CENTOGENE
THE RARE DISEASE COMPANY

Pricelist by gene

Centogene AG :: Schillingallee 68 :: 18057 Rostock :: Germany
Phone: +49 (0)381 203 652 222
E-mail: testing@centogene.com

Gene	Disease	OMIM gene	Full gene sequencing	Deletion/ duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
A2M	Alpha-2-macroglobulin deficiency	103950	1.802,00 €					
AAAS	Achalasia addisonianism alacrimia syndrome	605378	862,00 €	424,00 €	1.136,00 €			x
AAGAB	Keratoderma, palmoplantar, punctate type 1A	614888	729,00 €					
AARS2	Combined oxidative phosphorylation deficiency type 8	612035	1.127,00 €					
AARS	CMT2N	601065	1.326,00 €					x
AASS	Hyperlysinemia type 1	605113	1.326,00 €					
ABAT	GABA-transaminase deficiency	137150	1.061,00 €					
ABCA1	Coronary artery disease in familial hypercholesterolemia, protection against	600046	2.028,00 €					x
ABCA3	Surfactant metabolism dysfunction type 3	601615	1.622,00 €	695,00 €	2.167,00 €			x
ABCA4	Cone-rod dystrophy type 3	601691	2.301,00 €	655,00 €	2.806,00 €			x
ABCA12	Ichthyosis congenital, Harlequin fetus type	607800	2.106,00 €	827,00 €	2.783,00 €			x
ABCB1	Colchicine resistance	171050	1.572,00 €					
ABCB4	Cholestasis intrahepatic, of pregnancy, type 3	171060	1.704,00 €	328,00 €	1.882,00 €			x
ABCB6	Dyschromatosis universalis hereditaria type 3	605452	1.260,00 €					
ABCB7	Anemia, sideroblastic, with ataxia	300135	1.061,00 €					
ABCB11	Cholestasis benign recurrent intrahepatic type 2	603201	1.704,00 €	590,00 €	2.144,00 €			x
ABCC2	Dubin-Johnson syndrome	601107	1.673,00 €	674,00 €	2.197,00 €			x
ABCC6	Arterial calcification type 2, generalized, infantile	603234	1.572,00 €	328,00 €	1.750,00 €			x
ABCC8	Diabetes mellitus, noninsulin-dependent	600509	2.028,00 €	328,00 €	2.206,00 €			x
ABCC9	Atrial fibrillation type 12	601439	2.028,00 €	905,00 €	2.783,00 €			x
ABCD1	Adrenoleukodystrophy, x-linked	300371	862,00 €	328,00 €	1.040,00 €			x
ABCD4	Methylmalonic aciduria CblJ type	603214	1.278,00 €					x
ABHD1	Lung alpha-beta hydrolase deficiency type 1	612195	819,00 €					x
ABHD5	Chanarin-Dorfman syndrome	604780	655,00 €	437,00 €	942,00 €			x
ACACA	Acetyl-CoA carboxylase deficiency	200350	1.955,00 €					
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	604773	729,00 €					
ACAD9	Leigh syndrome and mitochondrial encephalopathy	611103	1.260,00 €					x
ACADL	LCAD deficiency	609576	796,00 €					
ACADM	Acyl-CoA medium-chain dehydrogenase deficiency	607008	862,00 €	426,00 €	1.138,00 €			x
ACADS	Acyl-CoA short-chain dehydrogenase deficiency	606885	819,00 €	429,00 €	1.098,00 €			x
ACADSB	2-methylbutyrylglycinuria	600301	796,00 €					
ACADVL	Acyl-CoA very long-chain dehydrogenase deficiency	609575	1.127,00 €	328,00 €	1.305,00 €			x
ACAT1	Methylacetoacetic aciduria	607809	862,00 €	421,00 €	1.133,00 €			x
ACD	Dyskeratosis congenita, autosomal recessive type 7	609377	995,00 €					
ACE	Renal tubular dysgenesis	106180	1.704,00 €	527,00 €	2.081,00 €			x
ACHE	Acetycholinesterase deficiency	100740	573,00 €					
ACO2	Cerebellar-retinal degeneration, infantile	100850	1.127,00 €					
ACOX1	Acyl-CoA peroxisomal oxidase deficiency	609751	796,00 €					
ACP2	Lysosomal acid phosphatase deficiency	171650	862,00 €					
ACP5	Spondyloenchondrodysplasia with immune dysregulation	171640	468,00 €					

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ACSF3	Combined malonic and methylmalonic aciduria	614245	729,00 €					
ACSL4	Mental retardation, X-linked type 63	300157	995,00 €					
ACTA1	Myopathy with fiber-type disproportion type 1	102610	655,00 €	437,00 €	942,00 €			x
ACTA2	Aortic aneurysm, familial thoracic type 6	102620	737,00 €	421,00 €	1.008,00 €			x
ACTB	Baraitser-Winter syndrome type 1	102630	573,00 €	421,00 €	844,00 €			x
ACTC1	Atrial septal defect type 5	102540	737,00 €					x
ACTG1	Baraitser-Winter syndrome type 2	102560	468,00 €	421,00 €	739,00 €			x
ACTG2	Visceral myopathy	102545	737,00 €	421,00 €	1.008,00 €			
ACTN1	Bleeding disorder, platelet-type 15	102575	1.399,00 €					
ACTN2	Cardiomyopathy, dilated type 1AA	102573	1.460,00 €					
ACTN4	Focal segmental glomerulosclerosis type 1	604638	1.399,00 €					
ACVR1	Fibrodysplasia ossificans progressiva	102576	819,00 €	429,00 €	1.098,00 €			x
ACVR2B	Heterotaxy, visceral type 4	602730	729,00 €	328,00 €	907,00 €			
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2	601284	819,00 €	328,00 €	997,00 €			x
ACY1	Aminoacylase deficiency	104620	796,00 €					
ADA	Severe combined immunodeficiency due to ADA deficiency	608958	796,00 €					
ADAM9	Cone-rod dystrophy type 9	602713	1.460,00 €					
ADAM17	Inflammatory skin and bowel disease, neonatal, type 1	603639	1.260,00 €					
ADAM22	Neurodevelopmental disorder, ADAM22 related	603709	1.673,00 €	328,00 €	1.851,00 €			
ADAMTS2	Ehlers-Danlos syndrome type 7C	604539	1.521,00 €	463,00 €	1.834,00 €			x
ADAMTS10	Weill-Marchesani syndrome - AR	608990	1.260,00 €	548,00 €	1.658,00 €			x
ADAMTS13	Thrombotic thrombocytopenic purpura	604134	1.572,00 €	611,00 €	2.033,00 €			x
ADAMTSL2	Geleophysic dysplasia type 1	612277	1.278,00 €	430,00 €	1.558,00 €			x
ADAMTSL4	Ectopia lentis et pupillae	610113	1.326,00 €	437,00 €	1.613,00 €			x
ADAR	Aicardi-Goutieres syndrome type 6	146920	1.326,00 €					
ADAT3	Mental retardation, autosomal recessive type 36	615302	374,00 €					
ADCK4	Nephrotic syndrome type 9	615567	819,00 €					
ADCY5	Dyskinesia, familial, with facial myokymia	600293	1.521,00 €	442,00 €	1.813,00 €			x
ADD2	Hypertension, ADD2 related	102681	1.127,00 €					
ADGRG1	Polymicrogyria bilateral frontoparietal	604110	995,00 €	445,00 €	1.290,00 €			x
ADGRV1	Febrile seizures, familial, type 4	602851	1.955,00 €	905,00 €	2.710,00 €			
ADK	Hypermethioninemia due to adenosine kinase deficiency	102750	862,00 €					
ADNP	Helsmoortel-van der Aa syndrome	611386	729,00 €					
ADSL	Adenylosuccinase deficiency	608222	995,00 €					
AFF2	Mental retardation, X-linked, associated with fragile site FRAXE	300806	1.704,00 €	328,00 €	1.882,00 €			x
AFF3	Rheumatoid arthritis, susceptibility to	601464	1.460,00 €					
AFG3L2	Spastic ataxia type 5, autosomal recessive	604581	1.127,00 €	424,00 €	1.401,00 €			x
AGA	Aspartylglucosaminuria	613228	819,00 €					
AGK	Cataract, autosomal recessive type 38	610345	1.061,00 €	424,00 €	1.335,00 €			x
AGL	Glycogen storage disease type 3	610860	1.622,00 €	716,00 €	2.188,00 €			x

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AGPAT2	Lipodystrophy generalized type 1	603100	573,00 €	421,00 €	844,00 €			x
AGPS	Rhizomelic chondrodysplasia punctata type 3	603051	1.521,00 €					
AGRN	Myasthenic syndrome, congenital	103320	2.028,00 €	821,00 €	2.699,00 €			
AGT	Renal tubular dysgenesis	106150	655,00 €					
AGTR1	Renal tubular dysgenesis	106165	491,00 €					
AGTR2	Mental retardation, X-linked type 88, AGTR2 related	300034	491,00 €					
AGXT	Hyperoxaluria type 1	604285	796,00 €	328,00 €	974,00 €			x
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	180960	729,00 €					
AHDC1	Mental retardation, autosomal dominant type 25	615790	1.260,00 €					
AHI1	Joubert syndrome type 3	608894	1.572,00 €					
AHNAK2	Autism spectrum disorder	608570	2.016,00 €					
AICDA	Immunodeficiency type 2, with hyper-IgM	605257	468,00 €					
AIFM1	CMTX4	300169	1.193,00 €	424,00 €	1.467,00 €			x
AIMP1	Leukodystrophy hypomyelinating type 3	603605	655,00 €	437,00 €	942,00 €			x
AIP	Pituitary adenoma, ACTH-secreting, due to AIP germline mutation	605555	573,00 €	328,00 €	751,00 €			x
AIPL1	Cone-rod dystrophy	604392	655,00 €	328,00 €	833,00 €			x
AIRE	Autoimmune polyendocrinopathy syndrome type 1	607358	995,00 €	437,00 €	1.282,00 €			x
AK2	Reticular dysgenesis	103020	491,00 €					
AKAP1	Mitochondrial Disorders, AKAP1 related	602449	995,00 €	429,00 €	1.274,00 €			x
AKAP9	Long QT syndrome type 11	604001	2.141,00 €	905,00 €	2.896,00 €			
AKR1C4	46,XY sex reversal type 8, modifier of	600451	819,00 €					
AKR1D1	Bile acid synthesis defect type 2, congenital	604741	819,00 €					
AKT1	Cowden syndrome type 6	164730	995,00 €	445,00 €	1.290,00 €			x
AKT2	Diabetes mellitus, noninsulin-dependent	164731	796,00 €					
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome type 2	611223	995,00 €					
ALAS2	Anemia, sideroblastic, X-linked	301300	796,00 €					
ALDH1A2	Tetralogy of Fallot	603687	862,00 €					
ALDH1A3	Microphthalmia syndromic type 8	600463	928,00 €	426,00 €	1.204,00 €			x
ALDH3A2	Sjogren-Larsson syndrome	609523	796,00 €	429,00 €	1.075,00 €			x
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	610045	862,00 €					
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency	603178	862,00 €					
ALDH7A1	Pyridoxine-dependent epilepsy	107323	1.278,00 €	421,00 €	1.549,00 €			x
ALDH18A1	Cutis laxa type 3A, autosomal recessive	138250	1.127,00 €					
ALDOA	Glycogen storage disease type 12	103850	737,00 €					
ALDOB	Fructose intolerance	612724	737,00 €	328,00 €	915,00 €			x
ALG1	Glycosylation disorder type 1K	605907	862,00 €	426,00 €	1.138,00 €			
ALG2	Glycosylation disorder type 1I	607905	468,00 €					
ALG3	Glycosylation disorder type 1D	608750	655,00 €					
ALG6	Glycosylation disorder type 1C	604566	995,00 €					
ALG8	Glycosylation disorder type 1H	608103	862,00 €					

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ALG9	Glycosylation disorder type 1L	606941	928,00 €					
ALG11	Congenital disorder of glycosylation, type Ip	613666	573,00 €					
ALG12	Glycosylation disorder type 1G	607144	819,00 €					
ALG13	Glycosylation disorder type 1S	300776	1.764,00 €					
ALK	Neuroblastoma type 3, susceptibility to, familial	105590	1.673,00 €					
ALMS1	Alstrom syndrome	606844	2.028,00 €	484,00 €	2.362,00 €			x
ALOX12B	Ichthyosis, congenital, autosomal recessive, type 2	603741	995,00 €	445,00 €	1.290,00 €			x
ALOXE3	Ichthyosiform erythroderma, congenital, nonbullous type 1	607206	995,00 €	424,00 €	1.269,00 €			
ALPL	Hypophosphatasia, adult	171760	862,00 €	421,00 €	1.133,00 €			x
ALS2	Amyotrophic lateral sclerosis type 2, juvenile	606352	1.724,00 €	716,00 €	2.290,00 €			x
ALX3	Frontonasal dysplasia type 1	606014	468,00 €	328,00 €	646,00 €			x
ALX4	Frontonasal dysplasia type 2	605420	573,00 €	328,00 €	751,00 €			x
AMACR	Alpha-methylacyl CoA racemase deficiency	604489	655,00 €	421,00 €	926,00 €			x
AMBN	Amelogenesis imperfecta type 1F	601259	729,00 €					
AMELX	Amelogenesis imperfecta type 1E	300391	491,00 €					
AMER1	Osteopathia striata with cranial sclerosis	300647	796,00 €	343,00 €	989,00 €			x
AMH	Persistent Mullerian duct syndrome type 1	600957	573,00 €	390,00 €	813,00 €			x
AMHR2	Persistent Mullerian duct syndrome type 2	600956	995,00 €	429,00 €	1.274,00 €			
AMN	Megaloblastic anemia type 1	605799	819,00 €	421,00 €	1.090,00 €			x
AMPD1	Myopathy due to myoadenylate deaminase deficiency	102770	1.127,00 €	424,00 €	1.401,00 €			x
AMPD2	Pontocerebellar hypoplasia, type 9	102771	1.278,00 €					
AMPD3	AMP deaminase deficiency, erythrocytic	102772	1.127,00 €					
AMT	Glycine encephalopathy	238310	819,00 €	328,00 €	997,00 €			x
AMTN	Amelotin deficiency	610912	737,00 €					
ANG	Amyotrophic lateral sclerosis type 9	105850	386,00 €					x
ANK1	Spherocytosis type 1	612641	1.973,00 €					
ANK2	Long QT syndrome type 4	106410	2.176,00 €	772,00 €	2.798,00 €			x
ANK3	Mental retardation, autosomal recessive type 37	600465	1.955,00 €					x
ANKH	Chondrocalcinosis type 2	605145	796,00 €					
ANKRD11	KBG syndrome	611192	1.622,00 €	437,00 €	1.909,00 €			x
ANKRD26	Thrombocytopenia type 2	610855	1.876,00 €					
ANKS3	Autism spectrum disorder		995,00 €					
ANKS6	Nephronophthisis type 16	615370	1.127,00 €					
ANLN	Focal segmental glomerulosclerosis type 8	616027	1.521,00 €					
ANO3	DYT24	610110	1.764,00 €	590,00 €	2.204,00 €			
ANO5	Gnathodiaphyseal dysplasia	608662	1.338,00 €	328,00 €	1.516,00 €			x
ANO10	Spinocerebellar ataxia type 10, autosomal recessive	613726	995,00 €					x
ANOS1	Kallmann syndrome type 1	300836	995,00 €	328,00 €	1.173,00 €			x
ANTXR1	Hemangioma capillary infantile	606410	1.399,00 €	421,00 €	1.670,00 €			
ANTXR2	Hyaline fibromatosis syndrome	608041	1.127,00 €	424,00 €	1.401,00 €			x

