	1.572,00 €					
Epstein syndrome	MYH9	160775	2.028,00 €	328,00 €	2.206,00 €			x
Erythralgia, primary	SCN9A	603415	1.572,00 €	569,00 €	1.991,00 €			x
Erythrocyte lactate transporter defect	SLC16A1	600682	655,00 €	390,00 €	895,00 €			x
Erythrocytosis, familial type 1	EPOR	133171	819,00 €	437,00 €	1.106,00 €			
Erythrocytosis, familial type 3	EGLN1	606425	655,00 €	390,00 €	895,00 €			x
Erythrocytosis, familial type 4	EPAS1	603349	1.193,00 €	424,00 €	1.467,00 €			
Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis and hyper IgE	DSG1	125670	1.193,00 €					
Erythrokeratoderma variabilis et progressive	GJB3	603324	374,00 €	328,00 €	552,00 €			x
Erythrokeratoderma variabilis et progressive	GJB4	605425	374,00 €					
Esophageal cancer, somatic	TGFBR2	190182					1.900,00 €	
Esophageal carcinoma, somatic	DCC	120470					1.900,00 €	
Estrogen resistance	ESR1	133430	737,00 €					

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Ethylmalonic encephalopathy	ETHE1	608451	655,00 €	437,00 €	942,00 €			x
Exfoliation syndrome, susceptibility to	LOXL1	153456	796,00 €					
Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	COX4I2	607976	468,00 €					
Exostoses, multiple, type 1	EXT1	608177	928,00 €	328,00 €	1.106,00 €			x
Exostoses, multiple, type 2	EXT2	608210	1.127,00 €	328,00 €	1.305,00 €			x
Exudative vitreoretinopathy	FZD4	604579	573,00 €	328,00 €	751,00 €			x
Exudative vitreoretinopathy type 2	NDP	300658	491,00 €	374,00 €	715,00 €			x
Exudative vitreoretinopathy type 5	TSPAN12	613138	737,00 €					
Fabry disease	GLA	300644	819,00 €	328,00 €	997,00 €			x
Fabry disease	GLA	300644	819,00 €	328,00 €	997,00 €			x
Facial paresis type 3	HOXB1	142968	468,00 €					x
Facio-genital dysplasia	FGD1	300546	1.193,00 €	328,00 €	1.371,00 €			x
Facioscapulohumeral dystrophy-like phenotype, FAT1 related	FAT1	600976	1.802,00 €					
Factor II deficiency	F2	176930	1.061,00 €					
Factor II deficiency	F2	176930	1.061,00 €					
Factor V deficiency	F5	612309	1.673,00 €					x
Factor VII deficiency	F7	613878	729,00 €	328,00 €	907,00 €			
Factor X deficiency	F10	613872	819,00 €	328,00 €	997,00 €			x
Factor XI deficiency	F11	264900	928,00 €	328,00 €	1.106,00 €			
Factor XII deficiency	F12	610619	928,00 €	437,00 €	1.215,00 €			x
Factor XIII A deficiency	F13A1	134570	995,00 €	445,00 €	1.290,00 €			x
Factor XIII B deficiency	F13B	134580	928,00 €					
Familial adenomatous polyposis coli	APC	611731	1.927,00 €	328,00 €	2.105,00 €			x
Familial adenomatous polyposis type 2	MUTYH	604933	862,00 €	328,00 €	1.040,00 €			
Familial adenomatous polyposis type 3	NTHL1	602656	491,00 €	421,00 €	762,00 €			
Familial adenomatous polyposis type 4	MSH3	600887	1.521,00 €	505,00 €	1.876,00 €			
Familial atrial fibrillation type 13	SCN1B	600235	573,00 €	374,00 €	797,00 €			x
Familial hemiplegic migraine type 1	CACNA1A	601011	2.067,00 €	328,00 €	2.245,00 €			x
Familial hemiplegic migraine type 2	ATP1A2	182340	1.399,00 €	328,00 €	1.577,00 €			x
Familial hemiplegic migraine type 3	SCN1A	182389	1.572,00 €	328,00 €	1.750,00 €			x
Familial infantile myoclonic epilepsy	TBC1D24	613577	819,00 €	437,00 €	1.106,00 €			x
Fanconi anemia type A	FANCA	607139	1.888,00 €	655,00 €	2.393,00 €			x
Fanconi anemia type B	FANCB	300515	995,00 €	328,00 €	1.173,00 €			x
Fanconi anemia type C	FANCC	613899	1.061,00 €					
Fanconi anemia type D1	BRC A2	600185	525,00 €	330,00 €	705,00 €			
Fanconi anemia type D2	FANCD2	613984	1.845,00 €	328,00 €	2.023,00 €			x
Fanconi anemia type E	FANCE	613976	796,00 €					
Fanconi anemia type F	FANCF	613897	374,00 €					x
Fanconi anemia type G	FANCG	602956	729,00 €	437,00 €	1.016,00 €			
Fanconi anemia type I	FANCI	611360	1.876,00 €					

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Fanconi anemia type J	BRIP1	605882	1.399,00 €	328,00 €	1.577,00 €			x
Fanconi anemia type L	FANCL	608111	995,00 €					
Fanconi anemia type M	FANCM	609644	1.622,00 €					
Fanconi anemia type N	PALB2	610355	1.193,00 €	328,00 €	1.371,00 €			x
Fanconi anemia type P	SLX4	613278	1.338,00 €					x
Fanconi anemia, complementation group Q	ERCC4	133520	862,00 €					
Fanconi anemia, complementation group Q	ERCC4	133520	862,00 €					
Fanconi anemia, XRCCR2 related	XRCC2	600375	468,00 €	374,00 €	692,00 €			x
Fanconi renotubular syndrome type 2	SLC34A1	182309	729,00 €					x
Fanconi-Bickel syndrome	SLC2A2	138160	796,00 €	429,00 €	1.075,00 €			x
Fanconi-Bickel syndrome	SLC2A2	138160	796,00 €	429,00 €	1.075,00 €			x
Farber disease	ASAH1	613468	1.061,00 €	437,00 €	1.348,00 €			x
Fatal familial insomnia	PRNP	176640	386,00 €	343,00 €	579,00 €	200,00 €		
Favism, susceptibility to	G6PD	305900	862,00 €	426,00 €	1.138,00 €			x
Favism, susceptibility to	G6PD	305900	862,00 €	426,00 €	1.138,00 €			x
Fazio-Londe disease	SLC52A3	613350	573,00 €	390,00 €	813,00 €			x
Febrile seizures, familial, type 4	ADGRV1	602851	1.955,00 €	905,00 €	2.710,00 €			
Feingold syndrome	MYCN	164840	573,00 €	328,00 €	751,00 €			x
Feingold syndrome type 2	MIR17HG	609415	468,00 €					
Fetal akinesia deformation sequence	DOK7	610285	796,00 €	429,00 €	1.075,00 €			x
Fetal akinesia deformation sequence	RAPSN	601592	655,00 €	437,00 €	942,00 €			x
FG syndrome type 1	MED12	300188	1.888,00 €	772,00 €	2.510,00 €			x
FG syndrome type 2	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x
FG syndrome type 4	CASK	300172	1.704,00 €	328,00 €	1.882,00 €			x
Fibrochondrogenesis 2	COL11A2	120290	1.802,00 €					
Fibrochondrogenesis type 1	COL11A1	120280	1.955,00 €	655,00 €	2.460,00 €			x
Fibrochondrogenesis type 1	COL11A1	120280	1.955,00 €	655,00 €	2.460,00 €			x
Fibrodysplasia ossificans progressiva	ACVR1	102576	819,00 €	429,00 €	1.098,00 €			x
Fibrosis of extraocular muscles, congenital type 1	KIF21A	608283	1.927,00 €					
Fibrosis of extraocular muscles, congenital type 2	PHOX2A	602753	491,00 €	374,00 €	715,00 €			x
Fibrosis of extraocular muscles, congenital type 3a	TUBB3	602661	573,00 €					
Fibular aplasia or hypoplasia, femoral bowing and poly-, syn-, and oligodactyly	WNT7A	601570	468,00 €	437,00 €	755,00 €			x
Filaminopathy	FLNC	102565	1.845,00 €					x
FILS syndrome	POLE	174762	2.059,00 €	905,00 €	2.814,00 €			
Fish eye disease	LCAT	606967	655,00 €	421,00 €	926,00 €			x
Fleck retina, familial benign	PLA2G5	601192	491,00 €					
Floating-Harbor syndrome	SRCAP	611421	2.028,00 €	716,00 €	2.594,00 €			x
Focal dermal hypoplasia	PORCN	300651	729,00 €	445,00 €	1.024,00 €			x
Focal segmental glomerulosclerosis and dilated cardiomyopath, MT-TY related	MT-TY	590100	296,00 €					
Focal segmental glomerulosclerosis type 1	ACTN4	604638	1.399,00 €					

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Focal segmental glomerulosclerosis type 2	TRPC6	603652	995,00 €					
Focal segmental glomerulosclerosis type 3	CD2AP	604241	1.278,00 €					
Focal segmental glomerulosclerosis type 4, susceptibility to	APOL1	603743	655,00 €					
Focal segmental glomerulosclerosis type 5	INF2	610982	1.643,00 €	484,00 €	1.977,00 €			x
Focal segmental glomerulosclerosis type 6	MYO1E	601479	1.764,00 €					
Focal segmental glomerulosclerosis type 7	PAX2	167409	862,00 €	429,00 €	1.141,00 €			x
Focal segmental glomerulosclerosis type 8	ANLN	616027	1.521,00 €					
Focal segmental glomerulosclerosis type 9	CRB2	609720	1.260,00 €					
Focal segmental glomerulosclerosis, LAMA5 related	LAMA5	601033	2.282,00 €					
Folate malabsorption, hereditary	SLC46A1	611672	573,00 €					
Follicle-stimulating hormone deficiency, isolated	FSHB	136530	386,00 €	328,00 €	564,00 €			
Foveal hypoplasia type 1	PAX6	607108	1.061,00 €	328,00 €	1.239,00 €			x
Fragile X syndrome	FMR1	309550	1.399,00 €	328,00 €	1.577,00 €	400,00 €		x
Fragile X tremor/ataxia syndrome	FMR1	309550	1.399,00 €	328,00 €	1.577,00 €	400,00 €		x
Frank-ter Haar syndrome	SH3PXD2B	613293	1.127,00 €					
Fraser syndrome	FRAS1	607830	1.955,00 €	905,00 €	2.710,00 €			x
Fraser syndrome	FREM2	608945	1.724,00 €					
Fraser syndrome	GRIP1	604597	1.460,00 €	505,00 €	1.815,00 €			x
Friedreich ataxia	FXN	606829	573,00 €	328,00 €	751,00 €	200,00 €		x
Frontometaphyseal dysplasia	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x
Frontonasal dysplasia type 1	ALX3	606014	468,00 €	328,00 €	646,00 €			x
Frontonasal dysplasia type 2	ALX4	605420	573,00 €	328,00 €	751,00 €			x
Fructose-1,6-bisphosphatase deficiency	FBP1	611570	655,00 €	437,00 €	942,00 €			x
Fructose intolerance	ALDOB	612724	737,00 €	328,00 €	915,00 €			x
Fructose uptake deficiency, SLC2A5 related	SLC2A5	138230	796,00 €					
Fructosuria essential	KHK	614058	655,00 €					
FSH releasing protein deficiency	INHBA	147290	468,00 €					
Fucosidosis	FUCA1	612280	819,00 €	437,00 €	1.106,00 €			x
Fucosidosis	FUCA1	612280	819,00 €	437,00 €	1.106,00 €			x
Fukuyama congenital muscular dystrophy	FKTN	607440	928,00 €	328,00 €	1.106,00 €			x
Fumarase deficiency	FH	136850	729,00 €	328,00 €	907,00 €			x
Fumarase deficiency	FH	136850	729,00 €	328,00 €	907,00 €			x
Fundus albipunctatus	PRPH2	179605	468,00 €	328,00 €	646,00 €			x
Fundus albipunctatus	RDH5	601617	491,00 €					
GABA-transaminase deficiency	ABAT	137150	1.061,00 €					
Galactokinase deficiency	GALK1	604313	655,00 €	437,00 €	942,00 €			x
Galactose epimerase deficiency	GALE	606953	729,00 €	421,00 €	1.000,00 €			x
Galactosemia	GALT	606999	819,00 €	328,00 €	997,00 €			x
Galactosialidosis	CTSA	613111	928,00 €	445,00 €	1.223,00 €			x
Gallbladder disease type 1	ABCB4	171060	1.704,00 €	328,00 €	1.882,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Galloway-Mowat syndrome	WDR73	616144	737,00 €					
Gastric cancer, BLM related, somatic	BLM	604610					1.900,00 €	
Gastric cancer, hereditary diffuse	CDH1	192090	1.127,00 €	328,00 €	1.305,00 €			
Gastric cancer, somatic	APC	611731					1.900,00 €	
Gastric cancer, somatic	ERBB2	164870					1.900,00 €	
Gastric cancer, somatic	FGFR2	176943					1.900,00 €	
Gastric cancer, somatic	KLF6	602053					1.900,00 €	
Gastric cancer, somatic	KRAS	190070					950,00 €	
Gastric cancer, somatic	MUTYH	604933					1.900,00 €	
Gastrointestinal stromal tumor, familial	KIT	164920	1.326,00 €	328,00 €	1.504,00 €			x
Gastrointestinal stromal tumor, somatic	PDGFRA	173490					1.900,00 €	
Gaucher disease type 1	GBA	606463	796,00 €	421,00 €	1.067,00 €			x
Gaucher disease type 2	GBA	606463	796,00 €	421,00 €	1.067,00 €			x
Gaucher disease type 3C	GBA	606463	796,00 €	421,00 €	1.067,00 €			x
Gaucher disease type 3	GBA	606463	796,00 €	421,00 €	1.067,00 €			x
Gaucher disease, atypical	PSAP	176801	1.061,00 €	445,00 €	1.356,00 €			x
Gaucher disease, perinatal lethal	GBA	606463	796,00 €	421,00 €	1.067,00 €			x
Gaze palsy, horizontal, with progressive scoliosis	ROBO3	608630	1.582,00 €					
Gaze palsy, horizontal, with progressive scoliosis	ROBO3	608630	1.582,00 €					
Geleophysic dysplasia type 1	ADAMTSL2	612277	1.278,00 €	430,00 €	1.558,00 €			x
Geleophysic dysplasia type 2	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			x
Generalized epilepsy and paroxysmal dyskinesia	KCNMA1	600150	1.643,00 €					
Generalized epilepsy with febrile seizures plus type 1	SCN1B	600235	573,00 €	374,00 €	797,00 €			x
Generalized epilepsy with febrile seizures plus type 2	SCN1A	182389	1.572,00 €	328,00 €	1.750,00 €			x
Generalized epilepsy with febrile seizures plus type 3	GABRG2	137164	796,00 €	429,00 €	1.075,00 €			x
Generalized epilepsy with febrile seizures plus type 7	SCN9A	603415	1.572,00 €	569,00 €	1.991,00 €			x
Genitopatellar syndrome	KAT6B	605880	1.582,00 €	421,00 €	1.853,00 €			x
Germ cell tumors, somatic	KIT	164920					1.900,00 €	
Geroderma osteodysplasticum	GORAB	607983	655,00 €					
Gerstmann-Straussler disease	PRNP	176640	386,00 €	343,00 €	579,00 €	200,00 €		
Ghosal hematodiaphyseal syndrome	TBXAS1	274180	1.061,00 €					
Giant axonal neuropathy type 1	GAN	605379	729,00 €	429,00 €	1.008,00 €			x
Gilbert syndrome	UGT1A1	191740	737,00 €	390,00 €	977,00 €			x
Gillespie syndrome	ITPR1	147265	1.955,00 €					x
Gillespie syndrome	ITPR1	147265	1.955,00 €					x
Gitelman syndrome	SLC12A3	600968	1.825,00 €	328,00 €	2.003,00 €			x
Glass syndrome	SATB2	612313	862,00 €					
Glaucoma, open angle type 1A	MYOC	601652	655,00 €					
Glaucoma, open angle type 1E	OPTN	602432	928,00 €	424,00 €	1.202,00 €			x
Glaucoma, open angle type 1F	ASB10	615054	573,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Glaucoma, open angle type 1F	NTF4	162662	386,00 €					
Glaucoma, open angle type 1G	WDR36	609669	1.460,00 €					x
Glaucoma, primary type 3A	CYP1B1	601771	737,00 €	328,00 €	915,00 €			x
Glaucoma, primary type 3D	LTBP2	602091	1.724,00 €					x
Glioblastoma type 3, susceptibility to, due to BRCA2 germline mutation	BRCA2	600185	525,00 €	330,00 €	705,00 €			
Glioblastoma, somatic	ERBB2	164870					1.900,00 €	
Glioma susceptibility 1, susceptibility to, somatic	TP53	191170					1.900,00 €	
Glioma, susceptibility to, somatic	IDH1	147700					1.900,00 €	
Glomerulocystic kidney disease with hyperuricemia and isosthenuria	UMOD	191845	862,00 €	421,00 €	1.133,00 €			x
Glucocorticoid deficiency type 1	MC2R	607397	374,00 €					
Glucocorticoid deficiency type 2	MRAP	609196	468,00 €					
Glucocorticoid resistance, generalized	NR3C1	138040	862,00 €	421,00 €	1.133,00 €			
Glucose/Galactose malabsorption	SLC5A1	182380	1.061,00 €	445,00 €	1.356,00 €			x
GLUT1 deficiency syndrome type 1	SLC2A1	138140	819,00 €	328,00 €	997,00 €			x
Glutamate formiminotransferase deficiency	FTCD	606806	862,00 €					
Glutamine deficiency, congenital	GLUL	138290	573,00 €					
Glutaric acidemia type 1	GCDH	608801	729,00 €	421,00 €	1.000,00 €			x
Glutaric acidemia type 2C	ETFDH	231675	928,00 €	426,00 €	1.204,00 €			x
Glutaric aciduria type 3	SUGCT	609187	1.061,00 €					
Glutathione S-transferase theta-1 deficiency	GSTT1	600436	491,00 €	328,00 €	669,00 €			
Glutathione synthetase deficiency	GSS	601002	862,00 €	426,00 €	1.138,00 €			x
Glycerol kinase deficiency	GK	300474	1.399,00 €	442,00 €	1.691,00 €			x
Glycine encephalopathy	AMT	238310	819,00 €	328,00 €	997,00 €			x
Glycine encephalopathy	AMT	238310	819,00 €	328,00 €	997,00 €			x
Glycine encephalopathy	GCSH	238330	491,00 €	328,00 €	669,00 €			x
Glycine encephalopathy	GLDC	238300	1.643,00 €	328,00 €	1.821,00 €			x
Glycine N-methyltransferase deficiency	GNMT	606628	573,00 €					
Glycogen storage disease of heart (lethal)	PRKAG2	602743	1.127,00 €	424,00 €	1.401,00 €			x
Glycogen storage disease type 0 muscle	GYS1	138570	1.061,00 €	424,00 €	1.335,00 €			x
Glycogen storage disease type 0	GYS2	138571	1.127,00 €	424,00 €	1.401,00 €			x
Glycogen storage disease type 1A	G6PC	613742	655,00 €	390,00 €	895,00 €			x
Glycogen storage disease type 1B	SLC37A4	602671	737,00 €	429,00 €	1.016,00 €			x
Glycogen storage disease type 1C	SLC37A4	602671	737,00 €	429,00 €	1.016,00 €			x
Glycogen storage disease type 2	GAA	606800	1.260,00 €	328,00 €	1.438,00 €			x
Glycogen storage disease type 3	AGL	610860	1.622,00 €	716,00 €	2.188,00 €			x
Glycogen storage disease type 4	GBE1	607839	1.127,00 €	424,00 €	1.401,00 €			x
Glycogen storage disease type 5	PYGM	608455	1.326,00 €	437,00 €	1.613,00 €			x
Glycogen storage disease type 6B	PYGL	613741	1.278,00 €	437,00 €	1.565,00 €			x
Glycogen storage disease type 7	PFKM	610681	1.338,00 €	527,00 €	1.715,00 €			x
Glycogen storage disease type 9A	PHKA2	300798	1.673,00 €	695,00 €	2.218,00 €			x

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Glycogen storage disease type 9B	PHKB	172490	1.622,00 €	653,00 €	2.125,00 €			x
Glycogen storage disease type 9C	PHKG2	172471	796,00 €					x
Glycogen storage disease type 10	PGAM2	612931	468,00 €	374,00 €	692,00 €			x
Glycogen storage disease type 11	LDHA	150000	819,00 €	437,00 €	1.106,00 €			x
Glycogen storage disease type 12	ALDOA	103850	737,00 €					
Glycogen storage disease type 13	ENO3	131370	729,00 €					x
Glycogen storage disease type 14	PGM1	171900	995,00 €	421,00 €	1.266,00 €			
Glycogen storage disease type 15	GYG1	603942	729,00 €					x
Glycoprotein Ia C807T polymorphism	ITGA2	192974	1.278,00 €					
Glycosylation disorder type 2A	MGAT2	602616	468,00 €					
Glycosylation disorder type 2C	SLC35C1	605881	491,00 €					
Glycosylation disorder type 1A	PMM2	601785	819,00 €	437,00 €	1.106,00 €			
Glycosylation disorder type 1B	MPI	154550	737,00 €					x
Glycosylation disorder type 1C	ALG6	604566	995,00 €					
Glycosylation disorder type 1C	ALG6	604566	995,00 €					
Glycosylation disorder type 1D	ALG3	608750	655,00 €					
Glycosylation disorder type 1E	DPM1	603503	655,00 €					x
Glycosylation disorder type 1E	DPM1	603503	655,00 €					x
Glycosylation disorder type 1F	MPDU1	604041	468,00 €					
Glycosylation disorder type 1G	ALG12	607144	819,00 €					
Glycosylation disorder type 1H	ALG8	608103	862,00 €					
Glycosylation disorder type 1I	ALG2	607905	468,00 €					
Glycosylation disorder type 1J	DPAGT1	191350	819,00 €					
Glycosylation disorder type 1J	DPAGT1	191350	819,00 €					
Glycosylation disorder type 1K	ALG1	605907	862,00 €	426,00 €	1.138,00 €			
Glycosylation disorder type 1L	ALG9	606941	928,00 €					
Glycosylation disorder type 1M	DOLK	610746	468,00 €					
Glycosylation disorder type 1M	DOLK	610746	468,00 €					
Glycosylation disorder type 1N	RFT1	611908	928,00 €					
Glycosylation disorder type 1O	DPM3	605951	386,00 €					
Glycosylation disorder type 1S	ALG13	300776	1.764,00 €					
Glycosylation disorder type 1U	DPM2	603564	468,00 €					
Glycosylation disorder type 2A	MGAT2	602616	468,00 €					
Glycosylation disorder type 2B	MOGS	601336	819,00 €					
Glycosylation disorder type 2C	SLC35C1	605881	491,00 €					
Glycosylation disorder type 2D	B4GALT1	137060	655,00 €					
Glycosylation disorder type 2D	B4GALT1	137060	655,00 €					
Glycosylation disorder type 2E	COG7	606978	1.193,00 €					x
Glycosylation disorder type 2E	COG7	606978	1.193,00 €					x
Glycosylation disorder type 2F	SLC35A1	605634	655,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Glycosylation disorder type 2F	SLC35A1	605634	655,00 €					
Glycosylation disorder type 2G	COG1	606973	995,00 €	437,00 €	1.282,00 €			
Glycosylation disorder type 2G	COG1	606973	995,00 €	437,00 €	1.282,00 €			
Glycosylation disorder type 2H	COG8	606979	655,00 €					
Glycosylation disorder type 2H	COG8	606979	655,00 €					
Glycosylation disorder type 2I	COG5	606821	1.704,00 €					
Glycosylation disorder type 2J	COG4	606976	1.460,00 €					
Glycosylation disorder type 2K	TMEM165	614726	655,00 €	421,00 €	926,00 €			x
Glycosylation disorder type 2M	SLC35A2	314375	819,00 €					
Glycosylation disorder type 3	COG6	606977	1.460,00 €	430,00 €	1.740,00 €			x
Glycosylation disorder type IR	DDOST	602202	729,00 €					
Glycosylation disorder x-linked	SSR4	300090	655,00 €					
GM1-gangliosidosis	GLB1	611458	1.127,00 €	424,00 €	1.401,00 €			x
GM1-gangliosidosis type 1	GLB1	611458	1.127,00 €	424,00 €	1.401,00 €			x
GM1-gangliosidosis type 2	GLB1	611458	1.127,00 €	424,00 €	1.401,00 €			x
GM2-gangliosidosis type 2	HEXB	606873	995,00 €	437,00 €	1.282,00 €			x
Gnathodiaphyseal dysplasia	ANO5	608662	1.338,00 €	328,00 €	1.516,00 €			x
Goitre, multinodular	KEAP1	606016	655,00 €					
Goldberg-Shprintzen megacolon syndrome	KIF1BP	609367	729,00 €					
Gracile bone dysplasia	FAM111A	615292	655,00 €					
GRACILE syndrome	BCS1L	603647	819,00 €	421,00 €	1.090,00 €			x
Granulomatous disease, chronic, autosomal recessive, cytochrome b- positive, type 1	NCF1	608512	862,00 €	429,00 €	1.141,00 €			x
Granulomatous disease, chronic, autosomal recessive, cytochrome b- positive, type 1	NCF1	608512	862,00 €	429,00 €	1.141,00 €			x
Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative	CYBA	608508	573,00 €					
Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative	CYBA	608508	573,00 €					
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 2	NCF2	608515	995,00 €					
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 2	NCF2	608515	995,00 €					
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 3	NCF4	601488	729,00 €					
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 3	NCF4	601488	729,00 €					
Granulomatous disease, chronic, X-linked	CYBB	300481	1.127,00 €	426,00 €	1.403,00 €			x
Granulomatous disease, chronic, X-linked	CYBB	300481	1.127,00 €	426,00 €	1.403,00 €			x
Greenberg skeletal dysplasia	LBR	600024	995,00 €					
Greenberg skeletal dysplasia	LBR	600024	995,00 €					
Greig cephalopolysyndactyly syndrome	GLI3	165240	1.278,00 €	328,00 €	1.456,00 €			x
Griselli syndrome type 1	MYO5A	160777	2.262,00 €	704,00 €	2.816,00 €			x
Griselli syndrome type 2	RAB27A	603868	491,00 €	437,00 €	778,00 €			x
Griselli syndrome type 3	MLPH	606526	1.061,00 €					x
Growth hormone deficiency	GH1	139250	491,00 €	328,00 €	669,00 €			x
Growth hormone deficiency	GHRHR	139191	862,00 €	328,00 €	1.040,00 €			x
Growth hormone insensitivity with immunodeficiency	STAT5B	604260	1.260,00 €	328,00 €	1.438,00 €			



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Growth hormone insensitivity, partial	GHR	600946	1.061,00 €	328,00 €	1.239,00 €			x
Growth retardation with deafness and mental retardation due to IGF1 deficiency	IGF1	147440	819,00 €	328,00 €	997,00 €			x
Guanidinoacetate methyltransferase deficiency	GAMT	601240	491,00 €	390,00 €	731,00 €			x
Guttmacher syndrome	HOXA13	142959	491,00 €					
Guttmacher syndrome	HOXA13	142959	491,00 €					
Gyrate atrophy of choroid and retina with or without ornithinemia	OAT	613349	796,00 €	429,00 €	1.075,00 €			x
Haim-Munk syndrome	CTSC	602365	796,00 €	437,00 €	1.083,00 €			x
Hamamy syndrome	IRX5	606195	491,00 €					
Hand-foot-uterus syndrome	HOXA13	142959	491,00 €					
Hand-foot-uterus syndrome	HOXA13	142959	491,00 €					
Hartnup disorder	SLC6A19	608893	796,00 €	421,00 €	1.067,00 €			x
Hartsfield syndrome	FGFR1	136350	1.193,00 €	328,00 €	1.371,00 €			x
Hawkinsinuria	HPD	609695	729,00 €					
HDL deficiency, type 2	ABCA1	600046	2.028,00 €					x
Hearing loss, MAP1A related	MAP1A	600178	1.521,00 €					
Hearing loss, MYH7B related	MYH7B	609928	1.775,00 €					
Heart block, progressive, familial, type 1A	SCN5A	600163	1.673,00 €	328,00 €	1.851,00 €			x
Heart-hand syndrome, Slovenian type	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Heart-hand syndrome, Slovenian type	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Heimler syndrome type 1	PEX1	602136	1.643,00 €	505,00 €	1.998,00 €			x
Heimler syndrome type 1	PEX1	602136	1.643,00 €	505,00 €	1.998,00 €			x
Helsmoortel-van der Aa syndrome	ADNP	611386	729,00 €					
Hemangioblastoma, cerebellar, somatic	VHL	608537					1.900,00 €	
Hemangioma capillary infantile	ANTXR1	606410	1.399,00 €	421,00 €	1.670,00 €			
Hemangioma, capillary infantile, familial, susceptibility to	KDR	191306	1.572,00 €					
Hemochromatosis classical	HFE	613609	573,00 €	328,00 €	751,00 €			x
Hemochromatosis type 2A	HFE2	608374	491,00 €	328,00 €	669,00 €			x
Hemochromatosis type 2B	HAMP	606464	374,00 €	328,00 €	552,00 €			x
Hemochromatosis type 3	TFR2	604720	1.061,00 €	328,00 €	1.239,00 €			x
Hemochromatosis type 4	SLC40A1	604653	819,00 €	328,00 €	997,00 €			x
Hemolytic anemia due to G6PD deficiency	G6PD	305900	862,00 €	426,00 €	1.138,00 €			x
Hemolytic anemia due to G6PD deficiency	G6PD	305900	862,00 €	426,00 €	1.138,00 €			x
Hemolytic anemia due to triosephosphate isomerase deficiency	TPI1	190450	491,00 €					
Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	CD59	107271	374,00 €					
Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	CD59	107271	374,00 €					
Hemolytic anemia, Kell-system related	KEL	613883	1.260,00 €					
Hemolytic uremic syndrome	CFB	138470	1.260,00 €					
Hemolytic uremic syndrome	CFH	134370	1.399,00 €	328,00 €	1.577,00 €			x
Hemolytic uremic syndrome	CFHR1	134371	819,00 €	328,00 €	997,00 €			x
Hemolytic uremic syndrome	CFHR2	600889	491,00 €	328,00 €	669,00 €			x