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AP1S1	MEDNIK syndrome	603531	491,00 €					
AP1S2	Mental retardation, X-linked type 59	300629	491,00 €					x
AP2S1	Hypocalciuric hypercalcemia, familial type 3	602242	573,00 €					
AP3B1	Hermansky-Pudlak syndrome type 2	603401	1.825,00 €					
AP4B1	SPG47	607245	862,00 €					
AP4E1	SPG51	607244	1.260,00 €					
AP4M1	Microcephaly, AP4M1 related	602296	729,00 €					
AP4S1	SPG52	607243	655,00 €					
AP5Z1	SPG48	613653	1.260,00 €					
APC2	Neurodevelopmental disorder, APC2-related	612034	1.825,00 €	445,00 €	2.120,00 €			
APC	Adenoma, periampullary, somatic	611731					1.900,00 €	
APCDD1	Hypotrichosis type 1	607479	655,00 €					
APOA1	Amyloidosis, familial visceral	107680	491,00 €	437,00 €	778,00 €			
APOA5	Hyperchylomicronemia type 5	606368	737,00 €					
APOB	Hypercholesterolemia type B autosomanl dominant	107730	1.989,00 €	611,00 €	2.450,00 €			x
APOC2	Apolipoprotein C-II deficiency	608083	386,00 €	437,00 €	673,00 €			x
APOE	Alzheimer disease type 2	107741	655,00 €	437,00 €	942,00 €			
APOL1	Focal segmental glomerulosclerosis type 4, susceptibility to	603743	655,00 €					
APOPT1	Mitochondrial complex IV deficiency	616003	655,00 €					
APP	Alzheimer disease type 1	104760	1.278,00 €	328,00 €	1.456,00 €			x
APRT	Adenine phosphoribosyltransferase deficiency	102600	491,00 €					
APTX	Ataxia-oculomotor apraxia type 1	606350	819,00 €	328,00 €	997,00 €			x
AQP2	Diabetes insipidus, nephrogenic, autosomal	107777	468,00 €	437,00 €	755,00 €			x
AR	Androgen insensitivity	313700	1.061,00 €	328,00 €	1.239,00 €	200,00 €		x
ARFGEF2	Periventricular heterotopia with microcephaly	605371	1.759,00 €	821,00 €	2.430,00 €			x
ARG1	Arginase deficiency	608313	655,00 €					
ARHGAP31	Adams-Oliver syndrome type 1	610911	1.326,00 €	421,00 €	1.597,00 €			x
ARHGDIA	Nephrotic syndrome type 8	601925	573,00 €					
ARHGEF6	Mental retardation, X-linked type 46	300267	1.338,00 €					
ARHGEF9	Early infantile epileptic encephalopathy type 8	300429	796,00 €					
ARHGEF10	Slowed nerve conduction velocity, autosomanal dominant	608136	1.622,00 €					x
ARID1A	Endometrioid carcinoma, ARID1A related, somatic	603024					1.900,00 €	
ARID1B	Mental retardation, autosomal dominant type 12	614556	1.460,00 €	328,00 €	1.638,00 €			
ARL6IP1	SPG61	607669	573,00 €					
ARL6	Bardet-Biedl syndrome type 3	608845	655,00 €					
ARL11	Tumor predisposition syndrome, ARL11 related	609351	386,00 €					
ARL13B	Joubert syndrome type 8	608922	796,00 €					
ARMC4	Primary ciliary dyskinesia type 23	615408	1.278,00 €					
ARMS2	Macular degeneration, age-related type 8, association with	611313	386,00 €	328,00 €	564,00 €			
ARNT2	Webb-Dattani syndrome	606036	1.326,00 €					

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ARSA	Metachromatic Leukodystrophy	607574	819,00 €	437,00 €	1.106,00 €			x
ARSB	Mucopolysaccharidosis type 6	611542	729,00 €	437,00 €	1.016,00 €			x
ARSE	Chondrodysplasia punctata, X-linked recessive	300180	862,00 €					
ARSI	SPG66, ARSI related	610009	468,00 €					
ARX	Corpus callosum, agenesis of, with abnormal genitalia	300382	737,00 €	328,00 €	915,00 €			x
ASAH1	Farber disease	613468	1.061,00 €	437,00 €	1.348,00 €			x
ASB10	Glaucoma, open angle type 1F	615054	573,00 €					
ASCL1	Central hypoventilation syndrome, congenital	100790	386,00 €	328,00 €	564,00 €			x
ASL	Argininosuccinic aciduria	608310	862,00 €	424,00 €	1.136,00 €			x
ASNS	Asparaginesynthetase deficiency	108370	796,00 €					
ASPA	Canavan disease	608034	573,00 €	328,00 €	751,00 €			x
ASPM	Microcephaly, autosomal recessive type 5	605481	2.067,00 €	328,00 €	2.245,00 €			x
ASS1	Citrullinemia	603470	995,00 €	424,00 €	1.269,00 €			x
ASXL1	Bohring-Opitz syndrome	612990	1.399,00 €	426,00 €	1.675,00 €			
ASXL3	Bainbridge-Ropers syndrome	615115	1.572,00 €					
ATL1	Neuropathy, hereditary sensory, type ID	606439	1.061,00 €	328,00 €	1.239,00 €			x
ATM	Ataxia-telangiectasia	607585	1.955,00 €	655,00 €	2.460,00 €			
ATN1	Dentatorubral-pallidoluysian atrophy	607462	1.193,00 €			200,00 €		x
ATOH7	Retinal nonattachment nonsyndromic congenital	609875	386,00 €					
ATP1A2	Alternating hemiplegia of childhood type 1	182340	1.399,00 €	328,00 €	1.577,00 €			x
ATP1A3	Alternating hemiplegia of childhood type 2	182350	1.326,00 €	328,00 €	1.504,00 €			x
ATP1B4	Autism, ATP1B4 related		737,00 €					
ATP2A1	Brody myopathy	108730	1.399,00 €	463,00 €	1.712,00 €			x
ATP2B3	Spinocerebellar ataxia type 1, X-linked	300014	1.278,00 €					
ATP5A1	Combined oxidative phosphorylation deficiency type 22	164360	729,00 €					
ATP5E	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3	606153	386,00 €					
ATP6AP2	Mental retardation, X-linked with epilepsy	300556	819,00 €					
ATP6V0A2	Cutis laxa type 2A, autosomal recessive	611716	1.278,00 €					
ATP6V0A4	Renal tubular acidosis, distal, autosomal recessive	605239	1.399,00 €	463,00 €	1.712,00 €			x
ATP6V1B1	Renal tubular acidosis with deafness	192132	928,00 €	437,00 €	1.215,00 €			x
ATP7A	Menkes disease	300011	1.704,00 €	328,00 €	1.882,00 €			x
ATP7B	Wilson disease	606882	1.582,00 €	328,00 €	1.760,00 €			x
ATP8A2	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome type 4	605870	1.927,00 €	905,00 €	2.682,00 €			
ATP8B1	Cholestasis progressive intrahepatic type 1	602397	1.521,00 €	590,00 €	1.961,00 €			x
ATP10A	Autism/Mental retardation/Angelman syndrome, susceptibility to, ATP10A related	605855	1.460,00 €					
ATP13A2	PARK9 Parkinson	610513	1.764,00 €	611,00 €	2.225,00 €			x
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	608918	737,00 €					
ATR	Cutaneous telangiectasia and cancer syndrome, familial	601215	1.955,00 €					x
ATRIP	Seckel syndrome	606605	995,00 €					
ATRX	Alpha-thalassemia/mental retardation syndrome	300032	1.845,00 €	328,00 €	2.023,00 €			x

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E-mail: testing@centogene.com

Gene	Disease	OMIM gene	Full gene sequencing	Deletion/ duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
ATXN1	Spinocerebellar ataxia type 1, autosomal dominant	601556				200,00 €		x
ATXN2	Spinocerebellar ataxia type 2, autosomal dominant	601517				200,00 €		x
ATXN3	Spinocerebellar ataxia type 3, autosomal dominant	607047				200,00 €		x
ATXN7	Spinocerebellar ataxia type 7, autosomal dominant	607640				200,00 €		x
ATXN8OS	Spinocerebellar ataxia type 8, autosomal dominant	613289				200,00 €		x
ATXN10	Spinocerebellar ataxia type 10, autosomal dominant	611150				200,00 €		x
AUH	3-methylglutaconic aciduria type 1	600529	729,00 €	429,00 €	1.008,00 €			x
AURKC	SPGF5	603495	655,00 €					
AVP	Diabetes insipidus, neurohypophyseal	192340	386,00 €	374,00 €	610,00 €			
AVPR1A	Autism, AVPR1A related	600821	573,00 €					
AVPR2	Diabetes insipidus, nephrogenic, X-linked	300538	491,00 €	374,00 €	715,00 €			x
AZF region	Azoospermia induced by Y chromosome microdeletions			328,00 €				
B3GALNT2	Congenital muscular dystrophy and hypoglycosylation of α-dystroglycan	610194	1.061,00 €					
B3GALT6	Ehlers-Danlos syndrome, progeroid type, type 2	615291	468,00 €					
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	606374	655,00 €					x
B3GLCT	Peters-Plus syndrome	610308	1.061,00 €	445,00 €	1.356,00 €			x
B4GALNT1	SPG26	601873	819,00 €					
B4GALT1	Glycosylation disorder type 2D	137060	655,00 €					
B4GALT7	Ehlers-Danlos syndrome, progeroid type 1	604327	491,00 €	421,00 €	762,00 €			x
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A13	605517	374,00 €					
B9D1	Meckel syndrome type 9	614144	796,00 €					
B9D2	Meckel syndrome type 10	611951	468,00 €					
BAAT	Hypercholanemia	602938	468,00 €					
BAG3	Cardiomyopathy, dilated type 1HH	603883	737,00 €	328,00 €	915,00 €			x
BAP1	Tumor predisposition syndrome	603089	1.127,00 €	328,00 €	1.305,00 €			
BARD1	Breast cancer, susceptibility to	601593	928,00 €					
BBS1	Bardet-Biedl syndrome type 1	209901	928,00 €					x
BBS2	Bardet-Biedl syndrome type 2	606151	1.127,00 €	424,00 €	1.401,00 €			x
BBS4	Bardet-Biedl syndrome type 4	600374	1.127,00 €					
BBS5	Bardet-Biedl syndrome type 5	603650	928,00 €	421,00 €	1.199,00 €			x
BBS7	Bardet-Biedl syndrome type 7	607590	1.260,00 €					
BBS9	Bardet-Biedl syndrome type 9	607968	1.338,00 €					
BBS10	Bardet-Biedl syndrome type 10	610148	573,00 €					x
BBS12	Bardet-Biedl syndrome type 12	610683	819,00 €					
BCAP31	Deafness, dystonia, and cerebral hypomyelination, X-linked	300398	737,00 €					
BCAT1	Branched-chain aminotransferase 1 deficiency	113520	862,00 €					
BCAT2	Branched-chain aminotransferase 2 deficiency	113530	796,00 €					
BCHE	Butyrylcholinesterase deficiency	177400	819,00 €	437,00 €	1.106,00 €			
BCKDHA	Maple syrup urine disease type 1a	608348	819,00 €	421,00 €	1.090,00 €			x
BCKDHB	Maple syrup urine disease type 1b	248611	729,00 €	429,00 €	1.008,00 €			x

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BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency	614901	737,00 €					
BCL6	Lymphoma, B-cell type	109565					1.900,00 €	
BCL9L	Heterotaxy, visceral, BCL9L related	609004	1.061,00 €					
BCL10	Lymphoma, follicular, somatic	603517					1.900,00 €	
BCOR	Microphthalmia syndromic type 2	300485	1.338,00 €	445,00 €	1.633,00 €			
BCS1L	Bjornstad syndrome	603647	819,00 €	421,00 €	1.090,00 €			x
BDNF	Central hypoventilation syndrome, congenital	113505	573,00 €	328,00 €	751,00 €			
BEAN1	Spinocerebellar ataxia type 31, autosomal dominant	612051				200,00 €		
BEST1	Bestrophinopathy	607854	928,00 €	328,00 €	1.106,00 €			x
BEST2	Macular dystrophy, BEST2-related	607335	729,00 €					
BEST3	Macular dystrophy, BEST3-related	607337	928,00 €					
BEST4	Macular dystrophy, BEST4-related	607336	655,00 €					
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction	615416	374,00 €	296,00 €	520,00 €			
BICC1	Renal cystic dysplasia, cystic, susceptibility to	614295	1.399,00 €					x
BICD2	Spinal muscular atrophy, lower extremity, autosomal dominant, type 2	609797	819,00 €					
BIN1	Myopathy, centronuclear	601248	1.399,00 €					
BLK	Maturity-onset diabetes of the young type 11	191305	862,00 €					
BLM	Bloom syndrome	604610	1.460,00 €	484,00 €	1.794,00 €			x
BLNK	Agammaglobulinemia type 4, autosomal recessive	604515	1.193,00 €					
BLOC1S3	Hermansky-Pudlak syndrome type 8	609762	655,00 €					
BMP1	Osteogenesis imperfecta type 13	112264	1.127,00 €	437,00 €	1.414,00 €			
BMP2	Brachydactyly type A2	112261	468,00 €					
BMP4	Microphthalmia syndromic type 6	112262	655,00 €	328,00 €	833,00 €			x
BMP15	Ovarian dysgenesis type 2	300247	468,00 €					
BMPR1A	Juvenile polyposis syndrome	601299	819,00 €	328,00 €	997,00 €			x
BMPR1B	Brachydactyly type A2	603248	796,00 €					
BMPR2	Pulmonary hypertension, primary type	600799	1.193,00 €	328,00 €	1.371,00 €			x
BOLA3	Multiple mitochondrial dysfunctions syndrome type 2	613183	468,00 €					
BPIFA3	Autism spectrum disorder		655,00 €					
BRAF	Adenocarcinoma of lung, somatic	164757					1.290,00 €	
BRCA1	Breast-ovarian cancer	113705	525,00 €	330,00 €	705,00 €			
BRCA2	Breast cancer, male, susceptibility to	600185	525,00 €	330,00 €	705,00 €			
BRIP1	Fanconi anemia type J	605882	1.399,00 €	328,00 €	1.577,00 €			x
BRWD3	Mental retardation, X-linked type 93	300553	1.888,00 €	704,00 €	2.442,00 €			x
BSCL2	Encephalopathy, progressive, with or without lipodystrophy	606158	819,00 €	429,00 €	1.098,00 €			x
BSND	Bartter syndrome type 4a	606412	468,00 €					
BTD	Biotinidase deficiency	609019	729,00 €	437,00 €	1.016,00 €			x
BTK	Agammaglobulinemia and isolated hormone deficiency	300300	1.338,00 €	328,00 €	1.516,00 €			x
C1QA	C1q deficiency	120550	374,00 €					
C1QTNF5	Retinal degeneration, late-onset, autosomal dominant	608752	374,00 €					

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C2CD3	Orofaciodigital syndrome type 14	615944	1.775,00 €					
C2ORF71	Retinitis pigmentosa type 54, autosomal recessive	613425	1.061,00 €					
C2	C2 deficiency	613927	1.061,00 €					
C3	C3 deficiency	120700	1.622,00 €	704,00 €	2.176,00 €			x
C4BPA	Pregnancy loss, recurrent, C4BPA related	120830	862,00 €					
C4orf26	Amelogenesis imperfecta type 2A4	614829	468,00 €					
C5orf42	Joubert syndrome type 17	614571	2.223,00 €	905,00 €	2.978,00 €			x
C5	C5 deficiency	120900	1.845,00 €	905,00 €	2.600,00 €			
C7orf43	Autism, C7orf43 related		729,00 €					
C7	C7 deficiency	217070	1.260,00 €					
C8ORF37	Retinitis pigmentosa type 64, autosomal recessive	614477	819,00 €					
C9orf72	Amyotrophic lateral sclerosis with frontotemporal dementia	614260				200,00 €		
C10ORF11	Albinism, oculocutaneous type 5	614537	819,00 €					x
C12orf57	Temtamy syndrome	615140	374,00 €					
C12ORF65	Combined oxidative phosphorylation deficiency type 7	613541	386,00 €					
C15orf41	Dyserythropoietic anemia, congenital, type 1B	615626	862,00 €	421,00 €	1.133,00 €			x
C19orf12	Neurodegeneration with brain iron accumulation type 4	614297	374,00 €	374,00 €	598,00 €			x
C21orf2	Cone-rod dystrophy, C21orf2 related	603191	737,00 €					
C21orf59	Primary ciliary dyskinesia type 26	615494	655,00 €					
CA2	Osteopetrosis, autosomal recessive type 3	611492	655,00 €	437,00 €	942,00 €			
CA4	Retinitis pigmentosa type 17, autosomal dominant	114760	655,00 €					
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion type 3	114815	819,00 €					
CABP2	Deafness, autosomal recessive type 93	614899	655,00 €					
CABP4	Night blindness, congenital stationary type 2B	608965	491,00 €	421,00 €	762,00 €			x
CACNA1A	Episodic ataxia type 2	601011	2.067,00 €	328,00 €	2.245,00 €	200,00 €		x
CACNA1B	DYT23	601012	1.989,00 €					
CACNA1C	Brugada syndrome type 3	114205	2.106,00 €	905,00 €	2.861,00 €			
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities	114206	2.145,00 €	905,00 €	2.900,00 €			
CACNA1F	Aland Island eye disease	300110	1.802,00 €					
CACNA1H	Epilepsy, childhood absence type 6, susceptibility to	607904	1.977,00 €					
CACNA1S	Hypokalemic periodic paralysis type 1	114208	1.888,00 €	328,00 €	2.066,00 €			x
CACNA2D4	Retinal cone dystrophy type 4	608171	2.028,00 €					
CACNB2	Brugada syndrome type 4	600003	1.338,00 €					
CACNB4	Episodic ataxia type 5	601949	1.127,00 €	437,00 €	1.414,00 €			
CALCRL	Vascular system defects due to CALCRL deficiency	114190	819,00 €					
CALM1	Ventricular tachycardia, catecholaminergic polymorphic type 4	114180	729,00 €	421,00 €	1.000,00 €			
CALM2	Long QT syndrome type 15	114182	655,00 €	437,00 €	942,00 €			x
CALR3	Cardiomyopathy, familial hypertrophic type 19	611414	796,00 €					
CALR	CALR, selective sequencing of exon 9	109091						
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation	611501	1.643,00 €	484,00 €	1.977,00 €			

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CANT1	Desbuquois dysplasia type 1	613165	491,00 €					
CAPN1	Muscular-skeletal disorder, CAPN1 related	114220	1.338,00 €					
CAPN3	Limb-girdle muscular dystrophy, autosomal recessive type 2A	114240	1.460,00 €	328,00 €	1.638,00 €			x
CARD11	B-cell expansion with NFKB and T-cell anergy	607210	1.521,00 €					
CARD14	Pityriasis rubra pilaris	607211	1.338,00 €					
CASC5	Microcephaly, autosomal recessive type 4	609173	2.028,00 €					
CASK	FG syndrome type 4	300172	1.704,00 €	328,00 €	1.882,00 €			x
CASP8	Autoimmune lymphoproliferative syndrome type 2B	601763	729,00 €					
CASP10	Autoimmune lymphoproliferative syndrome type 2A	601762	862,00 €					
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic type 2	114251	796,00 €					
CASR	Hyperparathyroidism, neonatal severe	601199	796,00 €	328,00 €	974,00 €			x
CATSPER1	SPGF7	606389	796,00 €					
CATSPER2	Deafness and male infertility, CATSPER2 related	607249	862,00 €					
CAV3	Cardiomyopathy, familial hypertrophic	601253	386,00 €	328,00 €	564,00 €			x
CBFB	Skeletal abnormalities, CBFB related	121360	491,00 €					
CBL	Juvenile myelomonocytic leukemia, due to CBL germline mutation	165360	1.127,00 €	424,00 €	1.401,00 €			x
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	613381	1.127,00 €	424,00 €	1.401,00 €			x
CC2D1A	Mental retardation, autosomal recessive type 3	610055	1.673,00 €					
CC2D2A	COACH syndrome	612013	1.977,00 €	800,00 €	2.627,00 €			x
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome type 1	612753	796,00 €					
CCDC8	Three M syndrome type 3	614145	491,00 €					
CCDC28B	Bardet-Biedl syndrome, modifier of, CCDC28B related	610162	491,00 €					x
CCDC39	Primary ciliary dyskinesia type 14	613798	1.338,00 €					
CCDC40	Primary ciliary dyskinesia type 15	613799	1.338,00 €					
CCDC50	Deafness, autosomal dominant type 44	611051	862,00 €					x
CCDC65	Primary ciliary dyskinesia type 27	611088	737,00 €					
CCDC78	Centronuclear myopathy type 4	614666	862,00 €					
CCDC88C	Hydrocephalus, nonsyndromic, autosomal recessive type 1	611204	1.572,00 €					
CCDC103	Primary ciliary dyskinesia type 17	614677	491,00 €					
CCDC114	Primary ciliary dyskinesia type 20	615038	1.061,00 €					
CCM2	Cerebral cavernous malformations type 2	607929	796,00 €	328,00 €	974,00 €			
CCND1	Colorectal cancer, hereditary, susceptibility to	168461	491,00 €	328,00 €	669,00 €			
CCNO	Primary ciliary dyskinesia type 29	607752	468,00 €					
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia	610150	819,00 €					
CD2AP	Focal segmental glomerulosclerosis type 3	604241	1.278,00 €					
CD3D	Immunodeficiency type 19	186790	491,00 €					
CD3E	Immunodeficiency type 18	186830	737,00 €					
CD3G	Cone-rod dystrophy type 17	186740	573,00 €					
CD19	Immunodeficiency common variable type 3	107265	862,00 €					
CD27	Lymphoproliferative syndrome type 2	186711	491,00 €					

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CD36	Platelet glycoprotein IV deficiency	173510	928,00 €	437,00 €	1.215,00 €			
CD40LG	Immunodeficiency, X-linked with hyper-IgM	300386	655,00 €	390,00 €	895,00 €			x
CD40	Immunodeficiency type 3, with hyper-IgM	109535	819,00 €					
CD46	Hemolytic uremic syndrome, atypical type 2, susceptibility to	120920	928,00 €	328,00 €	1.106,00 €			x
CD59	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	107271	374,00 €					
CD79A	Agammaglobulinemia type 3, autosomal recessive	112205	374,00 €					
CD79B	Agammaglobulinemia type 6, autosomal recessive	147245	468,00 €					
CD81	Immunodeficiency common variable type 6	186845	737,00 €					
CD96	C syndrome	606037	1.061,00 €	445,00 €	1.356,00 €			x
CD247	Immunodeficiency type 25	186780	737,00 €					
CD320	Methylmalonic aciduria CblR type	606475	491,00 €	390,00 €	731,00 €			x
CDAN1	Anemia dyserythropoietic type 1A	607465	1.825,00 €	590,00 €	2.265,00 €			x
CDC20	Cell cycle disorder, CDC20 related	603618	655,00 €					
CDC73	Hyperparathyroidism type 1, familial	607393	1.127,00 €	328,00 €	1.305,00 €			
CDH1	Endometrial carcinoma, somatic	192090					1.900,00 €	
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy	114021	1.193,00 €					
CDH23	Deafness, autosomal recessive, type 12	605516	1.955,00 €					x
CDHR1	Cone-rod dystrophy type 15	609502	1.260,00 €					
CDK4	Melanoma, cutaneous malignant, familial, CDK4 related	123829	573,00 €	328,00 €	751,00 €			
CDK5RAP2	Microcephaly, autosomal recessive type 3	608201	1.802,00 €	328,00 €	1.980,00 €			x
CDK6	Microcephaly, autosomal recessive type 12	603368	655,00 €					
CDKL5	Angelman-like syndrome	300203	1.460,00 €	328,00 €	1.638,00 €			x
CDKN1B	Multiple endocrine neoplasia type 4	600778	386,00 €	328,00 €	564,00 €			
CDKN1C	Beckwith-Wiedemann syndrome	600856	374,00 €	328,00 €	552,00 €			x
CDKN2A	Melanoma and neural system tumor syndrome, familial	600160	737,00 €	328,00 €	915,00 €			
CDKN2B	Multiple endocrine neoplasia type 1, CDKN2B related	600431	729,00 €	328,00 €	907,00 €			
CDON	Holoprosencephaly type 11	608707	1.399,00 €					
CDSN	Hypotrichosis type 2	602593	573,00 €					x
CDT1	Meier-Gorlin syndrome 4	605525	729,00 €					
CEACAM16	Deafness, autosomal dominant type 4B	614591	655,00 €					
CEBPA	Leukemia, acute myeloid, somatic	116897					1.900,00 €	
CECR1	Polyarteritis nodosa, childhood-onset	607575	729,00 €	429,00 €	1.008,00 €			x
CEL	Maturity-onset diabetes of the young type 8	114840	862,00 €	328,00 €	1.040,00 €			
CELF6	Autism, CELF6 related	612681	862,00 €					
CELSR2	Schizophrenia, CELSR2 related	604265	1.973,00 €					
CENPE	Microcephaly, autosomal recessive type 13	117143	1.973,00 €					
CENPJ	Microcephaly, autosomal recessive type 6	609279	1.643,00 €	328,00 €	1.821,00 €			x
CEP41	Joubert syndrome type 15	610523	862,00 €					
CEP57	Mosaic variegated aneuploidy syndrome type 2	607951	796,00 €					
CEP63	Microcephaly, CEP63 related	614724	1.193,00 €					

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CEP135	Microcephaly, autosomal recessive type 8	611423	1.521,00 €	548,00 €	1.919,00 €			x
CEP152	Microcephaly, autosomal recessive type 9	613529	1.764,00 €					
CEP164	Nephronophthisis type 15	614848	1.724,00 €	695,00 €	2.269,00 €			x
CEP290	Bardet-Biedl syndrome type 14	610142	2.262,00 €	842,00 €	2.954,00 €			x
CERKL	Retinitis pigmentosa type 26, autosomal recessive	608381	995,00 €	437,00 €	1.282,00 €			x
CERS1	Progressive myoclonus epilepsy type 8	606919	737,00 €					
CERS3	Ichthyosis, congenital, autosomal recessive, type 9	615276	796,00 €					
CFAP53	Heterotaxy, visceral type 6	614759	737,00 €					
CFB	Hemolytic uremic syndrome	138470	1.260,00 €					
CFC1	Heterotaxy, visceral type 2	605194	573,00 €	328,00 €	751,00 €			
CFH	Hemolytic uremic syndrome	134370	1.399,00 €	328,00 €	1.577,00 €			x
CFHR1	Hemolytic uremic syndrome	134371	819,00 €	328,00 €	997,00 €			x
CFHR2	Hemolytic uremic syndrome	600889	491,00 €	328,00 €	669,00 €			x
CFHR3	Hemolytic uremic syndrome	605336	573,00 €	328,00 €	751,00 €			x
CFHR4	Hemolytic uremic syndrome	605337	729,00 €					
CFHR5	Hemolytic uremic syndrome	608593	729,00 €	328,00 €	907,00 €			
CFI	Hemolytic uremic syndrome	217030	928,00 €	328,00 €	1.106,00 €			x
CFL1	Corticobasal Degeneration, CFL1 related	601442	468,00 €					
CFL2	Nemaline myopathy type 7	601443	491,00 €					
CFTR	Congenital bilateral absence of vas deferens	602421	950,00 €	328,00 €	1.128,00 €			x
CHAT	Myasthenic syndrome, congenital	118490	1.061,00 €	445,00 €	1.356,00 €			x
CHD2	Epileptic encephalopathy, childhood-onset	602119	1.802,00 €	905,00 €	2.557,00 €			
CHD7	CHARGE syndrome	608892	2.028,00 €	328,00 €	2.206,00 €			x
CHEK2	Li-Fraumeni syndrome type 2	604373	1.061,00 €	328,00 €	1.239,00 €			
CHGB	Amyotrophic lateral sclerosis risk factor	118920	729,00 €					
CHKB	Muscular dystrophy, congenital, megaconial type	612395	737,00 €					
CHM	Choroideremia	300390	1.193,00 €	328,00 €	1.371,00 €			x
CHMP1A	Pontocerebellar hypoplasia type 8	164010	655,00 €					x
CHMP2B	Amyotrophic lateral sclerosis type 17	609512	573,00 €					
CHN1	Duane Retraction syndrome	118423	928,00 €					
chr. 7q11.23	Williams-Beuren syndrome			328,00 €				
chr. 11p15	Beckwith-Wiedemann syndrome			328,00 €				x
chr. 15q11	Angelman syndrome			328,00 €				x
chr. 22q13.3	Phelan-McDermid syndrome			328,00 €				x
CHRDL1	Megalocornea, X-linked	300350	796,00 €	421,00 €	1.067,00 €			x
CHRM3	Prune belly syndrome	118494	491,00 €					
CHRNA1	Multiple pterygium syndrome lethal type	100690	862,00 €	421,00 €	1.133,00 €			x
CHRNA2	Epilepsy, nocturnal frontal lobe type 4	118502	737,00 €	437,00 €	1.024,00 €			x
CHRNA4	Epilepsy, nocturnal frontal lobe type 1	118504	819,00 €	328,00 €	997,00 €			x
CHRNB1	Myasthenic syndrome, congenital	100710	796,00 €	429,00 €	1.075,00 €			x