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Hemolytic uremic syndrome	CFHR3	605336	573,00 €	328,00 €	751,00 €			x
Hemolytic uremic syndrome	CFHR4	605337	729,00 €					
Hemolytic uremic syndrome	CFHR5	608593	729,00 €	328,00 €	907,00 €			
Hemolytic uremic syndrome	CFI	217030	928,00 €	328,00 €	1.106,00 €			x
Hemolytic uremic syndrome	THBD	188040	573,00 €					x
Hemolytic uremic syndrome, atypical type 2, susceptibility to	CD46	120920	928,00 €	328,00 €	1.106,00 €			x
Hemophagocytic lymphohistiocytosis type 2	PRF1	170280	491,00 €	328,00 €	669,00 €			x
Hemophagocytic lymphohistiocytosis type 3	UNC13D	608897	1.278,00 €	328,00 €	1.456,00 €			x
Hemophagocytic lymphohistiocytosis type 4	STX11	605014	374,00 €	328,00 €	552,00 €			x
Hemophagocytic lymphohistiocytosis type 5	STXBP2	601717	995,00 €					
Hemophilia A	F8	300841	1.845,00 €	328,00 €	2.023,00 €			x
Hemophilia B	F9	300746	729,00 €	328,00 €	907,00 €			x
Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	JAM3	606871	737,00 €					
Hennekam lymphangiectasia-lymphedema syndrome type 1	CCBE1	612753	796,00 €					
Hennekam lymphangiectasia-lymphedema syndrome type 2	FAT4	612411	1.802,00 €					
Hepatic failure, early onset, and neurologic disorder	SCO1	603644	737,00 €					
Hepatic venoocclusive disease with immunodeficiency	SP110	604457	1.643,00 €					
Hepatic venoocclusive disease with immunodeficiency	SP110	604457	1.643,00 €					
Hepatoblastoma, somatic	APC	611731					1.900,00 €	
Hepatocellular carcinoma, somatic	CTNNB1	116806					1.900,00 €	
Hepatocellular carcinoma, somatic	IGF2R	147280					1.900,00 €	
Hepatocellular carcinoma, somatic	TP53	191170					1.900,00 €	
Hereditary breast and ovarian cancer syndrome, RAD50 related	RAD50	604040	1.582,00 €	328,00 €	1.760,00 €			
Hereditary motor and sensory neuropathy, Okinawa type	TFG	602498	655,00 €					
Hereditary myopathy with early respiratory failure	TTN	188840	2.225,00 €	905,00 €	2.980,00 €			
Hereditary persistence of fetal hemoglobin	HBG2	142250	468,00 €	328,00 €	646,00 €			x
Hereditary Retinoblastoma	RB1	614041	1.673,00 €	328,00 €	1.851,00 €			x
Hermansky Pudlak syndrome type 4	HPS4	606682	1.326,00 €					
Hermansky-Pudlak syndrome type 1	HPS1	604982	1.127,00 €					
Hermansky-Pudlak syndrome type 2	AP3B1	603401	1.825,00 €					
Hermansky-Pudlak syndrome type 3	HPS3	606118	1.260,00 €					
Hermansky-Pudlak syndrome type 5	HPS5	607521	1.582,00 €					
Hermansky-Pudlak syndrome type 6	HPS6	607522	491,00 €					
Hermansky-Pudlak syndrome type 7	DTNBP1	607145	729,00 €					
Hermansky-Pudlak syndrome type 8	BLOC1S3	609762	655,00 €					
Herpes simplex encephalitis type 2, susceptibility to	TLR3	603029	737,00 €					
Heterotaxy, visceral type 1	ZIC3	300265	491,00 €	328,00 €	669,00 €			x
Heterotaxy, visceral type 2	CFC1	605194	573,00 €	328,00 €	751,00 €			
Heterotaxy, visceral type 4	ACVR2B	602730	729,00 €	328,00 €	907,00 €			
Heterotaxy, visceral type 5	NODAL	601265	468,00 €	328,00 €	646,00 €			

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Heterotaxy, visceral type 6	CFAP53	614759	737,00 €					
Heterotaxy, visceral type 7	MMP21	608416	655,00 €					
Heterotaxy, visceral, BCL9L related	BCL9L	609004	1.061,00 €					
Heterotopia, periventricular, ED variant	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x
Heterotopia, periventricular, X-linked dominant	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x
Hippocampal longterm potentiation, RNF39 related	RNF39	607524	573,00 €					
Hirschsprung disease	ECE1	600423	1.278,00 €					
Hirschsprung disease	EDN3	131242	573,00 €	328,00 €	751,00 €			x
Hirschsprung disease	EDNRB	131244	862,00 €	328,00 €	1.040,00 €			x
Hirschsprung disease	KIF1BP	609367	729,00 €					
Hirschsprung disease	NRG1	142445	995,00 €					
Hirschsprung disease	NRTN	602018	468,00 €	328,00 €	646,00 €			
Hirschsprung disease	RET	164761	1.338,00 €	328,00 €	1.516,00 €			x
Hirschsprung disease	ZEB2	605802	862,00 €	328,00 €	1.040,00 €			x
Hirschsprung disease, type 3, susceptibility to	GDNF	600837	491,00 €	328,00 €	669,00 €			x
Histiocytosis-lymphadenopathy plus syndrome	SLC29A3	612373	573,00 €					
Holocarboxylase synthetase deficiency	HLCS	609018	928,00 €	421,00 €	1.199,00 €			x
Holoprosencephaly type 2	SIX3	603714	491,00 €	328,00 €	669,00 €			x
Holoprosencephaly type 3	SHH	600725	796,00 €	328,00 €	974,00 €			x
Holoprosencephaly type 4	TGIF1	602630	468,00 €	328,00 €	646,00 €			
Holoprosencephaly type 5	ZIC2	603073	737,00 €	328,00 €	915,00 €			x
Holoprosencephaly type 11	CDON	608707	1.399,00 €					
Holoprosencephaly-type 9	GLI2	165230	1.326,00 €	328,00 €	1.504,00 €			x
Holt-Oram syndrome	TBX5	601620	729,00 €	328,00 €	907,00 €			x
Homocystinuria	MTHFR	607093	729,00 €	328,00 €	907,00 €			x
Homocystinuria due to cystathionine beta-synthase deficiency	CBS	613381	1.127,00 €	424,00 €	1.401,00 €			x
Homocystinuria-megaloblastic anemia, cbl E type	MTRR	602568	1.127,00 €					
Hoyeraal-Hreidarsson syndrome	DKC1	300126	1.061,00 €	328,00 €	1.239,00 €			x
HSAN1	SPTLC1	605712	1.127,00 €	328,00 €	1.305,00 €			x
HSAN2A	WNK1	605232	1.802,00 €	590,00 €	2.242,00 €			x
HSAN2B	FAM134B	613114	796,00 €	421,00 €	1.067,00 €			x
HSAN3	IKBKAP	603722	1.724,00 €	779,00 €	2.353,00 €			x
HSAN4	NTRK1	191315	1.193,00 €	424,00 €	1.467,00 €			x
HSAN5	NGF	162030	386,00 €	374,00 €	610,00 €			x
HSAN8	PRDM12	616458	573,00 €					
HSN2C	KIF1A	601255	2.016,00 €	841,00 €	2.707,00 €			x
Huntington disease	HTT	613004				200,00 €		
Huntington disease, ZDHHC17 related	ZDHHC17	607799	1.193,00 €					
Huntington disease-like type 1	PRNP	176640	386,00 €	343,00 €	579,00 €	200,00 €		
Huntington disease-like type 2	JPH3	605268				200,00 €		

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Hurler syndrome	IDUA	252800	928,00 €	437,00 €	1.215,00 €			x
Hurler-Scheie syndrome	IDUA	252800	928,00 €	437,00 €	1.215,00 €			x
Hurthle cell thyroid carcinoma, due to germline NDUFA13 mutation	NDUFA13	609435	491,00 €					
Hutchinson-Gilford progeria	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Hyaline fibromatosis syndrome	ANTXR2	608041	1.127,00 €	424,00 €	1.401,00 €			x
Hydatidiform mole	NLRP7	609661	796,00 €	429,00 €	1.075,00 €			
Hydatidiform mole, recurrent, type 2	KHDC3L	611687	374,00 €	374,00 €	598,00 €			
Hydranencephaly with abnormal genitalia/Lissencephaly X-linked 2	ARX	300382	737,00 €	328,00 €	915,00 €			x
Hydranencephaly, Fowler type	FLVCR2	610865	729,00 €					
Hydrocephalus with aqueductal stenosis and congenital intestinal pseudoobstruction	L1CAM	308840	1.582,00 €	611,00 €	2.043,00 €			x
Hydrocephalus, nonsyndromic, autosomal recessive type 1	CCDC88C	611204	1.572,00 €					
Hydrocephalus, nonsyndromic, autosomal recessive type 2	MPDZ	603785	2.059,00 €	905,00 €	2.814,00 €			
Hydrolethals syndrome	HYLS1	610693	374,00 €					
Hyperaldosteronism type 3	KCNJ5	600734	491,00 €					
Hyperbilirubinemia, familial transient neonatal	UGT1A1	191740	737,00 €	390,00 €	977,00 €			x
Hyperbilirubinemia, Rotor type	SLCO1B1	604843	1.061,00 €	445,00 €	1.356,00 €			x
Hyperbilirubinemia, Rotor type	SLCO1B3	605495	995,00 €	424,00 €	1.269,00 €			x
Hypercalcemia infantile type	CYP24A1	126065	796,00 €	421,00 €	1.067,00 €			x
Hypercholanemia	BAAT	602938	468,00 €					
Hypercholanemia	TJP2	607709	1.521,00 €					
Hypercholesterolemia autosomal dominant type 3	PCSK9	607786	862,00 €	421,00 €	1.133,00 €			x
Hypercholesterolemia autosomal recessive	LDLRAP1	605747	729,00 €					
Hypercholesterolemia due to LDL-receptor-disorder autosomal dominant	LDLR	606945	1.326,00 €	328,00 €	1.504,00 €			x
Hypercholesterolemia type B autosomal dominant	APOB	107730	1.989,00 €	611,00 €	2.450,00 €			x
Hyperchylomicronemia type 5	APOA5	606368	737,00 €					
Hyperekplexia	GLRA1	138491	819,00 €	328,00 €	997,00 €			x
Hyperekplexia	GLRB	138492	819,00 €	328,00 €	997,00 €			x
Hyperekplexia	SLC6A5	604159	1.193,00 €	328,00 €	1.371,00 €			x
Hyperekplexia, EIEE8 related	ARHGEF9	300429	796,00 €					
Hypereosinophilic syndrome, idiopathic, resistant to imatinib	PDGFRA	173490					1.900,00 €	
Hyperferritinemia-cataract syndrome	FTL	134790	468,00 €	437,00 €	755,00 €			x
Hyper-IgE recurrent infection syndrome	STAT3	102582	1.260,00 €	328,00 €	1.438,00 €			x
Hyper-IgE recurrent infection syndrome, autosomal recessive	DOCK8	611432	2.102,00 €	655,00 €	2.607,00 €			x
Hyperinsulinaemia, association with, G6PC2 related	G6PC2	608058	655,00 €					
Hyperinsulinemic hypoglycemia type 1	ABCC8	600509	2.028,00 €	328,00 €	2.206,00 €			x
Hyperinsulinemic hypoglycemia type 2	KCNJ11	600937	468,00 €	296,00 €	614,00 €			x
Hyperinsulinemic hypoglycemia type 3	GCK	138079	862,00 €	328,00 €	1.040,00 €			x
Hyperinsulinemic hypoglycemia type 6	GLUD1	138130	862,00 €					x
Hyperinsulinemic hypoglycemia type 7	SLC16A1	600682	655,00 €	390,00 €	895,00 €			x
Hyperinsulinism, UCP2 related	UCP2	601693	491,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Hyperkalemic periodic paralysis	SCN4A	603967	1.764,00 €	328,00 €	1.942,00 €			x
Hyperlipidemia, familial combined, susceptibility to	USF1	191523	737,00 €					
Hyperlipoproteinemia type 1	LPL	609708	729,00 €	328,00 €	907,00 €			x
Hyperlysinemia type 1	AASS	605113	1.326,00 €					
Hypermanganesemia with dystonia, polycythemia and cirrhosis	SLC30A10	611146	573,00 €	437,00 €	860,00 €			x
Hypermethioninemia due to adenosine kinase deficiency	ADK	102750	862,00 €					
Hypermethioninemia due to adenosine kinase deficiency	ADK	102750	862,00 €					
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	AHCY	180960	729,00 €					
Hyperornithinemia- Hyperammonemia - Homocitrullinuria syndrome	SLC25A15	603861	573,00 €	437,00 €	860,00 €			
Hyperoxaluria type 1	AGXT	604285	796,00 €	328,00 €	974,00 €			x
Hyperoxaluria type 2	GRHPR	604296	819,00 €	328,00 €	997,00 €			x
Hyperoxaluria type 3	HOGA1	613597	655,00 €	437,00 €	942,00 €			x
Hyperoxaluria, SLC26A6 related	SLC26A6	610068	1.278,00 €					
Hyperparathyroidism type 1, familial	CDC73	607393	1.127,00 €	328,00 €	1.305,00 €			
Hyperparathyroidism type 2, familial	CDC73	607393	1.127,00 €	328,00 €	1.305,00 €			
Hyperparathyroidism, neonatal severe	CASR	601199	796,00 €	328,00 €	974,00 €			x
Hyperphenylalaninemia, BH4 deficient, type C	QDPR	612676	737,00 €					x
Hyperphenylalaninemia, BH4 deficient, type D	PCBD1	126090	374,00 €					
Hyperphenylalaninemia, BH4-deficient, B	GCH1	600225	737,00 €	328,00 €	915,00 €			x
Hyperphenylalaninemia, BH4-deficient, type A	PTS	612719	819,00 €	421,00 €	1.090,00 €			x
Hyperphosphatasia with mental retardation syndrome type 1	PIGV	610274	573,00 €					
Hyperphosphatasia with mental retardation syndrome type 2	PIGO	614730	928,00 €					
Hyperphosphatasia with mental retardation syndrome type 3	PGAP2	615187	729,00 €					
Hyperphosphatasia with mental retardation syndrome type 4	PGAP3	611801	737,00 €					
Hyperphosphatasia with mental retardation syndrome type 5	PIGW	610275	468,00 €					
Hyperprolinemia type 1	PRODH	606810	1.061,00 €					
Hypertension early onset	NR3C2	600983	928,00 €	421,00 €	1.199,00 €			x
Hypertension, ADD2 related	ADD2	102681	1.127,00 €					
Hypertension, salt-sensitive essential, susceptibility to	CYP3A5	605325	1.061,00 €	328,00 €	1.239,00 €			
Hypertriglyceridemia, susceptibility to	LIPI	609252	796,00 €					
Hypertriglyceridemia, transient infantile	GPD1	138420	655,00 €	437,00 €	942,00 €			
Hypertrophic osteoarthropathy type 1	HPGD	601688	655,00 €					
Hypertrophic osteoarthropathy type 2	SLCO2A1	601460	1.061,00 €					
Hyperuricemic nephropathy, familial juvenile type 1	UMOD	191845	862,00 €	421,00 €	1.133,00 €			x
Hypoaldosteronism congenital due to CMO I deficiency	CYP11B2	124080	737,00 €	421,00 €	1.008,00 €			x
Hypoaldosteronism, congenital, due to CMO II deficiency	CYP11B2	124080	737,00 €	421,00 €	1.008,00 €			x
Hypoalphalipoproteinemia	APOA1	107680	491,00 €	437,00 €	778,00 €			
Hypobetalipoproteinemia type 1	APOB	107730	1.989,00 €	611,00 €	2.450,00 €			x
Hypocalcemia, autosomal dominant 2	GNA11	605573	737,00 €					
Hypocalcemia, autosomal dominant, with Bartter syndrome	CASR	601199	796,00 €	328,00 €	974,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Hypocalciuric hypercalcemia, familial type 3	AP2S1	602242	573,00 €					
Hypocalciuric hypercalcemia, type 1	CASR	601199	796,00 €	328,00 €	974,00 €			x
Hypochondroplasia	FGFR3	134934	995,00 €	421,00 €	1.266,00 €			x
Hypoglycemia of infancy, leucine-sensitive	ABCC8	600509	2.028,00 €	328,00 €	2.206,00 €			x
Hypogonadism, alopecia, Diabetes mellitus, mental retardation and extrapyramidal syndrome	DCAF17	612515	995,00 €	437,00 €	1.282,00 €			x
Hypogonadotropic hypogonadism	KISS1R	604161	737,00 €	328,00 €	915,00 €			x
Hypogonadotropic hypogonadism	KISS1	603286	374,00 €	328,00 €	552,00 €			
Hypogonadotropic hypogonadism	LHB	152780	491,00 €					
Hypogonadotropic hypogonadism	NSMF	608137	995,00 €	424,00 €	1.269,00 €			x
Hypogonadotropic hypogonadism	NSMF	608137	995,00 €	424,00 €	1.269,00 €			x
Hypogonadotropic hypogonadism type 3 with or without anosmia	PROKR2	607123	468,00 €	328,00 €	646,00 €			x
Hypogonadotropic hypogonadism type 6 with or without anosmia	FGF8	600483	468,00 €	421,00 €	739,00 €			x
Hypogonadotropic hypogonadism type 6 with or without anosmia	FGF8	600483	468,00 €	421,00 €	739,00 €			x
Hypogonadotropic hypogonadism type 7 with or without anosmia	GNRHR	138850	573,00 €	328,00 €	751,00 €			
Hypogonadotropic hypogonadism type 10 with or without anosmia	TAC3	162330	737,00 €					
Hypogonadotropic hypogonadism type 11 with or without anosmia	TACR3	162332	573,00 €	390,00 €	813,00 €			x
Hypogonadotropic hypogonadism type 12 with or without anosmia	GNRH1	152760	374,00 €	328,00 €	552,00 €			x
Hypogonadotropic hypogonadism type 15 with or without anosmia	HS6ST1	604846	468,00 €	343,00 €	661,00 €			x
Hypogonadotropic hypogonadism type 14	WDR11	606417	1.764,00 €	611,00 €	2.225,00 €			x
Hypoinsulinemic hypoglycemia with hemihypertrophy	AKT2	164731	796,00 €					
Hypokalemic periodic paralysis type 1	CACNA1S	114208	1.888,00 €	328,00 €	2.066,00 €			x
Hypomagnesemia type 1	TRPM6	607009	1.888,00 €	821,00 €	2.559,00 €			x
Hypomagnesemia type 2	FXD2	601814	374,00 €					
Hypomagnesemia type 3	CLDN16	603959	491,00 €	390,00 €	731,00 €			x
Hypomagnesemia type 4	EGF	131530	1.521,00 €	505,00 €	1.876,00 €			x
Hypomagnesemia type 5	CLDN19	610036	491,00 €					
Hypomagnesemia type 6	CNNM2	607803	928,00 €	437,00 €	1.215,00 €			x
Hypomyelination with brainstem and spinal cord involvement and leg spasticity	DARS	603084	1.061,00 €					
Hypoparathyroidism	PTH	168450	374,00 €					
Hypoparathyroidism, familial isolated	GCM2	603716	737,00 €	390,00 €	977,00 €			x
Hypoparathyroidism, sensorineural deafness, and renal dysplasia	GATA3	131320	655,00 €	328,00 €	833,00 €			
Hypoparathyroidism-retardation-dysmorphism syndrome	TBCE	604934	1.193,00 €	421,00 €	1.464,00 €			x
Hypophosphatasia, adult	ALPL	171760	862,00 €	421,00 €	1.133,00 €			x
Hypophosphatasia, childhood	ALPL	171760	862,00 €	421,00 €	1.133,00 €			x
Hypophosphatasia, infantile	ALPL	171760	862,00 €	421,00 €	1.133,00 €			x
Hypophosphatemic rickets	CLCN5	300008	1.061,00 €	445,00 €	1.356,00 €			x
Hypophosphatemic rickets with hypercalciuria	SLC34A3	609826	862,00 €	426,00 €	1.138,00 €			x
Hypophosphatemic rickets with hypercalciuria	SLC34A3	609826	862,00 €	426,00 €	1.138,00 €			x
Hypophosphatemic rickets, autosomal dominant	FGF23	605380	468,00 €	328,00 €	646,00 €			x
Hypophosphatemic rickets, autosomal dominant	FGF23	605380	468,00 €	328,00 €	646,00 €			x

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Hypophosphatemic rickets, autosomal recessive type 1	DMP1	600980	573,00 €					x
Hypophosphatemic rickets, autosomal recessive type 2	ENPP1	173335	1.582,00 €	527,00 €	1.959,00 €			x
Hypophosphatemic rickets, X-linked	PHEX	300550	1.521,00 €	328,00 €	1.699,00 €			x
Hypospadias type 1, X-linked	AR	313700	1.061,00 €	328,00 €	1.239,00 €	200,00 €		x
Hypospadias type 1, X-linked	AR	313700	1.061,00 €	328,00 €	1.239,00 €	200,00 €		x
Hypospadias type 2, X-linked	MAMLD1	300120	1.061,00 €	390,00 €	1.301,00 €			x
Hypospadias type 2, X-linked	MAMLD1	300120	1.061,00 €	390,00 €	1.301,00 €			x
Hypothyroidism congenital nongoitrous type 1	TSHR	603372	1.193,00 €	328,00 €	1.371,00 €			x
Hypothyroidism congenital nongoitrous type 2, familial	PAX8	167415	928,00 €	328,00 €	1.106,00 €			
Hypothyroidism congenital nongoitrous type 4	TSHB	188540	386,00 €					
Hypothyroidism congenital nongoitrous type 6	THRA	190120	819,00 €	429,00 €	1.098,00 €			x
Hypothyroidism, isolated, TRHR related	TRHR	188545	374,00 €					
Hypotonia-cystinuria syndrome	PREPL	609557	1.127,00 €	328,00 €	1.305,00 €			x
Hypotrichosis type 1	APCDD1	607479	655,00 €					
Hypotrichosis type 2	CDSN	602593	573,00 €					x
Hypotrichosis type 3	KRT74	608248	729,00 €					
Hypotrichosis type 4	HR	602302	1.193,00 €	430,00 €	1.473,00 €			
Hypotrichosis type 6	DSG4	607892	1.260,00 €					
Hypotrichosis type 7	LIPH	607365	796,00 €					
Hypotrichosis type 8	LPAR6	609239	468,00 €					
Hypotrichosis type 11	SNRPE	128260	573,00 €					
Hypotrichosis type 12	RPL21	603636	491,00 €					
Hypotrichosis type 13	KRT71	608245	729,00 €					
Hypotrichosis-lymphedema-telangiectasia syndrome	SOX18	601618	468,00 €					
Hypouricemia, renal type 1	SLC22A12	607096	796,00 €					
Hypouricemia, renal type 1	SLC22A12	607096	796,00 €					
Hypouricemia, renal type 2	SLC2A9	606142	928,00 €					
Hypouricemia, renal type 2	SLC2A9	606142	928,00 €					
Ichthyosiform erythroderma, congenital, nonbullous type 1	ALOXE3	607206	995,00 €	424,00 €	1.269,00 €			
Ichthyosiform erythroderma, congenital, nonbullous type 1	NIPAL4	609383	737,00 €					
Ichthyosis congenital, autosomal recessive, PNPLA1 related	PNPLA1	612121	819,00 €					
Ichthyosis congenital, Harlequin fetus type	ABCA12	607800	2.106,00 €	827,00 €	2.783,00 €			x
Ichthyosis follicularis, atricia, and photophobia syndrome	MBTPS2	300294	862,00 €					
Ichthyosis prematurity syndrome	SLC27A4	604194	729,00 €					
Ichthyosis vulgaris	FLG	135940	1.622,00 €	374,00 €	1.846,00 €			x
Ichthyosis, bullous type	KRT2	600194	729,00 €	421,00 €	1.000,00 €			x
Ichthyosis, congenital, autosomal recessive type 1	TGM1	190195	1.127,00 €	445,00 €	1.422,00 €			x
Ichthyosis, congenital, autosomal recessive, type 2	ALOX12B	603741	995,00 €	445,00 €	1.290,00 €			x
Ichthyosis, congenital, autosomal recessive, type 9	CERS3	615276	796,00 €					
Ichthyosis, congenital, autosomal recessive, type 11	ST14	606797	928,00 €					

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Ichthyosis, lamellar type 2	ABCA12	607800	2.106,00 €	827,00 €	2.783,00 €			x
Ichthyosis, lamellar type 3	CYP4F22	611495	928,00 €					
Ichthyosis, lamellar type 4	LIPN	613924	819,00 €	421,00 €	1.090,00 €			x
Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	CLDN1	603718	468,00 €					
Ichthyosis, spastic quadriplegia, and mental retardation	ELOVL4	605512	573,00 €					
Ichthyosis, X-linked	STS	300747	729,00 €	328,00 €	907,00 €			x
IDH1, selective sequencing of exon 4	IDH1	147700						
IDH2, selective sequencing of exon 4	IDH2	147650						
IMAGE syndrome	CDKN1C	600856	374,00 €	328,00 €	552,00 €			x
Immunodeficiency common variable type 1	ICOS	604558	573,00 €	390,00 €	813,00 €			x
Immunodeficiency common variable type 2	TNFRSF13B	604907	573,00 €	390,00 €	813,00 €			
Immunodeficiency common variable type 3	CD19	107265	862,00 €					
Immunodeficiency common variable type 4	TNFRSF13C	606269	374,00 €					
Immunodeficiency common variable type 6	CD81	186845	737,00 €					
Immunodeficiency common variable type 8	LRBA	606453	2.176,00 €	905,00 €	2.931,00 €			
Immunodeficiency common variable type 10	NFKB2	164012	1.338,00 €					
Immunodeficiency due to defect in MAPBP-interacting protein	LAMTOR2	610389	468,00 €					
Immunodeficiency due to purine nucleoside phosphorylase deficiency	PNP	164050	491,00 €					
Immunodeficiency type 2, with hyper-IgM	AICDA	605257	468,00 €					
Immunodeficiency type 3, with hyper-IgM	CD40	109535	819,00 €					
Immunodeficiency type 5, with hyper IgM	UNG	191525	737,00 €					
Immunodeficiency type 8	CORO1A	605000	819,00 €					
Immunodeficiency type 9	ORAI1	610277	386,00 €					
Immunodeficiency type 10	STIM1	605921	928,00 €					
Immunodeficiency type 11	CARD11	607210	1.521,00 €					
Immunodeficiency type 12	MALT1	604860	1.193,00 €					
Immunodeficiency type 14	PIK3CD	602839	1.326,00 €					
Immunodeficiency type 15	IKBKB	603258	1.326,00 €					
Immunodeficiency type 18	CD3E	186830	737,00 €					
Immunodeficiency type 19	CD3D	186790	491,00 €					
Immunodeficiency type 21	GATA2	137295	655,00 €	328,00 €	833,00 €			
Immunodeficiency type 22	LCK	153390	729,00 €					
Immunodeficiency type 24	CTPS1	123860	1.127,00 €					
Immunodeficiency type 25	CD247	186780	737,00 €					
Immunodeficiency type 26, with or without neurologic abnormalities	PRKDC	600899	1.955,00 €	905,00 €	2.710,00 €			x
Immunodeficiency type 32A, mycobacteriosis, autosomal dominant	IRF8	601565	819,00 €					
Immunodeficiency type 32B, monocyte and dendritic cell deficiency, autosomal recessive	IRF8	601565	819,00 €					
Immunodeficiency type 34	CYBB	300481	1.127,00 €	426,00 €	1.403,00 €			x
Immunodeficiency type 35	TYK2	176941	1.460,00 €	527,00 €	1.837,00 €			x
Immunodeficiency type 36	PIK3R1	171833	1.260,00 €					



Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Immunodeficiency type 38	ISG15	147571	386,00 €					
Immunodeficiency with natural killer cell deficiency	MCM4	602638	1.061,00 €					
Immunodeficiency, common variable type 7	CR2	120650	1.326,00 €					
Immunodeficiency, isolated	IKBKG	300248	796,00 €					x
Immunodeficiency, primary, autosomal recessive, IL21R-related	IL21R	605383	729,00 €					
Immunodeficiency, X-linked with hyper-IgM	CD40LG	300386	655,00 €	390,00 €	895,00 €			x
Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia	MAGT1	300715	729,00 €					
Immunodeficiency-centromeric instability-facial anomalies syndrome type 1	DNMT3B	602900	1.326,00 €	484,00 €	1.660,00 €			
Immunodeficiency-centromeric instability-facial anomalies syndrome type 2	ZBTB24	614064	819,00 €					
Immunodeficiency-centromeric instability-facial anomalies syndrome type 2	ZBTB24	614064	819,00 €					
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	FOXP3	300292	928,00 €					
Immunological disorder, PECAM1 related	PECAM1	173445	1.061,00 €					
Immunological disorder, PICALM related	PICALM	603025	1.338,00 €					
Inclusion body myopathy	GNE	603824	928,00 €	421,00 €	1.199,00 €			x
Inclusion body myopathy	MYH2	160740	1.825,00 €					
Incontinentia pigmenti type 2	IKBKG	300248	796,00 €					x
Infantile liver failure syndrome type 2	NBAS	608025	2.145,00 €					
Infantile neuroaxonal dystrophy type 1	PLA2G6	603604	1.127,00 €	328,00 €	1.305,00 €			x
Inflammatory bowel disease type 13	ABCB1	171050	1.572,00 €					
Inflammatory skin and bowel disease, neonatal, type 1	ADAM17	603639	1.260,00 €					
Insensitivity to pain, channelopathy-associated	SCN9A	603415	1.572,00 €	569,00 €	1.991,00 €			x
Insulin-like growth factor resistance	IGF1R	147370	1.460,00 €	328,00 €	1.638,00 €			x
Intellectual disability nonsyndromic, CIC related	CIC	612082	1.704,00 €					
Intellectual disability nonsyndromic, CNKSR2 related	CNKSR2	300724	1.521,00 €					
Intellectual disability, TBR1 related	TBR1	604616	737,00 €					
Interleukin 2 receptor deficiency	IL2RA	147730	796,00 €					
Interleukin 12A deficiency	IL12A	161560	573,00 €					
Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	ITGA3	605025	1.521,00 €					
Interstitial nephritis karyomegalic	FAN1	613534	1.127,00 €					
Intestinal atresia, multiple	TTC7A	609332	1.399,00 €	442,00 €	1.691,00 €			x
Intestinal pseudoobstruction, neuronal	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x
Intrahepatic cholestasis of pregnancy, NR1H4 related	NR1H4	603826	796,00 €					
Intrinsic factor deficiency	GIF	609342	729,00 €					
Invasive pneumococcal disease, recurrent isolated type 1	IRAK4	606883	862,00 €					
Invasive pneumococcal disease, recurrent isolated type 2	IKBKG	300248	796,00 €					x
IRAK4 deficiency	IRAK4	606883	862,00 €					
Iridogoniodysgenesis type 1	FOXC1	601090	491,00 €	328,00 €	669,00 €			x
Iron-refractory iron deficiency anemia	TMPRSS6	609862	1.326,00 €	430,00 €	1.606,00 €			
Isobutyryl-CoA dehydrogenase deficiency	ACAD8	604773	729,00 €					
Isovaleric acidemia	IVD	607036	796,00 €	421,00 €	1.067,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
IVIC syndrome	SALL4	607343	729,00 €	437,00 €	1.016,00 €			x
Jackson-Weiss syndrome	FGFR1	136350	1.193,00 €	328,00 €	1.371,00 €			x
Jackson-Weiss syndrome	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
JAK2, selective sequencing of exons 12, 14 and 16	JAK2	147796						
Jalili syndrome	CNNM4	607805	737,00 €					
Jawad syndrome	RBBP8	604124	1.278,00 €					x
Jensen syndrome	TIMM8A	300356	374,00 €	343,00 €	567,00 €			x
Jervell and Lange-Nielsen syndrome type 1	KCNQ1	607542	1.061,00 €	328,00 €	1.239,00 €			x
Jervell and Lange-Nielsen syndrome type 2	KCNE1	176261	296,00 €	328,00 €	474,00 €			x
Johanson Blizzard syndrome	UBR1	605981	2.059,00 €	807,00 €	2.716,00 €			x
Joubert syndrome type 1	INPP5E	613037	862,00 €					x
Joubert syndrome type 2	TMEM216	613277	491,00 €					x
Joubert syndrome type 3	AHI1	608894	1.572,00 €					
Joubert syndrome type 4	NPHP1	607100	1.278,00 €	328,00 €	1.456,00 €			x
Joubert syndrome type 5	CEP290	610142	2.262,00 €	842,00 €	2.954,00 €			x
Joubert syndrome type 6	TMEM67	609884	1.704,00 €					
Joubert syndrome type 7	RPGRIP1L	610937	1.704,00 €					x
Joubert syndrome type 8	ARL13B	608922	796,00 €					
Joubert syndrome type 9	CC2D2A	612013	1.977,00 €	800,00 €	2.627,00 €			x
Joubert syndrome type 10	OFD1	300170	1.521,00 €	484,00 €	1.855,00 €			x
Joubert syndrome type 13	TCTN1	609863	1.193,00 €					
Joubert syndrome type 14	TMEM237	614423	995,00 €					
Joubert syndrome type 15	CEP41	610523	862,00 €					
Joubert syndrome type 16	TMEM138	614459	468,00 €					
Joubert syndrome type 17	C5orf42	614571	2.223,00 €	905,00 €	2.978,00 €			x
Joubert syndrome type 18	TCTN3	613847	928,00 €	437,00 €	1.215,00 €			
Joubert syndrome type 20	TMEM231	614949	729,00 €					
Joubert syndrome type 21	CSPP1	611654	1.825,00 €					
Joubert syndrome type 22	PDE6D	602676	491,00 €					
Joubert syndrome type 23	KIAA0586	610178	1.825,00 €					
Joubert syndrome type 24	TCTN2	613846	1.260,00 €					
Joubert syndrome, EXOC8 related	EXOC8	615283	573,00 €					x
Joubert syndrome, EXOSC8 related	EXOSC8	606019	796,00 €					
Juvenile myelomonocytic leukemia, due to CBL germline mutation	CBL	165360	1.127,00 €	424,00 €	1.401,00 €			x
Juvenile polyposis syndrome	BMPR1A	601299	819,00 €	328,00 €	997,00 €			x
Juvenile polyposis syndrome	SMAD4	600993	729,00 €	328,00 €	907,00 €			x
Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	SMAD4	600993	729,00 €	328,00 €	907,00 €			x
Kabuki syndrome type 1	KMT2D	602113	2.106,00 €	328,00 €	2.284,00 €			x
Kabuki syndrome type 2	KDM6A	300128	1.572,00 €	328,00 €	1.750,00 €			x
Kabuki syndrome type 2	KDM6A	300128	1.572,00 €	328,00 €	1.750,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Kagami-Ogata syndrome	paternal UPD chr. 14			328,00 €				
Kallmann syndrome type 1	ANOS1	300836	995,00 €	328,00 €	1.173,00 €			x
Kallmann syndrome type 2	FGFR1	136350	1.193,00 €	328,00 €	1.371,00 €			x
Kallmann syndrome type 2	FGFR1	136350	1.193,00 €	328,00 €	1.371,00 €			x
Kallmann syndrome type 4	PROK2	607002	468,00 €	328,00 €	646,00 €			x
Kallmann syndrome type 4	PROK2	607002	468,00 €	328,00 €	646,00 €			x
Kallmann syndrome type 5	CHD7	608892	2.028,00 €	328,00 €	2.206,00 €			x
Kallmann syndrome, SEMA3A related	SEMA3A	603961	1.193,00 €	424,00 €	1.467,00 €			x
KBG syndrome	ANKRD11	611192	1.622,00 €	437,00 €	1.909,00 €			x
Kenny-Caffey syndrome type 2	FAM111A	615292	655,00 €					
Keratitis ichthyosis deafness syndrome autosomal dominant	GJB2	121011	374,00 €	328,00 €	552,00 €			x
Keratoconus type 1	VSX1	605020	655,00 €					
Keratoderma, palmoplantar, punctate type 1A	AAGAB	614888	729,00 €					
Keratoderma, palmoplantar, with deafness	GJB2	121011	374,00 €	328,00 €	552,00 €			x
Keratosis follicularis spinulosa declavans, X-linked	MBTPS2	300294	862,00 €					
Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	POMP	613386	573,00 €					
Keratosis palmoplantaris striata type 1	DSG1	125670	1.193,00 €					
Keratosis palmoplantaris striata type 2	DSP	125647	1.927,00 €	328,00 €	2.105,00 €			x
Keutel syndrome	MGP	154870	491,00 €					
Kindler syndrome	FBLIM1	607747	655,00 €					
Kindler syndrome	FERMT1	607900	1.061,00 €					
King-Denborough syndrome	RYR1	180901	1.955,00 €	655,00 €	2.460,00 €			x
KIT, selective sequencing of exons 8, 9, 11, 13 and 17	KIT	164920						
Kleefstra syndrome	EHMT1	607001	1.572,00 €	328,00 €	1.750,00 €			x
Klippel-Feil syndrome type 1, autosomal dominant	GDF6	601147	491,00 €	343,00 €	684,00 €			x
Klippel-Feil syndrome type 2, autosomal dominant	MEOX1	600147	374,00 €	374,00 €	598,00 €			
Klippel-Feil syndrome type 3, autosomal dominant	GDF3	606522	468,00 €	343,00 €	661,00 €			x
Klippel-Feil syndrome type 4, autosomal dominant, with myopathy and facial dysmorphism	MYO18B	607295	2.145,00 €	905,00 €	2.900,00 €			
Kniest dysplasia	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Knobloch syndrome type 1	COL18A1	120328	2.184,00 €					
Knuckle pads and leukonychia sensorineural deafness	GJB2	121011	374,00 €	328,00 €	552,00 €			x
Kohlschütter Tonz syndrome	ROGDI	614574	655,00 €	429,00 €	934,00 €			x
Koolen syndrome	KANSL1	612452	1.127,00 €	328,00 €	1.305,00 €			
Krabbe disease	GALC	606890	1.260,00 €	328,00 €	1.438,00 €			x
Krabbe disease	GALC	606890	1.260,00 €	328,00 €	1.438,00 €			x
Krabbe disease, atypical	PSAP	176801	1.061,00 €	445,00 €	1.356,00 €			x
KRAS somatic Hotspot: c.35G>A, p.G12D	KRAS	190070						
KRAS somatic Hotspot: c.35G>T p.G12V	KRAS	190070						
KRAS somatic Hotspot: c.38G>A, p.G13D	KRAS	190070						
KRAS, selective sequencing of exon 2 and 3	KRAS	190070						

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
L-2-hydroxyglutaric aciduria	L2HGDH	609584	796,00 €	328,00 €	974,00 €			x
Lactase deficiency, congenital	LCT	603202	1.582,00 €					
Lactate dehydrogenase-B deficiency	LDHB	150100	655,00 €					
Lacticacidemia due to PDX1 deficiency	PDHX	608769	796,00 €	429,00 €	1.075,00 €			
Lactose intolerance, adult type	MCM6	601806	1.193,00 €					
LADD syndrome	FGF10	602115	374,00 €	328,00 €	552,00 €			x
LADD syndrome	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Langer-Giedion syndrome	EXT1	608177	928,00 €	328,00 €	1.106,00 €			x
Langer-Giedion syndrome	TRPS1	604386	995,00 €	328,00 €	1.173,00 €			x
Laron syndrome	GHR	600946	1.061,00 €	328,00 €	1.239,00 €			x
Larsen syndrome	FLNB	603381	2.059,00 €	328,00 €	2.237,00 €			x
Laryngoonychocutaneous syndrome	LAMA3	600805	1.955,00 €	905,00 €	2.710,00 €			
LCAD deficiency	ACADL	609576	796,00 €					
LCAT DEFICIENCY	LCAT	606967	655,00 €	421,00 €	926,00 €			x
Leber congenital amaurosis type 1	GUCY2D	600179	1.193,00 €	328,00 €	1.371,00 €			x
Leber congenital amaurosis type 3	SPATA7	609868	928,00 €					
Leber congenital amaurosis type 5	LCA5	611408	729,00 €	328,00 €	907,00 €			x
Leber congenital amaurosis type 6	RPGRIP1	605446	1.582,00 €	328,00 €	1.760,00 €			x
Leber congenital amaurosis type 7	CRX	602225	491,00 €	328,00 €	669,00 €			x
Leber congenital amaurosis type 8	CRB1	604210	1.460,00 €	328,00 €	1.638,00 €			x
Leber congenital amaurosis type 9	NMNAT1	608700	491,00 €	437,00 €	778,00 €			
Leber congenital amaurosis type 10	CEP290	610142	2.262,00 €	842,00 €	2.954,00 €			x
Leber congenital amaurosis type 11	IMPDH1	146690	1.127,00 €	328,00 €	1.305,00 €			
Leber congenital amaurosis type 12	RD3	180040	386,00 €					
Leber congenital amaurosis type 16	KCNJ13	603208	491,00 €					
Leber congenital amaurosis type 17	GDF6	601147	491,00 €	343,00 €	684,00 €			x
Leber congenital amaurosis with myopathy	DTHD1		729,00 €					
Leber optic atrophy	MT-ATP6	516060	296,00 €					x
Leber optic atrophy	MT-CO1	516030	296,00 €					x
Leber optic atrophy	MT-CO3	516050	296,00 €					x
Leber optic atrophy	MT-CYB	516020	296,00 €					x
Leber optic atrophy	MT-ND1	516000	296,00 €					x
Leber optic atrophy	MT-ND2	516001	296,00 €					x
Leber optic atrophy	MT-ND4L	516004	296,00 €					
Leber optic atrophy	MT-ND4	516003	296,00 €					x
Leber optic atrophy	MT-ND5	516005	296,00 €					x
Leber optic atrophy	MT-ND6	516006	296,00 €					x
Left-right axis malformations	LEFTY2	601877	468,00 €					
Legg-Calve-Perthes disease	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Legius syndrome	SPRED1	609291	737,00 €	328,00 €	915,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Leigh syndrome	BCS1L	603647	819,00 €	421,00 €	1.090,00 €			x
Leigh syndrome	COX15	603646	796,00 €	421,00 €	1.067,00 €			x
Leigh syndrome	FOXRED1	613622	796,00 €					
Leigh syndrome	NDUFA2	602137	374,00 €					
Leigh syndrome	NDUFA9	603834	796,00 €					
Leigh syndrome	NDUFA10	603835	729,00 €					
Leigh syndrome	NDUFAF1	606934	374,00 €					
Leigh syndrome	NDUFAF2	609653	573,00 €	437,00 €	860,00 €			
Leigh syndrome	NDUFAF3	612911	468,00 €					
Leigh syndrome	NDUFAF6	612392	819,00 €	421,00 €	1.090,00 €			
Leigh syndrome	NDUFS3	603846	573,00 €					
Leigh syndrome	NDUFS4	602694	491,00 €					x
Leigh syndrome	NDUFS7	601825	737,00 €	437,00 €	1.024,00 €			x
Leigh syndrome	NDUFS8	602141	374,00 €					
Leigh syndrome	NUBPL	613621	796,00 €					
Leigh syndrome	SDHA	600857	1.061,00 €	328,00 €	1.239,00 €			x
Leigh syndrome and mitochondrial encephalopathy	ACAD9	611103	1.260,00 €					x
Leigh syndrome due to COX deficiency	SURF1	185620	729,00 €	421,00 €	1.000,00 €			x
Leigh syndrome due to mitochondrial complex I deficiency	MT-ND3	516002	296,00 €					x
Leigh syndrome due to mitochondrial complex I deficiency	MT-ND5	516005	296,00 €					x
Leigh syndrome due to mitochondrial complex I deficiency	MT-ND6	516006	296,00 €					x
Leigh syndrome due to mitochondrial complex I deficiency	NDUFA12	614530	468,00 €					
Leigh syndrome due to pyruvate and alpha-ketoglutarate dehydrogenase deficiencies, LIPT1 related	LIPT1	610284	468,00 €					
Leigh syndrome due to pyruvate carboxylase deficiency	PC	608786	1.278,00 €	484,00 €	1.612,00 €			x
Leigh syndrome due to the mitochondrial complex IV deficiency	TACO1	612958	655,00 €					
Leigh syndrome, French-Canadian type	LRPPRC	607544	1.977,00 €					
Leigh syndrome, X-linked	PDHA1	300502	862,00 €	328,00 €	1.040,00 €			x
Lenz-Majewski hyperostotic dwarfism	PTDSS1	612792	995,00 €					
LEOPARD syndrome type 3	BRAF	164757	1.260,00 €					x
Lesch-Nyham syndrome	HPRT1	308000	729,00 €	328,00 €	907,00 €			x
Lethal congenital contracture syndrome type 1	GLE1	603371	1.127,00 €					
Lethal congenital contracture syndrome type 4	MYBPC1	160794	1.724,00 €	632,00 €	2.206,00 €			
Leukemia, acute lymphoblastic	IKZF1	603023					1.900,00 €	
Leukemia, acute lymphoblastic, susceptibility to	PAX5	167414					1.900,00 €	
Leukemia, acute lymphoblastic, susceptibility to, due to PAX5 germline mutation	PAX5	167414	729,00 €	328,00 €	907,00 €			
Leukemia, acute myelogenous	JAK2	147796					1.900,00 €	
Leukemia, acute myelogenous	KRAS	190070					950,00 €	
Leukemia, acute myeloid	KIT	164920					1.900,00 €	
Leukemia, acute myeloid	RUNX1	151385					1.900,00 €	
Leukemia, acute myeloid form, susceptible due to TERT germline mutation	TERT	187270	1.260,00 €	328,00 €	1.438,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Leukemia, acute myeloid, somatic	CEBPA	116897					1.900,00 €	
Leukemia, juvenile myelomonocytic	PTPN11	176876					1.900,00 €	
Leukemia, lymphoblastic and myeloid, EZH2 related	EZH2	601573					1.900,00 €	
Leukemia, megakaryoblastic, with or without Down syndrome, somatic	GATA1	305371					1.900,00 €	
Leukemia, myeloid acute form, due to CEBPA germline mutation	CEBPA	116897	573,00 €	328,00 €	751,00 €			x
Leukocyte adhesion deficiency	ITGB1	135630	1.061,00 €					
Leukocyte adhesion deficiency	ITGB2	600065	1.061,00 €					
Leukocyte adhesion deficiency type 3	FERMT3	607901	796,00 €					
Leukocyte adhesion deficiency type 3	FERMT3	607901	796,00 €					
Leukodystrophy demyelinating adult-onset, autosomal dominant	LMNB1	150340	862,00 €	328,00 €	1.040,00 €			x
Leukodystrophy hypomyelinating	GJC2	608803	573,00 €	343,00 €	766,00 €			x
Leukodystrophy hypomyelinating type 3	AIMP1	603605	655,00 €	437,00 €	942,00 €			x
Leukodystrophy hypomyelinating type 4	HSPD1	118190	796,00 €	421,00 €	1.067,00 €			x
Leukodystrophy hypomyelinating type 5	FAM126A	610531	796,00 €	429,00 €	1.075,00 €			x
Leukodystrophy hypomyelinating type 6	TUBB4A	602662	655,00 €	390,00 €	895,00 €			x
Leukodystrophy hypomyelinating type 7	POLR3A	614258	1.572,00 €	653,00 €	2.075,00 €			x
Leukodystrophy hypomyelinating type 8	POLR3B	614366	1.704,00 €	590,00 €	2.144,00 €			x
Leukodystrophy hypomyelinating type 9	RARS	107820	1.061,00 €					
Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation	DARS2	610956	1.193,00 €	424,00 €	1.467,00 €			x
Leukoencephalopathy with dystonia and motor neuropathy	SCP2	184755	1.193,00 €					
Leukoencephalopathy with vanishing white matter	EIF2B1	606686	819,00 €					x
Leukoencephalopathy with vanishing white matter	EIF2B2	606454	737,00 €					
Leukoencephalopathy with vanishing white matter	EIF2B3	606273	796,00 €					x
Leukoencephalopathy with vanishing white matter	EIF2B4	606687	862,00 €					x
Leukoencephalopathy with vanishing white matter	EIF2B5	603945	1.127,00 €					x
Leukoencephalopathy, cystic without megalencephaly	RNASET2	612944	819,00 €	421,00 €	1.090,00 €			x
Leukoencephalopathy, diffuse hereditary, with spheroids	CSF1R	164770	1.193,00 €	463,00 €	1.506,00 €			x
Leukoencephalopathy, progressive, with ovarian failure	AARS2	612035	1.127,00 €					
Lewy body dementia, susceptibility to	GBA	606463	796,00 €	421,00 €	1.067,00 €			x
Leydig cell hypoplasia type 1	LHCGR	152790	1.061,00 €	429,00 €	1.340,00 €			x
Liddle syndrome	SCNN1B	600760	796,00 €	426,00 €	1.072,00 €			x
Liddle syndrome	SCNN1G	600761	796,00 €	426,00 €	1.072,00 €			x
Li-Fraumeni syndrome type 1	TP53	191170	819,00 €	328,00 €	997,00 €			x
Li-Fraumeni syndrome type 2	CHEK2	604373	1.061,00 €	328,00 €	1.239,00 €			
LIG4 syndrome	LIG4	601837	862,00 €					
Limb-girdle muscular dystrophy, autosomal dominant type 1A	MYOT	604103	819,00 €	328,00 €	997,00 €			x
Limb-girdle muscular dystrophy, autosomal dominant type 1B	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Limb-girdle muscular dystrophy, autosomal dominant type 1C	CAV3	601253	386,00 €	328,00 €	564,00 €			x
Limb-girdle muscular dystrophy, autosomal dominant type 1E	DNAJB6	611332	819,00 €					
Limb-girdle muscular dystrophy, autosomal recessive type 2F	SGCD	601411	737,00 €	328,00 €	915,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Limb-girdle muscular dystrophy, autosomal recessive type 2A	CAPN3	114240	1.460,00 €	328,00 €	1.638,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2B	DYSF	603009	2.223,00 €	328,00 €	2.401,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2C	SGCG	608896	655,00 €	328,00 €	833,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2D	SGCA	600119	819,00 €	328,00 €	997,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2E	SGCB	600900	573,00 €	328,00 €	751,00 €			x
Limb-Girdle Muscular Dystrophy, autosomal recessive type 2G	TCAP	604488	386,00 €	343,00 €	579,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2H	TRIM32	602290	819,00 €					x
Limb-girdle muscular dystrophy, autosomal recessive type 2I	FKRP	606596	491,00 €	328,00 €	669,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2J	TTN	188840	2.225,00 €	905,00 €	2.980,00 €			
Limb-girdle muscular dystrophy, autosomal recessive type 2K	POMT1	607423	1.193,00 €	328,00 €	1.371,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2L	ANO5	608662	1.338,00 €	328,00 €	1.516,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2M	FKTN	607440	928,00 €	328,00 €	1.106,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2N	POMT1	607423	1.193,00 €	328,00 €	1.371,00 €			x
Limb-girdle muscular dystrophy, autosomal recessive type 2S	TRAPPC11	614138	1.825,00 €					
Limb-girdle muscular dystrophy, autosomal recessive type 12C	POMK	615247	386,00 €					
Limb-mammary syndrome	TP63	603273	1.061,00 €	424,00 €	1.335,00 €			x
Lipodystrophy generalized type 1	AGPAT2	603100	573,00 €	421,00 €	844,00 €			x
Lipodystrophy generalized type 2	BSCL2	606158	819,00 €	429,00 €	1.098,00 €			x
Lipodystrophy generalized type 4	PTRF	603198	468,00 €	343,00 €	661,00 €			x
Lipodystrophy type 2, familial partial	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Lipodystrophy, familial partial, type 3	PPARG	601487	655,00 €	328,00 €	833,00 €			
Lipoid congenital adrenal hyperplasia	STAR	600617	655,00 €	437,00 €	942,00 €			x
Lipoma, somatic	MEN1	613733					1.900,00 €	
Lipoprotein glomerulopathy	APOE	107741	655,00 €	437,00 €	942,00 €			
Lissencephaly type 1	PAFAH1B1	601545	729,00 €	328,00 €	907,00 €			x
Lissencephaly type 2 (Norman-Roberts type)	RELN	600514	1.955,00 €					x
Lissencephaly type 3	TUBA1A	602529	655,00 €					
Lissencephaly type 4 with microcephaly	NDE1	609449	737,00 €					
Lissencephaly type 5	LAMB1	150240	1.927,00 €	716,00 €	2.493,00 €			
Lissencephaly, X-linked type 1	DCX	300121	819,00 €	328,00 €	997,00 €			x
Lissencephaly, X-linked type 2	ARX	300382	737,00 €	328,00 €	915,00 €			x
Lissencephaly/Subcortical laminal heteropia, X-linked	DCX	300121	819,00 €	328,00 €	997,00 €			x
Liver failure transient infantile	TRMU	610230	796,00 €					x
Loeys-Dietz syndrome type 1A	TGFBR1	190181	819,00 €	328,00 €	997,00 €			x
Loeys-Dietz syndrome type 1B	TGFBR2	190182	862,00 €	328,00 €	1.040,00 €			x
Loeys-Dietz syndrome type 1C	SMAD3	603109	729,00 €					
Loeys-Dietz syndrome type 2A	TGFBR1	190181	819,00 €	328,00 €	997,00 €			x
Loeys-Dietz syndrome type 2B	TGFBR2	190182	862,00 €	328,00 €	1.040,00 €			x
Loeys-Dietz syndrome type 4	TGFB2	190220	737,00 €	437,00 €	1.024,00 €			x
Long QT syndrome type 1	KCNQ1	607542	1.061,00 €	328,00 €	1.239,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Long QT syndrome type 2	KCNH2	152427	1.193,00 €	328,00 €	1.371,00 €			x
Long QT syndrome type 3	SCN5A	600163	1.673,00 €	328,00 €	1.851,00 €			x
Long QT syndrome type 4	ANK2	106410	2.176,00 €	772,00 €	2.798,00 €			x
Long QT syndrome type 5	KCNE1	176261	296,00 €	328,00 €	474,00 €			x
Long QT syndrome type 6	KCNE2	603796	386,00 €	328,00 €	564,00 €			x
Long QT syndrome type 8	CACNA1C	114205	2.106,00 €	905,00 €	2.861,00 €			
Long QT syndrome type 9	CAV3	601253	386,00 €	328,00 €	564,00 €			x
Long QT syndrome type 10	SCN4B	608256	491,00 €					
Long QT syndrome type 11	AKAP9	604001	2.141,00 €	905,00 €	2.896,00 €			
Long QT syndrome type 12	SNTA1	601017	737,00 €					
Long QT syndrome type 13	KCNJ5	600734	491,00 €					
Long QT syndrome type 15	CALM2	114182	655,00 €	437,00 €	942,00 €			x
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA	600890	1.260,00 €	437,00 €	1.547,00 €			x
Lowe oculocerebrorenal syndrome	OCRL	300535	1.460,00 €	505,00 €	1.815,00 €			x
Lowe oculocerebrorenal syndrome	OCRL	300535	1.460,00 €	505,00 €	1.815,00 €			x
Lujan-Fryns syndrome	MED12	300188	1.888,00 €	772,00 €	2.510,00 €			x
Lung alpha-beta hydrolase deficiency type 1	ABHD1	612195	819,00 €					x
Lung cancer, somatic	KRAS	190070					950,00 €	
Lung cancer, SOX2 related, somatic	SOX2	184429					1.900,00 €	
Lutheran inhibitor blood group	KLF1	600599	468,00 €	374,00 €	692,00 €			x
Lymphangioliomyomatosis, somatic	TSC2	191092					1.900,00 €	
Lymphedema, hereditary, type 1A	FLT4	136352	1.582,00 €					
Lymphedema, hereditary, type IC	GJC2	608803	573,00 €	343,00 €	766,00 €			x
Lymphedema-distichiasis syndrome	FOXC2	602402	468,00 €	328,00 €	646,00 €			x
Lymphoma, B-cell type	BCL6	109565					1.900,00 €	
Lymphoma, follicular, somatic	BCL10	603517					1.900,00 €	
Lymphoma, MALT, somatic	BCL10	603517					1.900,00 €	
Lymphoproliferative syndrome type 1	ITK	186973	1.127,00 €	328,00 €	1.305,00 €			
Lymphoproliferative syndrome type 2	CD27	186711	491,00 €					
Lymphoproliferative syndrome, autoimmune, type 5	CTLA4	123890	374,00 €	437,00 €	661,00 €			x
Lymphoproliferative syndrome, X-linked type 1	SH2D1A	300490	468,00 €	328,00 €	646,00 €			
Lymphoproliferative syndrome, X-linked type 2	XIAP	300079	737,00 €	328,00 €	915,00 €			
Lynch syndrome-like tumors, MLH1 related, somatic	MLH1	120436					1.900,00 €	
LYSINURIC PROTEIN INTOLERANCE	SLC7A7	603593	796,00 €	429,00 €	1.075,00 €			x
Lysosomal acid phosphatase deficiency	ACP2	171650	862,00 €					
Macrocephaly, alopecia, cutis laxa, and scoliosis	RIN2	610222	1.127,00 €					
Macroglobulinemia, Waldenstrom, somatic	MYD88	602170					1.900,00 €	
Macular degeneration, age-related type 3	FBLN5	604580	796,00 €					
Macular degeneration, age-related type 6	RAX2	610362	374,00 €					
Macular degeneration, age-related type 8, association with	ARMS2	611313	386,00 €	328,00 €	564,00 €			



Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Macular degeneration, age-related type 11	CST3	604312	374,00 €	437,00 €	661,00 €			x
Macular degeneration, early-onset	FBN2	612570	1.955,00 €	905,00 €	2.710,00 €			
Macular dystrophy retinal type 2	PROM1	604365	1.704,00 €	569,00 €	2.123,00 €			x
Macular dystrophy with central cone involvement	MFSD8	611124	862,00 €	426,00 €	1.138,00 €			x
Macular dystrophy, BEST2-related	BEST2	607335	729,00 €					
Macular dystrophy, BEST3-related	BEST3	607337	928,00 €					
Macular dystrophy, BEST4-related	BEST4	607336	655,00 €					
Macular dystrophy, vitelliform	BEST1	607854	928,00 €	328,00 €	1.106,00 €			x
Macular dystrophy, vitelliform	PRPH2	179605	468,00 €	328,00 €	646,00 €			x
Mainzer Saldino syndrome	IFT140	614620	1.764,00 €					
Majeed syndrome	LPIN2	605519	1.326,00 €					x
Major affective disorder	CUX2	610648	1.521,00 €					
Major histocompatibility complex 1 deficiency	MR1	600764	573,00 €	437,00 €	860,00 €			x
Mal de Meleda	SLURP1	606119	374,00 €					
Male germ cell tumor, somatic	BCL10	603517					1.900,00 €	
Malignant hyperthermia type 5	CACNA1S	114208	1.888,00 €	328,00 €	2.066,00 €			x
Malonyl-CoA decarboxylase deficiency	MLYCD	606761	655,00 €	328,00 €	833,00 €			x
Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome	POLD1	174761	1.127,00 €					
Mandibuloacral dysplasia	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Mandibuloacral dysplasia with type B lipodystrophy	ZMPSTE24	606480	729,00 €	328,00 €	907,00 €			x
Mandibulofacial dysostosis with microcephaly	EFTUD2	603892	1.764,00 €	590,00 €	2.204,00 €			x
Mandibulofacial dysostosis with microcephaly	EFTUD2	603892	1.764,00 €	590,00 €	2.204,00 €			x
Mannose-binding protein deficiency	MBL2	154545	491,00 €	437,00 €	778,00 €			x
Mannosidosis, beta A, lysosomal-like	MANBAL		386,00 €					
Mannosidosis-alpha	MAN2B1	609458	1.326,00 €	505,00 €	1.681,00 €			x
Mannosidosis-beta	MANBA	609489	1.193,00 €	424,00 €	1.467,00 €			x
Maple syrup urine disease type 1a	BCKDHA	608348	819,00 €	421,00 €	1.090,00 €			x
Maple syrup urine disease type 1b	BCKDHB	248611	729,00 €	429,00 €	1.008,00 €			x
Maple syrup urine disease type 2	DBT	248610	862,00 €	429,00 €	1.141,00 €			x
Maple syrup urine disease type 3	DLD	238331	928,00 €	437,00 €	1.215,00 €			x
Maple syrup urine disease, mild variant	PPM1K	611065	573,00 €					
Marden-Walker syndrome	PIEZO2	613629	2.184,00 €	811,00 €	2.845,00 €			
Marden-Walker syndrome	PIEZO2	613629	2.184,00 €	811,00 €	2.845,00 €			
Marfan lipodystrophy syndrome	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			x
Marfan syndrome	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			
Marfan syndrome	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			
Marfan syndrome, TGFBR1 related	TGFBR1	190181	819,00 €	328,00 €	997,00 €			x
Marfan syndrome, TGFBR2 related	TGFBR2	190182	862,00 €	328,00 €	1.040,00 €			x
Marshall syndrome	COL11A1	120280	1.955,00 €	655,00 €	2.460,00 €			x
Marshall syndrome	COL11A1	120280	1.955,00 €	655,00 €	2.460,00 €			x

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Martsolf syndrome	RAB3GAP2	609275	1.775,00 €	905,00 €	2.530,00 €			
MASA syndrome	L1CAM	308840	1.582,00 €	611,00 €	2.043,00 €			x
MASS syndrome	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			
MASS syndrome	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			
Maturity-onset diabetes of the young type 1	HNF4A	600281	1.193,00 €	328,00 €	1.371,00 €			x
Maturity-onset diabetes of the young type 2	GCK	138079	862,00 €	328,00 €	1.040,00 €			x
Maturity-onset diabetes of the young type 3	HNF1A	142410	729,00 €	328,00 €	907,00 €			x
Maturity-onset diabetes of the young type 4	PDX1	600733	468,00 €	328,00 €	646,00 €			x
Maturity-onset diabetes of the young type 5	HNF1B	189907	819,00 €	328,00 €	997,00 €			x
Maturity-onset diabetes of the young type 6	NEUROD1	601724	491,00 €	328,00 €	669,00 €			x
Maturity-onset diabetes of the young type 7	KLF11	603301	573,00 €	328,00 €	751,00 €			x
Maturity-onset diabetes of the young type 8	CEL	114840	862,00 €	328,00 €	1.040,00 €			
Maturity-onset diabetes of the young type 9	PAX4	167413	729,00 €	328,00 €	907,00 €			x
Maturity-onset diabetes of the young type 10	INS	176730	491,00 €	343,00 €	684,00 €			x
Maturity-onset diabetes of the young type 11	BLK	191305	862,00 €					
Maturity-onset diabetes of the young, NKX2-2 related	NKX2-2	604612	491,00 €					
Maturity-onset diabetes of the young, RFX6 related	RFX6	612659	1.278,00 €	430,00 €	1.558,00 €			x
Maturity-onset diabetes of the young, ZFP57 related	ZFP57	612192	655,00 €	328,00 €	833,00 €			x
McKusick-Kaufman syndrome	MKKS	604896	655,00 €	421,00 €	926,00 €			x
McKusick-Kaufman syndrome	MKKS	604896	655,00 €	421,00 €	926,00 €			x
McKusick-Kaufman syndrome	MKKS	604896	655,00 €	421,00 €	926,00 €			x
McLeod syndrome with or without chronic granulomatous disease	XK	314850	491,00 €	374,00 €	715,00 €			x
McLeod syndrome with or without chronic granulomatous disease	XK	314850	491,00 €	374,00 €	715,00 €			x
Meckel syndrome type 1	MKS1	609883	1.127,00 €	424,00 €	1.401,00 €			
Meckel syndrome type 3	TMEM67	609884	1.704,00 €					
Meckel syndrome type 4	CEP290	610142	2.262,00 €	842,00 €	2.954,00 €			x
Meckel syndrome type 8	TCTN2	613846	1.260,00 €					
Meckel syndrome type 9	B9D1	614144	796,00 €					
Meckel syndrome type 10	B9D2	611951	468,00 €					
Meconium ileus	GUCY2C	601330	1.704,00 €					
Mediterranean fever	MEFV	608107	862,00 €	328,00 €	1.040,00 €			x
MEDNIK syndrome	AP1S1	603531	491,00 €					
Medullary cystic kidney disease type 2	UMOD	191845	862,00 €	421,00 €	1.133,00 €			x
Medullary thyroid carcinoma, somatic	RET	164761					1.900,00 €	
Medulloblastoma, desmoplastic, familial	SUFU	607035	928,00 €	421,00 €	1.199,00 €			x
Medulloblastoma, due to BRCA2 germline mutation	BRCA2	600185	525,00 €	330,00 €	705,00 €			
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	605908	862,00 €	328,00 €	1.040,00 €			x
Megalencephalic leukoencephalopathy with subcortical cysts 2A	HEPACAM	611642	862,00 €	437,00 €	1.149,00 €			x
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	PIK3R2	603157	995,00 €	424,00 €	1.269,00 €			x
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome type 2	AKT3	611223	995,00 €					