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CHRNB2	Epilepsy, nocturnal frontal lobe type 3	118507	655,00 €	328,00 €	833,00 €			x
CHRND	Multiple pterygium syndrome lethal type	100720	862,00 €	421,00 €	1.133,00 €			x
CHRNE	Myasthenic syndrome, congenital	100725	729,00 €	421,00 €	1.000,00 €			x
CHRNG	Pterygium syndrome	100730	928,00 €	421,00 €	1.199,00 €			x
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations	603799	491,00 €	374,00 €	715,00 €			x
CHST8	Peeling skin syndrome type 3	610190	655,00 €					
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1	608429	468,00 €	296,00 €	614,00 €			x
CIB2	Deafness, autosomal recessive type 48	605564	573,00 €					
CIC	Intellectual disability nonsyndromic, CIC related	612082	1.704,00 €					
CIITA	Bare lymphocyte syndrome, type 2, complementation group A	600005	1.460,00 €					
CISD2	Wolfram syndrome type 2	611507	374,00 €	374,00 €	598,00 €			x
CITED2	Atrial septal defect type 8	602937	386,00 €					
CIZ1	Cervical dystonia	611420	1.326,00 €	421,00 €	1.597,00 €			x
CLCF1	Cold-induced sweating syndrome type 2	607672	468,00 €					
CLCN1	Myotonia congenita	118425	1.278,00 €	328,00 €	1.456,00 €			x
CLCN2	Epilepsy, idiopathic generalized type 11	600570	1.326,00 €	505,00 €	1.681,00 €			x
CLCN5	Dent disease	300008	1.061,00 €	445,00 €	1.356,00 €			x
CLCN7	Osteopetrosis, autosomal dominant type 1	602727	1.399,00 €	527,00 €	1.776,00 €			x
CLCNKA	Bartter syndrome type 4b	602024	1.326,00 €					
CLCNKB	Bartter syndrome type 3	602023	1.193,00 €	328,00 €	1.371,00 €			x
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	603718	468,00 €					
CLDN14	Deafness, autosomal recessive type 29	605608	386,00 €					
CLDN16	Hypomagnesemia type 3	603959	491,00 €	390,00 €	731,00 €			x
CLDN19	Hypomagnesemia type 5	610036	491,00 €					
CLIC2	Mental retardation, X-linked type 32	300138	655,00 €					
CLMP	Congenital short-bowel syndrome	611693	655,00 €					
CLN3	Ceroid lipofuscinosis neuronal type 3	607042	655,00 €	328,00 €	833,00 €			x
CLN5	Ceroid lipofuscinosis neuronal type 5	608102	491,00 €	437,00 €	778,00 €			x
CLN6	Ceroid lipofuscinosis neuronal type 6	606725	655,00 €	437,00 €	942,00 €			x
CLN8	Ceroid lipofuscinosis neuronal type 8	607837	386,00 €	374,00 €	610,00 €			x
CLP1	Pontocerebellar hypoplasia, type 10	608757	491,00 €					
CLRN1	Retinitis pigmentosa type 61, autosomal recessive	606397	573,00 €	437,00 €	860,00 €			x
CNBP	Myotonic dystrophy type 2	116955				200,00 €		x
CNGA1	Retinitis pigmentosa type 49, autosomal recessive	123825	928,00 €					
CNGA3	Achromatopsia type 2	600053	928,00 €					
CNGB1	Retinitis pigmentosa type 45, autosomal recessive	600724	1.673,00 €					
CNGB3	Achromatopsia type 3	605080	1.260,00 €	421,00 €	1.531,00 €			x
CNKSR2	Intellectual disability nonsyndromic, CNKSR2 related	300724	1.521,00 €					
CNNM2	Hypomagnesemia type 6	607803	928,00 €	437,00 €	1.215,00 €			x
CNNM4	Jalili syndrome	607805	737,00 €					

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CNOT3	Cardiac defects, CNOT3 related	604910	1.326,00 €					
CNTN1	Compton-North congenital myopathy	600016	1.521,00 €					
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome	604569	1.582,00 €	505,00 €	1.937,00 €			
CNTNAP4	Neurodevelopmental disorder, CNTNAP4 related	610518	1.582,00 €					
COA5	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 3	613920	374,00 €					
COASY	Neurodegeneration with brain iron accumulation type 6	609855	796,00 €					
COCH	Deafness, autosomal dominant type 9	603196	819,00 €	429,00 €	1.098,00 €			x
COG1	Glycosylation disorder type 2G	606973	995,00 €	437,00 €	1.282,00 €			
COG4	Glycosylation disorder type 2J	606976	1.460,00 €					
COG5	Glycosylation disorder type 2I	606821	1.704,00 €					
COG6	Glycosylation disorder type 3	606977	1.460,00 €	430,00 €	1.740,00 €			x
COG7	Glycosylation disorder type 2E	606978	1.193,00 €					x
COG8	Glycosylation disorder type 2H	606979	655,00 €					
COL1A1	Ehlers-Danlos syndrome type 7A	120150	1.825,00 €	328,00 €	2.003,00 €			x
COL1A2	Ehlers-Danlos syndrome type 7B	120160	2.016,00 €	328,00 €	2.194,00 €			x
COL2A1	Achondrogenesis type 2	120140	1.973,00 €	328,00 €	2.151,00 €			x
COL3A1	Ehlers-Danlos syndrome type 3	120180	1.989,00 €	328,00 €	2.167,00 €			x
COL4A1	Porencephaly, familial	120130	1.931,00 €	811,00 €	2.592,00 €			x
COL4A2	Porencephaly type 2	120090	1.845,00 €	824,00 €	2.519,00 €			x
COL4A3	Alport syndrome, autosomal recessive	120070	2.067,00 €	328,00 €	2.245,00 €			x
COL4A4	Alport syndrome, autosomal recessive	120131	1.973,00 €	328,00 €	2.151,00 €			x
COL4A5	Alport syndrome, X-Linked	303630	2.067,00 €	655,00 €	2.572,00 €			x
COL4A6	Deafness, X-linked type 6	303631	2.016,00 €	905,00 €	2.771,00 €			
COL5A1	Ehlers-Danlos syndrome type 1/2	120215	2.141,00 €	655,00 €	2.646,00 €			x
COL5A2	Ehlers-Danlos syndrome type 1/2	120190	1.989,00 €	842,00 €	2.681,00 €			x
COL6A1	Bethlem myopathy	120220	1.643,00 €	737,00 €	2.230,00 €			x
COL6A2	Bethlem myopathy	120240	1.825,00 €	590,00 €	2.265,00 €			x
COL6A3	Bethlem myopathy type 1	120250	2.106,00 €	755,00 €	2.711,00 €			x
COL6A6	Myopathy, COL6A6 related		1.931,00 €					
COL7A1	Epidermolysis bullosa dystrophica	120120	2.176,00 €	328,00 €	2.354,00 €			x
COL9A1	Stickler syndrome, autosomal recessive	120210	1.876,00 €					
COL9A2	Stickler syndrome type 5	120260	1.825,00 €					
COL9A3	Epiphyseal dysplasia, multiple, type 3	120270	1.673,00 €					
COL10A1	Metaphyseal chondrodysplasia, Schmid type	120110	655,00 €	374,00 €	879,00 €			x
COL11A1	Fibrochondrogenesis type 1	120280	1.955,00 €	655,00 €	2.460,00 €			x
COL11A2	Deafness, autosomal dominant type 13	120290	1.802,00 €					
COL12A1	Bethlem myopathy type 2	120320	1.955,00 €	905,00 €	2.710,00 €			x
COL15A1	Early onset glaucoma, phenotype modifier of, COL15A1 related	120325	1.802,00 €	905,00 €	2.557,00 €			
COL17A1	Epidermolysis bullosa, junctional	113811	2.028,00 €	874,00 €	2.752,00 €			x
COL18A1	Knobloch syndrome type 1	120328	2.184,00 €					

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COLEC11	3MC syndrome type 2	612502	729,00 €					x
COLQ	Endplate acetylcholinesterase deficiency	603033	1.260,00 €					
COMP	Epiphyseal dysplasia, multiple, type 1	600310	995,00 €	430,00 €	1.275,00 €			x
COMT	Catechol-o-methyltransferase deficiency	116790	468,00 €					
COQ2	Coenzyme Q10 deficiency type 1	609825	655,00 €					
COQ8A	Spinocerebellar ataxia type 9, autosomal rececssive	606980	928,00 €	445,00 €	1.223,00 €			x
COQ9	Coenzyme Q10 deficiency type 5	612837	819,00 €	421,00 €	1.090,00 €			x
CORIN	Preeclampsia/eclampsia type 5	605236	1.399,00 €					
CORO1A	Immunodeficiency type 8	605000	819,00 €					
COX4I2	Dyserythropoietic anemia	607976	468,00 €					
COX6A1	CMTRID	602072	374,00 €					
COX6B1	Mitochondrial complex IV deficiency	124089	468,00 €					
COX10	Encephalopathy mitochondrial with proximal renal tubulopathy due to cytochrome c oxidase deficiency	602125	737,00 €	328,00 €	915,00 €			
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 2	603646	796,00 €	421,00 €	1.067,00 €			x
COX20	Ataxia and muscle hypotonia	614698	468,00 €					
CP	Cerebellar ataxia	117700	1.326,00 €					x
CPA1	Pancreatitis, chronic, early onset	114850	729,00 €					
CPA6	Epilepsy, familial temporal lobe type 5	609562	796,00 €					
CPOX	Coproporphyria	612732	737,00 €	328,00 €	915,00 €			
CPS1	Carbamoylphosphate synthetase I deficiency	608307	1.802,00 €	821,00 €	2.473,00 €			x
CPT1A	Carnitine palmitoyltransferase 1A deficiency	600528	1.278,00 €	430,00 €	1.558,00 €			x
CPT1B	Carnitine palmitoyltransferase 1B deficiency	601987	1.193,00 €					
CPT1C	SPG73	608846	1.260,00 €					
CPT2	Carnitine palmitoyltransferase 2 deficiency, infantile	600650	819,00 €	390,00 €	1.059,00 €			x
CR2	Immunodeficiency, common variable type 7	120650	1.326,00 €					
CRB1	Leber congenital amaurosis type 8	604210	1.460,00 €	328,00 €	1.638,00 €			x
CRB2	Focal segmental glomerulosclerosis type 9	609720	1.260,00 €					
CRBN	Mental retardation, autosomal recessive type 2	609262	796,00 €					
CREB3L1	Osteogenesis disorders, CREB3L1 related	258480	862,00 €					
CREBBP	Rubinstein-Taybi syndrome	600140	1.572,00 €	328,00 €	1.750,00 €			x
CRELD1	Atrioventricular septal defect, partial with heterotaxy syndrome	607170	796,00 €					
CRHR1	Pulmonary newborn hypertension	122561	995,00 €	328,00 €	1.173,00 €			x
CRLF1	Cold-induced sweating syndrome	604237	819,00 €					
CROCC	Neurodevelopmental disorder, CROCC related	615776	1.977,00 €					
CRTAP	Osteogenesis imperfecta type 7	605497	729,00 €					
CRX	Cone-rod dystrophy type 2	602225	491,00 €	328,00 €	669,00 €			x
CRYAA	Cataract, autosomal recessive congenital type 1	123580	374,00 €					x
CRYAB	Cardiomyopathy, dilated type 1	123590	468,00 €					x
CRYBA4	Cataract type 23	123631	491,00 €					x
CRYBB1	Cataract type 17, multiple types	600929	491,00 €					x

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CRYBB3	Cataract, autosomal recessive congenital nuclear type 2	123630	491,00 €					
CRYM	Deafness, autosomal dominant type 40	123740	862,00 €					
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids	164770	1.193,00 €	463,00 €	1.506,00 €			x
CSF1	Osteogenesis and dental anomalies, CSF1 related	120420	796,00 €	421,00 €	1.067,00 €			x
CSF2RA	Surfactant metabolism dysfunction type 4	306250	928,00 €	328,00 €	1.106,00 €			x
CSF2RB	Surfactant metabolism dysfunction type 5	138981	995,00 €					x
CSF3R	Neutrophilia, hereditary	138971	1.127,00 €					x
CSPP1	Joubert syndrome type 21	611654	1.825,00 €					
CSRP3	Cardiomyopathy, dilated type 1M	600824	491,00 €					
CST3	Cerebral amyloid angiopathy	604312	374,00 €	437,00 €	661,00 €			x
CSTA	Peeling skin syndrome type 4	184600	374,00 €					
CSTB	Unverricht-Lundborg disease	601145	374,00 €	374,00 €	598,00 €	200,00 €		x
CTC1	Coat plus syndrome	613129	1.338,00 €	484,00 €	1.672,00 €			x
CTDP1	Cataracts with facial dysmorphism and neuropathy	604927	1.193,00 €	426,00 €	1.469,00 €			x
CTH	Cystathioninuria	219500	796,00 €					
CTLA4	Lymphoproliferative syndrome, autoimmune, type 5	123890	374,00 €	437,00 €	661,00 €			x
CTNNA2	Neuronal migration disorder	114025	1.278,00 €					
CTNNB1	Colorectal cancer, somatic	116806					1.900,00 €	
CTNS	Cystinosis, nephropathic	606272	729,00 €	426,00 €	1.005,00 €			x
CTPS1	Immunodeficiency type 24	123860	1.127,00 €					
CTRC	Pancreatitis	601405	655,00 €					
CTSA	Galactosialidosis	613111	928,00 €	445,00 €	1.223,00 €			x
CTSC	Haim-Munk syndrome	602365	796,00 €	437,00 €	1.083,00 €			x
CTSD	Ceroid lipofuscinosis neuronal type 10	116840	737,00 €	421,00 €	1.008,00 €			x
CTSK	Pycnodysostosis	601105	655,00 €	437,00 €	942,00 €			x
CUBN	Megaloblastic anemia type 1, Finnish type	602997	1.955,00 €					x
CUL3	Pseudohypoaldosteronism type 2E	603136	1.260,00 €					
CUL4B	Mental retardation, X-linked type 15	300304	1.460,00 €					
CUL7	Three M syndrome type 1	609577	1.764,00 €	548,00 €	2.162,00 €			x
CUX2	Major affective disorder	610648	1.521,00 €					
CWF19L1	Spinocerebellar ataxia type 17, autosomal recessive	616120	995,00 €					
CXCR4	WHIM syndrome	162643	491,00 €	328,00 €	669,00 €			x
CYB5R3	Methemoglobinemia type 1	613213	819,00 €	421,00 €	1.090,00 €			
CYBA	Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative	608508	573,00 €					
CYBB	Granulomatous disease, chronic, X-linked	300481	1.127,00 €	426,00 €	1.403,00 €			x
CYCS	Thrombocytopenia type 4	123970	296,00 €					
CYLD	Cylindromatosis, familial	605018	1.326,00 €	437,00 €	1.613,00 €			
CYP1A2	Cytochrome P450 deficiency	124060	737,00 €	328,00 €	915,00 €			
CYP1B1	Glaucoma, primary type 3A	601771	737,00 €	328,00 €	915,00 €			x
CYP2B6	Efavirenz, poor metabolism of	123930	796,00 €	328,00 €	974,00 €			

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CYP2C9	Coumarin/Warfarin resistance due to CYP2C9 variants	601130	796,00 €	328,00 €	974,00 €			
CYP2C19	CYP2C19 related poor drug metabolism	124020	796,00 €	328,00 €	974,00 €			x
CYP2R1	Rickets, vitamin D 25-hydroxylation-deficient, type 1B	608713	573,00 €					
CYP2U1	SPG56	610670	729,00 €	390,00 €	969,00 €			x
CYP3A5	Hypertension, salt-sensitive essential, susceptibility to	605325	1.061,00 €	328,00 €	1.239,00 €			
CYP4F22	Ichthyosis, lamellar type 3	611495	928,00 €					
CYP4V2	Bietti crystalline corneoretinal dystrophy	608614	729,00 €	328,00 €	907,00 €			
CYP7B1	Bile acid synthesis defect type 3, congenital	603711	655,00 €	421,00 €	926,00 €			x
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	118485	819,00 €					x
CYP11B1	Adrenal hyperplasia due to steroid 11-beta-hydroxylase deficiency	610613	819,00 €	421,00 €	1.090,00 €			x
CYP11B2	Hypoaldosteronism congenital due to CMO I deficiency	124080	737,00 €	421,00 €	1.008,00 €			x
CYP17A1	17-hydroxylation activity deficiency	609300	737,00 €	328,00 €	915,00 €			x
CYP19A1	Aromatase deficiency	107910	819,00 €	429,00 €	1.098,00 €			
CYP21A2	Adrenal hyperplasia due to 21-hydroxylase deficiency	613815	819,00 €	328,00 €	997,00 €			x
CYP24A1	Hypercalcemia infantile type	126065	796,00 €	421,00 €	1.067,00 €			x
CYP27A1	Cerebrotendinous xanthomatosis	606530	737,00 €	421,00 €	1.008,00 €			x
CYP27B1	Rickets, vitamin D dependent, type 1	609506	819,00 €					
D2HGDH	D-2-hydroxyglutaric aciduria type 1	609186	819,00 €	328,00 €	997,00 €			x
DAG1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C9	128239	819,00 €					
DARS2	Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation	610956	1.193,00 €	424,00 €	1.467,00 €			x
DARS	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	603084	1.061,00 €					
DBH	Dopamine beta-hydroxylase (DBH) deficiency	609312	928,00 €					
DBT	Maple syrup urine disease type 2	248610	862,00 €	429,00 €	1.141,00 €			x
DCAF17	Hypogonadism, alopecia, Diabetes mellitus, mental retardation and extrapyramidal syndrome	612515	995,00 €	437,00 €	1.282,00 €			x
DCC	Colorectal cancer, somatic	120470					1.900,00 €	
DCDC2	Deafness, autosomal recessive type 66	605755	862,00 €					
DCLRE1C	Omenn syndrome	605988	1.193,00 €	328,00 €	1.371,00 €			x
DCPS	Al-Raqad syndrome	610534	573,00 €					
DCTN1	Neuronopathy distal hereditary motor type 7B	601143	1.704,00 €	674,00 €	2.228,00 €			x
DCX	Lissencephaly, X-linked type 1	300121	819,00 €	328,00 €	997,00 €			x
DCXR	Pentosuria	608347	796,00 €					x
DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype	600811	729,00 €					
DDC	Aromatic L-amino acid decarboxylase deficiency	107930	1.061,00 €					
DDHD1	SPG28	614603	995,00 €					
DDHD2	SPG54	615003	1.127,00 €					
DDOST	Glycosylation disorder type IR	602202	729,00 €					
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type	191311	1.061,00 €					
DDX3X	Mental retardation, X-linked type 102	300160	796,00 €					
DDX11	Warsaw breakage syndrome	601150	1.521,00 €					
DDX59	Orofaciodigital syndrome type 5	615464	729,00 €	437,00 €	1.016,00 €			x

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DEAF1	Mental retardation, autosomal dominant type 24	602635	928,00 €					
DECR1	CoA-2 4-dienoyl reductase 1 deficiency	222745	737,00 €					
DEPDC5	Epilepsy, familial focal with variable foci	614191	1.931,00 €	738,00 €	2.519,00 €			
DES	Cardiomyopathy, dilated type 1I	125660	737,00 €	421,00 €	1.008,00 €			x
DFNA5	Deafness, autosomal dominant type 5	608798	737,00 €					
DFNB31	Deafness, autosomal recessive type 31	607928	995,00 €					
DFNB59	Deafness, autosomal recessive type 59	610219	737,00 €					
DGKE	Nephrotic syndrome type 7	601440	862,00 €					
DGUOK	Mitochondrial DNA depletion syndrome	601465	655,00 €	328,00 €	833,00 €			x
DHCR7	Smith-Lemli-Opitz syndrome	602858	737,00 €	328,00 €	915,00 €			x
DHCR24	Desmosterolosis	606418	819,00 €					
DHDDS	Retinitis pigmentosa type 59, autosomal recessive	608172	729,00 €					
DHH	46,XY gonadal dysgenesis, partial, with minifascicular neuropathy	605423	468,00 €	374,00 €	692,00 €			x
DHODH	Postaxial acrofacial dysostosis	126064	819,00 €	421,00 €	1.090,00 €			x
DHTKD1	2-aminoadipic 2-oxoadipic aciduria	614984	1.260,00 €					
DIABLO	Deafness, autosomal dominant type 64	605219	737,00 €					
DIAPH1	Deafness, autosomal dominant type 1	602121	1.673,00 €					
DIAPH3	Auditory neuropathy, autosomal dominant	614567	1.825,00 €					
DICER1	Pleuropulmonary blastoma	606241	1.764,00 €	611,00 €	2.225,00 €			
DIS3L2	Perlman Syndrome	614184	1.278,00 €	442,00 €	1.570,00 €			x
DISP1	Craniofacial and neuro-developmental abnormalities	607502	1.061,00 €					
DKC1	Dyskeratosis congenita, X-linked	300126	1.061,00 €	328,00 €	1.239,00 €			x
DLAT	Pyruvate dehydrogenase E2 deficiency	608770	995,00 €					
DLD	Maple syrup urine disease type 3	238331	928,00 €	437,00 €	1.215,00 €			x
DLG3	Mental retardation, X-linked type 90	300189	1.399,00 €	430,00 €	1.679,00 €			x
DLL3	Spondylocostal dysostosis, autosomal recessive type 1	602768	729,00 €	437,00 €	1.016,00 €			x
DLL4	Adams-Oliver syndrome type 6	605185	729,00 €					
DLX3	Amelogenesis imperfecta type 4	600525	374,00 €	374,00 €	598,00 €			x
DLX5	Split-hand/foot malformation type 1 with sensorineural hearing loss	600028	468,00 €	374,00 €	692,00 €			x
DMD	Cardiomyopathy, dilated type 3B	300377	950,00 €	655,00 €	1.455,00 €			x
DMGDH	Dimethylglycine dehydrogenase deficiency	605849	1.061,00 €					
DMP1	Hypophosphatemic rickets, autosomal recessive type 1	600980	573,00 €					x
DMPK	Myotonic dystrophy type 1	605377				200,00 €		x
DNA2	Progressive external ophthalmoplegia with mitochondrial deletions type 6	601810	1.399,00 €					
DNAAF1	Primary ciliary dyskinesia type 13	613190	928,00 €					
DNAAF2	Primary ciliary dyskinesia type 10	612517	737,00 €					
DNAAF3	Primary ciliary dyskinesia type 2	614566	995,00 €					
DNAAF5	Primary ciliary dyskinesia type 18	614864	995,00 €	426,00 €	1.271,00 €			
DNAH5	Primary ciliary dyskinesia type 3	603335	1.955,00 €	328,00 €	2.133,00 €			
DNAH9	Primary ciliary dyskinesia, DNAH9 related	603330	1.955,00 €	905,00 €	2.710,00 €			

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Phone: +49 (0)381 203 652 222
E-mail: testing@centogene.com

Gene	Disease	OMIM gene	Full gene sequencing	Deletion/ duplication testing	Package (sequencing + del/dup)	Repeat expansion testing	Somatic analysis	Prenatal testing offered
DNAH11	Primary ciliary dyskinesia type 7	603339	1.955,00 €	905,00 €	2.710,00 €			
DNAI1	Primary ciliary dyskinesia type 1	604366	1.460,00 €	328,00 €	1.638,00 €			
DNAI2	Primary ciliary dyskinesia type 9	605483	1.061,00 €					
DNAJB2	Spinal muscular atrophy type 5	604139	737,00 €	421,00 €	1.008,00 €			
DNAJB6	Limb-girdle muscular dystrophy, autosomal dominant type 1E	611332	819,00 €					
DNAJC5	Ceroid lipofuscinosis neuronal type 4	611203	468,00 €					
DNAJC6	PARK19 Parkinson, juvenile-onset	608375	1.399,00 €	430,00 €	1.679,00 €			x
DNAJC13	PARK21 Parkinson	614334	1.931,00 €	905,00 €	2.686,00 €			
DNAJC19	Cardiomyopathy, dilated with ataxia	608977	573,00 €					
DNAL1	Primary ciliary dyskinesia type 16	610062	729,00 €					
DNAL4	Mirror movements type 3	610565	374,00 €	437,00 €	661,00 €			
DNASE1L3	Systemic lupus erythematosus type 16	602244	737,00 €					
DNASE1	Systemic lupus erythematosus	125505	737,00 €					
DNM1L	Encephalopathy lethal, due to defective mitochondrial peroxisomal fission	603850	1.338,00 €					
DNM1	Early infantile epileptic encephalopathy type 31	602377	1.521,00 €					
DNM2	DI-CMTB	602378	1.399,00 €	442,00 €	1.691,00 €			x
DNMT1	Cerebellar ataxia with deafness and narcolepsy, autosomal recessive	126375	1.802,00 €	704,00 €	2.356,00 €			
DNMT3A	Acute myeloid leukemia, somatic, DNMT3A related	602769					1.900,00 €	
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome type 1	602900	1.326,00 €	484,00 €	1.660,00 €			
DOCK6	Adams-Oliver syndrome type 2	614194	2.028,00 €	824,00 €	2.702,00 €			x
DOCK7	Early infantile epileptic encephalopathy type 23	615730	1.973,00 €	905,00 €	2.728,00 €			
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive	611432	2.102,00 €	655,00 €	2.607,00 €			x
DOK7	Fetal akinesia deformation sequence	610285	796,00 €	429,00 €	1.075,00 €			x
DOLK	Glycosylation disorder type 1M	610746	468,00 €					
DPAGT1	Glycosylation disorder type 1J	191350	819,00 €					
DPM1	Glycosylation disorder type 1E	603503	655,00 €					x
DPM2	Glycosylation disorder type 1U	603564	468,00 €					
DPM3	Glycosylation disorder type 1O	605951	386,00 €					
DPY19L2	SPGF9	613893	1.399,00 €					
DPYD	Dihydropyrimidine dehydrogenase deficiency	612779	1.582,00 €	328,00 €	1.760,00 €			
DPYS	Dihydropyrimidinuria	613326	819,00 €					
DRD2	DYT11, DRD2 related	126450	737,00 €	437,00 €	1.024,00 €			x
DRD4	Attention deficit-hyperactivity disorder	126452	491,00 €					
DRD5	Attention deficit-hyperactivity disorder	126452	374,00 €					
DSC2	Arrhythmogenic right ventricular cardiomyopathy type 11	125645	1.278,00 €	328,00 €	1.456,00 €			x
DSE	Ehlers-Danlos syndrome, musculocontractural type 2	605942	729,00 €					
DSG1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis and hyper IgE	125670	1.193,00 €					
DSG2	Arrhythmogenic right ventricular cardiomyopathy type 10	125671	1.193,00 €	328,00 €	1.371,00 €			x
DSG4	Hypotrichosis type 6	607892	1.260,00 €					
DSP	Arrhythmogenic right ventricular cardiomyopathy type 8	125647	1.927,00 €	328,00 €	2.105,00 €			x

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DSPP	Deafness, autosomal dominant type 39, with dentinogenesis type 1	125485	491,00 €					
DST	Epidermolysis bullosa simplex, autosomal recessive type 2	113810	2.225,00 €					x
DTHD1	Leber congenital amaurosis with myopathy		729,00 €					
DTNBP1	Hermansky-Pudlak syndrome type 7	607145	729,00 €					
DUOX1	Thyroid dyshormonogenesis type 6	606758	1.775,00 €					
DUOX2	Thyroid dyshormonogenesis type 6	606759	1.775,00 €					
DUOXA2	Thyroid dyshormonogenesis type 5	612772	573,00 €					
DVL1	Robinow syndrome, autosomal dominant type 2	601365	928,00 €					
DYM	Dyggve-Melchior-Clausen disease	607461	1.193,00 €					
DYNC1H1	Charcot-Marie-Tooth disease, axonal type 20	600112	1.955,00 €					x
DYNC2H1	Short-rib thoracic dysplasia type 3 with or without polydactyly	603297	1.955,00 €	905,00 €	2.710,00 €			x
DYRK1A	Mental retardation, autosomal dominant type 7	600855	796,00 €					
DYSF	Limb-girdle muscular dystrophy, autosomal recessive type 2B	603009	2.223,00 €	328,00 €	2.401,00 €			x
DYX1C1	Primary ciliary dyskinesia type 25	608706	729,00 €					
EARS2	Combined oxidative phosphorylation deficiency type 12	612799	729,00 €	437,00 €	1.016,00 €			
EBP	Chondrodysplasia punctata, X-linked dominant	300205	491,00 €					
ECE1	Central hypoventilation syndrome, congenital	600423	1.278,00 €					
ECEL1	Arthrogryposis, distal, type 5D	605896	1.061,00 €	421,00 €	1.332,00 €			x
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	602292	737,00 €	437,00 €	1.024,00 €			
ECM1	Urbach-Wiethe disease	602201	737,00 €					
EDA	Ectodermal dysplasia, hypohidrotic, X-linked	300451	729,00 €	328,00 €	907,00 €			x
EDAR	Ectodermal dysplasia, hypohidrotic, autosomal recessive	604095	729,00 €	328,00 €	907,00 €			x
EDARADD	Ectodermal dysplasia, hypohidrotic, autosomal recessive	606603	655,00 €	328,00 €	833,00 €			x
EDN3	Central hypoventilation syndrome, congenital	131242	573,00 €	328,00 €	751,00 €			x
EDNRB	Hirschsprung disease	131244	862,00 €	328,00 €	1.040,00 €			x
EEF1A2	Early infantile epileptic encephalopathy type 33	602959	737,00 €					
EEF2	Spinocerebellar ataxia type 26, autosomal dominant	130610	1.061,00 €					
EFCAB13	Autism, EFCAB13 related		1.460,00 €					
EFEMP1	Doyne honeycob retinal dystrophy	601548	796,00 €	421,00 €	1.067,00 €			x
EFEMP2	Cutis laxa type 1B, autosomal recessive	604633	729,00 €	429,00 €	1.008,00 €			x
EFHC1	Epilepsy, juvenile absence type 1	608815	928,00 €	429,00 €	1.207,00 €			x
EFHC2	Mental retardation, x-linked, EFHC2 related	300817	1.061,00 €					
EFNB1	Craniofrontonasal syndrome	300035	491,00 €	328,00 €	669,00 €			x
EFTUD2	Mandibulofacial dysostosis with microcephaly	603892	1.764,00 €	590,00 €	2.204,00 €			x
EGF	Hypomagnesemia type 4	131530	1.521,00 €	505,00 €	1.876,00 €			x
EGFR	Colorectal Cancer, resistance to cetuximab, EGFR related, somatic	131550					1.900,00 €	
EGLN1	Erythrocytosis, familial type 3	606425	655,00 €	390,00 €	895,00 €			x
EGR2	CMT1D	129010	491,00 €	328,00 €	669,00 €			x
EHMT1	Kleefstra syndrome	607001	1.572,00 €	328,00 €	1.750,00 €			x
EIF2AK3	Wolcott-Rallison syndrome	604032	1.260,00 €					