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Megaloblastic anemia type 1, Finnish type	CUBN	602997	1.955,00 €					x
Megaloblastic anemia type 1	AMN	605799	819,00 €	421,00 €	1.090,00 €			x
Megalocornea, X-linked	CHRDL1	300350	796,00 €	421,00 €	1.067,00 €			x
Meier-Gorlin syndrome 4	CDT1	605525	729,00 €					
Meier-Gorlin syndrome type 1	ORC1	601902	1.127,00 €	424,00 €	1.401,00 €			x
Melanocytic nevus syndrome, congenital, somatic	NRAS	164790					950,00 €	
Melanoma and neural system tumor syndrome, familial	CDKN2A	600160	737,00 €	328,00 €	915,00 €			
Melanoma, cutaneous malignant	MC1R	155555	374,00 €	296,00 €	520,00 €			
Melanoma, cutaneous malignant	MITF	156845	1.061,00 €	328,00 €	1.239,00 €			x
Melanoma, cutaneous malignant, familial	CDKN2A	600160	737,00 €	328,00 €	915,00 €			
Melanoma, cutaneous malignant, familial type 6, susceptibility to	XRCC3	600675	729,00 €					
Melanoma, cutaneous malignant, familial type 10, susceptibility to	POT1	606478	1.326,00 €					
Melanoma, cutaneous malignant, familial, CDK4 related	CDK4	123829	573,00 €	328,00 €	751,00 €			
Melanoma, malignant, somatic	BRAF	164757					1.290,00 €	
Melanoma, malignant, somatic	STK11	602216					1.900,00 €	
MELAS syndrome	MT-TC	590020	296,00 €					
MELAS syndrome	MT-TF	590070	296,00 €					
MELAS syndrome, MT-TL1 related	MT-TL1	590050	296,00 €					
MELAS syndrome, MT-TL1 related	MT-TL1	590050	296,00 €					
MELAS syndrome, MT-TL1 related	MT-TL1	590050	296,00 €					
MELAS syndrome, MT-TL1 related	MT-TL1	590050	296,00 €					
Melnick-Needles syndrome	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x
Meningioma, familial, PDGFB related	PDGFB	190040	655,00 €	437,00 €	942,00 €			x
Meningioma, familial, susceptibility to	SMARCE1	603111	729,00 €	429,00 €	1.008,00 €			
Meningioma, familial, susceptibility to	SUFU	607035	928,00 €	421,00 €	1.199,00 €			x
Meningioma, MN1 deficiency related	MN1	156100	862,00 €					
Meningioma, NF2-related, somatic	NF2	607379					1.900,00 €	
Menkes disease	ATP7A	300011	1.704,00 €	328,00 €	1.882,00 €			x
Mental retardation and distinctive facial features with or without cardiac defects	MED13L	608771	1.775,00 €					
Mental retardation and microcephaly with pontine and cerebellar hypoplasia	CASK	300172	1.704,00 €	328,00 €	1.882,00 €			x
Mental retardation non-syndromic	ELK1	311040	737,00 €					
Mental retardation non-syndromic	KLF8	300286	655,00 €					
Mental retardation non-syndromic	NXF5	300319	1.260,00 €					
Mental retardation non-syndromic	ZCCHC12	300701	468,00 €					
Mental retardation with Cerebellar ataxia and dysequilibrium syndrome type 2	WDR81	614218	1.338,00 €					
Mental retardation with hypotonic facies syndrome, X-linked	ATRX	300032	1.845,00 €	328,00 €	2.023,00 €			x
Mental retardation with language impairment and autistic features	FOXP1	605515	1.278,00 €					
Mental retardation X-linked, SMARCA1 related	SMARCA1	300012	1.643,00 €					
Mental retardation X-linked, syndromic, Claes-Jensen type	KDM5C	314690	1.582,00 €	548,00 €	1.980,00 €			
Mental retardation X-linked, syndromic, Lubs type	MECP2	300005	468,00 €	328,00 €	646,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Mental retardation, autosomal dominant type 1	MBD5	611472	1.127,00 €					
Mental retardation, autosomal dominant type 2	DOCK8	611432	2.102,00 €	655,00 €	2.607,00 €			x
Mental retardation, autosomal dominant type 5	SYNGAP1	603384	1.278,00 €					
Mental retardation, autosomal dominant type 6	GRIN2B	138252	1.278,00 €	328,00 €	1.456,00 €			x
Mental retardation, autosomal dominant type 7	DYRK1A	600855	796,00 €					
Mental retardation, autosomal dominant type 8	GRIN1	138249	1.278,00 €					
Mental retardation, autosomal dominant type 9	KIF1A	601255	2.016,00 €	841,00 €	2.707,00 €			x
Mental retardation, autosomal dominant type 12	ARID1B	614556	1.460,00 €	328,00 €	1.638,00 €			
Mental retardation, autosomal dominant type 13	DYNC1H1	600112	1.955,00 €					
Mental retardation, autosomal dominant type 13	DYNC1H1	600112	1.955,00 €					
Mental retardation, autosomal dominant type 13	TRAPPC9	611966	1.521,00 €					
Mental retardation, autosomal dominant type 14	ARID1A	603024	1.460,00 €	328,00 €	1.638,00 €			
Mental retardation, autosomal dominant type 15	SMARCB1	601607	819,00 €	328,00 €	997,00 €			x
Mental retardation, autosomal dominant type 16	SMARCA4	603254	1.876,00 €	758,00 €	2.484,00 €			
Mental retardation, autosomal dominant type 17	PACS1	607492	1.278,00 €					
Mental retardation, autosomal dominant type 18	GATAD2B	614998	729,00 €					
Mental retardation, autosomal dominant type 19	CTNNB1	116806	995,00 €	445,00 €	1.290,00 €			x
Mental retardation, autosomal dominant type 20	MEF2C	600662	862,00 €	328,00 €	1.040,00 €			x
Mental retardation, autosomal dominant type 23	SETD5	615743	1.521,00 €					
Mental retardation, autosomal dominant type 24	DEAF1	602635	928,00 €					
Mental retardation, autosomal dominant type 25	AHDC1	615790	1.260,00 €					
Mental retardation, autosomal dominant type 27	SOX11	600898	374,00 €					
Mental retardation, autosomal dominant type 28	ADNP	611386	729,00 €					
Mental retardation, autosomal dominant type 31	PURA	600473	386,00 €					
Mental retardation, autosomal dominant type 32	KAT6A	601408	1.399,00 €					
Mental retardation, autosomal dominant type 37	POGZ	614787	1.521,00 €					
Mental retardation, autosomal dominant type 38	EEF1A2	602959	737,00 €					
Mental retardation, autosomal recessive type 2	CRBN	609262	796,00 €					
Mental retardation, autosomal recessive type 3	CC2D1A	610055	1.673,00 €					
Mental retardation, autosomal recessive type 5	NSUN2	610916	1.326,00 €	430,00 €	1.606,00 €			x
Mental retardation, autosomal recessive type 7	TUSC3	601385	796,00 €	429,00 €	1.075,00 €			
Mental retardation, autosomal recessive type 12	ST3GAL3	606494	862,00 €					
Mental retardation, autosomal recessive type 14	TECR	610057	737,00 €					
Mental retardation, autosomal recessive type 15	MAN1B1	604346	862,00 €					
Mental retardation, autosomal recessive type 18	MED23	605042	1.673,00 €					
Mental retardation, autosomal recessive type 27	LINS1	610350	729,00 €					
Mental retardation, autosomal recessive type 36	ADAT3	615302	374,00 €					
Mental retardation, autosomal recessive type 37	ANK3	600465	1.955,00 €					x
Mental retardation, autosomal recessive type 38	HERC2	605837	1.955,00 €	905,00 €	2.710,00 €			
Mental retardation, autosomal recessive type 39	TTI2	614426	737,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Mental retardation, autosomal recessive type 41	KPTN	615620	862,00 €					
Mental retardation, autosomal recessive type 42	PGAP1	611655	1.704,00 €					
Mental retardation, autosomal recessive type 46	NDST1	600853	1.193,00 €					
Mental retardation, autosomal recessive type 49	GPT2	138210	796,00 €					
Mental retardation, truncal obesity, retinal dystrophy, and micropenis	INPP5E	613037	862,00 €					x
Mental retardation, X-linked	RAB40AL	300405	374,00 €					
Mental retardation, X-linked syndromic, Christianson type	SLC9A6	300231	1.127,00 €					
Mental retardation, X-linked syndromic, Nascimento-type	UBE2A	312180	655,00 €					
Mental retardation, X-linked syndromic, Raymond type	ZDHHC9	300646	819,00 €					
Mental retardation, X-linked syndromic, Turner type	HUWE1	300697	1.955,00 €					x
Mental retardation, X-linked syndromic, Turner type	HUWE1	300697	1.955,00 €					x
Mental retardation, X-linked type 1	IQSEC2	300522	1.338,00 €	445,00 €	1.633,00 €			
Mental retardation, X-linked type 3	HCFC1	300019	1.572,00 €					
Mental retardation, X-linked type 13	MECP2	300005	468,00 €	328,00 €	646,00 €			x
Mental retardation, X-linked type 14	UPF3B	300298	729,00 €	429,00 €	1.008,00 €			x
Mental retardation, X-linked type 15	CUL4B	300304	1.460,00 €					
Mental retardation, X-linked type 16	FGD1	300546	1.193,00 €	328,00 €	1.371,00 €			x
Mental retardation, X-linked type 17	HSD17B10	300256	573,00 €					
Mental retardation, X-linked type 19	RPS6KA3	300075	1.521,00 €	328,00 €	1.699,00 €			x
Mental retardation, X-linked type 21	IL1RAPL1	300206	729,00 €	429,00 €	1.008,00 €			
Mental retardation, X-linked type 29	ARX	300382	737,00 €	328,00 €	915,00 €			x
Mental retardation, X-linked type 30	PAK3	300142	1.193,00 €	328,00 €	1.371,00 €			
Mental retardation, X-linked type 32	CLIC2	300138	655,00 €					
Mental retardation, X-linked type 41	GDI1	300104	796,00 €					
Mental retardation, X-linked type 44	FTSJ1	300499	729,00 €					
Mental retardation, X-linked type 45	ZNF81	314998	819,00 €					
Mental retardation, X-linked type 46	ARHGEF6	300267	1.338,00 €					
Mental retardation, X-linked type 58	TSPAN7	300096	655,00 €	328,00 €	833,00 €			x
Mental retardation, X-linked type 59	AP1S2	300629	491,00 €					x
Mental retardation, X-linked type 63	ACSL4	300157	995,00 €					
Mental retardation, X-linked type 72	RAB39B	300774	374,00 €					
Mental retardation, X-linked type 88, AGTR2 related	AGTR2	300034	491,00 €					
Mental retardation, X-linked type 89	ZNF41	314995	862,00 €					
Mental retardation, X-linked type 90	DLG3	300189	1.399,00 €	430,00 €	1.679,00 €			x
Mental retardation, X-linked type 91	ZDHHC15	300576	862,00 €	421,00 €	1.133,00 €			x
Mental retardation, X-linked type 92	ZNF674	300573	737,00 €					
Mental retardation, X-linked type 93	BRWD3	300553	1.888,00 €	704,00 €	2.442,00 €			x
Mental retardation, X-linked type 94	GRIA3	305915	1.260,00 €					
Mental retardation, X-linked type 95	MAGT1	300715	729,00 €					
Mental retardation, X-linked type 96	SYP	313475	573,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/ duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Mental retardation, X-linked type 97	ZNF711	314990	729,00 €					
Mental retardation, X-linked type 99	USP9X	300072	1.931,00 €	905,00 €	2.686,00 €			
Mental retardation, X-linked type 101	MID2	300204	729,00 €	429,00 €	1.008,00 €			
Mental retardation, X-linked type 102	DDX3X	300160	796,00 €					
Mental retardation, X-linked with epilepsy	ATP6AP2	300556	819,00 €					
Mental retardation, X-linked, associated with fragile site FRAXE	AFF2	300806	1.704,00 €	328,00 €	1.882,00 €			x
Mental retardation, x-linked, EFHC2 related	EFHC2	300817	1.061,00 €					
Mental retardation, X-linked, nonsyndromic	KIAA2022	300524	1.061,00 €					
Mental retardation, X-linked, Siderius type	PHF8	300560	1.704,00 €					
Mental retardation, X-linked, Snyder-Robinson type	SMS	300105	796,00 €					
Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	OPHN1	300127	1.521,00 €	527,00 €	1.898,00 €			x
Mental retardation, X-linked, with isolated growth hormone deficiency	SOX3	313430	468,00 €	296,00 €	614,00 €			x
MERRF syndrome, MT-TK related	MT-TK	590060	296,00 €					
MERRF syndrome, MT-TK related	MT-TK	590060	296,00 €					
MERRF syndrome, MT-TK related	MT-TK	590060	296,00 €					
MERRF syndrome, MT-TK related	MT-TK	590060	296,00 €					
MERRF syndrome, MT-TP related	MT-TP	590075	296,00 €					
MERRF syndrome, MT-TP related	MT-TP	590075	296,00 €					
MERRF syndrome, MT-TP related	MT-TP	590075	296,00 €					
MERRF syndrome, MT-TP related	MT-TP	590075	296,00 €					
MERRF/MELAS overlap syndrome, MT-TS1 related	MT-TS1	590080	296,00 €					
MERRF/MELAS overlap syndrome, MT-TS1 related	MT-TS1	590080	296,00 €					
MERRF/MELAS overlap syndrome, MT-TS2 related	MT-TS2	590085	296,00 €					
MERRF/MELAS overlap syndrome, MT-TS2 related	MT-TS2	590085	296,00 €					
Mesothelioma, somatic	BCL10	603517					1.900,00 €	
Mesothelioma, somatic	WT1	607102					1.900,00 €	
Metachromatic Leukodystrophy	ARSA	607574	819,00 €	437,00 €	1.106,00 €			x
Metachromatic leukodystrophy due to Saposin B deficiency	PSAP	176801	1.061,00 €	445,00 €	1.356,00 €			x
Metaphyseal anadysplasia type 1	MMP13	600108	862,00 €					
Metaphyseal anadysplasia type 2	MMP9	120361	928,00 €					
Metaphyseal chondrodysplasia, Schmid type	COL10A1	120110	655,00 €	374,00 €	879,00 €			x
Metaphyseal chondromatosis with increased urinary excretion of D-2-hydroxyglutarate	IDH1	147700	655,00 €					
Metaphyseal dysplasia without hypotrichosis	RMRP	157660	296,00 €	296,00 €	442,00 €			x
Methemoglobinemia type 1	CYB5R3	613213	819,00 €	421,00 €	1.090,00 €			
Methionine adenosyltransferase deficiency, autosomal recessive	MAT1A	610550	819,00 €					
Methylacetoacetic aciduria	ACAT1	607809	862,00 €	421,00 €	1.133,00 €			x
Methylcobalamin deficiency CblG type	MTR	156570	1.876,00 €					
Methylmalonate semialdehyde dehydrogenase deficiency	ALDH6A1	603178	862,00 €					
Methylmalonic aciduria CblA type	MMAA	607481	655,00 €					
Methylmalonic aciduria CblB type	MMAB	607568	819,00 €					x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Methylmalonic aciduria CblC type	MMACHC	609831	468,00 €	437,00 €	755,00 €			x
Methylmalonic aciduria CblD type	MMADHC	611935	655,00 €	437,00 €	942,00 €			x
Methylmalonic aciduria CblF type	LMBRD1	612625	1.260,00 €					
Methylmalonic aciduria CblJ type	ABCD4	603214	1.278,00 €					x
Methylmalonic aciduria CblR type	CD320	606475	491,00 €	390,00 €	731,00 €			x
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	MUT	609058	995,00 €	426,00 €	1.271,00 €			x
Methylmalonyl-CoA epimerase deficiency	MCEE	608419	491,00 €					
Mevalonic aciduria	MVK	251170	729,00 €	429,00 €	1.008,00 €			x
Microcephalic osteodysplastic primordial dwarfism type 1	RNU4ATAC	601428	296,00 €					
Microcephalic osteodysplastic primordial dwarfism type 2	PCNT	605925	2.145,00 €	807,00 €	2.802,00 €			x
Microcephaly and chorioretinopathy with or without mental retardation	TUBGCP6	610053	1.673,00 €	527,00 €	2.050,00 €			x
Microcephaly with cortical malformations, autosomal recessive type 2	WDR62	613583	1.775,00 €	674,00 €	2.299,00 €			x
Microcephaly with epilepsy and diabetes syndrome	IER3IP1	609382	374,00 €					
Microcephaly with or without chorioretinopathy, Lymphedema, or Mental retardation, MCLMR	KIF11	148760	1.338,00 €					
Microcephaly with simplified gyral pattern and insulin-dependant diabetes	GFM2	606544	1.278,00 €					
Microcephaly, Amish type	SLC25A19	606521	573,00 €					
Microcephaly, AP4M1 related	AP4M1	602296	729,00 €					
Microcephaly, autosomal recessive type 1	MCPH1	607117	1.260,00 €	328,00 €	1.438,00 €			x
Microcephaly, autosomal recessive type 3	CDK5RAP2	608201	1.802,00 €	328,00 €	1.980,00 €			x
Microcephaly, autosomal recessive type 4	CASC5	609173	2.028,00 €					
Microcephaly, autosomal recessive type 5	ASPM	605481	2.067,00 €	328,00 €	2.245,00 €			x
Microcephaly, autosomal recessive type 6	CENPJ	609279	1.643,00 €	328,00 €	1.821,00 €			x
Microcephaly, autosomal recessive type 7	STIL	181590	1.278,00 €					x
Microcephaly, autosomal recessive type 8	CEP135	611423	1.521,00 €	548,00 €	1.919,00 €			x
Microcephaly, autosomal recessive type 9	CEP152	613529	1.764,00 €					
Microcephaly, autosomal recessive type 11	PHC1	602978	1.193,00 €					
Microcephaly, autosomal recessive type 12	CDK6	603368	655,00 €					
Microcephaly, autosomal recessive type 13	CENPE	117143	1.973,00 €					
Microcephaly, CEP63 related	CEP63	614724	1.193,00 €					
Microcephaly, MRE11A related	MRE11A	600814	1.326,00 €	437,00 €	1.613,00 €			x
Microcephaly, MSMO1 related	MSMO1	607545	573,00 €					
Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	QARS	603727	1.278,00 €					
Microcephaly, short stature, and polymicrogyria with seizures	RTTN	610436	2.106,00 €					
Microcephaly, TUBB2B related	TUBB2B	612850	491,00 €					
Microcephaly-capillary malformation syndrome	STAMBIP	606247	729,00 €					
Microphthalmia syndromic type 2	BCOR	300485	1.338,00 €	445,00 €	1.633,00 €			
Microphthalmia syndromic type 3	SOX2	184429	374,00 €	328,00 €	552,00 €			x
Microphthalmia syndromic type 5	OTX2	600037	468,00 €	390,00 €	708,00 €			x
Microphthalmia syndromic type 6	BMP4	112262	655,00 €	328,00 €	833,00 €			x
Microphthalmia syndromic type 6	SIX6	606326	374,00 €					

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Microphthalmia syndromic type 7	HCCS	300056	573,00 €	437,00 €	860,00 €			x
Microphthalmia syndromic type 8	ALDH1A3	600463	928,00 €	426,00 €	1.204,00 €			x
Microphthalmia syndromic type 9	STRA6	610745	1.399,00 €	430,00 €	1.679,00 €			x
Microphthalmia, isolated type 2	VSX2	142993	491,00 €	390,00 €	731,00 €			x
Microphthalmia, isolated type 3	RAX	601881	491,00 €					
Microphthalmia, isolated type 4	GDF6	601147	491,00 €	343,00 €	684,00 €			x
Microphthalmia, isolated type 5	MFRP	606227	995,00 €					
Microphthalmia, isolated type 6	PRSS56	613858	995,00 €					
Microphthalmia, isolated type 9	GDF3	606522	468,00 €	343,00 €	661,00 €			x
Microphthalmia, isolated with coloboma type 3	VSX2	142993	491,00 €	390,00 €	731,00 €			x
Microphthalmia, isolated with coloboma type 6, digenic	GDF3	606522	468,00 €	343,00 €	661,00 €			x
Microphthalmia, isolated with coloboma type 6, digenic	GDF6	601147	491,00 €	343,00 €	684,00 €			x
Microphthalmia, isolated with coloboma type 9	TENM3	610083	1.825,00 €					
Microphthalmia, syndromic type 1	NAA10	300013	737,00 €					
Microphthalmia, syndromic type 11	VAX1	604294	573,00 €					
Microphthalmia, VAX2 related	VAX2	604295	374,00 €					
Microspherophakia and/or megalocornea	LTBP2	602091	1.724,00 €					x
Microvascular complications of diabetes type 1	VEGFA	192240	819,00 €					
Microvascular complications of diabetes type 6, susceptibility to	SOD2	147460	573,00 €					
Miller Dieker lissencephaly syndrome	YWHAE	605066	819,00 €	328,00 €	997,00 €			
Minicore myopathy with external ophthalmoplegia	RYR1	180901	1.955,00 €	655,00 €	2.460,00 €			x
Mirror movements type 1	DCC	120470	1.775,00 €	611,00 €	2.236,00 €			
Mirror movements type 2	RAD51	179617	796,00 €	429,00 €	1.075,00 €			
Mirror movements type 3	DNAL4	610565	374,00 €	437,00 €	661,00 €			
Mismatch repair cancer syndrome	MLH1	120436	1.260,00 €	328,00 €	1.438,00 €			
Mismatch repair cancer syndrome	MSH2	609309	1.193,00 €	328,00 €	1.371,00 €			
Mismatch repair cancer syndrome	MSH6	600678	1.127,00 €	328,00 €	1.305,00 €			
Mismatch repair cancer syndrome	PMS2	600259	995,00 €	328,00 €	1.173,00 €			
Mitchell-Riley syndrome	RFX6	612659	1.278,00 €	430,00 €	1.558,00 €			x
Mitochondrial complex I deficiency	FOXRED1	613622	796,00 €					
Mitochondrial complex I deficiency	MT-ND1	516000	296,00 €					x
Mitochondrial complex I deficiency	MT-ND2	516001	296,00 €					x
Mitochondrial complex I deficiency	MT-ND3	516002	296,00 €					x
Mitochondrial complex I deficiency	MT-ND4L	516004	296,00 €					
Mitochondrial complex I deficiency	MT-ND4	516003	296,00 €					x
Mitochondrial complex I deficiency	MT-ND5	516005	296,00 €					x
Mitochondrial complex I deficiency	MT-ND6	516006	296,00 €					x
Mitochondrial complex I deficiency	NDUFA1	300078	374,00 €					
Mitochondrial complex I deficiency	NDUFA11	612638	573,00 €					
Mitochondrial complex I deficiency	NDUFAF1	606934	374,00 €					



Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Mitochondrial complex I deficiency	NDUFAF3	612911	468,00 €					
Mitochondrial complex I deficiency	NDUFAF4	611776	819,00 €					
Mitochondrial complex I deficiency	NDUFAF5	612360	796,00 €	429,00 €	1.075,00 €			x
Mitochondrial complex I deficiency	NDUFB3	603839	374,00 €					
Mitochondrial complex I deficiency	NDUFB3	603839	374,00 €					
Mitochondrial complex I deficiency	NDUFS1	157655	1.338,00 €	430,00 €	1.618,00 €			x
Mitochondrial complex I deficiency	NDUFS2	602985	862,00 €					
Mitochondrial complex I deficiency	NDUFS4	602694	491,00 €					x
Mitochondrial complex I deficiency	NDUFS6	603848	468,00 €					
Mitochondrial complex I deficiency	NDUFV1	161015	737,00 €					
Mitochondrial complex I deficiency	NDUFV2	600532	737,00 €	437,00 €	1.024,00 €			
Mitochondrial complex I deficiency, MT-TN related	MT-TN	590010	296,00 €					
Mitochondrial complex II deficiency	SDHAF1	612848	374,00 €	328,00 €	552,00 €			
Mitochondrial complex II deficiency	SDHD	602690	468,00 €	328,00 €	646,00 €			x
Mitochondrial complex III deficiency	BCS1L	603647	819,00 €	421,00 €	1.090,00 €			x
Mitochondrial complex III deficiency	UQCRB	191330	819,00 €					
Mitochondrial complex III deficiency	UQCRC2	191329	995,00 €	437,00 €	1.282,00 €			x
Mitochondrial complex III deficiency	UQCRQ	612080	374,00 €					
Mitochondrial complex III deficiency, nuclear type 2	TTC19	613814	796,00 €	429,00 €	1.075,00 €			
Mitochondrial complex III deficiency, nuclear type 7	UQCC2	614461	468,00 €					
Mitochondrial complex III deficiency, nuclear type 7	UQCC2	614461	468,00 €					
Mitochondrial complex III deficiency, nuclear type 7	UQCC2	614461	468,00 €					
Mitochondrial complex IV deficiency	APOPT1	616003	655,00 €					
Mitochondrial complex IV deficiency	COX6B1	124089	468,00 €					
Mitochondrial complex IV deficiency	FASTKD2	612322	1.399,00 €					
Mitochondrial complex IV deficiency	MT-CO3	516050	296,00 €					x
Mitochondrial complex IV deficiency	PET100	614770	374,00 €					
Mitochondrial complex V (ATP synthase) deficiency	MT-ATP6	516060	296,00 €					x
Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	ATPAF2	608918	737,00 €					
Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	TMEM70	612418	491,00 €	374,00 €	715,00 €			x
Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3	ATP5E	606153	386,00 €					
Mitochondrial complex V deficiency, nuclear type 4	ATP5A1	164360	729,00 €					
Mitochondrial Disorders, AKAP1 related	AKAP1	602449	995,00 €	429,00 €	1.274,00 €			x
Mitochondrial DNA depletion syndrome	DGUOK	601465	655,00 €	328,00 €	833,00 €			x
Mitochondrial DNA depletion syndrome	SUCLA2	603921	796,00 €	328,00 €	974,00 €			x
Mitochondrial DNA depletion syndrome	TK2	188250	862,00 €	328,00 €	1.040,00 €			x
Mitochondrial DNA depletion syndrome	TK2	188250	862,00 €	328,00 €	1.040,00 €			x
Mitochondrial DNA depletion syndrome 8B, MNGIE type	RRM2B	604712	729,00 €	328,00 €	907,00 €			x
Mitochondrial DNA depletion syndrome type 4A	POLG	174763	1.521,00 €	328,00 €	1.699,00 €			x
Mitochondrial DNA depletion syndrome type 4B	POLG	174763	1.521,00 €	328,00 €	1.699,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Mitochondrial DNA depletion syndrome type 6	MPV17	137960	491,00 €	328,00 €	669,00 €			x
Mitochondrial DNA depletion syndrome type 7	TWNK	606075	737,00 €	328,00 €	915,00 €			x
Mitochondrial DNA depletion syndrome type 11	MGME1	615076	491,00 €					
Mitochondrial DNA depletion syndrome type 13	FBXL4	605654	819,00 €					
Mitochondrial DNA depletion syndrome, encephalomyopathic type with methylmalonic aciduria	SUCLG1	611224	819,00 €	328,00 €	997,00 €			x
Mitochondrial encephalomyopathy	MFF	614785	491,00 €					
Mitochondrial encephalomyopathy	MT-CYB	516020	296,00 €					x
Mitochondrial modifier of deafness	TRMU	610230	796,00 €					x
Mitochondrial myopathy and sideroblastic anemia type 1	PUS1	608109	573,00 €					
Mitochondrial myopathy and sideroblastic anemia type 1	PUS1	608109	573,00 €					
Mitochondrial myopathy and sideroblastic anemia type 1	PUS1	608109	573,00 €					
Mitochondrial myopathy and sideroblastic anemia type 1	PUS1	608109	573,00 €					
Mitochondrial myopathy, infantile, transient, MT-TE related	MT-TE	590025	296,00 €					
Mitochondrial myopathy, isolated	MT-TD	590015	296,00 €					
Mitochondrial myopathy, MT-TA related	MT-TA	590000	296,00 €					
Mitochondrial myopathy, MT-TM related	MT-TM	590065	296,00 €					
Mitochondrial neurogastrointestinal encephalopathy syndrome without leukoencephalopathy	POLG	174763	1.521,00 €	328,00 €	1.699,00 €			x
Mitochondrial neurogastrointestinal encephalopathy syndrome without leukoencephalopathy	TYMP	131222	819,00 €	429,00 €	1.098,00 €			x
Mitochondrial pyruvate carrier deficiency	MPC1	614738	491,00 €					
Mitochondrial respiratory chain complex II deficiency	SDHA	600857	1.061,00 €	328,00 €	1.239,00 €			x
Mitochondrial respiratory chain disease, TIMM21 related	TIMM21	615180	573,00 €					
Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	ECHS1	602292	737,00 €	437,00 €	1.024,00 €			
Miyoshi muscular dystrophy type 3	ANO5	608662	1.338,00 €	328,00 €	1.516,00 €			x
Miyoshi myopathy	DYSF	603009	2.223,00 €	328,00 €	2.401,00 €			x
MMR genes methylation analysis	MMR genes			328,00 €				
Molybdenum cofactor deficiency type A	MOCS1	603707	729,00 €	429,00 €	1.008,00 €			x
Molybdenum cofactor deficiency type B	MOCS2	603708	737,00 €	437,00 €	1.024,00 €			
Molybdenum cofactor deficiency type C	GPHN	603930	1.460,00 €	484,00 €	1.794,00 €			x
Monocarboxylate transporter 1 deficiency	SLC16A1	600682	655,00 €	390,00 €	895,00 €			x
Mosaic variegated aneuploidy syndrome type 2	CEP57	607951	796,00 €					
Moyamoya disease type 2, susceptibility to	RNF213	613768	1.955,00 €	905,00 €	2.710,00 €			
Moyamoya disease type 5	ACTA2	102620	737,00 €	421,00 €	1.008,00 €			x
Moyamoya type 6 with achalasia	GUCY1A3	139396	796,00 €					
MPL, selective sequencing of exon 10	MPL	159530						
Muckle-wells syndrome	NLRP3	606416	995,00 €	429,00 €	1.274,00 €			x
Mucopolipidosis type 2 alpha/beta	GNPTAB	607840	1.521,00 €	442,00 €	1.813,00 €			x
Mucopolipidosis type 3 gamma	GNPTG	607838	928,00 €	429,00 €	1.207,00 €			
Mucopolipidosis type 3	GNPTAB	607840	1.521,00 €	442,00 €	1.813,00 €			x
Mucopolipidosis type 4	MCOLN1	605248	796,00 €	437,00 €	1.083,00 €			x
Mucopolysaccharidosis type 2	IDS	300823	862,00 €	328,00 €	1.040,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Mucopolysaccharidosis type 3A	SGSH	605270	819,00 €	437,00 €	1.106,00 €			x
Mucopolysaccharidosis type 3B	NAGLU	609701	819,00 €	421,00 €	1.090,00 €			x
Mucopolysaccharidosis type 3C	HGSNAT	610453	1.193,00 €	421,00 €	1.464,00 €			x
Mucopolysaccharidosis type 3D	GNS	607664	995,00 €	437,00 €	1.282,00 €			x
Mucopolysaccharidosis type 4A	GALNS	612222	1.061,00 €	445,00 €	1.356,00 €			x
Mucopolysaccharidosis type 4B	GLB1	611458	1.127,00 €	424,00 €	1.401,00 €			x
Mucopolysaccharidosis type 6	ARSB	611542	729,00 €	437,00 €	1.016,00 €			x
Mucopolysaccharidosis type 7	GUSB	611499	862,00 €	421,00 €	1.133,00 €			x
Mucopolysaccharidosis type 9	HYAL1	607071	737,00 €					
Mucopolysaccharidosis type IH	IDUA	252800	928,00 €	437,00 €	1.215,00 €			x
Muir-Torre syndrome	MLH1	120436	1.260,00 €	328,00 €	1.438,00 €			
Muir-Torre syndrome	MSH2	609309	1.193,00 €	328,00 €	1.371,00 €			
Mulibrey nanism	TRIM37	605073	1.582,00 €	527,00 €	1.959,00 €			x
Multicentric carpotarsal osteolysis syndrome	MAFB	608968	374,00 €					
Multicentric osteolysis, nodulosis, and arthropathy	MMP2	120360	995,00 €	426,00 €	1.271,00 €			x
Multiple congenital anomalies-hypotonia-seizures syndrome type 1	PIGN	606097	1.704,00 €	653,00 €	2.207,00 €			x
Multiple congenital anomalies-hypotonia-seizures syndrome type 3	PIGT	610272	796,00 €					
Multiple endocrine neoplasia type 1, CDKN2B related	CDKN2B	600431	729,00 €	328,00 €	907,00 €			
Multiple endocrine neoplasia type 1	MEN1	613733	729,00 €	328,00 €	907,00 €			x
Multiple endocrine neoplasia type 2A	RET	164761	1.338,00 €	328,00 €	1.516,00 €			x
Multiple endocrine neoplasia type 2B	RET	164761	1.338,00 €	328,00 €	1.516,00 €			x
Multiple endocrine neoplasia type 4	CDKN1B	600778	386,00 €	328,00 €	564,00 €			
Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	B3GAT3	606374	655,00 €					x
Multiple mitochondrial dysfunctions syndrome type 1	NFU1	608100	737,00 €					
Multiple mitochondrial dysfunctions syndrome type 2	BOLA3	613183	468,00 €					
Multiple mitochondrial dysfunctions syndrome type 3	IBA57	615316	573,00 €					
Multiple mitochondrial dysfunctions syndrome type 4	ISCA2	615317	468,00 €					
Multiple pterygium syndrome lethal type	CHRNA1	100690	862,00 €	421,00 €	1.133,00 €			x
Multiple pterygium syndrome lethal type	CHRND	100720	862,00 €	421,00 €	1.133,00 €			x
Multisystemic smooth muscle dysfunction syndrome	ACTA2	102620	737,00 €	421,00 €	1.008,00 €			x
Multisystemic smooth muscle dysfunction syndrome	ACTA2	102620	737,00 €	421,00 €	1.008,00 €			x
Muscle glycogenosis	PHKA1	311870	1.622,00 €	674,00 €	2.146,00 €			
Muscle hypertrophy	MSTN	601788	468,00 €					x
Muscle-eye-brain disease, POMK related	POMK	615247	386,00 €					
Muscular dystrophy type 1A	LAMA2	156225	2.282,00 €	655,00 €	2.787,00 €			x
Muscular dystrophy type 1C	FKRP	606596	491,00 €	328,00 €	669,00 €			x
Muscular dystrophy type 1D	LARGE	603590	995,00 €	328,00 €	1.173,00 €			x
Muscular dystrophy, Becker type	DMD	300377	950,00 €	655,00 €	1.455,00 €			x
Muscular dystrophy, congenital, LMNA related	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Muscular dystrophy, congenital, megalocornia type	CHKB	612395	737,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Muscular dystrophy, Duchenne type	DMD	300377	950,00 €	655,00 €	1.455,00 €			x
Muscular dystrophy, limb-girdle type 2A	CAPN3	114240	1.460,00 €	328,00 €	1.638,00 €			x
Muscular dystrophy, limb-girdle, type 2Q	PLEC	601282	1.955,00 €					x
Muscular dystrophy, oculopharyngeal	PABPN1	602279	655,00 €	437,00 €	942,00 €	200,00 €		
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A2	POMT2	607439	1.278,00 €	328,00 €	1.456,00 €			x
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A3	POMGNT1	606822	1.061,00 €	328,00 €	1.239,00 €			x
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A8	POMGNT2	614828	468,00 €					
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A10	TMEM5	605862	655,00 €					
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A13	B4GAT1	605517	374,00 €					
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B1	POMT1	607423	1.193,00 €	328,00 €	1.371,00 €			x
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B2	POMT2	607439	1.278,00 €	328,00 €	1.456,00 €			x
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B3	POMGNT1	606822	1.061,00 €	328,00 €	1.239,00 €			x
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C2	POMT2	607439	1.278,00 €	328,00 €	1.456,00 €			x
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C3	POMGNT1	606822	1.061,00 €	328,00 €	1.239,00 €			x
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C9	DAG1	128239	819,00 €					
Muscular-skeletal disorder, CAPN1 related	CAPN1	114220	1.338,00 €					
Myasthenia congenital with tubular aggregates 1	GFPT1	138292	1.399,00 €					
Myasthenic syndrome associated with acetylcholine receptor deficiency	MUSK	601296	1.260,00 €	445,00 €	1.555,00 €			x
Myasthenic syndrome due to mutation in SCN4A	SCN4A	603967	1.764,00 €	328,00 €	1.942,00 €			x
Myasthenic syndrome fast channel congenital	CHRNA1	100690	862,00 €	421,00 €	1.133,00 €			x
Myasthenic syndrome slow-channel congenital	CHRNA1	100690	862,00 €	421,00 €	1.133,00 €			x
Myasthenic syndrome type 10, congenital	DOK7	610285	796,00 €	429,00 €	1.075,00 €			x
Myasthenic syndrome type 11, congenital, associated with acetylcholine receptor deficiency	RAPSN	601592	655,00 €	437,00 €	942,00 €			x
Myasthenic syndrome, congenital	AGRN	103320	2.028,00 €	821,00 €	2.699,00 €			
Myasthenic syndrome, congenital	CHAT	118490	1.061,00 €	445,00 €	1.356,00 €			x
Myasthenic syndrome, congenital	CHRN1	100710	796,00 €	429,00 €	1.075,00 €			x
Myasthenic syndrome, congenital	CHRNE	100725	729,00 €	421,00 €	1.000,00 €			x
Myasthenic syndrome, congenital, type 3C, associated with acetylcholine receptor deficiency	CHRND	100720	862,00 €	421,00 €	1.133,00 €			x
Myasthenic syndrome, fast-channel, congenital, type 3B	CHRND	100720	862,00 €	421,00 €	1.133,00 €			x
Myasthenic syndrome, slow channel, congenital, type 3A	CHRND	100720	862,00 €	421,00 €	1.133,00 €			x
Mycobacterial infection, atypical, familial disseminated	IFNGR1	107470	819,00 €					
Myelodysplastic syndrome, somatic	TET2	612839					1.900,00 €	
Myelofibrosis, somatic	JAK2	147796					1.900,00 €	
Myeloproliferative disorder, chronic, with eosinophilia	PDGFRB	173410	1.399,00 €	484,00 €	1.733,00 €			x
Myhre syndrome	SMAD4	600993	729,00 €	328,00 €	907,00 €			x
Myoclonic dystonia, DRD2 related	DRD2	126450	737,00 €	437,00 €	1.024,00 €			x
Myoclonic epilepsy of Lafora	EPM2A	607566	468,00 €	328,00 €	646,00 €			x
Myoclonic epilepsy of Lafora	NHLRC1	608072	386,00 €	328,00 €	564,00 €			x
Myoclonus, familial cortical	NOL3	605235	468,00 €					
Myoglobinuria acute recurrent	LPIN1	605518	1.460,00 €	463,00 €	1.773,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Myoglobinuria acute recurrent	LPIN1	605518	1.460,00 €	463,00 €	1.773,00 €			x
Myopathy due to Integrin 7A deficiency	ITGA7	600536	1.704,00 €					
Myopathy due to myoadenylate deaminase deficiency	AMPD1	102770	1.127,00 €	424,00 €	1.401,00 €			x
Myopathy due to myoadenylate deaminase deficiency	AMPD1	102770	1.127,00 €	424,00 €	1.401,00 €			x
Myopathy with fiber-type disproportion	SELENON	606210	995,00 €	426,00 €	1.271,00 €			x
Myopathy with fiber-type disproportion type 1	ACTA1	102610	655,00 €	437,00 €	942,00 €			x
Myopathy with lactic acidosis hereditary	ISCU	611911	655,00 €					
Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	MEGF10	612453	1.582,00 €	548,00 €	1.980,00 €			
Myopathy, centronuclear	BIN1	601248	1.399,00 €					
Myopathy, centronuclear	DNM2	602378	1.399,00 €	442,00 €	1.691,00 €			x
Myopathy, COL6A6 related	COL6A6		1.931,00 €					
Myopathy, desmin related, associated with mutation in the CRYAB gene	CRYAB	123590	468,00 €					x
Myopathy, distal type 1	MYH7	160760	1.759,00 €	328,00 €	1.937,00 €			x
Myopathy, distal type 4	FLNC	102565	1.845,00 €					x
Myopathy, distal with anterior tibial onset	DYSF	603009	2.223,00 €	328,00 €	2.401,00 €			x
Myopathy, distal, Tateyama type	CAV3	601253	386,00 €	328,00 €	564,00 €			x
Myopathy, early-onset with fatal cardiomyopathy	TTN	188840	2.225,00 €	905,00 €	2.980,00 €			
Myopathy, lactic acidosis, and sideroblastic anemia type 2	YARS2	610957	737,00 €					
Myopathy, limb girdle with bone fragility	MTAP	156540	737,00 €	328,00 €	915,00 €			x
Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	GFER	600924	737,00 €					
Myopathy, MT-TQ related	MT-TQ	590030	296,00 €					
Myopathy, MT-TQ related	MT-TQ	590030	296,00 €					
Myopathy, myofibrillar type 6	BAG3	603883	737,00 €	328,00 €	915,00 €			x
Myopathy, myofibrillar, Desmin related	DES	125660	737,00 €	421,00 €	1.008,00 €			x
Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related	CRYAB	123590	468,00 €					x
Myopathy, myofibrillar, ZASP related	LDB3	605906	1.193,00 €					
Myopathy, scapulohumeroperoneal	ACTA1	102610	655,00 €	437,00 €	942,00 €			x
Myopathy, tubular aggregate, type 1	STIM1	605921	928,00 €					
Myopathy, tubular aggregate, type 2	ORAI1	610277	386,00 €					
Myosclerosis, autosomal recessive	COL6A2	120240	1.825,00 €	590,00 €	2.265,00 €			x
Myosin storage myopathy	MYH7	160760	1.759,00 €	328,00 €	1.937,00 €			x
Myotilinopathy	MYOT	604103	819,00 €	328,00 €	997,00 €			x
Myotonia congenita	CLCN1	118425	1.278,00 €	328,00 €	1.456,00 €			x
Myotonic dystrophy type 1	DMPK	605377				200,00 €		x
Myotonic dystrophy type 1	DMPK	605377				200,00 €		x
Myotonic dystrophy type 2	CNBP	116955				200,00 €		x
Myotonic dystrophy type 2	CNBP	116955				200,00 €		x
Myotubular myopathy X-linked	MTM1	300415	1.061,00 €	328,00 €	1.239,00 €			x
Myxoma, intracardiac	PRKAR1A	188830	796,00 €	429,00 €	1.075,00 €			x
N-acetylglutamate synthase deficiency	NAGS	608300	819,00 €	437,00 €	1.106,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Naegeli-Franceschetti-Jadassohn syndrome	KRT14	148066	862,00 €	437,00 €	1.149,00 €			x
Nail-Patella syndrome	LMX1B	602575	655,00 €	328,00 €	833,00 €			x
Nance-Horan syndrome	NHS	300457	1.399,00 €	421,00 €	1.670,00 €			x
Nanophthalmia type 2	MFRP	606227	995,00 €					
Narcolepsy	HCRT	602358	468,00 €					
Nasopharyngeal carcinoma, somatic	TP53	191170					1.900,00 €	
Native American myopathy	STAC3	615521	862,00 €					
Nemaline myopathy type 1	TPM3	191030	928,00 €	429,00 €	1.207,00 €			x
Nemaline myopathy type 2, autosomal recessive	NEB	161650	1.955,00 €					x
Nemaline myopathy type 3	ACTA1	102610	655,00 €	437,00 €	942,00 €			x
Nemaline myopathy type 4	TPM2	190990	819,00 €	421,00 €	1.090,00 €			x
Nemaline myopathy type 5	TNNT1	191041	862,00 €	437,00 €	1.149,00 €			x
Nemaline myopathy type 6	KBTD13	613727	468,00 €					
Nemaline myopathy type 7	CFL2	601443	491,00 €					
Neonatal death due Leigh syndrome, MT-TV related	MT-TV	590105	296,00 €					
Nephrogenic syndrome of inappropriate antidiuresis	AVPR2	300538	491,00 €	374,00 €	715,00 €			x
Nephrolithiasis type 1	CLCN5	300008	1.061,00 €	445,00 €	1.356,00 €			x
Nephrolithiasis/osteoporosis, hypophosphatemic, type 1	SLC34A1	182309	729,00 €					x
Nephrolithiasis/osteoporosis, hypophosphatemic, type 2	SLC9A3R1	604990	655,00 €					
Nephrolithiasis/osteoporosis, hypophosphatemic, type 2	SLC9A3R1	604990	655,00 €					
Nephronophthisis type 1	NPHP1	607100	1.278,00 €	328,00 €	1.456,00 €			x
Nephronophthisis type 2	INVS	243305	1.260,00 €					
Nephronophthisis type 3	NPHP3	608002	1.704,00 €					
Nephronophthisis type 4	NPHP4	607215	1.825,00 €	632,00 €	2.307,00 €			x
Nephronophthisis type 7	GLIS2	608539	737,00 €					
Nephronophthisis type 9	NEK8	609799	995,00 €					
Nephronophthisis type 12	TTC21B	612014	1.572,00 €	611,00 €	2.033,00 €			x
Nephronophthisis type 13	WDR19	608151	1.927,00 €					
Nephronophthisis type 14	ZNF423	604557	1.193,00 €					
Nephronophthisis type 15	CEP164	614848	1.724,00 €	695,00 €	2.269,00 €			x
Nephronophthisis type 16	ANKS6	615370	1.127,00 €					
Nephronophthisis type 19	DCDC2	605755	862,00 €					
Nephronophthisis-like nephropathy type 1	XPNPEP3	613553	729,00 €					
Nephrosis, Finnish type	NPHS1	602716	1.582,00 €	611,00 €	2.043,00 €			x
Nephrotic syndrome	NPHS2	604766	737,00 €	437,00 €	1.024,00 €			x
Nephrotic syndrome type 2	NPHS1	602716	1.582,00 €	611,00 €	2.043,00 €			x
Nephrotic syndrome type 3	PLCE1	608414	1.825,00 €	695,00 €	2.370,00 €			
Nephrotic syndrome type 5	LAMB2	150325	1.764,00 €					x
Nephrotic syndrome type 7	DGKE	601440	862,00 €					
Nephrotic syndrome type 8	ARHGDI4	601925	573,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Nephrotic syndrome type 9	ADCK4	615567	819,00 €					
Netherton syndrome	SPINK5	605010	1.724,00 €	716,00 €	2.290,00 €			x
Neuraminidase deficiency	NEU1	608272	573,00 €	421,00 €	844,00 €			x
Neuroaxonal neurodegeneration, infantile, with facial dysmorphism	NALCN	611549	1.888,00 €	905,00 €	2.643,00 €			
Neuroblastoma type 3, susceptibility to, familial	ALK	105590	1.673,00 €					
Neurocutaneous melanosis, somatic	NRAS	164790					950,00 €	
Neurodegeneration due to cerebral folate transport deficiency	FOLR1	136430	491,00 €	421,00 €	762,00 €			x
Neurodegeneration with brain iron acculation type 5	WDR45	300526	928,00 €	421,00 €	1.199,00 €			
Neurodegeneration with brain iron accumulation type 4	C19orf12	614297	374,00 €	374,00 €	598,00 €			x
Neurodegeneration with brain iron accumulation type 6	COASY	609855	796,00 €					
Neurodegeneration with brain iron accumulation, GTPBP2 related	GTPBP2	607434	862,00 €					
Neurodevelopmental disorder, ADAM22 related	ADAM22	603709	1.673,00 €	328,00 €	1.851,00 €			
Neurodevelopmental disorder, APC2-related	APC2	612034	1.825,00 €	445,00 €	2.120,00 €			
Neurodevelopmental disorder, CNTNAP4 related	CNTNAP4	610518	1.582,00 €					
Neurodevelopmental disorder, CROCC related	CROCC	615776	1.977,00 €					
Neurodevelopmental disorder, FRMPD4 related	FRMPD4	300838	1.521,00 €					
Neurodevelopmental disorder, KCTD3 related	KCTD3	613272	1.326,00 €					
Neurodevelopmental disorder, MACF1 related	MACF1	608271	1.955,00 €	905,00 €	2.710,00 €			
Neurodevelopmental disorder, MTOR related	MTOR	601231	2.067,00 €	905,00 €	2.822,00 €			
Neurodevelopmental disorder, NCAM1 related	NCAM1	116930	1.521,00 €					
Neurodevelopmental disorder, NGEF related	NGEF	605991	995,00 €					
Neurodevelopmental disorder, PIGQ related	PIGQ	605754	796,00 €					
Neurodevelopmental disorder, TUBB related	TUBB	191130	655,00 €					
Neurodevelopmental disorder, ZNF311 related	ZNF311		796,00 €					
Neurodevelopmental malformation and microcephaly	KIF2A	602591	1.399,00 €					
Neurodevelopmental malformation and microcephaly	KIF5C	604593	1.764,00 €					
Neurodevelopmental malformation and microcephaly	TUBG1	191135	862,00 €					
Neurofibromatosis type 1 -like syndrome	SPRED1	609291	737,00 €	328,00 €	915,00 €			x
Neurofibromatosis type 1 -like syndrome	SPRED1	609291	737,00 €	328,00 €	915,00 €			x
Neurofibromatosis type 1	NF1	613113	2.225,00 €	655,00 €	2.730,00 €			x
Neurofibromatosis type 1	NF1	613113	2.225,00 €	655,00 €	2.730,00 €			x
Neurofibromatosis type 2	NF2	607379	1.260,00 €	328,00 €	1.438,00 €			x
Neurofibromatosis type 2	NF2	607379	1.260,00 €	328,00 €	1.438,00 €			x
Neurogenic scapuloperoneal syndrome, Kaeser type	DES	125660	737,00 €	421,00 €	1.008,00 €			x
Neuromyotonia and axonal neuropathy, autosomal recessive	HINT1	601314	374,00 €	374,00 €	598,00 €			x
Neuronal migration disorder	CTNNA2	114025	1.278,00 €					
Neuronal migration disorder	EOMES	604615	573,00 €					
Neuronal migration disorder	SPTBN5	605916	1.955,00 €					
Neuronal migration disorder	SRGAP2	606524	1.764,00 €					
Neuronopathy distal hereditary motor type 2A	HSPB8	608014	374,00 €	328,00 €	552,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Neuronopathy distal hereditary motor type 2B	HSPB1	602195	374,00 €	328,00 €	552,00 €			x
Neuronopathy distal hereditary motor type 5	GARS	600287	1.193,00 €	328,00 €	1.371,00 €			x
Neuronopathy distal hereditary motor type 6	IGHMBP2	600502	1.193,00 €	328,00 €	1.371,00 €			x
Neuronopathy distal hereditary motor type 7B	DCTN1	601143	1.704,00 €	674,00 €	2.228,00 €			x
Neuropathy sensor type 1E	DNMT1	126375	1.802,00 €	704,00 €	2.356,00 €			
Neuropathy with liability to pressure palsies [HNPP]	PMP22	601097	468,00 €	328,00 €	646,00 €			x
Neuropathy with sensory ataxic, dysarthria, and ophthalmoparesis	POLG	174763	1.521,00 €	328,00 €	1.699,00 €			x
Neuropathy, distal hereditary motor, type 5A	BSC1L2	606158	819,00 €	429,00 €	1.098,00 €			x
Neuropathy, hereditary sensory and autonomic type 2	SCN9A	603415	1.572,00 €	569,00 €	1.991,00 €			x
Neuropathy, hereditary sensory and autonomic type 6	DST	113810	2.225,00 €					x
Neuropathy, hereditary sensory, type ID	ATL1	606439	1.061,00 €	328,00 €	1.239,00 €			x
Neuropathy, hereditary sensory, with spastic paraplegia	CCT5	610150	819,00 €					
Neuropathy, hereditary sensory, with spastic paraplegia	CCT5	610150	819,00 €					
Neutral lipid storage disease with myopathy	PNPLA2	609059	737,00 €					
Neutropenia, nonimmune chronic idiopathic, of adults	GFI1	600871	655,00 €	437,00 €	942,00 €			x
Neutropenia, severe congenital type 1	ELANE	130130	491,00 €	390,00 €	731,00 €			x
Neutropenia, severe congenital type 2, autosomal dominant	GFI1	600871	655,00 €	437,00 €	942,00 €			x
Neutropenia, severe congenital type 3	HAX1	605998	655,00 €	437,00 €	942,00 €			x
Neutropenia, severe congenital type 4, autosomal recessive	G6PC3	611045	655,00 €	421,00 €	926,00 €			x
Neutropenia, severe congenital type 5, autosomal recessive	VPS45	610035	1.127,00 €					
Neutropenia, severe congenital type 5, autosomal recessive	VPS45	610035	1.127,00 €					
Neutropenia, severe congenital type 6, autosomal recessive	JAGN1	616012	386,00 €					
Neutrophil immunodeficiency syndrome	RAC2	602049	655,00 €					
Neutrophilia, hereditary	CSF3R	138971	1.127,00 €					x
Nevus sebaceous, HRAS related, somatic	HRAS	190020					1.900,00 €	
Nevus, epidermal, somatic	FGFR3	134934					1.900,00 €	
Nicolaides Baraitser syndrome	SMARCA2	600014	1.927,00 €	716,00 €	2.493,00 €			x
Niemann-Pick disease type A/B	SMPD1	607608	655,00 €	328,00 €	833,00 €			x
Niemann-Pick disease type C1	NPC1	607623	1.643,00 €	328,00 €	1.821,00 €			x
Niemann-Pick disease type C1	NPC1	607623	1.643,00 €	328,00 €	1.821,00 €			x
Niemann-Pick disease type C2	NPC2	601015	491,00 €	328,00 €	669,00 €			x
Night blindness type 1, congenital stationary, autosomal dominant	RHO	180380	491,00 €	328,00 €	669,00 €			x
Night blindness, congenital stationar type 1B	GRM6	604096	862,00 €					
Night blindness, congenital stationar type 1C	TRPM1	603576	1.764,00 €	569,00 €	2.183,00 €			
Night blindness, congenital stationary type 1A	NYX	300278	491,00 €					
Night blindness, congenital stationary type 2A	CACNA1F	300110	1.802,00 €					
Night blindness, congenital stationary type 2B	CABP4	608965	491,00 €	421,00 €	762,00 €			x
Night blindness, congenital stationary type 3	GNAT1	139330	573,00 €					x
Night blindness, congenital stationary, autosomal dominant type 2	PDE6B	180072	1.521,00 €					
Night blindness, congenital stationary, type 1E	GPR179	614515	1.704,00 €					



Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Nijmegen breakage syndrome	NBN	602667	1.260,00 €					
Nonaka myopathy	GNE	603824	928,00 €	421,00 €	1.199,00 €			x
Nonarteritic anterior ischemic optic neuropathy	GP1BA	606672	655,00 €					
Nonpolyposis hereditary colon cancer, PMS1 related	PMS1	600258	1.193,00 €	426,00 €	1.469,00 €			x
Nonsmall cell lung cancer, familial, susceptibility to	EGFR	131550	1.572,00 €	328,00 €	1.750,00 €			
Nonsmall cell lung cancer, responsive to tyrosin kinase inhibitor, somatic, EGFR related	EGFR	131550					1.900,00 €	
Nonsmall cell lung cancer, somatic	BRAF	164757					1.290,00 €	
Noonan syndrom like	SHOC2	602775	737,00 €	421,00 €	1.008,00 €			
Noonan syndrome type 1	PTPN11	176876	995,00 €	424,00 €	1.269,00 €			x
Noonan syndrome type 3	KRAS	190070	491,00 €	421,00 €	762,00 €			x
Noonan syndrome type 4	SOS1	182530	1.582,00 €					x
Noonan syndrome type 5	RAF1	164760	1.061,00 €	424,00 €	1.335,00 €			x
Noonan syndrome type 6	NRAS	164790	468,00 €					
Noonan syndrome type 7	BRAF	164757	1.260,00 €					x
Noonan syndrome type 8	RIT1	609591	491,00 €					x
Noonan syndrome type 10	LZTR1	600574	1.338,00 €	328,00 €	1.516,00 €			
Noonan syndrome-like disorder with or without juvenile meylomonocytic leukemia	CBL	165360	1.127,00 €	424,00 €	1.401,00 €			x
Norrie disease	NDP	300658	491,00 €	374,00 €	715,00 €			x
NPM1, selective sequencing of exon 11	NPM1	164040						
NRAS somatic Hotspot: c.34G>T, p.G12C	NRAS	164790						
NRAS somatic Hotspot: c.35G>A, p.G12D	NRAS	164790						
NRAS somatic Hotspot: c.181C>A p.Q61K	NRAS	164790						
NRAS somatic Hotspot: c.182A>G, p.Q61R	NRAS	164790						
NRAS, selective sequencing of exons 2 and 3	NRAS	164790						
Nystagmus type 1	FRMD7	300628	862,00 €	328,00 €	1.040,00 €			
Nystagmus type 6	GPR143	300808	729,00 €	328,00 €	907,00 €			x
Obesity	MC4R	155541	491,00 €	328,00 €	669,00 €			x
Obesity due to leptin deficiency	LEP	164160	491,00 €	328,00 €	669,00 €			x
Obesity with adrenal insufficiency and red hair	POMC	176830	374,00 €	328,00 €	552,00 €			x
Obesity with impaired prohormone processing	PCSK1	162150	1.127,00 €					
Obesity, early-onset, susceptibility to	POMC	176830	374,00 €	328,00 €	552,00 €			x
Obesity, severe	PPARG	601487	655,00 €	328,00 €	833,00 €			
Obesity, susceptibility to, SLC6A14 related	SLC6A14	300444	995,00 €	437,00 €	1.282,00 €			
Occipital horn syndrome	ATP7A	300011	1.704,00 €	328,00 €	1.882,00 €			x
Occult macular dystrophy	RP1L1	608581	1.521,00 €	437,00 €	1.808,00 €			x
Oculodentodigital dysplasia	GJA1	121014	468,00 €	343,00 €	661,00 €			x
Odontohypophosphatasia	ALPL	171760	862,00 €	421,00 €	1.133,00 €			x
Odontoonychodermal dysplasia	WNT10A	606268	491,00 €	437,00 €	778,00 €			x
Ogden syndrome	NAA10	300013	737,00 €					
Oguchi disease	GRK1	180381	819,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Oguchi disease	SAG	181031	1.061,00 €					
Oligo-astheno-teratozoospermia	NANOS1	608226	729,00 €					
Olmsted syndrome	TRPV3	607066	1.193,00 €					
Olmsted syndrome	TRPV3	607066	1.193,00 €					
Omenn syndrome	DCLRE1C	605988	1.193,00 €	328,00 €	1.371,00 €			x
Omenn syndrome	RAG2	179616	491,00 €	328,00 €	669,00 €			x
Omodysplasia type 1	GPC6	604404	819,00 €	421,00 €	1.090,00 €			x
Oocyte maturation defect	ZP1	195000	928,00 €					
Oogenesis dysfunction	SOHLH1	610224	819,00 €					
Ophthalmoplegia, isolated, MT-TN related	MT-TN	590010	296,00 €					
Opitz G syndrome	MID1	300552	928,00 €	328,00 €	1.106,00 €			x
Opitz-Kaveggia syndrome	MED12	300188	1.888,00 €	772,00 €	2.510,00 €			x
Opsismodysplasia	INPPL1	600829	1.704,00 €					
Optic atrophy type 1	OPA1	605290	1.764,00 €	328,00 €	1.942,00 €			x
Optic atrophy type 3	OPA3	606580	468,00 €	343,00 €	661,00 €			x
Optic atrophy type 7	TMEM126A	612988	468,00 €					x
Optic atrophy type 9	ACO2	100850	1.127,00 €					
Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia, and neuropathy	OPA1	605290	1.764,00 €	328,00 €	1.942,00 €			x
Opticoacoustic nerve atrophy with dementia	TIMM8A	300356	374,00 €	343,00 €	567,00 €			x
Oral-facial-digital syndrome type 1	OFD1	300170	1.521,00 €	484,00 €	1.855,00 €			x
Ornithine transcarbamoylase deficiency	OTC	300461	862,00 €	328,00 €	1.040,00 €			x
Orofacial cleft type 5	MSX1	142983	468,00 €	343,00 €	661,00 €			x
Orofacial cleft type 6	IRF6	607199	729,00 €	328,00 €	907,00 €			x
Orofacial cleft type 7	PVRL1	600644	819,00 €					
Orofacial cleft type 10	SUMO1	601912	491,00 €					
Orofacial cleft type 11	BMP4	112262	655,00 €	328,00 €	833,00 €			x
Orofaciodigital syndrome type 4	TCTN3	613847	928,00 €	437,00 €	1.215,00 €			
Orofaciodigital syndrome type 5	DDX59	615464	729,00 €	437,00 €	1.016,00 €			x
Orofaciodigital syndrome type 6	C5orf42	614571	2.223,00 €	905,00 €	2.978,00 €			x
Orofaciodigital syndrome type 14	C2CD3	615944	1.775,00 €					
Orolaryngeal cancer, multiple, somatic	CDKN2A	600160					1.900,00 €	
Orotic aciduria	UMPS	613891	655,00 €	421,00 €	926,00 €			x
Orthostatic intolerance	SLC6A2	163970	1.127,00 €					
Osseous heteroplasia, progressive	GNAS	139320	1.338,00 €	328,00 €	1.516,00 €			
Osteoarthritis with mild chondrodysplasia	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Osteogenesis and dental anomalies, CSF1 related	CSF1	120420	796,00 €	421,00 €	1.067,00 €			x
Osteogenesis disorders, CREB3L1 related	CREB3L1	258480	862,00 €					
Osteogenesis imperfecta	COL1A1	120150	1.825,00 €	328,00 €	2.003,00 €			x
Osteogenesis imperfecta	COL1A2	120160	2.016,00 €	328,00 €	2.194,00 €			x
Osteogenesis imperfecta type 5	IFITM5	614757	386,00 €	343,00 €	579,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Osteogenesis imperfecta type 6	SERPINF1	172860	655,00 €					
Osteogenesis imperfecta type 7	CRTAP	605497	729,00 €					
Osteogenesis imperfecta type 8	P3H1	610339	1.061,00 €					
Osteogenesis imperfecta type 9	PPIB	123841	491,00 €					
Osteogenesis imperfecta type 10	SERPINH1	613848	573,00 €					
Osteogenesis imperfecta type 11	FKBP10	607063	729,00 €					
Osteogenesis imperfecta type 12	SP7	606633	491,00 €					x
Osteogenesis imperfecta type 13	BMP1	112264	1.127,00 €	437,00 €	1.414,00 €			
Osteogenesis imperfecta type 14	TMEM38B	611236	573,00 €	421,00 €	844,00 €			x
Osteogenesis imperfecta type 15	WNT1	164820	491,00 €					
Osteogenesis imperfecta with congenital joint contractures	PLOD2	601865	1.278,00 €					
Osteoglophonic dysplasia	FGFR1	136350	1.193,00 €	328,00 €	1.371,00 €			x
Osteolysis, familial expansile	TNFRSF11A	603499	928,00 €					
Osteomyelitis, sterile multifocal, with periostitis and pustulosis	IL1RN	147679	655,00 €	421,00 €	926,00 €			x
Osteopathia striata with cranial sclerosis	AMER1	300647	796,00 €	343,00 €	989,00 €			x
Osteopetrosis of infancy, malignant	SNX10	614780	573,00 €	437,00 €	860,00 €			x
Osteopetrosis, autosomal dominant type 1	CLCN7	602727	1.399,00 €	527,00 €	1.776,00 €			x
Osteopetrosis, autosomal recessive type 1	TCIRG1	604592	1.338,00 €					x
Osteopetrosis, autosomal recessive type 2	TNFSF11	602642	573,00 €					
Osteopetrosis, autosomal recessive type 3	CA2	611492	655,00 €	437,00 €	942,00 €			
Osteopetrosis, autosomal recessive type 4	CLCN7	602727	1.399,00 €	527,00 €	1.776,00 €			x
Osteopetrosis, autosomal recessive type 5	OSTM1	607649	1.127,00 €					
Osteopetrosis, autosomal recessive type 6	PLEKHM1	611466	928,00 €					
Osteopetrosis, autosomal recessive type 7	TNFRSF11A	603499	928,00 €					
Osteoporosis pseudoglioma syndrome	LRP5	603506	1.521,00 €	328,00 €	1.699,00 €			x
Otofaciocervical syndrome	EYA1	601653	1.127,00 €	328,00 €	1.305,00 €			x
Otopaladigital syndrome type 1	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x
Otopaladigital syndrome type 2	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x
Otospondylomegaepiphyseal dysplasia	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Otospondylomegaepiphyseal dysplasia	COL11A2	120290	1.802,00 €					
Ovalocytosis	SLC4A1	109270	1.260,00 €					
Ovarian cancer, somatic	CTNNB1	116806					1.900,00 €	
Ovarian cancer, somatic	ERBB2	164870					1.900,00 €	
Ovarian carcinoma, somatic	CDH1	192090					1.900,00 €	
Ovarian clear-cell carcinoma, ARID1A related, somatic	ARID1A	603024					1.900,00 €	
Ovarian dysgenesis type 1	FSHR	136435	729,00 €	429,00 €	1.008,00 €			
Ovarian dysgenesis type 2	BMP15	300247	468,00 €					
Pachyonychia congenita type 1	KRT16	148067	737,00 €					
Pachyonychia congenita type 2	KRT17	148069	737,00 €					
Pachyonychia congenita type 3	KRT6A	148041	737,00 €					x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Pachyonychia congenita type 4	KRT6B	148042	729,00 €					
Paget disease of bone	SQSTM1	601530	737,00 €					
Paget disease, juvenile	TNFRSF11B	602643	491,00 €					
Pallister-Hall syndrome	GLI3	165240	1.278,00 €	328,00 €	1.456,00 €			x
Palmoplantar keratoderma, nonepidermolytic, focal	KRT16	148067	737,00 €					
Pancreatic agenesis and congenital heart defects	GATA6	601656	819,00 €	437,00 €	1.106,00 €			x
Pancreatic agenesis type 2	PTF1A	607194	374,00 €	343,00 €	567,00 €			
Pancreatic and cerebellar agenesis	PTF1A	607194	374,00 €	343,00 €	567,00 €			
Pancreatic cancer type 2, susceptibility to	BRCA2	600185	525,00 €	330,00 €	705,00 €			
Pancreatic cancer type 3, susceptibility to	PALB2	610355	1.193,00 €	328,00 €	1.371,00 €			x
Pancreatic cancer, somatic	SMAD4	600993					1.900,00 €	
Pancreatic cancer, somatic	STK11	602216					1.900,00 €	
Pancreatic cancer, somatic	TP53	191170					1.900,00 €	
Pancreatic cancer, susceptibility to, type 4	BRCA1	113705	525,00 €	330,00 €	705,00 €			
Pancreatic cancer/melanoma syndrome, familial	CDKN2A	600160	737,00 €	328,00 €	915,00 €			
Pancreatic carcinoma, somatic	KRAS	190070					950,00 €	
Pancreatitis	CTRC	601405	655,00 €					
Pancreatitis	PRSS1	276000	573,00 €	328,00 €	751,00 €			x
Pancreatitis	SPINK1	167790	468,00 €	328,00 €	646,00 €			x
Pancreatitis, chronic, early onset	CPA1	114850	729,00 €					
Pancreatitis, chronic, protection against	PRSS2	601564	491,00 €					
Panhypopituitarism, X-linked	SOX3	313430	468,00 €	296,00 €	614,00 €			x
Pantothenate kinase-associated neurodegeneration	PANK2	606157	819,00 €	328,00 €	997,00 €			x
Papillon-Lefevre syndrome	CTSC	602365	796,00 €	437,00 €	1.083,00 €			x
Papillorenal syndrome	PAX2	167409	862,00 €	429,00 €	1.141,00 €			x
Paraganglioma and gastric stromal sarcoma	SDHD	602690	468,00 €	328,00 €	646,00 €			x
Paraganglioma and gastric stromal sarcoma	SDHD	602690	468,00 €	328,00 €	646,00 €			x
Paragangliomas type 1, with or without deafness	SDHD	602690	468,00 €	328,00 €	646,00 €			x
Paragangliomas type 4	SDHB	185470	737,00 €	328,00 €	915,00 €			
Paragangliomas type 5	SDHA	600857	1.061,00 €	328,00 €	1.239,00 €			x
Paramyotonia congenita of von Eulenburg	SCN4A	603967	1.764,00 €	328,00 €	1.942,00 €			x
Parathyroid adenoma with cystic changes, familial	CDC73	607393	1.127,00 €	328,00 €	1.305,00 €			
Parathyroid adenoma, somatic	MEN1	613733					1.900,00 €	
Parietal foramina type 1	MSX2	123101	491,00 €	343,00 €	684,00 €			x
Parietal foramina type 2	ALX4	605420	573,00 €	328,00 €	751,00 €			x
PARK1 Parkinson	SNCA	163890	573,00 €	328,00 €	751,00 €			
PARK2 Parkinson	PARK2	602544	862,00 €	328,00 €	1.040,00 €			x
PARK4 Parkinson	SNCA	163890	573,00 €	328,00 €	751,00 €			
PARK5 Parkinson	UCHL1	191342	655,00 €	328,00 €	833,00 €			x
PARK6 Parkinson	PINK1	608309	819,00 €	328,00 €	997,00 €			

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
PARK7 Parkinson	PARK7	602533	655,00 €	328,00 €	833,00 €			x
PARK8 Parkinson	LRRK2	609007	2.028,00 €	328,00 €	2.206,00 €			x
PARK9 Parkinson	ATP13A2	610513	1.764,00 €	611,00 €	2.225,00 €			x
PARK13 Parkinson	HTRA2	606441	737,00 €	437,00 €	1.024,00 €			
PARK14 Parkinson	PLA2G6	603604	1.127,00 €	328,00 €	1.305,00 €			x
PARK15 Parkinson	FBXO7	605648	729,00 €	421,00 €	1.000,00 €			x
PARK17 Parkinson	VPS35	601501	1.193,00 €	424,00 €	1.467,00 €			x
PARK19 Parkinson, juvenile-onset	DNAJC6	608375	1.399,00 €	430,00 €	1.679,00 €			x
PARK20 Parkinson	SYNJ1	604297	1.775,00 €	674,00 €	2.299,00 €			x
PARK21 Parkinson	DNAJC13	614334	1.931,00 €	905,00 €	2.686,00 €			
Parkes Weber syndrome	RASA1	139150	1.643,00 €	328,00 €	1.821,00 €			x
Parkinson disease, late-onset, susceptibility to	GBA	606463	796,00 €	421,00 €	1.067,00 €			x
Parkinson disease, susceptibility to, MT-TT related	MT-TT	590090	296,00 €					
Parkinsonism with spasticity, X-linked	ATP6AP2	300556	819,00 €					
Parkinsonism-Dystonia, infantile	SLC6A3	126455	995,00 €	445,00 €	1.290,00 €			x
Paroxysmal exercise-induced dyskinesia with epilepsy and/or hemolytic anemia	SLC2A1	138140	819,00 €	328,00 €	997,00 €			x
Paroxysmal nonkinesigenic dyskinesia	PNKD	609023	796,00 €	429,00 €	1.075,00 €			x
Partington syndrome	ARX	300382	737,00 €	328,00 €	915,00 €			x
Patterned dystrophy of retinal pigment epithelium	PRPH2	179605	468,00 €	328,00 €	646,00 €			x
PDGFRA, selective sequencing of exons 12, 14 and 18	PDGFRA	173490						
PDGFRA, selective sequencing of exons 12, 14 and 18	PDGFRA	173490						
Peeling skin syndrome type 1	CDSN	602593	573,00 €					x
Peeling skin syndrome type 2	TGM5	603805	928,00 €	426,00 €	1.204,00 €			x
Peeling skin syndrome type 3	CHST8	610190	655,00 €					
Peeling skin syndrome type 4	CSTA	184600	374,00 €					
Pelger-Huet anomaly	LBR	600024	995,00 €					
Pelizaeus-Merzbacher disease	PLP1	300401	737,00 €	328,00 €	915,00 €			x
Pelizaeus-Merzbacher disease	SLC16A2	300095	655,00 €	421,00 €	926,00 €			x
Pelvic organ prolapse, LAMC1 related	LAMC1	150290	1.704,00 €					
Pendred syndrome	SLC26A4	605646	1.326,00 €	328,00 €	1.504,00 €			x
Pendred syndrome	SLC26A4	605646	1.326,00 €	328,00 €	1.504,00 €			x
Pentosuria	DCXR	608347	796,00 €					x
Periodic fever autosomal dominant	TNFRSF1A	191190	819,00 €	429,00 €	1.098,00 €			x
Peripheral demyelinating neuropathy Waardenburg syndrome and Hirschsprung disease	SOX10	602229	573,00 €	437,00 €	860,00 €			x
Periventricular heterotopia with microcephaly	ARFGEF2	605371	1.759,00 €	821,00 €	2.430,00 €			x
Pertman Syndrome	DIS3L2	614184	1.278,00 €	442,00 €	1.570,00 €			x
Peroxisome biogenesis disorder 14B	PEX11B	603867	573,00 €	328,00 €	751,00 €			
Peroxisome biogenesis disorder type 1B	PEX1	602136	1.643,00 €	505,00 €	1.998,00 €			x
Peroxisome biogenesis disorder type 2A	PEX5	600414	1.061,00 €	424,00 €	1.335,00 €			x
Peroxisome biogenesis disorder type 2B	PEX5	600414	1.061,00 €	424,00 €	1.335,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Peroxisome biogenesis disorder type 10A	PEX3	603164	796,00 €					x
Perrault syndrome	HSD17B4	601860	1.521,00 €	527,00 €	1.898,00 €			x
Perrault syndrome type 5	TWNK	606075	737,00 €	328,00 €	915,00 €			x
Persistent Mullerian duct syndrome type 1	AMH	600957	573,00 €	390,00 €	813,00 €			x
Persistent Mullerian duct syndrome type 2	AMHR2	600956	995,00 €	429,00 €	1.274,00 €			
Peters Anomaly	CYP1B1	601771	737,00 €	328,00 €	915,00 €			x
Peters anomaly	PAX6	607108	1.061,00 €	328,00 €	1.239,00 €			x
Peters Anomaly	PITX2	601542	573,00 €	328,00 €	751,00 €			x
Peters-Plus syndrome	B3GLCT	610308	1.061,00 €	445,00 €	1.356,00 €			x
Peutz-Jeghers syndrome	STK11	602216	819,00 €	328,00 €	997,00 €			x
Peutz-Jeghers syndrome, somatic	STK11	602216					1.900,00 €	
Pfeiffer syndrome	FGFR1	136350	1.193,00 €	328,00 €	1.371,00 €			x
Pfeiffer syndrome	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Phelan-McDermid syndrome	chr. 22q13.3			328,00 €				x
Phelan-McDermid syndrome	SHANK3	606230	1.704,00 €	328,00 €	1.882,00 €			
Phenylketonuria	PAH	612349	928,00 €	328,00 €	1.106,00 €			x
Phenylketonuria modifier, SLC7A5 related	SLC7A5	600182	862,00 €					
Pheochromocytoma type 1	SDHD	602690	468,00 €	328,00 €	646,00 €			x
Pheochromocytoma type 2	SDHB	185470	737,00 €	328,00 €	915,00 €			
Pheochromocytoma type 3	SDHC	602413	573,00 €	328,00 €	751,00 €			x
Pheochromocytoma type 5	SDHAF2	613019	468,00 €	328,00 €	646,00 €			x
Pheochromocytoma type 8	TMEM127	613403	468,00 €					
Pheochromocytoma type 9	MAX	154950	737,00 €	328,00 €	915,00 €			
Phosphoenolpyruvate carboxykinase deficiency, cytosolic	PCK1	614168	729,00 €					
Phosphoenolpyruvate carboxykinase deficiency, mitochondrial	PCK2	614095	796,00 €					
Phosphoglycerate dehydrogenase deficiency	PHGDH	606879	796,00 €	421,00 €	1.067,00 €			x
Phosphoglycerate kinase 1 deficiency	PGK1	311800	862,00 €					
Phosphoribosylpyrophosphate synthetase superactivity	PRPS1	311850	655,00 €	437,00 €	942,00 €			x
Phosphoserine aminotransferase deficiency	PSAT1	610936	819,00 €					x
Phosphoserine phosphatase deficiency	PSPH	172480	573,00 €					
Pick disease	PSEN1	104311	928,00 €	328,00 €	1.106,00 €			x
Piebaldism	KIT	164920	1.326,00 €	328,00 €	1.504,00 €			x
Piebaldism	SNAI2	602150	468,00 €	328,00 €	646,00 €			x
Pigmented nodular adrenocortical disease type 1, primary	PRKAR1A	188830	796,00 €	429,00 €	1.075,00 €			x
Pigmented paravenous chorioretinal atrophy	CRB1	604210	1.460,00 €	328,00 €	1.638,00 €			x
PIK3CA related overgrowth spectrum, somatic	PIK3CA	171834					1.900,00 €	
Pitt-Hopkins syndrome	NRXN1	600565	1.825,00 €	328,00 €	2.003,00 €			x
Pitt-Hopkins syndrome	TCF4	602272	1.582,00 €	328,00 €	1.760,00 €			x
Pituitary adenoma, ACTH-secreting, due to AIP germline mutation	AIP	605555	573,00 €	328,00 €	751,00 €			x
Pituitary adenoma, growth hormone-secreting, due to AIP germline mutation	AIP	605555	573,00 €	328,00 €	751,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Pituitary adenoma, prolactin-secreting, due to AIP germline mutation	AIP	605555	573,00 €	328,00 €	751,00 €			x
Pituitary hormone deficiency type 1	POU1F1	173110	573,00 €	328,00 €	751,00 €			x
Pituitary hormone deficiency type 2	PROP1	601538	374,00 €	328,00 €	552,00 €			x
Pituitary hormone deficiency, combined type 3	LHX3	600577	737,00 €	328,00 €	915,00 €			x
Pituitary hormone deficiency, combined type 4	LHX4	602146	655,00 €	328,00 €	833,00 €			x
Pituitary stalk interruption syndrome, GPR161 related	GPR161		655,00 €					
Pityriasis rubra pilaris	CARD14	607211	1.338,00 €					
Plasminogen activator inhibitor type 1	SERPINE1	173360	655,00 €					x
Plasminogen deficiency type 1	PLG	173350	1.326,00 €					
Platelet aggregation disorder	PEAR1	610278	1.521,00 €					
Platelet dense granule secretion defect, excessive bleeding	FLI1	193067	796,00 €					
Platelet disorder with associated myeloid malignancy	RUNX1	151385	729,00 €	328,00 €	907,00 €			x
Platelet glycoprotein IV deficiency	CD36	173510	928,00 €	437,00 €	1.215,00 €			
Platyspondylic skeletal dysplasia, Torrance type	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Pleuropulmonary blastoma	DICER1	606241	1.764,00 €	611,00 €	2.225,00 €			
Pneumothorax, primary spontaneous	FLCN	607273	729,00 €	328,00 €	907,00 €			x
Poikiloderma with neutropenia	USB1	613276	737,00 €	437,00 €	1.024,00 €			
Polyarteritis nodosa, childhood-onset	CECR1	607575	729,00 €	429,00 €	1.008,00 €			x
Polycystic kidney and hepatic disease	PKHD1	606702	1.955,00 €	655,00 €	2.460,00 €			x
Polycystic kidney disease type 1, autosomal dominant	PKD1	601313	2.176,00 €	655,00 €	2.681,00 €			x
Polycystic kidney disease type 1, autosomal recessive	PKHD1	606702	1.955,00 €	655,00 €	2.460,00 €			x
Polycystic kidney disease type 2, autosomal dominant	PKD2	173910	1.127,00 €	328,00 €	1.305,00 €			x
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	TREM2	605086	491,00 €	390,00 €	731,00 €			x
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	TYROBP	604142	468,00 €	390,00 €	708,00 €			x
Polycystic liver disease	PRKCSH	177060	995,00 €					
Polycystic ovary syndrome type 1	SULT2A1	125263	573,00 €					x
Polycystic ovary syndrome type 1	SULT2B1	604125	655,00 €					
Polycythemia vera, somatic	JAK2	147796					1.900,00 €	
Polyglucosan body myopathy type 1 with or without immunodeficiency	RBCK1	610924	862,00 €					
Polymicrogyria asymmetric	TUBB2B	612850	491,00 €					
Polymicrogyria bilateral frontoparietal	ADGRG1	604110	995,00 €	445,00 €	1.290,00 €			x
Polymicrogyria bilateral occipital	NR2E1	603849	928,00 €					
Polymicrogyria bilateral occipital	NR2E1	603849	928,00 €					
Polymicrogyria with optic nerve hypoplasia	TUBA8	605742	573,00 €	390,00 €	813,00 €			x
Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	PI4KA	600286	2.145,00 €					
Polyposis syndrome, hereditary mixed	GREM1	603054	386,00 €	328,00 €	564,00 €			
Polyposis syndrome, hereditary mixed type 2	BMPR1A	601299	819,00 €	328,00 €	997,00 €			x
Pompe disease	GAA	606800	1.260,00 €	328,00 €	1.438,00 €			x
Pompe disease	GAA	606800	1.260,00 €	328,00 €	1.438,00 €			x
Pontocerebellar hypoplasia type 1A	VRK1	602168	862,00 €	426,00 €	1.138,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/ duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Pontocerebellar hypoplasia type 1B	EXOSC3	606489	468,00 €	437,00 €	755,00 €			x
Pontocerebellar hypoplasia type 2A	TSEN54	608755	862,00 €	429,00 €	1.141,00 €			x
Pontocerebellar hypoplasia type 2B	TSEN2	608753	862,00 €					x
Pontocerebellar hypoplasia type 2C	TSEN34	608754	491,00 €					
Pontocerebellar hypoplasia type 2D	SEPSECS	613009	819,00 €					
Pontocerebellar hypoplasia type 2E	VPS53	615850	1.399,00 €					
Pontocerebellar hypoplasia type 4	TSEN54	608755	862,00 €	429,00 €	1.141,00 €			x
Pontocerebellar hypoplasia type 5	TSEN54	608755	862,00 €	429,00 €	1.141,00 €			x
Pontocerebellar hypoplasia type 6	RARS2	611524	1.278,00 €					x
Pontocerebellar hypoplasia type 8	CHMP1A	164010	655,00 €					x
Pontocerebellar hypoplasia, type 9	AMPD2	102771	1.278,00 €					
Pontocerebellar hypoplasia, type 10	CLP1	608757	491,00 €					
Popliteal pterygium syndrome type 1	IRF6	607199	729,00 €	328,00 €	907,00 €			x
Popliteal pterygium syndrome, lethal type	RIPK4	605706	796,00 €					x
Porencephaly type 2	COL4A2	120090	1.845,00 €	824,00 €	2.519,00 €			x
Porencephaly, familial	COL4A1	120130	1.931,00 €	811,00 €	2.592,00 €			x
Poretti-Boltshauser syndrome	LAMA1	150320	1.955,00 €	905,00 €	2.710,00 €			
Porokeratosis type 3, disseminated superficial actinic	MVK	251170	729,00 €	429,00 €	1.008,00 €			x
Porphyria acute intermittent	HMBS	609806	928,00 €	328,00 €	1.106,00 €			x
Porphyria congenital erythropoietic	UROS	606938	819,00 €	328,00 €	997,00 €			x
Porphyria cutanea tarda	UROD	613521	737,00 €	328,00 €	915,00 €			x
Porphyria variegata	PPOX	600923	862,00 €	328,00 €	1.040,00 €			
Postaxial acrofacial dysostosis	DHODH	126064	819,00 €	421,00 €	1.090,00 €			x
Potassium-aggravated myotonia	SCN4A	603967	1.764,00 €	328,00 €	1.942,00 €			x
Prader-Willi syndrome	chr. 15q11			328,00 €				x
Prader-Willi syndrome	NDN	602117	468,00 €	328,00 €	646,00 €			x
Prader-Willi syndrome	SNRPN	182279	573,00 €	328,00 €	751,00 €			x
Precocious puberty, male	LHCGR	152790	1.061,00 €	429,00 €	1.340,00 €			x
Preeclampsia/eclampsia type 5	CORIN	605236	1.399,00 €					
Pregnancy loss, recurrent, C4BPA related	C4BPA	120830	862,00 €					
Premature ovarian failure type 1	FMR1	309550	1.399,00 €	328,00 €	1.577,00 €			x
Primary aldosteronism, seizures, and neurologic abnormalities	CACNA1D	114206	2.145,00 €	905,00 €	2.900,00 €			
Primary ciliary dyskinesia type 1	DNAI1	604366	1.460,00 €	328,00 €	1.638,00 €			
Primary ciliary dyskinesia type 2	DNAAF3	614566	995,00 €					
Primary ciliary dyskinesia type 3	DNAH5	603335	1.955,00 €	328,00 €	2.133,00 €			
Primary ciliary dyskinesia type 5	HYDIN	610812	1.955,00 €					
Primary ciliary dyskinesia type 6	NME8	607421	1.326,00 €					
Primary ciliary dyskinesia type 7	DNAH11	603339	1.955,00 €	905,00 €	2.710,00 €			
Primary ciliary dyskinesia type 9	DNAI2	605483	1.061,00 €					
Primary ciliary dyskinesia type 10	DNAAF2	612517	737,00 €					



Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Primary ciliary dyskinesia type 11	RSPH4A	612647	655,00 €					
Primary ciliary dyskinesia type 12	RSPH9	612648	655,00 €					
Primary ciliary dyskinesia type 13	DNAAF1	613190	928,00 €					
Primary ciliary dyskinesia type 14	CCDC39	613798	1.338,00 €					
Primary ciliary dyskinesia type 15	CCDC40	613799	1.338,00 €					
Primary ciliary dyskinesia type 16	DNAL1	610062	729,00 €					
Primary ciliary dyskinesia type 17	CCDC103	614677	491,00 €					
Primary ciliary dyskinesia type 18	DNAAF5	614864	995,00 €	426,00 €	1.271,00 €			
Primary ciliary dyskinesia type 19	LRRC6	614930	928,00 €					
Primary ciliary dyskinesia type 20	CCDC114	615038	1.061,00 €					
Primary ciliary dyskinesia type 23	ARMC4	615408	1.278,00 €					
Primary ciliary dyskinesia type 24	RSPH1	609314	819,00 €					
Primary ciliary dyskinesia type 25	DYX1C1	608706	729,00 €					
Primary ciliary dyskinesia type 26	C21orf59	615494	655,00 €					
Primary ciliary dyskinesia type 27	CCDC65	611088	737,00 €					
Primary ciliary dyskinesia type 28	SPAG1	603395	1.193,00 €					
Primary ciliary dyskinesia type 29	CCNO	607752	468,00 €					
Primary ciliary dyskinesia, DNAH9 related	DNAH9	603330	1.955,00 €	905,00 €	2.710,00 €			
Primary lateral sclerosis, juvenile	ALS2	606352	1.724,00 €	716,00 €	2.290,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 1	POLG	174763	1.521,00 €	328,00 €	1.699,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 1	POLG	174763	1.521,00 €	328,00 €	1.699,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 2	SLC25A4	103220	491,00 €	328,00 €	669,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 3	TWNK	606075	737,00 €	328,00 €	915,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 3	TWNK	606075	737,00 €	328,00 €	915,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 3	TWNK	606075	737,00 €	328,00 €	915,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 4	POLG2	604983	737,00 €	328,00 €	915,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 4	POLG2	604983	737,00 €	328,00 €	915,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 5	RRM2B	604712	729,00 €	328,00 €	907,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 5	RRM2B	604712	729,00 €	328,00 €	907,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 5	RRM2B	604712	729,00 €	328,00 €	907,00 €			x
Progressive external ophthalmoplegia with mitochondrial deletions type 6	DNA2	601810	1.399,00 €					
Progressive external ophthalmoplegia with mitochondrial deletions, autosomal recessive	POLG	174763	1.521,00 €	328,00 €	1.699,00 €			x
Progressive familial heart block	TRPM4	606936	1.704,00 €					
Progressive hearing loss	P2RX2	600844	862,00 €					
Progressive myoclonus epilepsy type 1A	PRICKLE1	608500	729,00 €	437,00 €	1.016,00 €			x
Progressive myoclonus epilepsy type 3	KCTD7	611725	491,00 €	437,00 €	778,00 €			x
Progressive myoclonus epilepsy type 6	GOSR2	604027	655,00 €					
Progressive myoclonus epilepsy type 8	CERS1	606919	737,00 €					
Prolidase deficiency	PEPD	613230	1.061,00 €					
Prolidase deficiency	PEPD	613230	1.061,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Propionic acidemia	PCCA	232000	1.521,00 €	328,00 €	1.699,00 €			x
Propionic acidemia	PCCB	232050	1.193,00 €	424,00 €	1.467,00 €			x
Prosaposin deficiency	PSAP	176801	1.061,00 €	445,00 €	1.356,00 €			x
Prostate cancer	BRCA2	600185	525,00 €	330,00 €	705,00 €			
Prostate cancer	SRD5A2	607306	468,00 €	328,00 €	646,00 €			x
Prostate cancer	STAG1	604358	1.825,00 €	905,00 €	2.580,00 €			
Prostate cancer	ZNF783		819,00 €					
Prostate cancer, familial, association with	HOXB13	604607	386,00 €					
Prostate cancer, hereditary type 1	RNASEL	180435	819,00 €					
Prostate cancer, hereditary type 2, susceptibility to	ELAC2	605367	1.278,00 €					
Prostate cancer, somatic	KLF6	602053					1.900,00 €	
Prostate tumor, AR related, somatic	AR	313700					1.900,00 €	
Protein C Deficiency, AD	PROC	612283	737,00 €	328,00 €	915,00 €			x
Protein S Deficiency, autosomal dominant	PROS1	176880	1.061,00 €	328,00 €	1.239,00 €			x
Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis	CLCN5	300008	1.061,00 €	445,00 €	1.356,00 €			x
Protoporphyrin, erythropoietic, X-linked	ALAS2	301300	796,00 €					
Protoporphyrin, erythropoietic, X-linked	ALAS2	301300	796,00 €					
Prune belly syndrome	CHRM3	118494	491,00 €					
Pseudoachondroplasia	COMP	600310	995,00 €	430,00 €	1.275,00 €			x
Pseudohermaphroditism with gynecomastia	HSD17B3	605573	796,00 €					
Pseudohermaphroditism with gynecomastia	HSD17B3	605573	796,00 €					
Pseudohypoaldosteronism type 2D	KLHL3	605775	1.061,00 €					x
Pseudohypoaldosteronism type 2E	CUL3	603136	1.260,00 €					
Pseudohypoaldosteronism, type 1, autosomal dominant	NR3C2	600983	928,00 €	421,00 €	1.199,00 €			x
Pseudohypoaldosteronism, type 1, autosomal recessive	SCNN1A	600228	862,00 €	421,00 €	1.133,00 €			x
Pseudohypoaldosteronism, type 1, autosomal recessive	SCNN1B	600760	796,00 €	426,00 €	1.072,00 €			x
Pseudohypoaldosteronism, type 1, autosomal recessive	SCNN1G	600761	796,00 €	426,00 €	1.072,00 €			x
Pseudohypoaldosteronism, type 2B	WNK4	601844	1.399,00 €					
Pseudohypoparathyroidism type 1A	GNAS	139320	1.338,00 €	328,00 €	1.516,00 €			
Pseudohypoparathyroidism type 1B	GNAS	139320	1.338,00 €	328,00 €	1.516,00 €			
Pseudohypoparathyroidism type 1C	GNAS	139320	1.338,00 €	328,00 €	1.516,00 €			
Pseudopseudohypoparathyroidism	GNAS	139320	1.338,00 €	328,00 €	1.516,00 €			
Pseudoxanthoma elasticum	ABCC6	603234	1.572,00 €	328,00 €	1.750,00 €			x
Pseudoxanthoma elasticum, forme fruste	ABCC6	603234	1.572,00 €	328,00 €	1.750,00 €			x
Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	GGCX	137167	1.061,00 €					
Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	GGCX	137167	1.061,00 €					
Psoriasis susceptibility type 11	IL12B	161561	737,00 €					
Psoriasis type 2	CARD14	607211	1.338,00 €					
Psoriasis, generalized pustular	IL36RN	605507	468,00 €	390,00 €	708,00 €			x
Psychomotor retardation	TANC1	611397	1.643,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Pterygium syndrome	CHRNA1	100730	928,00 €	421,00 €	1.199,00 €			x
Ptosis, congenital	ZFXH4	606940	1.622,00 €					
Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 1	TERT	187270	1.260,00 €	328,00 €	1.438,00 €			x
Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 2	PARN	604212	1.460,00 €					
Pulmonary fibrosis, idiopathic	SFTPA1	178630	573,00 €					
Pulmonary fibrosis, idiopathic	SFTPA2	178642	491,00 €					
Pulmonary hypertension, primary type	BMPR2	600799	1.193,00 €	328,00 €	1.371,00 €			x
Pulmonary newborn hypertension	CRHR1	122561	995,00 €	328,00 €	1.173,00 €			x
Pulmonary venoocclusive disease type 1	BMPR2	600799	1.193,00 €	328,00 €	1.371,00 €			x
Pulmonary venoocclusive disease type 2	EIF2AK4	609280	2.028,00 €					
Pycnodysostosis	CTSK	601105	655,00 €	437,00 €	942,00 €			x
Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	MYD88	602170	573,00 €					
Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	PSTPIP1	606347	1.061,00 €					
Pyridoxamine 5'-phosphate oxidase deficiency	PNPO	603287	655,00 €	437,00 €	942,00 €			x
Pyridoxine-dependent epilepsy	ALDH7A1	107323	1.278,00 €	421,00 €	1.549,00 €			x
Pyruvate carboxylase deficiency	PC	608786	1.278,00 €	484,00 €	1.612,00 €			x
Pyruvate carboxylase deficiency	PC	608786	1.278,00 €	484,00 €	1.612,00 €			x
Pyruvate dehydrogenase E1-alpha deficiency	PDHA1	300502	862,00 €	328,00 €	1.040,00 €			x
Pyruvate dehydrogenase E1-beta deficiency	PDHB	179060	819,00 €	429,00 €	1.098,00 €			x
Pyruvate dehydrogenase E2 deficiency	DLAT	608770	995,00 €					
Pyruvate dehydrogenase lipoic acid synthetase deficiency	LIAS	607031	729,00 €					
Pyruvate dehydrogenase phosphatase deficiency	PDP1	605993	491,00 €					
Pyruvate kinase deficiency with hemolytic anemia	PKLR	609712	796,00 €	328,00 €	974,00 €			x
Radioulnar synostosis, FGFR3 related	FGFR3	605830	655,00 €					
Raine syndrome	FAM20C	611061	796,00 €					
RAPADILINO syndrome	RECQL4	603780	1.193,00 €	442,00 €	1.485,00 €			
Rapp-Hodgkin syndrome	TP63	603273	1.061,00 €	424,00 €	1.335,00 €			x
Refsum disease	PEX7	601757	729,00 €	429,00 €	1.008,00 €			x
Refsum disease	PHYH	602026	819,00 €	421,00 €	1.090,00 €			x
Renal cancer, KDM6A related, somatic	KDM6A	300128					1.900,00 €	
Renal carcinoma, chromophobe, somatic	FLCN	607273					1.900,00 €	
Renal carcinoma, Tuberous sclerosis-associated, somatic	TSC1	605284					1.900,00 €	
Renal cell carcinoma, due to HNF1A germline mutation	HNF1A	142410	729,00 €	328,00 €	907,00 €			x
Renal cell carcinoma, papillary type 1, familial	MET	164860	1.399,00 €	328,00 €	1.577,00 €			
Renal cell carcinoma, papillary type 1, somatic	MET	164860					1.900,00 €	
Renal cell carcinoma, somatic	VHL	608537					1.900,00 €	
Renal cystic dysplasia, cystic, susceptibility to	BICC1	614295	1.399,00 €					x
Renal dysfunction due to SLC26A1 deficiency	SLC26A1	610130	573,00 €					
Renal glucosuria	SLC5A2	182381	928,00 €	437,00 €	1.215,00 €			x
Renal tubular acidosis with deafness	ATP6V1B1	192132	928,00 €	437,00 €	1.215,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Renal tubular acidosis, distal, autosomal recessive	ATP6V0A4	605239	1.399,00 €	463,00 €	1.712,00 €			x
Renal tubular acidosis, proximal, with ocular abnormalities	SLC4A4	603345	1.582,00 €	527,00 €	1.959,00 €			x
Renal tubular acidosis, SLC4A5 related	SLC4A5	606757	1.643,00 €	548,00 €	2.041,00 €			x
Renal tubular dysgenesis	ACE	106180	1.704,00 €	527,00 €	2.081,00 €			x
Renal tubular dysgenesis	AGT	106150	655,00 €					
Renal tubular dysgenesis	AGTR1	106165	491,00 €					
Renal tubular dysgenesis	REN	179820	729,00 €	429,00 €	1.008,00 €			x
Renpenning syndrome	PQBP1	300463	491,00 €	328,00 €	669,00 €			x
Restrictive dermopathy, lethal	LMNA	150330	995,00 €	328,00 €	1.173,00 €			x
Restrictive dermopathy, lethal	ZMPSTE24	606480	729,00 €	328,00 €	907,00 €			x
RET, selective sequencing of exons 5, 8, 10, 11 and 13-16	RET	164761						
Reticular dysgenesis	AK2	103020	491,00 €					
Retinal cone dystrophy type 3B	KCNV2	607604	573,00 €	343,00 €	766,00 €			x
Retinal cone dystrophy type 4	CACNA2D4	608171	2.028,00 €					
Retinal degeneration, late-onset, autosomal dominant	C1QTNF5	608752	374,00 €					
Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities	ITM2B	603904	573,00 €					
Retinal nonattachment nonsyndromic congenital	ATOH7	609875	386,00 €					
Retinitis pigmentosa juvenile	LRAT	604863	374,00 €					
Retinitis pigmentosa SEMA4C related	SEMA4C	604462	1.193,00 €					
Retinitis pigmentosa type 1, autosomal dominant	RP1	603937	1.775,00 €	328,00 €	1.953,00 €			x
Retinitis pigmentosa type 2 X-linked	RP2	300757	573,00 €	328,00 €	751,00 €			x
Retinitis pigmentosa type 3 X-linked	RPGR	312610	1.521,00 €	328,00 €	1.699,00 €			x
Retinitis pigmentosa type 4, autosomal dominant/recessive	RHO	180380	491,00 €	328,00 €	669,00 €			x
Retinitis pigmentosa type 7, autosomal dominant	PRPH2	179605	468,00 €	328,00 €	646,00 €			x
Retinitis pigmentosa type 7	ROM1	180721	573,00 €					
Retinitis pigmentosa type 9, autosomal dominant	RP9	607331	491,00 €					
Retinitis pigmentosa type 10, autosomal dominant	IMPDH1	146690	1.127,00 €	328,00 €	1.305,00 €			
Retinitis pigmentosa type 11, autosomal dominant	PRPF31	606419	796,00 €	328,00 €	974,00 €			x
Retinitis pigmentosa type 12, autosomal recessive	CRB1	604210	1.460,00 €	328,00 €	1.638,00 €			x
Retinitis pigmentosa type 13, autosomal dominant	PRPF8	607300	1.876,00 €	905,00 €	2.631,00 €			x
Retinitis pigmentosa type 14, autosomal recessive	TULP1	602280	862,00 €	445,00 €	1.157,00 €			x
Retinitis pigmentosa type 17, autosomal dominant	CA4	114760	655,00 €					
Retinitis pigmentosa type 18, autosomal dominant	PRPF3	607301	1.061,00 €					x
Retinitis pigmentosa type 19, autosomal recessive	ABCA4	601691	2.301,00 €	655,00 €	2.806,00 €			x
Retinitis pigmentosa type 20, autosomal recessive	RPE65	180069	796,00 €	328,00 €	974,00 €			x
Retinitis pigmentosa type 23 X-linked	OFD1	300170	1.521,00 €	484,00 €	1.855,00 €			x
Retinitis pigmentosa type 25	EYS	612424	1.955,00 €	328,00 €	2.133,00 €			
Retinitis pigmentosa type 26, autosomal recessive	CERKL	608381	995,00 €	437,00 €	1.282,00 €			x
Retinitis pigmentosa type 27, autosomal dominant	NRL	162080	386,00 €					
Retinitis pigmentosa type 28, autosomal recessive	FAM161A	613596	819,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Retinitis pigmentosa type 30, autosomal dominant	FSCN2	607643	491,00 €					
Retinitis pigmentosa type 31, autosomal dominant	TOPORS	609507	796,00 €					
Retinitis pigmentosa type 33, autosomal dominant	SNRNP200	601664	1.931,00 €					
Retinitis pigmentosa type 35, autosomal dominant/recessive	SEMA4A	607292	1.127,00 €					
Retinitis pigmentosa type 36, autosomal recessive	PRCD	610598	374,00 €					
Retinitis pigmentosa type 37, autosomal dominant/recessive	NR2E3	604485	737,00 €					
Retinitis pigmentosa type 38, autosomal recessive	MERTK	604705	1.399,00 €	430,00 €	1.679,00 €			x
Retinitis pigmentosa type 39	USH2A	608400	1.955,00 €	655,00 €	2.460,00 €			x
Retinitis pigmentosa type 40, autosomal recessive	PDE6B	180072	1.521,00 €					
Retinitis pigmentosa type 41, autosomal recessive	PROM1	604365	1.704,00 €	569,00 €	2.123,00 €			x
Retinitis pigmentosa type 42, autosomal dominant	KLHL7	611119	729,00 €					
Retinitis pigmentosa type 43, autosomal recessive	PDE6A	180071	1.460,00 €					
Retinitis pigmentosa type 44, autosomal dominant/recessive	RGR	600342	655,00 €					
Retinitis pigmentosa type 45, autosomal recessive	CNGB1	600724	1.673,00 €					
Retinitis pigmentosa type 46, autosomal recessive	IDH3B	604526	796,00 €					
Retinitis pigmentosa type 47, autosomal recessive	SAG	181031	1.061,00 €					
Retinitis pigmentosa type 48, autosomal dominant	GUCA1B	602275	374,00 €					
Retinitis pigmentosa type 49, autosomal recessive	CNGA1	123825	928,00 €					
Retinitis pigmentosa type 50, autosomal dominant	BEST1	607854	928,00 €	328,00 €	1.106,00 €			x
Retinitis pigmentosa type 51, autosomal recessive	TTC8	608132	1.193,00 €					
Retinitis pigmentosa type 53, autosomal recessive	RDH12	608830	737,00 €	328,00 €	915,00 €			x
Retinitis pigmentosa type 54, autosomal recessive	C2ORF71	613425	1.061,00 €					
Retinitis pigmentosa type 55, autosomal recessive	ARL6	608845	655,00 €					
Retinitis pigmentosa type 56, autosomal recessive	IMP2	607056	1.521,00 €	430,00 €	1.801,00 €			
Retinitis pigmentosa type 57, autosomal recessive	PDE6G	180073	374,00 €					
Retinitis pigmentosa type 58, autosomal recessive	ZNF513	613598	655,00 €					
Retinitis pigmentosa type 59, autosomal recessive	DHDDS	608172	729,00 €					
Retinitis pigmentosa type 60	PRPF6	613979	1.643,00 €					
Retinitis pigmentosa type 61, autosomal recessive	CLRN1	606397	573,00 €	437,00 €	860,00 €			x
Retinitis pigmentosa type 62, autosomal recessive	MAK	154235	1.061,00 €					
Retinitis pigmentosa type 64, autosomal recessive	C8ORF37	614477	819,00 €					
Retinitis pigmentosa type 66, autosomal recessive	RBP3	180290	862,00 €					
Retinitis pigmentosa type 74, autosomal recessive	BBS2	606151	1.127,00 €	424,00 €	1.401,00 €			x
Retinitis pigmentosa, juvenile, autosomal recessive	SPATA7	609868	928,00 €					
Retinitis punctata albescens	RHO	180380	491,00 €	328,00 €	669,00 €			x
Retinoschisis	RS1	300839	573,00 €	421,00 €	844,00 €			
Rett syndrome	MECP2	300005	468,00 €	328,00 €	646,00 €			x
Rett syndrome preserved speech variant	MECP2	300005	468,00 €	328,00 €	646,00 €			x
Rett syndrome, congenital variant	FOXP1	164874	468,00 €	328,00 €	646,00 €			x
Revesz syndrome	TINF2	604319	737,00 €					x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Rhabdoid tumors, somatic	SMARCB1	601607					1.900,00 €	
Rheumatoid arthritis, susceptibility to	AFF3	601464	1.460,00 €					
Rheumatoid arthritis, TNFAIP3 related	TNFAIP3	191163	729,00 €					
Rhizomelic chondrodysplasia punctata type 2	GNPAT	602744	1.061,00 €					
Rhizomelic chondrodysplasia punctata type 3	AGPS	603051	1.521,00 €					
Rhizomelic chondrodysplasia punctata type 5	PEX5	600414	1.061,00 €	424,00 €	1.335,00 €			x
Riboflavin deficiency	SLC52A1	607883	491,00 €					
Rickets, vitamin D 25-hydroxylation-deficient, type 1B	CYP2R1	608713	573,00 €					
Rickets, vitamin D 25-hydroxylation-deficient, type 1B	CYP2R1	608713	573,00 €					
Rickets, vitamin D dependent, type 1	CYP27B1	609506	819,00 €					
Rickets, vitamin D dependent, type 1	CYP27B1	609506	819,00 €					
Rickets, vitamin D-resistant, type 2A	VDR	601769	729,00 €	429,00 €	1.008,00 €			x
Rigid spine muscular dystrophy	SELENON	606210	995,00 €	426,00 €	1.271,00 €			x
Ring dermoid of cornea	PITX2	601542	573,00 €	328,00 €	751,00 €			x
Rippling muscle disease	CAV3	601253	386,00 €	328,00 €	564,00 €			x
Ritscher-Schinzel syndrome type 1	KIAA0196	610657	1.643,00 €	611,00 €	2.104,00 €			x
RNA processing related disorders	HNRNPU	602869	1.061,00 €					
Roberts syndrome	ESCO2	609353	796,00 €	429,00 €	1.075,00 €			x
Robinow syndrome, autosomal dominant type 1	WNT5A	164975	819,00 €	390,00 €	1.059,00 €			x
Robinow syndrome, autosomal dominant type 2	DVL1	601365	928,00 €					
Robinow syndrome, autosomal recessive	ROR2	602337	862,00 €	328,00 €	1.040,00 €			x
Robinow-Sorauf syndrome	TWIST1	601622	386,00 €	328,00 €	564,00 €			x
Rolandic epilepsy, mental retardation, and speech dyspraxia	SRPX2	300642	729,00 €					x
Rothmund-Thomson syndrome	RECQL4	603780	1.193,00 €	442,00 €	1.485,00 €			
Roussy-Levy syndrome	PMP22	601097	468,00 €	328,00 €	646,00 €			x
Rubinstein-Taybi syndrome	CREBBP	600140	1.572,00 €	328,00 €	1.750,00 €			x
Rubinstein-Taybi syndrome	EP300	602700	1.977,00 €	328,00 €	2.155,00 €			x
Saccharopinuria	AASS	605113	1.326,00 €					
Saethre-Chotzen syndrome	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Saethre-Chotzen syndrome	TWIST1	601622	386,00 €	328,00 €	564,00 €			x
Salih ataxia	RUBCN	613516	1.338,00 €					
Sandhoff disease	HEXB	606873	995,00 €	437,00 €	1.282,00 €			x
Sarcoidosis, early-onset	NOD2	605956	1.326,00 €	421,00 €	1.597,00 €			x
Sarcosinemia	SARDH	604455	1.338,00 €					
SC Phocomelia syndrome	ESCO2	609353	796,00 €	429,00 €	1.075,00 €			x
Scaphocephaly, maxillary retrusion, and mental retardation	FGFR2	176943	1.460,00 €	328,00 €	1.638,00 €			x
Scapuloperoneal myopathy, MYH7 related	MYH7	160760	1.759,00 €	328,00 €	1.937,00 €			x
Schaaf-Yang syndrome	MAGEL2	605283	819,00 €	328,00 €	997,00 €			
Scheie syndrome	IDUA	252800	928,00 €	437,00 €	1.215,00 €			x
Schimke immunoosseous dysplasia	SMARCAL1	606622	1.260,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Schindler disease	NAGA	104170	819,00 €					x
Schinzel-Giedion midface retraction syndrome	SETBP1	611060	1.127,00 €					
Schizencephaly	EMX2	600035	468,00 €	374,00 €	692,00 €			x
Schizophrenia, CALR related	CALR	109091	655,00 €					
Schizophrenia, CELSR2 related	CELSR2	604265	1.973,00 €					
Schizophrenia, GRID2 related	GRID2	602368	1.193,00 €	424,00 €	1.467,00 €			x
Schizophrenia, NOTCH4 related	NOTCH4	164951	1.673,00 €					
Schneckenbecken dysplasia	SLC35D1	610804	796,00 €					
Schwartz-Jampel syndrome type 1	HSPG2	142461	1.955,00 €					x
SCID autosomal recessive T negative B positive type	JAK3	600173	1.460,00 €					
Sclerosteosis type 1	SOST	605740	386,00 €					
Sea-blue histiocyte disease	APOE	107741	655,00 €	437,00 €	942,00 €			
Seckel syndrome	ATRIP	606605	995,00 €					
Seckel syndrome type 1	ATR	601215	1.955,00 €					x
Seckel syndrome type 2	RBBP8	604124	1.278,00 €					x
Seckel syndrome type 4	CENPJ	609279	1.643,00 €	328,00 €	1.821,00 €			x
Seckel syndrome type 5	CEP152	613529	1.764,00 €					
Seckel syndrome type 6	CEP63	614724	1.193,00 €					
Seckel syndrome type 7	NIN	608684	1.825,00 €					
SED congenita	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Segawa syndrome, autosomal recessive	TH	191290	796,00 €	437,00 €	1.083,00 €			x
Seizures, benign familial infantile, type 2	PRRT2	614386	468,00 €	374,00 €	692,00 €			x
Seizures, benign neonatal, type 1	KCNQ2	602235	1.260,00 €	328,00 €	1.438,00 €			x
Seizures, benign neonatal, type 2	KCNQ3	602232	1.127,00 €	328,00 €	1.305,00 €			x
Seizures, scoliosis, and macrocephaly syndrome	EXT2	608210	1.127,00 €	328,00 €	1.305,00 €			x
Selective T-cell defect	ZAP70	176947	862,00 €					
Sengers syndrome	AGK	610345	1.061,00 €	424,00 €	1.335,00 €			x
Senior-Loken syndrome type 5	IQCB1	609237	928,00 €					
Senior-Loken syndrome type 6	CEP290	610142	2.262,00 €	842,00 €	2.954,00 €			x
Senior-Loken syndrome type 7	SDCCAG8	613524	1.338,00 €					
Senior-Loken syndrome type 8	WDR19	608151	1.927,00 €					
Septooptic dysplasia	HESX1	601802	374,00 €	328,00 €	552,00 €			x
Serine hydrolase deficiency, SERHL2 related	SERHL2		862,00 €					
SERKAL syndrome	WNT4	603490	491,00 €	328,00 €	669,00 €			x
SERKAL syndrome	WNT4	603490	491,00 €	328,00 €	669,00 €			x
SESAME syndrome	KCNJ10	602208	374,00 €	343,00 €	567,00 €			x
Severe combined immunodeficiency due to ADA deficiency	ADA	608958	796,00 €					
Severe combined immunodeficiency due to IL2 deficiency	IL2	147680	374,00 €					
Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing ra	NHEJ1	611290	655,00 €	437,00 €	942,00 €			x
Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing ra	NHEJ1	611290	655,00 €	437,00 €	942,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Severe combined immunodeficiency, Athabascan type	DCLRE1C	605988	1.193,00 €	328,00 €	1.371,00 €			x
Severe combined immunodeficiency, B cell-negative	RAG1	179615	819,00 €					
Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	PTPRC	151460	1.724,00 €	695,00 €	2.269,00 €			
Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	IL7R	146661	819,00 €					
Short QT syndrome type 1	KCNH2	152427	1.193,00 €	328,00 €	1.371,00 €			x
Short QT syndrome type 2	KCNQ1	607542	1.061,00 €	328,00 €	1.239,00 €			x
Short QT syndrome type 3	KCNJ2	600681	491,00 €	328,00 €	669,00 €			x
Short stature syndrome	SHOX	312865	655,00 €	328,00 €	833,00 €			x
Short stature, microcephaly, and endocrine dysfunction	XRCC4	194363	655,00 €	437,00 €	942,00 €			
Short stature, optic nerve atrophy, and Pelger-Huet anomaly	NBAS	608025	2.145,00 €					
SHORT syndrome	PIK3R1	171833	1.260,00 €					
Short-rib thoracic dysplasia type 2 with or without polydactyly	IFT80	611177	1.278,00 €					
Short-rib thoracic dysplasia type 3 with or without polydactyly	DYNC2H1	603297	1.955,00 €	905,00 €	2.710,00 €			x
Short-rib thoracic dysplasia type 4 with or without polydactyly	TTC21B	612014	1.572,00 €	611,00 €	2.033,00 €			x
Short-rib thoracic dysplasia type 5 with or without polydactyly	WDR19	608151	1.927,00 €					
Short-rib thoracic dysplasia type 6 with or without polydactyly	NEK1	604588	1.825,00 €					
Short-rib thoracic dysplasia type 7 with or without polydactyly	WDR35	613602	1.825,00 €					
Short-rib thoracic dysplasia type 8 with or without polydactyly	WDR60	615462	1.582,00 €					
Short-rib thoracic dysplasia type 10 with or without polydactyly	IFT172	607386	2.102,00 €					
Short-rib thoracic dysplasia type 11 with or without polydactyly	WDR34	613363	796,00 €					
Shprintzen-Goldberg syndrome	SKI	164780	737,00 €	437,00 €	1.024,00 €			x
Shwachman-Diamond syndrome	SBDS	607444	491,00 €	390,00 €	731,00 €			x
Sialuria, finish type	SLC17A5	604322	796,00 €	429,00 €	1.075,00 €			x
Sick sinus syndrome type 1	SCN5A	600163	1.673,00 €	328,00 €	1.851,00 €			x
Sick sinus syndrome type 3	MYH6	160710	1.673,00 €					
Sickle cell anemia	HBB	141900	491,00 €	328,00 €	669,00 €			x
Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	TRNT1	612907	737,00 €					
Silver-Russell syndrome	chr. 11p15			328,00 €				x
Silver-Russell syndrome	maternal UPD chr. 7			328,00 €				
Simpson-Golabi-Behmel syndrome type 1	GPC3	300037	796,00 €	328,00 €	974,00 €			x
Sinoatrial node dysfunction and deafness	CACNA1D	114206	2.145,00 €	905,00 €	2.900,00 €			
Sinoatrial node dysfunction and deafness	CACNA1D	114206	2.145,00 €	905,00 €	2.900,00 €			
Sjogren-Larsson syndrome	ALDH3A2	609523	796,00 €	429,00 €	1.075,00 €			x
Skeletal abnormalities, CFBF related	CBFB	121360	491,00 €					
Skin fragility-woolly hair syndrome	DSP	125647	1.927,00 €	328,00 €	2.105,00 €			x
Skin hair eye pigmentation type 6	SLC24A4	609840	1.338,00 €					
Slowed nerve conduction velocity, autosomal dominant	ARHGEF10	608136	1.622,00 €					x
Small cell ovarian carcinoma, hypercalcemic type, SMARCA4 related, somatic	SMARCA4	603254					1.900,00 €	
SMED Strudwick type	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Smith-Lemli-Opitz syndrome	DHCR7	602858	737,00 €	328,00 €	915,00 €			x



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Smith-Magenis syndrome	RAI1	607642	1.193,00 €	328,00 €	1.371,00 €			x
Smith-Magenis syndrome, ULK2 related	ULK2	613653	1.704,00 €					
Smith-McCort dysplasia	DYM	607461	1.193,00 €					
Sneddon syndrome	CECR1	607575	729,00 €	429,00 €	1.008,00 €			x
Sorsby fundus dystrophy	TIMP3	188826	573,00 €					
Sotos syndrome type 1	NSD1	606681	1.775,00 €	328,00 €	1.953,00 €			x
Sotos-like syndrome	NFIX	164005	928,00 €	429,00 €	1.207,00 €			x
Spastic ataxia Charlevoix-Saguenay type	SACS	604490	1.802,00 €	328,00 €	1.980,00 €			x
Spastic ataxia type 1, autosomal dominant	VAMP1	185880	573,00 €	390,00 €	813,00 €			x
Spastic ataxia type 2, autosomal recessive	KIF1C	603060	1.460,00 €					
Spastic ataxia type 3, autosomal recessive	MARS2	609728	491,00 €					
Spastic ataxia type 5, autosomal recessive	AFG3L2	604581	1.127,00 €	424,00 €	1.401,00 €			x
Spastic paralysis, infantile onset ascending	ALS2	606352	1.724,00 €	716,00 €	2.290,00 €			x
Spastic paraplegia type 74, autosomal recessive	IBA57	615316	573,00 €					
Spastic paraplegia type 74, autosomal recessive	IBA57	615316	573,00 €					
Speech-language disorder type 1	FOXP2	605317	1.260,00 €	424,00 €	1.534,00 €			x
Spermatocytic seminoma, somatic	FGFR3	134934					1.900,00 €	
SPG1	L1CAM	308840	1.582,00 €	611,00 €	2.043,00 €			x
SPG2	PLP1	300401	737,00 €	328,00 €	915,00 €			x
SPG3A	ATL1	606439	1.061,00 €	328,00 €	1.239,00 €			x
SPG4	SPAST	604277	1.193,00 €	328,00 €	1.371,00 €			x
SPG5A	CYP7B1	603711	655,00 €	421,00 €	926,00 €			x
SPG6	NIPA1	608145	573,00 €	328,00 €	751,00 €			x
SPG7	SPG7	602783	1.260,00 €	328,00 €	1.438,00 €			x
SPG8	KIAA0196	610657	1.643,00 €	611,00 €	2.104,00 €			x
SPG10	KIF5A	602821	1.764,00 €	611,00 €	2.225,00 €			x
SPG11	SPG11	610844	1.931,00 €	328,00 €	2.109,00 €			x
SPG12	RTN2	603183	796,00 €	429,00 €	1.075,00 €			x
SPG13	HSPD1	118190	796,00 €	421,00 €	1.067,00 €			x
SPG15	ZFYVE26	612012	1.931,00 €	721,00 €	2.502,00 €			x
SPG17	BSC12	606158	819,00 €	429,00 €	1.098,00 €			x
SPG18	ERLIN2	611605	862,00 €					
SPG20	SPG20	607111	796,00 €	421,00 €	1.067,00 €			x
SPG21	SPG21	608181	737,00 €	421,00 €	1.008,00 €			
SPG26	B4GALNT1	601873	819,00 €					
SPG28	DDHD1	614603	995,00 €					
SPG30	KIF1A	601255	2.016,00 €	841,00 €	2.707,00 €			x
SPG31	REEP1	609139	737,00 €	328,00 €	915,00 €			x
SPG33	ZFYVE27	610243	796,00 €	421,00 €	1.067,00 €			x
SPG35	FA2H	611026	737,00 €	437,00 €	1.024,00 €			x

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SPG39	PNPLA6	603197	1.622,00 €	716,00 €	2.188,00 €			x
SPG42	SLC33A1	603690	655,00 €	421,00 €	926,00 €			x
SPG43	C19orf12	614297	374,00 €	374,00 €	598,00 €			x
SPG44	GJC2	608803	573,00 €	343,00 €	766,00 €			x
SPG45	NT5C2	600417	995,00 €					
SPG47	AP4B1	607245	862,00 €					
SPG48	AP5Z1	613653	1.260,00 €					
SPG49	TECPR2	615000	1.326,00 €					
SPG50	AP4M1	602296	729,00 €					
SPG51	AP4E1	607244	1.260,00 €					
SPG52	AP4S1	607243	655,00 €					
SPG53	VPS37A	609927	796,00 €					
SPG54	DDHD2	615003	1.127,00 €					
SPG55	C12ORF65	613541	386,00 €					
SPG56	CYP2U1	610670	729,00 €	390,00 €	969,00 €			x
SPG57	TFG	602498	655,00 €					
SPG59, USP8 related	USP8	603158	1.326,00 €					
SPG60, WDR48 related	WDR48	612167	1.260,00 €					
SPG61	ARL6IP1	607669	573,00 €					
SPG62, ERLIN1 related	ERLIN1	611604	796,00 €					
SPG63	AMPD2	102771	1.278,00 €					
SPG64	ENTPD1	601752	928,00 €					
SPG66, ARSI related	ARSI	610009	468,00 €					
SPG68, FLRT1 related	FLRT1	604806	491,00 €					
SPG71, ZFR related	ZFR	615635	1.326,00 €					
SPG72	REEP2	609347	655,00 €					
SPG72	REEP2	609347	655,00 €					
SPG73	CPT1C	608846	1.260,00 €					
SPGF4	SYCP3	604759	737,00 €					
SPGF5	AURKC	603495	655,00 €					
SPGF6	SPATA16	609856	796,00 €	429,00 €	1.075,00 €			
SPGF7	CATSPER1	606389	796,00 €					
SPGF8	NR5A1	184757	737,00 €	328,00 €	915,00 €			x
SPGF9	DPY19L2	613893	1.399,00 €					
Spherocytosis type 1	ANK1	612641	1.973,00 €					
Spherocytosis type 2	SPTB	182870	1.927,00 €					
Spherocytosis type 3	SPTA1	182860	2.145,00 €	905,00 €	2.900,00 €			
Spherocytosis type 5	EPB42	177070	995,00 €					
Spheroid body myopathy	MYOT	604103	819,00 €	328,00 €	997,00 €			x
Spiegler-Brooke syndrome	CYLD	605018	1.326,00 €	437,00 €	1.613,00 €			