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EIF2AK4	Pulmonary venoocclusive disease type 2	609280	2.028,00 €					
EIF2B1	Leukoencephalopathy with vanishing white matter	606686	819,00 €					x
EIF2B2	Leukoencephalopathy with vanishing white matter	606454	737,00 €					
EIF2B3	Leukoencephalopathy with vanishing white matter	606273	796,00 €					x
EIF2B4	Leukoencephalopathy with vanishing white matter	606687	862,00 €					x
EIF2B5	Leukoencephalopathy with vanishing white matter	603945	1.127,00 €					x
ELAC2	Combined oxidative phosphorylation deficiency type 17	605367	1.278,00 €					
ELANE	Neutropenia, severe congenital type 1	130130	491,00 €	390,00 €	731,00 €			x
ELK1	Mental retardation non-syndromic	311040	737,00 €					
ELN	Cutis laxa, autosomal dominant	130160	1.775,00 €	328,00 €	1.953,00 €			
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation	605512	573,00 €					
EMD	Emery-Dreifuss muscular dystrophy type 1	300384	573,00 €	421,00 €	844,00 €			x
EMX2	Schizencephaly	600035	468,00 €	374,00 €	692,00 €			x
EN2	Autism spectrum disorder	131310	491,00 €					
ENAM	Amelogenesis imperfecta type 1B	606585	1.127,00 €					
ENG	Telangiectasia, hereditary hemorrhagic, of Rendu, Osler and Weber type 1	131195	995,00 €	328,00 €	1.173,00 €			x
ENO3	Glycogen storage disease type 13	131370	729,00 €					x
ENPP1	Arterial calcification type 1, generalized, infantile	173335	1.582,00 €	527,00 €	1.959,00 €			x
ENTPD1	SPG64	601752	928,00 €					
EOGT	Adams-Oliver syndrome type 4	614789	1.127,00 €	421,00 €	1.398,00 €			
EOMES	Neuronal migration disorder	604615	573,00 €					
EP300	Colorectal cancer, somatic	602700					1.900,00 €	
EPAS1	Erythrocytosis, familial type 4	603349	1.193,00 €	424,00 €	1.467,00 €			
EPB42	Spherocytosis type 5	177070	995,00 €					
EPCAM	Colorectal cancer, hereditary nonpolyposis type 8	185535	819,00 €	328,00 €	997,00 €			
EPG5	Vici syndrome	615068	2.016,00 €	905,00 €	2.771,00 €			
EPM2A	Myoclonic epilepsy of Lafora	607566	468,00 €	328,00 €	646,00 €			x
EPOR	Erythrocytosis, familial type 1	133171	819,00 €	437,00 €	1.106,00 €			
ERBB2	Adenocarcinoma of lung, somatic	164870					1.900,00 €	
ERCC1	Cerebrooculofacioskeletal syndrome type 4	126380	819,00 €					
ERCC2	Trichothiodystrophy	126340	1.193,00 €					
ERCC3	Trichothiodystrophy	133510	1.061,00 €					
ERCC4	Fanconi anemia, complementation group Q	133520	862,00 €					
ERCC5	Xeroderma pigmentosum, group G	133530	1.193,00 €					
ERCC6L2	Bone marrow failure syndrome type 2	615667	1.399,00 €					
ERCC6	Cerebrooculofacioskeletal syndrome type 1	609413	1.521,00 €	442,00 €	1.813,00 €			x
ERCC8	Cockayne syndrome type A		1.127,00 €	421,00 €	1.398,00 €			x
ERF	Craniosynostosis type 4	611888	655,00 €					
ERLIN1	SPG62, ERLIN1 related	611604	796,00 €					
ERLIN2	SPG18	611605	862,00 €					

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ESCO2	Roberts syndrome	609353	796,00 €	429,00 €	1.075,00 €			x
ESPN	Deafness, autosomal recessive type 36	606351	1.061,00 €					
ESR1	Estrogen resistance	133430	737,00 €					
ESRRB	Deafness, autosomal recessive type 35	602167	819,00 €	429,00 €	1.098,00 €			
ETFA	Acyl-CoA multiple dehydrogenase deficiency	608053	995,00 €	421,00 €	1.266,00 €			x
ETFB	Acyl-CoA multiple dehydrogenase deficiency	130410	819,00 €	390,00 €	1.059,00 €			x
ETFDH	Glutaric acidemia type 2C	231675	928,00 €	426,00 €	1.204,00 €			x
ETHE1	Ethylmalonic encephalopathy	608451	655,00 €	437,00 €	942,00 €			x
ETV6	Thrombocytopenia type 5	600618	737,00 €	328,00 €	915,00 €			
EVC2	Ellis-van Creveld syndrome	607261	1.460,00 €	463,00 €	1.773,00 €			x
EVC	Ellis-van Creveld syndrome	604831	1.460,00 €	442,00 €	1.752,00 €			
EXOC8	Joubert syndrome, EXOC8 related	615283	573,00 €					x
EXOSC3	Pontocerebellar hypoplasia type 1B	606489	468,00 €	437,00 €	755,00 €			x
EXOSC8	Joubert syndrome, EXOSC8 related	606019	796,00 €					
EXPH5	Epidermolysis bullosa, nonspecific, autosomal recessive	612878	1.399,00 €					
EXT1	Chondrosarcoma, familial	608177	928,00 €	328,00 €	1.106,00 €			x
EXT2	Exostoses, multiple, type 2	608210	1.127,00 €	328,00 €	1.305,00 €			x
EYA1	Branchiootic syndrome type 1	601653	1.127,00 €	328,00 €	1.305,00 €			x
EYA4	Cardiomyopathy, dilated type 1J	603550	1.326,00 €	437,00 €	1.613,00 €			
EYS	Retinitis pigmentosa type 25	612424	1.955,00 €	328,00 €	2.133,00 €			
EZH2	Leukemia, lymphoblastic and myeloid, EZH2 related	601573					1.900,00 €	
F2	Dysprothrombinemia	176930	1.061,00 €					
F5	Budd-Chiari syndrome	612309	1.673,00 €					x
F7	Factor VII deficiency	613878	729,00 €	328,00 €	907,00 €			
F8	Hemophilia A	300841	1.845,00 €	328,00 €	2.023,00 €			x
F9	Hemophilia B	300746	729,00 €	328,00 €	907,00 €			x
F10	Factor X deficiency	613872	819,00 €	328,00 €	997,00 €			x
F11	Factor XI deficiency	264900	928,00 €	328,00 €	1.106,00 €			
F12	Factor XII deficiency	610619	928,00 €	437,00 €	1.215,00 €			x
F13A1	Factor XIIIA deficiency	134570	995,00 €	445,00 €	1.290,00 €			x
F13B	Factor XIIIB deficiency	134580	928,00 €					
FA2H	SPG35	611026	737,00 €	437,00 €	1.024,00 €			x
FAAH2	Autism, FAAH2 related	300654	862,00 €					
FAH	Tyrosinemia type 1	613871	995,00 €	437,00 €	1.282,00 €			x
FAM20A	Amelogenesis imperfecta type 1G	611062	862,00 €					
FAM20C	Raine syndrome	611061	796,00 €					
FAM58A	Toe syndactyly, telecanthus, and anogenital and renal malformations	300708	573,00 €					
FAM83H	Amelogenesis imperfecta type 3	611927	796,00 €					
FAM111A	Gracile bone dysplasia	615292	655,00 €					
FAM126A	Leukodystrophy hypomyelinating type 5	610531	796,00 €	429,00 €	1.075,00 €			x

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FAM134B	HSAN2B	613114	796,00 €	421,00 €	1.067,00 €			x
FAM161A	Retinitis pigmentosa type 28, autosomal recessive	613596	819,00 €					
FAN1	Interstitial nephritis karyomegalic	613534	1.127,00 €					
FANCA	Fanconi anemia type A	607139	1.888,00 €	655,00 €	2.393,00 €			x
FANCB	Fanconi anemia type B	300515	995,00 €	328,00 €	1.173,00 €			x
FANCC	Fanconi anemia type C	613899	1.061,00 €					
FANCD2	Fanconi anemia type D2	613984	1.845,00 €	328,00 €	2.023,00 €			x
FANCE	Fanconi anemia type E	613976	796,00 €					
FANCF	Fanconi anemia type F	613897	374,00 €					x
FANCG	Fanconi anemia type G	602956	729,00 €	437,00 €	1.016,00 €			
FANCI	Fanconi anemia type I	611360	1.876,00 €					
FANCL	Fanconi anemia type L	608111	995,00 €					
FANCM	Fanconi anemia type M	609644	1.622,00 €					
FARS2	Combined oxidative phosphorylation deficiency type 14	611592	573,00 €					
FAS	Autoimmune lymphoproliferative syndrome type 1A	134637	819,00 €	421,00 €	1.090,00 €			x
FASLG	Autoimmune lymphoproliferative syndrome type 1B	134638	573,00 €	437,00 €	860,00 €			
FASTKD2	Mitochondrial complex IV deficiency	612322	1.399,00 €					
FAT1	Facioscapulohumeral dystrophy-like phenotype, FAT1 related	600976	1.802,00 €					
FAT4	Hennekam lymphangiectasia-lymphedema syndrome type 2	612411	1.802,00 €					
FBLIM1	Kindler syndrome	607747	655,00 €					
FBLN5	Cutis laxa type 1A, autosomal recessive	604580	796,00 €					
FBN1	Acromicric dysplasia	134797	1.955,00 €	655,00 €	2.460,00 €			x
FBN2	Contractural arachnodactyly, congenital	612570	1.955,00 €	905,00 €	2.710,00 €			
FBP1	Fructose-1,6-bisphosphatase deficiency	611570	655,00 €	437,00 €	942,00 €			x
FBXL4	Mitochondrial DNA depletion syndrome type 13	605654	819,00 €					
FBXO7	PARK15 Parkinson	605648	729,00 €	421,00 €	1.000,00 €			x
FCRL6	Autism, FCRL6 related	613562	796,00 €					
FERMT1	Kindler syndrome	607900	1.061,00 €					
FERMT3	Leukocyte adhesion deficiency type 3	607901	796,00 €					
FGA	Afibrinogenemia, congenital	134820	655,00 €					
FGB	Afibrinogenemia, congenital	134830	819,00 €	437,00 €	1.106,00 €			
FGD1	Faciogenital dysplasia	300546	1.193,00 €	328,00 €	1.371,00 €			x
FGD4	CMT4H	611104	1.326,00 €	424,00 €	1.600,00 €			x
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia	602292	468,00 €	374,00 €	692,00 €			x
FGF8	Hypogonadotropic hypogonadism type 6 with or without anosmia	600483	468,00 €	421,00 €	739,00 €			x
FGF10	LADD syndrome	602115	374,00 €	328,00 €	552,00 €			x
FGF12	Early infantile epileptic encephalopathy type 47	601513	655,00 €					
FGF14	Spinocerebellar ataxia type 27, autosomal dominant	601515	729,00 €	390,00 €	969,00 €			x
FGF23	Hypophosphatemic rickets, autosomal dominant	605380	468,00 €	328,00 €	646,00 €			x
FGFR1	Craniosynostosis, FGFR1 related	136350	1.193,00 €	328,00 €	1.371,00 €			x

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FGFR2	Antley-Bixler syndrome	176943	1.460,00 €	328,00 €	1.638,00 €			x
FGFR3	Achondroplasia	134934	995,00 €	421,00 €	1.266,00 €			x
FGFRL1	Radioulnar synostosis, FGFRL1 related	605830	655,00 €					
FGG	Afibrinogenemia, congenital	134850	928,00 €					
FH	Fumarase deficiency	136850	729,00 €	328,00 €	907,00 €			x
FHL1	Emery-Dreifuss muscular dystrophy type 6	300163	737,00 €	437,00 €	1.024,00 €			x
FIG4	Amyotrophic lateral sclerosis type 11	609390	1.460,00 €	484,00 €	1.794,00 €			x
FKBP10	Osteogenesis imperfecta type 11	607063	729,00 €					
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss	614505	468,00 €					
FKRP	Limb-girdle muscular dystrophy, autosomal recessive type 2I	606596	491,00 €	328,00 €	669,00 €			x
FKTN	Cardiomyopathy, dilated type 1X	607440	928,00 €	328,00 €	1.106,00 €			x
FLCN	Birt-Hogg-Dube syndrome	607273	729,00 €	328,00 €	907,00 €			x
FLG	Dermatitis, atopic type 2	135940	1.622,00 €	374,00 €	1.846,00 €			x
FLI1	Platelet dense granule secretion defect, excessive bleeding	193067	796,00 €					
FLNA	Cardiac valvular dysplesia, X-linked	300017	1.825,00 €	824,00 €	2.499,00 €			x
FLNB	Atelosteogenesis type 1	603381	2.059,00 €	328,00 €	2.237,00 €			x
FLNC	Filaminopathy	102565	1.845,00 €					x
FLRT1	SPG68, FLRT1 related	604806	491,00 €					
FLT4	Lymphedema, hereditary, type 1A	136352	1.582,00 €					
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa	609144	995,00 €	429,00 €	1.274,00 €			x
FLVCR2	Hydranencephaly, Fowler type	610865	729,00 €					
FMO3	Trimethylaminuria	136132	819,00 €	421,00 €	1.090,00 €			x
FMR1	Fragile X syndrome	309550	1.399,00 €	328,00 €	1.577,00 €	400,00 €		x
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	136430	491,00 €	421,00 €	762,00 €			x
FOXC1	Axenfeld-Rieger syndrome type 3	601090	491,00 €	328,00 €	669,00 €			x
FOXC2	Lymphedema-distichiasis syndrome	602402	468,00 €	328,00 €	646,00 €			x
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins	601089	491,00 €	328,00 €	669,00 €			x
FOXF2	Disorders of sex development with cleft palate	603250	573,00 €	328,00 €	751,00 €			
FOXG1	Rett syndrome, congenital variant	164874	468,00 €	328,00 €	646,00 €			x
FOXH1	Congenital heart disease and transposition of the great arteries	603621	573,00 €					
FOXI1	Deafness, autosomal recessive type 4	601093	468,00 €	343,00 €	661,00 €			x
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis	605597	491,00 €	328,00 €	669,00 €			x
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	600838	729,00 €					
FOXP1	Mental retardation with language impairment and autistic features	605515	1.278,00 €					
FOXP2	Speech-language disorder type 1	605317	1.260,00 €	424,00 €	1.534,00 €			x
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	300292	928,00 €					
FOXRED1	Leigh syndrome	613622	796,00 €					
FRAS1	Fraser syndrome	607830	1.955,00 €	905,00 €	2.710,00 €			x
FREM1	Bifid nose	608944	2.028,00 €	800,00 €	2.678,00 €			x
FREM2	Fraser syndrome	608945	1.724,00 €					

Pricelist by gene

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FRMD7	Nystagmus type 1	300628	862,00 €	328,00 €	1.040,00 €			
FRMPD4	Neurodevelopmental disorder, FRMPD4 related	300838	1.521,00 €					
FRRS1L	Dysautonomia, FRRS1L-related	604574	491,00 €					
FSCN2	Retinitis pigmentosa type 30, autosomal dominant	607643	491,00 €					
FSHB	Follicle-stimulating hormone deficiency, isolated	136530	386,00 €	328,00 €	564,00 €			
FSHR	Ovarian dysgenesis type 1	136435	729,00 €	429,00 €	1.008,00 €			
FTCD	Glutamate formiminotransferase deficiency	606806	862,00 €					
FTL	Hyperferritinemia-cataract syndrome	134790	468,00 €	437,00 €	755,00 €			x
FTSJ1	Mental retardation, X-linked type 44	300499	729,00 €					
FUCA1	Fucosidosis	612280	819,00 €	437,00 €	1.106,00 €			x
FUS	Amyotrophic lateral sclerosis type 6	137070	995,00 €	445,00 €	1.290,00 €			x
FXN	Friedreich ataxia	606829	573,00 €	328,00 €	751,00 €	200,00 €		x
FXYD2	Hypomagnesemia type 2	601814	374,00 €					
FYCO1	Cataract, autosomal recessive congenital type 2	607182	1.521,00 €					
FZD4	Exudative vitreoretinopathy	604579	573,00 €	328,00 €	751,00 €			x
G6PC2	Hyperinsulinaemia, association with, G6PC2 related	608058	655,00 €					
G6PC3	Neutropenia, severe congenital type 4, autosomal recessive	611045	655,00 €	421,00 €	926,00 €			x
G6PC	Glycogen storage disease type 1A	613742	655,00 €	390,00 €	895,00 €			x
G6PD	Favism, susceptibility to	305900	862,00 €	426,00 €	1.138,00 €			x
GAA	Glycogen storage disease type 2	606800	1.260,00 €	328,00 €	1.438,00 €			x
GABRA1	Early infantile epileptic encephalopathy type 19	137160	796,00 €					
GABRB1	Early infantile epileptic encephalopathy type 45	137190	729,00 €					
GABRB3	Epilepsy, childhood absence type 5	137192	862,00 €	328,00 €	1.040,00 €			x
GABRD	Epilepsy, idiopathic generalized type 10	137163	737,00 €	421,00 €	1.008,00 €			x
GABRG2	Dravet syndrome	137164	796,00 €	429,00 €	1.075,00 €			x
GAD1	Cerebral palsy type 1, spastic quadriplegic	605363	1.326,00 €					
GALC	Krabbe disease	606890	1.260,00 €	328,00 €	1.438,00 €			x
GALE	Galactose epimerase deficiency	606953	729,00 €	421,00 €	1.000,00 €			x
GALK1	Galactokinase deficiency	604313	655,00 €	437,00 €	942,00 €			x
GALNS	Mucopolysaccharidosis type 4A	612222	1.061,00 €	445,00 €	1.356,00 €			x
GALT	Galactosemia	606999	819,00 €	328,00 €	997,00 €			x
GAMT	Guanidinoacetate methyltransferase deficiency	601240	491,00 €	390,00 €	731,00 €			x
GAN	Giant axonal neuropathy type 1	605379	729,00 €	429,00 €	1.008,00 €			x
GARS	CMT2D	600287	1.193,00 €	328,00 €	1.371,00 €			x
GATA1	Anemia, X-linked	305371	655,00 €	421,00 €	926,00 €			x
GATA2	Emberger syndrome	137295	655,00 €	328,00 €	833,00 €			
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	131320	655,00 €	328,00 €	833,00 €			
GATA4	Atrial septal defect type 2	600576	729,00 €	328,00 €	907,00 €			x
GATA6	Atrial septal defect type 9	601656	819,00 €	437,00 €	1.106,00 €			x
GATAD1	Cardiomyopathy, dilated type 2B	614518	491,00 €					

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GATAD2B	Mental retardation, autosomal dominant type 18	614998	729,00 €					
GATM	Arginine-glycine amidinotransferase deficiency	602360	819,00 €	421,00 €	1.090,00 €			x
GBA2	Cerebellar ataxia with spasticity	609471	1.193,00 €	424,00 €	1.467,00 €			x
GBA	Gaucher disease type 1	606463	796,00 €	421,00 €	1.067,00 €			x
GBE1	Andersen disease	607839	1.127,00 €	424,00 €	1.401,00 €			x
GCDH	Glutaric acidemia type 1	608801	729,00 €	421,00 €	1.000,00 €			x
GCH1	DYT5A	600225	737,00 €	328,00 €	915,00 €			x
GCK	Hyperinsulinemic hypoglycemia type 3	138079	862,00 €	328,00 €	1.040,00 €			x
GCM2	Hypoparathyroidism, familial isolated	603716	737,00 €	390,00 €	977,00 €			x
GCNT2	Cataract, autosomal dominant	600429	995,00 €					
GCSH	Glycine encephalopathy	238330	491,00 €	328,00 €	669,00 €			x
GDAP1	CMT2K	606598	573,00 €	328,00 €	751,00 €			x
GDF1	Transposition of great arteries, dextro-looped 3	602880	573,00 €	437,00 €	860,00 €			
GDF2	Telangiectasia hereditary hemorrhagic type 5	605120	491,00 €	343,00 €	684,00 €			
GDF3	Klippel-Feil syndrome type 3, autosomal dominant	606522	468,00 €	343,00 €	661,00 €			x
GDF5	Brachydactyly type A1C	601146	573,00 €	343,00 €	766,00 €			x
GDF6	Klippel-Feil syndrome type 1, autosomal dominant	601147	491,00 €	343,00 €	684,00 €			x
GDI1	Mental retardation, X-linked type 41	300104	796,00 €					
GDNF	Central hypoventilation syndrome, congenital	600837	491,00 €	328,00 €	669,00 €			x
GFAP	Alexander disease	137780	796,00 €	437,00 €	1.083,00 €			x
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	600924	737,00 €					
GFI1B	Bleeding disorder, platelet-type 17	604383	491,00 €					
GFI1	Neutropenia, nonimmune chronic idiopathic, of adults	600871	655,00 €	437,00 €	942,00 €			x
GFM1	Combined oxidative phosphorylation deficiency type 1	606639	1.278,00 €					
GFM2	Microcephaly with symplified gyral pattern and insulin-dependant diabetes	606544	1.278,00 €					
GFPT1	Myasthenia congenital with tubular aggregates 1	138292	1.399,00 €					
GFRA1	Central hypoventilation syndrome, congenital	601496	491,00 €	328,00 €	669,00 €			x
GGCX	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	137167	1.061,00 €					
GH1	Growth hormone deficiency	139250	491,00 €	328,00 €	669,00 €			x
GHR	Growth hormone insensitivity, partial	600946	1.061,00 €	328,00 €	1.239,00 €			x
GHRHR	Growth hormone deficiency	139191	862,00 €	328,00 €	1.040,00 €			x
GIF	Intrinsic factor deficiency	609342	729,00 €					
GIPC3	Deafness, autosomal recessive type 15	608792	573,00 €					
GJA1	Oculodentodigital dysplasia	121014	468,00 €	343,00 €	661,00 €			x
GJA5	Atrial fibrillation type 11	121013	729,00 €					
GJA8	Cataract-microcornea syndrome	600897	468,00 €	343,00 €	661,00 €			x
GJB1	CMTX1	304040	491,00 €	328,00 €	669,00 €			x
GJB2	Deafness with keratopachydermia and constrictions of fingers and toes	121011	374,00 €	328,00 €	552,00 €			x
GJB3	Deafness, autosomal dominant type 2B	603324	374,00 €	328,00 €	552,00 €			x
GJB4	Erythrokeratodermia variabilis et progressive	605425	374,00 €					

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GJB6	Deafness, autosomal dominant type 3B	604418	374,00 €	328,00 €	552,00 €			x
GJC2	Leukodystrophy hypomyelinating	608803	573,00 €	343,00 €	766,00 €			x
GK	Glycerol kinase deficiency	300474	1.399,00 €	442,00 €	1.691,00 €			x
GLA	Fabry disease	300644	819,00 €	328,00 €	997,00 €			x
GLB1	GM1-gangliosidosis	611458	1.127,00 €	424,00 €	1.401,00 €			x
GLDC	Glycine encephalopathy	238300	1.643,00 €	328,00 €	1.821,00 €			x
GLE1	Lethal congenital contracture syndrome type 1	603371	1.127,00 €					
GLI2	Holoprosencephaly-type 9	165230	1.326,00 €	328,00 €	1.504,00 €			x
GLI3	Greig cephalopolysyndactyly syndrome	165240	1.278,00 €	328,00 €	1.456,00 €			x
GLIS2	Nephronophthisis type 7	608539	737,00 €					
GLIS3	Diabetes mellitus, neonatal	610192	995,00 €					x
GLRA1	Hyperekplexia	138491	819,00 €	328,00 €	997,00 €			x
GLRB	Hyperekplexia	138492	819,00 €	328,00 €	997,00 €			x
GLRX5	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive	609588	386,00 €					
GLUD1	Hyperinsulinemic hypoglycemia type 6	138130	862,00 €					x
GLUL	Glutamine deficiency, congenital	138290	573,00 €					
GLYCTK	D-glyceric aciduria	610516	573,00 €					
GM2A	Tay-Sachs disease AB variant	613109	491,00 €	437,00 €	778,00 €			x
GMPPA	Alacrima, achalasia and mental retardation syndrome	615495	796,00 €					
GNA11	Hypocalcemia, autosomal dominant 2	605573	737,00 €					
GNA13	Vascular system defects due to GNA13 deficiency	604406	655,00 €					
GNAI3	Auriculocondylar syndrome type 1	139370	737,00 €	421,00 €	1.008,00 €			
GNAL	DYT25	139312	928,00 €	421,00 €	1.199,00 €			x
GNAO1	Early infantile epileptic encephalopathy type 17	139311	729,00 €					
GNAQ	Developmental delay, GNAQ related	600998	655,00 €	437,00 €	942,00 €			
GNAS	Osseous heteroplasia, progressive	139320	1.338,00 €	328,00 €	1.516,00 €			
GNAT1	Night blindness, congenital stationary type 3	139330	573,00 €					x
GNAT2	Achromatopsia type 4	139340	737,00 €					
GNB4	CMTDIF	610863	819,00 €					
GNE	Inclusion body myopathy	603824	928,00 €	421,00 €	1.199,00 €			x
GNMT	Glycine N-methyltransferase deficiency	606628	573,00 €					
GNPAT	Rhizomelic chondrodysplasia punctata type 2	602744	1.061,00 €					
GNPTAB	Mucolipidosis type 2 alpha/beta	607840	1.521,00 €	442,00 €	1.813,00 €			x
GNPTG	Mucolipidosis type 3 gamma	607838	928,00 €	429,00 €	1.207,00 €			
GNRH1	Hypogonadotropic hypogonadism type 12 with or without anosmia	152760	374,00 €	328,00 €	552,00 €			x
GNRHR	Hypogonadotropic hypogonadism type 7 with or without anosmia	138850	573,00 €	328,00 €	751,00 €			
GNS	Mucopolysaccharidosis type 3D	607664	995,00 €	437,00 €	1.282,00 €			x
GORAB	Geroderma osteodysplasticum	607983	655,00 €					
GOSR2	Progressive myoclonus epilepsy type 6	604027	655,00 €					
GP1BA	Bernard Soulier syndrome type A1	606672	655,00 €					

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GP1BB	Bernard Soulier syndrome type B	138720	374,00 €					
GP9	Bernard Soulier syndrome type C	173515	386,00 €					
GPC3	Simpson-Golabi-Behmel syndrome type 1	300037	796,00 €	328,00 €	974,00 €			x
GPC6	Omodysplasia type 1	604404	819,00 €	421,00 €	1.090,00 €			x
GPD1L	Brugada syndrome type 2	611778	737,00 €					
GPD1	Hypertriglyceridemia, transient infantile	138420	655,00 €	437,00 €	942,00 €			
GPHN	Molybdenum cofactor deficiency type C	603930	1.460,00 €	484,00 €	1.794,00 €			x
GPR101	Acromegaly, predisposition to, due to germline GPR101 mutation	300393	491,00 €					
GPR143	Albinism, ocular type I, Nettleship-Falls type	300808	729,00 €	328,00 €	907,00 €			x
GPR161	Pituitary stalk interruption syndrome, GPR161 related		655,00 €					
GPR179	Night blindness, congenital stationary, type 1E	614515	1.704,00 €					
GPSM2	Chudley-McCullough syndrome	609245	1.260,00 €	445,00 €	1.555,00 €			
GPT2	Mental retardation, autosomal recessive type 49	138210	796,00 €					
GREM1	Polyposis syndrome, hereditary mixed	603054	386,00 €	328,00 €	564,00 €			
GRHL2	Deafness, autosomal dominant type 28	608576	1.326,00 €					
GRHL3	van der Woude syndrome type 2	608317	1.193,00 €					
GRHPR	Hyperoxaluria type 2	604296	819,00 €	328,00 €	997,00 €			x
GRIA3	Mental retardation, X-linked type 94	305915	1.260,00 €					
GRID2	Schizophrenia, GRID2 related	602368	1.193,00 €	424,00 €	1.467,00 €			x
GRIN1	Mental retardation, autosomal dominant type 8	138249	1.278,00 €					
GRIN2A	Epilepsy with neurodevelopmental defects	138253	1.326,00 €	328,00 €	1.504,00 €			x
GRIN2B	Early infantile epileptic encephalopathy type 27	138252	1.278,00 €	328,00 €	1.456,00 €			x
GRIP1	Fraser syndrome	604597	1.460,00 €	505,00 €	1.815,00 €			x
GRK1	Oguchi disease	180381	819,00 €					
GRM1	Spinocerebellar ataxia type 13, autosomal recessive	604473	995,00 €	437,00 €	1.282,00 €			x
GRM6	Night blindness, congenital stationar type 1B	604096	862,00 €					
GRM7	Autism spectrum/ hyperactivity/ bipolar disorder, GRM7 related	604101	729,00 €	429,00 €	1.008,00 €			
GRN	Ceroid lipofuscinosis neuronal type 11	138945	796,00 €	328,00 €	974,00 €			x
GRXCR1	Deafness, autosomal recessive type 25	613283	468,00 €					
GSN	Amyloidosis, finnish type	137350	1.260,00 €					x
GSS	Glutathione synthetase deficiency	601002	862,00 €	426,00 €	1.138,00 €			x
GSTT1	Glutathione S-transferase theta-1 defficiency	600436	491,00 €	328,00 €	669,00 €			
GSTZ1	Tyrosinemia type 1B	603758	819,00 €	421,00 €	1.090,00 €			x
GTF2H5	Trichothiodystrophy	608780	386,00 €					
GTPBP2	Neurodegeneration with brain iron accumulation, GTPBP2 related	607434	862,00 €					
GTPBP3	Combined oxidative phosphorylation deficiency type 23	608536	655,00 €					
GUCA1A	Cone-rod dystrophy type 14	600364	573,00 €					
GUCA1B	Retinitis pigmentosa type 48, autosomal dominant	602275	374,00 €					
GUCY1A3	Moyamoya type 6 with achalasia	139396	796,00 €					
GUCY2C	Diarrhea type 6	601330	1.704,00 €					