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Spina bifida folate sensitive	MTRR	602568	1.127,00 €					
Spinal and bulbar muscular atrophy X-linked	AR	313700	1.061,00 €	328,00 €	1.239,00 €	200,00 €		x
Spinal muscular atrophy (SMA), NAIP related	NAIP	600355	1.061,00 €					
Spinal muscular atrophy distal, autosomal recessive type 4	PLEKHG5	611101	1.338,00 €	463,00 €	1.651,00 €			x
Spinal muscular atrophy type 1	SMN1	600354	819,00 €	655,00 €	1.324,00 €			x
Spinal muscular atrophy type 2	SMN1	600354	819,00 €	655,00 €	1.324,00 €			x
Spinal muscular atrophy type 3, modifier of	SMN2	601627		655,00 €				x
Spinal muscular atrophy type 3	SMN1	600354	819,00 €	655,00 €	1.324,00 €			x
Spinal muscular atrophy type 4	SMN1	600354	819,00 €	655,00 €	1.324,00 €			x
Spinal muscular atrophy type 5	DNAJB2	604139	737,00 €	421,00 €	1.008,00 €			
Spinal muscular atrophy with progressive myoclonic epilepsy	ASAH1	613468	1.061,00 €	437,00 €	1.348,00 €			x
Spinal muscular atrophy, distal, X-linked	ATP7A	300011	1.704,00 €	328,00 €	1.882,00 €			x
Spinal muscular atrophy, lower extremity, autosomal dominant, type 2	BICD2	609797	819,00 €					
Spinal muscular atrophy, lower extremity-predominant type 1, autosomal dominant	DYNC1H1	600112	1.955,00 €					x
Spinocerebellar ataxia type 1, autosomal dominant	ATXN1	601556				200,00 €		x
Spinocerebellar ataxia type 1, X-linked	ATP2B3	300014	1.278,00 €					
Spinocerebellar ataxia type 2, autosomal dominant	ATXN2	601517				200,00 €		x
Spinocerebellar ataxia type 3, autosomal dominant	ATXN3	607047				200,00 €		x
Spinocerebellar ataxia type 4, autosomal dominant	PLEKHG4	609526	1.260,00 €	484,00 €	1.594,00 €			x
Spinocerebellar ataxia type 5, autosomal dominant	SPTBN2	604985	1.977,00 €	779,00 €	2.606,00 €			x
Spinocerebellar ataxia type 6, autosomal dominant	CACNA1A	601011	2.067,00 €	328,00 €	2.245,00 €	200,00 €		x
Spinocerebellar ataxia type 7, autosomal dominant	ATXN7	607640				200,00 €		x
Spinocerebellar ataxia type 7, autosomal recessive	TPP1	607998	862,00 €	426,00 €	1.138,00 €			x
Spinocerebellar ataxia type 8, autosomal dominant	ATXN8OS	613289				200,00 €		x
Spinocerebellar ataxia type 8, autosomal recessive	SYNE1	608441	1.955,00 €	905,00 €				x
Spinocerebellar ataxia type 9, autosomal recessive	COQ8A	606980	928,00 €	445,00 €	1.223,00 €			x
Spinocerebellar ataxia type 10, autosomal dominant	ATXN10	611150				200,00 €		x
Spinocerebellar ataxia type 10, autosomal recessive	ANO10	613726	995,00 €					x
Spinocerebellar ataxia type 11, autosomal dominant	TTBK2	611695	1.278,00 €	445,00 €	1.573,00 €			x
Spinocerebellar ataxia type 12, autosomal dominant	PPP2R2B	604325				200,00 €		x
Spinocerebellar ataxia type 12, autosomal recessive	WWOX	605131	729,00 €	328,00 €	907,00 €			x
Spinocerebellar ataxia type 13, autosomal dominant	KCNC3	176264	737,00 €	390,00 €	977,00 €			x
Spinocerebellar ataxia type 13, autosomal recessive	GRM1	604473	995,00 €	437,00 €	1.282,00 €			x
Spinocerebellar ataxia type 14, autosomal dominant	PRKCG	176980	1.260,00 €	430,00 €	1.540,00 €			x
Spinocerebellar ataxia type 15	ITPR1	147265	1.955,00 €					x
Spinocerebellar ataxia type 17, autosomal dominant	TBP	600075				200,00 €		x
Spinocerebellar ataxia type 17, autosomal recessive	CWF19L1	616120	995,00 €					
Spinocerebellar ataxia type 18, autosomal dominant	IFRD1	603502	729,00 €	421,00 €	1.000,00 €			x
Spinocerebellar ataxia type 18, autosomal recessive	GRID2	602368	1.193,00 €	424,00 €	1.467,00 €			x
Spinocerebellar ataxia type 21, autosomal dominant	TMEM240	616101	374,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Spinocerebellar ataxia type 22, autosomal dominant	KCND3	605411	796,00 €	437,00 €	1.083,00 €			x
Spinocerebellar ataxia type 23, autosomal dominant	PDYN	131340	374,00 €	328,00 €	552,00 €			x
Spinocerebellar ataxia type 26, autosomal dominant	EEF2	130610	1.061,00 €					
Spinocerebellar ataxia type 27, autosomal dominant	FGF14	601515	729,00 €	390,00 €	969,00 €			x
Spinocerebellar ataxia type 28, autosomal dominant	AFG3L2	604581	1.127,00 €	424,00 €	1.401,00 €			x
Spinocerebellar ataxia type 29, congenital nonprogressive	ITPR1	147265	1.955,00 €					x
Spinocerebellar ataxia type 31, autosomal dominant	BEAN1	612051				200,00 €		
Spinocerebellar ataxia type 35, autosomal dominant	TGM6	613900	995,00 €					
Spinocerebellar ataxia type 36, autosomal dominant	NOP56	614154				200,00 €		
Spinocerebellar ataxia with axonal neuropathy, autosomal recessive	TDP1	607198	1.127,00 €	424,00 €	1.401,00 €			x
Spinocerebellar ataxia with axonal neuropathy, autosomal recessive	TDP1	607198	1.127,00 €	424,00 €	1.401,00 €			x
Spinocerebellar ataxia, infantile-onset	TWNK	606075	737,00 €	328,00 €	915,00 €			x
Split-hand/foot malformation type 1 with sensorineural hearing loss	DLX5	600028	468,00 €	374,00 €	692,00 €			x
Split-hand/foot malformation type 6	WNT10B	601906	491,00 €	390,00 €	731,00 €			x
Spondylocarpotarsal synostosis syndrome	FLNB	603381	2.059,00 €	328,00 €	2.237,00 €			x
Spondylocheirodysplasia, Ehlers-Danlos syndrome-like	SLC39A13	608735	729,00 €					
Spondylocostal dysostosis type 5	TBX6	602427	655,00 €					
Spondylocostal dysostosis, autosomal recessive type 1	DLL3	602768	729,00 €	437,00 €	1.016,00 €			x
Spondylocostal dysostosis, autosomal recessive type 2	MESP2	605195	468,00 €	343,00 €	661,00 €			x
Spondylocostal dysostosis, autosomal recessive type 3	LFNG	602576	928,00 €	437,00 €	1.215,00 €			x
Spondyloenchondrodysplasia with immune dysregulation	ACP5	171640	468,00 €					
Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	B3GALT6	615291	468,00 €					
Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	B3GALT6	615291	468,00 €					
Spondyloepimetaphyseal dysplasia, MATN3 related	MATN3	602109	819,00 €					
Spondyloepiphyseal dysplasia with congenital joint dislocations	CHST3	603799	491,00 €	374,00 €	715,00 €			x
Spondylo-megaepiphyseal-metaphyseal dysplasia	NKX3-2	602183	573,00 €	343,00 €	766,00 €			x
Spondylometaepiphyseal dysplasia, short limb-hand type	DDR2	191311	1.061,00 €					
Spondyloperipheral dysplasia	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Squamous cell carcinoma, burn scar-related, somatic	FAS	134637					1.900,00 €	
Stargardt Disease type 1	ABCA4	601691	2.301,00 €	655,00 €	2.806,00 €			x
Stargardt Disease type 1	CNGB3	605080	1.260,00 €	421,00 €	1.531,00 €			x
Stargardt Disease type 3	ELOVL4	605512	573,00 €					
Stargardt Disease type 4	PROM1	604365	1.704,00 €	569,00 €	2.123,00 €			x
Steatocystoma multiplex	KRT17	148069	737,00 €					
Stickler syndrome type 1, nonsyndromic ocular	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Stickler syndrome type 1	COL2A1	120140	1.973,00 €	328,00 €	2.151,00 €			x
Stickler syndrome type 2	COL11A1	120280	1.955,00 €	655,00 €	2.460,00 €			x
Stickler syndrome type 3	COL11A2	120290	1.802,00 €					
Stickler syndrome type 5	COL9A2	120260	1.825,00 €					
Stickler syndrome, autosomal recessive	COL9A1	120210	1.876,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Stiff skin syndrome	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			
Stiff skin syndrome	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			
Stocco dos Santos X-linked mental retardation syndrome	SHROOM4	300579	1.399,00 €					
Stormorken syndrome	STIM1	605921	928,00 €					
Stormorken syndrome	STIM1	605921	928,00 €					
Striatal degeneration	PDE8B	603390	1.521,00 €					
Stuve-Wiedemann syndrome	LIFR	151443	1.278,00 €	437,00 €	1.565,00 €			x
Succinic semialdehyde dehydrogenase deficiency	ALDH5A1	610045	862,00 €					
Succinyl CoA:3-oxoacid CoA transferase deficiency	OXCT1	601424	1.193,00 €	424,00 €	1.467,00 €			x
Sucrase-isomaltase deficiency	SI	609845	1.888,00 €	905,00 €	2.643,00 €			
Sudden infant death syndrome, susceptibility to	SCN5A	600163	1.673,00 €	328,00 €	1.851,00 €			x
Sudden infant death with dysgenesis of the testes syndrome	TSPYL1	604714	374,00 €					
Sulfatase deficiency	SUMF1	607939	819,00 €	421,00 €	1.090,00 €			x
Sulfite oxidase deficiency	SUOX	606887	655,00 €	421,00 €	926,00 €			x
Supravalvar aortic stenosis	ELN	130160	1.775,00 €	328,00 €	1.953,00 €			
Surfactant metabolism dysfunction	SFTPD	178635	655,00 €					
Surfactant metabolism dysfunction type 1	SFTPB	178640	729,00 €	421,00 €	1.000,00 €			x
Surfactant metabolism dysfunction type 2	SFTPC	178620	491,00 €	421,00 €	762,00 €			x
Surfactant metabolism dysfunction type 3	ABCA3	601615	1.622,00 €	695,00 €	2.167,00 €			x
Surfactant metabolism dysfunction type 4	CSF2RA	306250	928,00 €	328,00 €	1.106,00 €			x
Surfactant metabolism dysfunction type 5	CSF2RB	138981	995,00 €					x
Sveinsson choreoretinal atrophy	TEAD1	189967	796,00 €					
Syndactyly type 1	HOXD13	142989	374,00 €	328,00 €	552,00 €			x
Syndactyly type 5	HOXD13	142989	374,00 €	328,00 €	552,00 €			x
Syndactyly, mesoaxial synostotic, with phalangeal reduction	BHLHA9	615416	374,00 €	296,00 €	520,00 €			
Systemic lupus erythematosus	DNASE1	125505	737,00 €					
Systemic lupus erythematosus type 16	DNASE1L3	602244	737,00 €					
Systemic lupus erythematosus, susceptibility to	ITGAM	120980	1.582,00 €					
TANC2 related brain disorders	TANC2	615047	1.582,00 €					
Tangier disease	ABCA1	600046	2.028,00 €					x
Tay-Sachs disease	HEXA	606869	995,00 €	328,00 €	1.173,00 €			x
Tay-Sachs disease AB variant	GM2A	613109	491,00 €	437,00 €	778,00 €			x
T-cell immunodeficiency, congenital alopecia, and nail dystrophy	FOXM1	600838	729,00 €					
Telangiectasia hereditary hemorrhagic type 5	GDF2	605120	491,00 €	343,00 €	684,00 €			
Telangiectasia, hereditary hemorrhagic, of Rendu, Osler and Weber type 1	ENG	131195	995,00 €	328,00 €	1.173,00 €			x
Telangiectasia, hereditary hemorrhagic, type 2	ACVRL1	601284	819,00 €	328,00 €	997,00 €			x
Temple syndrome	maternal UPD chr. 1			328,00 €				
Temple-Baraitser syndrome	KCNH1	603305	928,00 €					
Temtamy syndrome	C12orf57	615140	374,00 €					
Terminal osseous dysplasia	FLNA	300017	1.825,00 €	824,00 €	2.499,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Testicular anomalies with or without congenital heart disease	GATA4	600576	729,00 €	328,00 €	907,00 €			x
Testicular anomalies with or without congenital heart disease	GATA4	600576	729,00 €	328,00 €	907,00 €			x
Testicular tumor, somatic	STK11	602216					1.900,00 €	
Tetraamelia, autosomal recessive	WNT3	165330	491,00 €					
Tetralogy of Fallot	ALDH1A2	603687	862,00 €					
Tetralogy of Fallot	GATA4	600576	729,00 €	328,00 €	907,00 €			x
Tetralogy of Fallot	GATA6	601656	819,00 €	437,00 €	1.106,00 €			x
Tetralogy of Fallot	ZFPM2	603693	928,00 €					
Thalassemia, alpha	HBA1	141800	374,00 €	328,00 €	552,00 €			x
Thalassemia, alpha	HBA2	141850	374,00 €	328,00 €	552,00 €			x
Thalassemia, delta	HBD	142000	468,00 €	328,00 €	646,00 €			
Thiamine metabolism dysfunction syndrome 4 progressive polyneuropathy type	SLC25A19	606521	573,00 €					
Thiamine metabolism dysfunction syndrome type 5	TPK1	606370	737,00 €					
Thiamine-responsive megaloblastic anemia syndrome	SLC19A2	603941	655,00 €					
Thoracic aortic aneurysm dissection	SMAD2	601366	729,00 €					
Three M syndrome type 1	CUL7	609577	1.764,00 €	548,00 €	2.162,00 €			x
Three M syndrome type 2	OBSL1	610991	1.704,00 €	442,00 €	1.996,00 €			x
Three M syndrome type 3	CCDC8	614145	491,00 €					
Thrombocythemia type 3, somatic	JAK2	147796					1.900,00 €	
Thrombocytopenia congenital amegakaryocytic	MPL	159530	995,00 €					
Thrombocytopenia type 2	ANKRD26	610855	1.876,00 €					
Thrombocytopenia type 2	MASTL	608221	1.061,00 €					
Thrombocytopenia type 4	CYCS	123970	296,00 €					
Thrombocytopenia type 5	ETV6	600618	737,00 €	328,00 €	915,00 €			
Thrombocytopenia with beta thalassemia X-linked	GATA1	305371	655,00 €	421,00 €	926,00 €			x
Thrombocytopenia, neonatal alloimmune	ITGA2B	607759	1.572,00 €	632,00 €	2.054,00 €			x
Thrombocytopenia, neonatal alloimmune	ITGB3	173470	1.127,00 €	445,00 €	1.422,00 €			x
Thrombocytopenia, X-linked	GATA1	305371	655,00 €	421,00 €	926,00 €			x
Thrombocytopenia, X-linked, intermittent	WAS	300392	928,00 €	421,00 €	1.199,00 €			x
Thrombocytosis, familial, JAK2 related	JAK2	147796	1.460,00 €	328,00 €	1.638,00 €			x
Thrombophilia due to thrombin defect	F2	176930	1.061,00 €					
Thrombophilia due to thrombin defect	F2	176930	1.061,00 €					
Thrombophilia, X-linked, due to factor IX defect	F9	300746	729,00 €	328,00 €	907,00 €			x
Thrombotic thrombocytopenic purpura	ADAMTS13	604134	1.572,00 €	611,00 €	2.033,00 €			x
Thromboxane synthase deficiency	TBXAS1	274180	1.061,00 €					
Thrombocytopenia-Absent-Radius-Syndrome	RBM8A	605313	491,00 €	421,00 €	762,00 €			x
Thyroid adenoma, hyperfunctioning, somatic	TSHR	603372					1.900,00 €	
Thyroid cancer type 2, nonmedullary, susceptibility to	SRGAP1	606523	1.399,00 €					
Thyroid carcinoma with thyrotoxicosis	TSHR	603372					1.900,00 €	
Thyroid carcinoma, follicular, HRAS related, somatic	HRAS	190020					1.900,00 €	

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Thyroid carcinoma, follicular, somatic	NRAS	164790					950,00 €	
Thyroid dysmorphogenesis type 1	SLC5A5	601843	1.061,00 €					
Thyroid dysmorphogenesis type 2A	TPO	606765	1.193,00 €	328,00 €	1.371,00 €			
Thyroid dysmorphogenesis type 3	TG	188450	2.106,00 €	905,00 €	2.861,00 €			
Thyroid dysmorphogenesis type 4	IYD	612025	737,00 €					
Thyroid dysmorphogenesis type 5	DUOXA2	612772	573,00 €					
Thyroid dysmorphogenesis type 6	DUOX1	606758	1.775,00 €					
Thyroid dysmorphogenesis type 6	DUOX2	606759	1.775,00 €					
Thyroid hormone metabolism abnormal	SECISBP2	607693	1.193,00 €					
Thyroid hormone resistance	THRB	190160	819,00 €	421,00 €	1.090,00 €			x
Thyrotoxic periodic paralysis type 1	CACNA1S	114208	1.888,00 €	328,00 €	2.066,00 €			x
Thyrotoxic periodic paralysis type 2	KCNJ18	613236	573,00 €	374,00 €	797,00 €			x
Tibial muscular dystrophy, tardive	TTN	188840	2.225,00 €	905,00 €	2.980,00 €			
Tietz albinism-deafness syndrome	MITF	156845	1.061,00 €	328,00 €	1.239,00 €			x
TJP1 deficiency	TJP1	601009	1.622,00 €					
Toe syndactyly, telecanthus, and anogenital and renal malformations	FAM58A	300708	573,00 €					
Tooth agenesis, selective type 1	MSX1	142983	468,00 €	343,00 €	661,00 €			x
Tooth agenesis, selective type 3	PAX9	167416	573,00 €	390,00 €	813,00 €			x
Tourette syndrome	SLITRK1	609678	737,00 €	296,00 €	883,00 €			x
Townes-Brocks syndrome	SALL1	602218	862,00 €	374,00 €	1.086,00 €			x
TPMT deficiency	TPMT	187680	737,00 €	328,00 €	915,00 €			
Transaldolase deficiency	TALDO1	602063	737,00 €	437,00 €	1.024,00 €			x
Transcobalamin II deficiency	TCN2	613441	729,00 €					x
Transposition of great arteries, dextro-looped 3	GDF1	602880	573,00 €	437,00 €	860,00 €			
Transposition of the great arteries, dextro-looped 1	MED13L	608771	1.775,00 €					
Transposition of the great arteries, dextro-looped 1	MED13L	608771	1.775,00 €					
Transposition of the great arteries, dextro-looped 1	MED13L	608771	1.775,00 €					
Treacher Collins syndrome type 1	TCOF1	606847	1.764,00 €	328,00 €	1.942,00 €			x
Treacher Collins syndrome type 2	POLR1D	613715	468,00 €	343,00 €	661,00 €			x
Treacher Collins syndrome type 3	POLR1C	610060	655,00 €					
Tremor essential type 4	FUS	137070	995,00 €	445,00 €	1.290,00 €			x
Trichodontoosseous syndrome	DLX3	600525	374,00 €	374,00 €	598,00 €			x
Trichoepithelioma, multiple familial, type 1	CYLD	605018	1.326,00 €	437,00 €	1.613,00 €			
Trichohepatoenteric syndrome type 1	TTC37	614589	1.927,00 €	738,00 €	2.515,00 €			x
Trichohepatoenteric syndrome type 2	SKIV2L	600478	1.338,00 €	590,00 €	1.778,00 €			x
Trichorhinophalangeal syndrome type 1	TRPS1	604386	995,00 €	328,00 €	1.173,00 €			x
Trichothiodystrophy	ERCC2	126340	1.193,00 €					
Trichothiodystrophy	ERCC3	133510	1.061,00 €					
Trichothiodystrophy	GTF2H5	608780	386,00 €					
Trichothiodystrophy, nonphotosensitive type 1	MPLKIP	609188	386,00 €	343,00 €	579,00 €			x

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Trifunctional protein deficiency	HADHA	600890	1.260,00 €	437,00 €	1.547,00 €			x
Trifunctional protein deficiency	HADHB	143450	1.061,00 €	424,00 €	1.335,00 €			x
Trigonocephaly type 1	FGFR1	136350	1.193,00 €	328,00 €	1.371,00 €			x
Trimethylaminuria	FMO3	136132	819,00 €	421,00 €	1.090,00 €			x
Triosephosphate isomerase deficiency	TPI1	190450	491,00 €					
Tuberous sclerosis	TSC1	605284	1.460,00 €	328,00 €	1.638,00 €			x
Tuberous sclerosis type 2	TSC2	191092	1.888,00 €	328,00 €	2.066,00 €			x
Tuftelin deficiency	TUFT1	600087	995,00 €					
Tumor predisposition syndrome	BAP1	603089	1.127,00 €	328,00 €	1.305,00 €			
Tumor predisposition syndrome, ARL11 related	ARL11	609351	386,00 €					
Tylosis with esophageal cancer	RHBDF2	614404	1.260,00 €					
Tylosis with esophageal cancer	RHBDF2	614404	1.260,00 €					
Tyrosine kinase 2 deficiency	TYK2	176941	1.460,00 €	527,00 €	1.837,00 €			x
Tyrosinemia type 1B	GSTZ1	603758	819,00 €	421,00 €	1.090,00 €			x
Tyrosinemia type 1	FAH	613871	995,00 €	437,00 €	1.282,00 €			x
Tyrosinemia type 2	TAT	613018	729,00 €	421,00 €	1.000,00 €			
Tyrosinemia type 3	HPD	609695	729,00 €					
Ullrich congenital muscular dystrophy	COL6A1	120220	1.643,00 €	737,00 €	2.230,00 €			x
Ullrich congenital muscular dystrophy	COL6A2	120240	1.825,00 €	590,00 €	2.265,00 €			x
Ullrich congenital muscular dystrophy type 1	COL6A3	120250	2.106,00 €	755,00 €	2.711,00 €			x
Ullrich congenital muscular dystrophy type 2	COL12A1	120320	1.955,00 €	905,00 €	2.710,00 €			x
Ulna and fibula, absence of, with severe limb deficiency	WNT7A	601570	468,00 €	437,00 €	755,00 €			x
Ulnar-Mammary syndrome	TBX3	601621	729,00 €	437,00 €	1.016,00 €			x
Unverricht-Lundborg disease	CSTB	601145	374,00 €	374,00 €	598,00 €	200,00 €		x
Urbach-Wiethe disease	ECM1	602201	737,00 €					
Urocanase deficiency	UROC1	613012	1.399,00 €					
Urofacial syndrome	LRIG2	608869	1.326,00 €					
Usher syndrome type 1D/F	PCDH15	605514	2.059,00 €	328,00 €	2.237,00 €			
Usher syndrome type 1G	USH1G	607696	573,00 €					
Usher syndrome type 1J	CIB2	605564	573,00 €					
Usher syndrome type 2C	ADGRV1	602851	1.955,00 €	905,00 €	2.710,00 €			
Usher syndrome type 2C	PDZD7	612971	1.061,00 €					
Usher syndrome type 3A	CLRN1	606397	573,00 €	437,00 €	860,00 €			x
Usher syndrome, type 1D	CDH23	605516	1.955,00 €					x
Uveal melanoma, GNAQ related, somatic	GNAQ	600998					1.900,00 €	
UV-sensitive syndrome type 1	ERCC6	609413	1.521,00 €	442,00 €	1.813,00 €			x
UV-sensitive syndrome type 3	UVSSA	614632	928,00 €					
Van Buchem disease	SOST	605740	386,00 €					
Van den Ende-Gupta syndrome	SCARF2	613619	928,00 €					
van der Woude syndrome type 1	IRF6	607199	729,00 €	328,00 €	907,00 €			x



Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
van der Woude syndrome type 2	GRHL3	608317	1.193,00 €					
Van Maldergem syndrome type 2	FAT4	612411	1.802,00 €					
Various cancers, DICER1 related, somatic	DICER1	606241					1.900,00 €	
Vascular system defects due to CALCRL deficiency	CALCRL	114190	819,00 €					
Vascular system defects due to GNA13 deficiency	GNA13	604406	655,00 €					
Vasculopathy, infantile-onset, TMEM173/STING related	TMEM173	612374	573,00 €	437,00 €	860,00 €			x
Vater association	HOXD13	142989	374,00 €	328,00 €	552,00 €			x
Ventricular fibrillation, paroxysmal familial type 1	SCN5A	600163	1.673,00 €	328,00 €	1.851,00 €			x
Ventricular septal defect type 1	GATA4	600576	729,00 €	328,00 €	907,00 €			x
Ventricular septal defect type 2	CITED2	602937	386,00 €					
Ventricular tachycardia, catecholaminergic polymorphic type 1	RYR2	180902	1.955,00 €					x
Ventricular tachycardia, catecholaminergic polymorphic type 2	CASQ2	114251	796,00 €					
Ventricular tachycardia, catecholaminergic polymorphic type 4	CALM1	114180	729,00 €	421,00 €	1.000,00 €			
Ventricular tachycardia, catecholaminergic polymorphic type 5	TRDN	603283	1.888,00 €	905,00 €	2.643,00 €			
Ventriculomegaly with cystic kidney disease	CRB2	609720	1.260,00 €					
Ventriculomegaly with cystic kidney disease	CRB2	609720	1.260,00 €					
Vesicoureteral reflux type 2	ROBO2	602431	1.704,00 €	590,00 €	2.144,00 €			x
Vesicoureteral reflux type 3	SOX17	610928	573,00 €	343,00 €	766,00 €			x
Vici syndrome	EPG5	615068	2.016,00 €	905,00 €	2.771,00 €			
Visceral myopathy	ACTG2	102545	737,00 €	421,00 €	1.008,00 €			
Vitamin E familial deficiency	TTPA	600415	491,00 €	390,00 €	731,00 €			x
Vitamin K-dependent clotting factors combined deficiency type 1	GGCX	137167	1.061,00 €					
Vitiligo-associated multiple autoimmune disease	NLRP1	606636	1.460,00 €	424,00 €	1.734,00 €			x
Vitreoretinchoroidopathy	BEST1	607854	928,00 €	328,00 €	1.106,00 €			x
Vohwinkel syndrome with ichthyosis	LOR	152445	374,00 €	343,00 €	567,00 €			x
von Hippel-Lindau syndrome	VHL	608537	374,00 €	328,00 €	552,00 €			x
von Willebrand disease	VWF	613160	2.028,00 €	655,00 €	2.533,00 €			
von Willebrand disease platelet type	GP1BA	606672	655,00 €					
Von-Gierke disease	G6PC	613742	655,00 €	390,00 €	895,00 €			x
Waardenburg syndrome type 1	PAX3	606597	796,00 €	328,00 €	974,00 €			x
Waardenburg syndrome type 2D	SNAI2	602150	468,00 €	328,00 €	646,00 €			x
Waardenburg syndrome type 2E	SOX10	602229	573,00 €	437,00 €	860,00 €			x
Waardenburg syndrome type 4C	SOX10	602229	573,00 €	437,00 €	860,00 €			x
Waardenburg syndrome/albinism	MITF	156845	1.061,00 €	328,00 €	1.239,00 €			x
Waardenburg syndrome/albinism	TYR	606933	655,00 €	328,00 €	833,00 €			x
Waardenburg syndrome/Hirschsprung disease	EDNRB	131244	862,00 €	328,00 €	1.040,00 €			x
Wagner syndrome	VCAN	118661	1.775,00 €					
Walker-Warburg syndrome	FKTN	607440	928,00 €	328,00 €	1.106,00 €			x
Walker-Warburg syndrome	ISPD	614631	729,00 €	429,00 €	1.008,00 €			x
Walker-Warburg syndrome or muscle-eye-brain disease, FKRP related	FKRP	606596	491,00 €	328,00 €	669,00 €			x

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Warburg micro syndrome 3	RAB18	602207	655,00 €					
Warburg micro syndrome type 1	RAB3GAP1	602536	1.460,00 €	527,00 €	1.837,00 €			x
Warburg micro syndrome type 1	RAB3GAP1	602536	1.460,00 €	527,00 €	1.837,00 €			x
Warburg micro syndrome type 1	RAB3GAP1	602536	1.460,00 €	527,00 €	1.837,00 €			x
Warburg micro syndrome type 2	RAB3GAP2	609275	1.775,00 €	905,00 €	2.530,00 €			
Warsaw breakage syndrome	DDX11	601150	1.521,00 €					
WDR27-related brain disorders	WDR27		1.582,00 €					
Weaver syndrome	EZH2	601573	1.193,00 €	437,00 €	1.480,00 €			x
Webb-Dattani syndrome	ARNT2	606036	1.326,00 €					
Weill-Marchesani syndrome - AR	ADAMTS10	608990	1.260,00 €	548,00 €	1.658,00 €			x
Weill-Marchesani syndrome type 3	LTBP2	602091	1.724,00 €					x
Weill-Marchesani syndrome, dominant type 2	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			
Weill-Marchesani syndrome, dominant type 2	FBN1	134797	1.955,00 €	655,00 €	2.460,00 €			
Werner syndrome	WRN	604611	1.927,00 €	905,00 €	2.682,00 €			
WHIM syndrome	CXCR4	162643	491,00 €	328,00 €	669,00 €			x
Wieacker-Wolff syndrome	ZC4H2	300897	491,00 €					
Wiedemann-Steiner syndrome	KMT2A	159555	2.262,00 €	758,00 €	2.870,00 €			x
Williams-Beuren syndrome	chr. 7q11.23			328,00 €				
Wilms tumor type 1, familial	WT1	607102	729,00 €	328,00 €	907,00 €			x
Wilms tumor, familial, due to BRCA2 mutation	BRCA2	600185	525,00 €	330,00 €	705,00 €			
Wilms tumor, IGF2 related, somatic	IGF2	147470					1.900,00 €	
Wilson disease	ATP7B	606882	1.582,00 €	328,00 €	1.760,00 €			x
Winchester Syndrome	MMP14	600754	729,00 €	429,00 €	1.008,00 €			x
Witkop syndrome	MSX1	142983	468,00 €	343,00 €	661,00 €			x
Wolcott-Rallison syndrome	EIF2AK3	604032	1.260,00 €					
Wolcott-Rallison syndrome	EIF2AK3	604032	1.260,00 €					
Wolff -Parkinson-White syndrome	PRKAG2	602743	1.127,00 €	424,00 €	1.401,00 €			x
Wolfram syndrome type 1	WFS1	606201	796,00 €	328,00 €	974,00 €			x
Wolfram syndrome type 1	WFS1	606201	796,00 €	328,00 €	974,00 €			x
Wolfram syndrome type 2	CISD2	611507	374,00 €	374,00 €	598,00 €			x
Wolfram syndrome type 2	CISD2	611507	374,00 €	374,00 €	598,00 €			x
Wolfram-like syndrome, autosomal dominant	WFS1	606201	796,00 €	328,00 €	974,00 €			x
Wolfram-like syndrome, autosomal dominant	WFS1	606201	796,00 €	328,00 €	974,00 €			x
Wolman disease	LIPA	613497	729,00 €	429,00 €	1.008,00 €			x
Wrinkly skin syndrome	ATP6V0A2	611716	1.278,00 €					
Xanthinuria type 1	XDH	607633	1.876,00 €					x
Xeroderma pigmentosum, group A	XPA	611153	573,00 €					
Xeroderma pigmentosum, group C	XPC	613208	1.326,00 €					
Xeroderma pigmentosum, group D	ERCC2	126340	1.193,00 €					
Xeroderma pigmentosum, group E, DDB-negative subtype	DDB2	600811	729,00 €					

Disease	Gene	OMIM gene	Full gene sequencing	Deletion/duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
Xeroderma pigmentosum, group F	ERCC4	133520	862,00 €					
Xeroderma pigmentosum, group G	ERCC5	133530	1.193,00 €					
Xeroderma pigmentosum, variant type	POLH	603968	928,00 €					x
XFE progeroid syndrome	ERCC4	133520	862,00 €					
XFE progeroid syndrome	ERCC4	133520	862,00 €					
Zellweger syndrome	PEX1	602136	1.643,00 €	505,00 €	1.998,00 €			x
Zellweger syndrome	PEX2	170993	468,00 €					x
Zellweger syndrome	PEX6	601498	1.193,00 €	424,00 €	1.467,00 €			
Zellweger syndrome	PEX10	602859	655,00 €					x
Zellweger syndrome	PEX12	601758	468,00 €	374,00 €	692,00 €			x
Zellweger syndrome	PEX13	601789	491,00 €	437,00 €	778,00 €			
Zellweger syndrome	PEX14	601791	729,00 €					
Zellweger syndrome	PEX16	603360	819,00 €					
Zellweger syndrome	PEX19	600279	655,00 €	437,00 €	942,00 €			x
Zellweger syndrome	PEX26	608666	491,00 €					x
ZIC5 related brain disorders	ZIC5		491,00 €					