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GUCY2D	Leber congenital amaurosis type 1	600179	1.193,00 €	328,00 €	1.371,00 €			x
GUF1	Early infantile epileptic encephalopathy type 40	617064	1.193,00 €					x
GUSB	Mucopolysaccharidosis type 7	611499	862,00 €	421,00 €	1.133,00 €			x
GYG1	Glycogen storage disease type 15	603942	729,00 €					x
GYG2	Autism, GYG2 related	300198	796,00 €					
GYS1	Glycogen storage disease type 0 muscle	138570	1.061,00 €	424,00 €	1.335,00 €			x
GYS2	Glycogen storage disease type 0	138571	1.127,00 €	424,00 €	1.401,00 €			x
H19	Beckwith-Wiedemann syndrome	103280	928,00 €	328,00 €	1.106,00 €			x
HADH	CoA-3-hydroxyacyl dehydrogenase deficiency	601609	729,00 €					
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	600890	1.260,00 €	437,00 €	1.547,00 €			x
HADHB	Trifunctional protein deficiency	143450	1.061,00 €	424,00 €	1.335,00 €			x
HAMP	Hemochromatosis type 2B	606464	374,00 €	328,00 €	552,00 €			x
HAX1	Neutropenia, severe congenital type 3	605998	655,00 €	437,00 €	942,00 €			x
HBA1	Thalassemia, alpha	141800	374,00 €	328,00 €	552,00 €			x
HBA2	Thalassemia, alpha	141850	374,00 €	328,00 €	552,00 €			x
HBB	Delta-beta thalassemia	141900	491,00 €	328,00 €	669,00 €			x
HBD	Thalassemia, delta	142000	468,00 €	328,00 €	646,00 €			
HBG2	Cyanosis, transient neonatal	142250	468,00 €	328,00 €	646,00 €			x
HCCS	Microphthalmia syndromic type 7	300056	573,00 €	437,00 €	860,00 €			x
HCFC1	Mental retardation, X-linked type 3	300019	1.572,00 €					
HCN1	Early infantile epileptic encephalopathy type 24	602780	819,00 €					
HCN2	Epilepsy, HCN2 related	602781	729,00 €					
HCN4	Brugada syndrome type 8	605206	729,00 €					
HCRT	Narcolepsy	602358	468,00 €					
HDAC4	Brachydactyly-mental retardation syndrome	605314	1.825,00 €					
HDAC8	Cornelia de Lange syndrome type 5	300269	928,00 €	429,00 €	1.207,00 €			x
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A	611642	862,00 €	437,00 €	1.149,00 €			x
HERC2	Mental retardation, autosomal recessive type 38	605837	1.955,00 €	905,00 €	2.710,00 €			
HESX1	Septooptic dysplasia	601802	374,00 €	328,00 €	552,00 €			x
HEXA	Tay-Sachs disease	606869	995,00 €	328,00 €	1.173,00 €			x
HEXB	GM2-gangliosidosis type 2	606873	995,00 €	437,00 €	1.282,00 €			x
HFE2	Hemochromatosis type 2A	608374	491,00 €	328,00 €	669,00 €			x
HFE	Hemochromatosis classical	613609	573,00 €	328,00 €	751,00 €			x
HGD	Alkaptonuria	607474	995,00 €	437,00 €	1.282,00 €			x
HGF	Deafness, autosomal recessive type 39	142409	1.326,00 €					
HGSNAT	Mucopolysaccharidosis type 3C	610453	1.193,00 €	421,00 €	1.464,00 €			x
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency	610690	995,00 €					
HINT1	Neuromyotonia and axonal neuropathy, autosomal recessive	601314	374,00 €	374,00 €	598,00 €			x
HLCS	Holocarboxylase synthetase deficiency	609018	928,00 €	421,00 €	1.199,00 €			x
HMBS	Porphyria acute intermittent	609806	928,00 €	328,00 €	1.106,00 €			x

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HMG20B	Dysmorphism, HMG20B related	605535	819,00 €					
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	613898	737,00 €	421,00 €	1.008,00 €			
HMGCS2	3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency	600234	819,00 €	328,00 €	997,00 €			
HNF1A	Diabetes mellitus, insulin-dependent type 20	142410	729,00 €	328,00 €	907,00 €			x
HNF1B	Maturity-onset diabetes of the young type 5	189907	819,00 €	328,00 €	997,00 €			x
HNF4A	Maturity-onset diabetes of the young type 1	600281	1.193,00 €	328,00 €	1.371,00 €			x
HNRNPU	RNA processing related disorders	602869	1.061,00 €					
HOGA1	Hyperoxaluria type 3	613597	655,00 €	437,00 €	942,00 €			x
HOXA1	Athabaskan brainstem dysgenesis syndrome	142955	468,00 €	343,00 €	661,00 €			x
HOXA13	Guttmacher syndrome	142959	491,00 €					
HOXB1	Facial paresis type 3	142968	468,00 €					x
HOXB13	Prostate cancer, familial, association with	604607	386,00 €					
HOXD13	Brachydactyly type E1	142989	374,00 €	328,00 €	552,00 €			x
HP	Anhaptoglobinemia	140100	737,00 €					
HPCA	DYT2	142622	468,00 €					
HPD	Hawkinsinuria	609695	729,00 €					
HPGD	Hypertrophic osteoarthropathy type 1	601688	655,00 €					
HPRT1	Lesch-Nyham syndrome	308000	729,00 €	328,00 €	907,00 €			x
HPS1	Hermansky-Pudlak syndrome type 1	604982	1.127,00 €					
HPS3	Hermansky-Pudlak syndrome type 3	606118	1.260,00 €					
HPS4	Hermansky Pudlak syndrome type 4	606682	1.326,00 €					
HPS5	Hermansky-Pudlak syndrome type 5	607521	1.582,00 €					
HPS6	Hermansky-Pudlak syndrome type 6	607522	491,00 €					
HR	Alopecia universalis	602302	1.193,00 €	430,00 €	1.473,00 €			
HRAS	Bladder cancer, HRAS related, somatic	190020					1.900,00 €	
HS6ST1	Hypogonadotropic hypogonadism type 15 with or without anosmia	604846	468,00 €	343,00 €	661,00 €			x
HSD3B2	3-beta-hydroxysteroid dehydrogenase deficiency type 2	613890	468,00 €	437,00 €	755,00 €			x
HSD11B2	Apparent mineralocorticoid excess	614232	573,00 €	390,00 €	813,00 €			x
HSD17B3	Pseudohermaphroditism with gynecomastia	605573	796,00 €					
HSD17B4	D-bifunctional protein deficiency	601860	1.521,00 €	527,00 €	1.898,00 €			x
HSD17B10	17-beta hydroxysteroid dehydrogenase X deficiency	300256	573,00 €					
HSF4	Cataract, lamellar	602438	862,00 €					
HSPB1	CMT2F	602195	374,00 €	328,00 €	552,00 €			x
HSPB8	CMT2L	608014	374,00 €	328,00 €	552,00 €			x
HSPD1	Leukodystrophy hypomyelinating type 4	118190	796,00 €	421,00 €	1.067,00 €			x
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type	142461	1.955,00 €					x
HTRA1	CARASIL	602194	729,00 €	421,00 €	1.000,00 €			x
HTRA2	PARK13 Parkinson	606441	737,00 €	437,00 €	1.024,00 €			
HTT	Huntington disease	613004				200,00 €		
HUWE1	Mental retardation, X-linked syndromic, Turner type	300697	1.955,00 €					x

Pricelist by gene

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HYAL1	Mucopolysaccharidosis type 9	607071	737,00 €					
HYDIN	Primary ciliary dyskinesia type 5	610812	1.955,00 €					
HYLS1	Hydrolethalus syndrome	610693	374,00 €					
IBA57	Multiple mitochondrial dysfunctions syndrome type 3	615316	573,00 €					
ICOS	Immunodeficiency common variable type 1	604558	573,00 €	390,00 €	813,00 €			x
IDH1	Glioma, susceptibility to, somatic	147700					1.900,00 €	
IDH2	D-2-hydroxyglutaric aciduria type 2	147650	796,00 €	429,00 €	1.075,00 €			x
IDH3B	Retinitis pigmentosa type 46, autosomal recessive	604526	796,00 €					
IDS	Mucopolysaccharidosis type 2	300823	862,00 €	328,00 €	1.040,00 €			x
IDUA	Hurler syndrome	252800	928,00 €	437,00 €	1.215,00 €			x
IER3IP1	Microcephaly with epilepsy and diabetes syndrome	609382	374,00 €					
IFIH1	Aicardi-Goutieres syndrome type 7	606951	1.127,00 €					
IFITM5	Osteogenesis imperfecta type 5	614757	386,00 €	343,00 €	579,00 €			x
IFNGR1	Mycobacterial infection, atypical, familial disseminated	107470	819,00 €					
IFNGR2	Atypical Mycobacterial infection	147569	655,00 €					x
IFRD1	Spinocerebellar ataxia type 18, autosomal dominant	603502	729,00 €	421,00 €	1.000,00 €			x
IFT43	Cranioectodermal dysplasia type 3	614068	737,00 €					
IFT80	Short-rib thoracic dysplasia type 2 with or without polydactyly	611177	1.278,00 €					
IFT122	Cranioectodermal dysplasia type 1	606045	1.673,00 €					
IFT140	Mainzer Saldino syndrome	614620	1.764,00 €					
IFT172	Short-rib thoracic dysplasia type 10 with or without polydactyly	607386	2.102,00 €					
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia	300139	737,00 €					
IGF1R	Insulin-like growth factor resistance	147370	1.460,00 €	328,00 €	1.638,00 €			x
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency	147440	819,00 €	328,00 €	997,00 €			x
IGF2R	Hepatocellular carcinoma, somatic	147280					1.900,00 €	
IGF2	Diabetes, IGF2 related	147470	468,00 €	328,00 €	646,00 €			x
IGHM	Agammaglobulinemia type 1, autosomal recessive	147020	468,00 €					
IGHMBP2	Neuronopathy distal hereditary motor type 6	600502	1.193,00 €	328,00 €	1.371,00 €			x
IGLL1	Agammaglobulinemia type 2, autosomal recessive	146770	374,00 €					
IKBKAP	HSAN3	603722	1.724,00 €	779,00 €	2.353,00 €			x
IKBKB	Immunodeficiency type 15	603258	1.326,00 €					
IKBKG	Atypical Mycobacterial infection	300248	796,00 €					x
IKZF1	Leukemia, acute lymphoblastic	603023					1.900,00 €	
IL1RAPL1	Mental retardation, X-linked type 21	300206	729,00 €	429,00 €	1.008,00 €			
IL1RN	Osteomyelitis, sterile multifocal, with periostitis and pustulosis	147679	655,00 €	421,00 €	926,00 €			x
IL2RA	Interleukin 2 receptor deficiency	147730	796,00 €					
IL2RG	Combined immunodeficiency, X-linked, moderate	308380	819,00 €					
IL2	Severe combined immunodeficiency due to IL2 deficiency	147680	374,00 €					
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	146661	819,00 €					
IL11RA	Craniosynostosis and dental anomalies	600939	729,00 €					

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IL12A	Interleukin 12A deficiency	161560	573,00 €					
IL12B	Psoriasis susceptibility type 11	161561	737,00 €					
IL12RB1	Atypical Mycobacterial infection	601604	1.193,00 €					
IL12RB2	Atypical Mycobacterial infection, IL12RB2 related	601642	1.127,00 €					
IL21R	Immunodeficiency, primary, autosomal recessive, IL21R-related	605383	729,00 €					
IL31RA	Amyloidosis, primary localized cutaneous, type 2	609510	1.326,00 €					
IL36RN	Psoriasis, generalized pustular	605507	468,00 €	390,00 €	708,00 €			x
ILDR1	Deafness, autosomal recessive type 42	609739	819,00 €					
IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type	614010	573,00 €					
IMPDH1	Leber congenital amaurosis type 11	146690	1.127,00 €	328,00 €	1.305,00 €			
IMPG2	Retinitis pigmentosa type 56, autosomal recessive	607056	1.521,00 €	430,00 €	1.801,00 €			
INF2	Focal segmental glomerulosclerosis type 5	610982	1.643,00 €	484,00 €	1.977,00 €			x
INHBA	FSH releasing protein deficiency	147290	468,00 €					
INPP5E	Joubert syndrome type 1	613037	862,00 €					x
INPPL1	Opsismodysplasia	600829	1.704,00 €					
INS	Diabetes mellitus type 1	176730	491,00 €	343,00 €	684,00 €			x
INSR	Diabetes mellitus, insulin-resistant with acanthosis nigricans	147670	1.399,00 €	463,00 €	1.712,00 €			x
INVS	Nephronophthisis type 2	243305	1.260,00 €					
IQCB1	Senior-Loken syndrome type 5	609237	928,00 €					
IQCE	Autism, IQCE related		1.338,00 €					
IQSEC2	Mental retardation, X-linked type 1	300522	1.338,00 €	445,00 €	1.633,00 €			
IRAK4	Invasive pneumococcal disease, recurrent isolated type 1	606883	862,00 €					
IRF6	Orofacial cleft type 6	607199	729,00 €	328,00 €	907,00 €			x
IRF8	Immunodeficiency type 32A, mycobacteriosis, autosomal dominant	601565	819,00 €					
IRX5	Hamamy syndrome	606195	491,00 €					
ISCA2	Multiple mitochondrial dysfunctions syndrome type 4	615317	468,00 €					
ISCU	Myopathy with lactic acidosis hereditary	611911	655,00 €					
ISG15	Immunodeficiency type 38	147571	386,00 €					
ISPD	Walker-Warburg syndrome	614631	729,00 €	429,00 €	1.008,00 €			x
ITGA2B	Thrombocytopenia, neonatal alloimmune	607759	1.572,00 €	632,00 €	2.054,00 €			x
ITGA2	Glycoprotein Ia C807T polymorphism	192974	1.278,00 €					
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	605025	1.521,00 €					
ITGA6	Epidermolysis bullosa junctionalis with pyloric atresia	147556	1.521,00 €	527,00 €	1.898,00 €			
ITGA7	Myopathy due to Integrin 7A deficiency	600536	1.704,00 €					
ITGAM	Systemic lupus erythematosus, susceptibility to	120980	1.582,00 €					
ITGB1	Leukocyte adhesion deficiency	135630	1.061,00 €					
ITGB2	Leukocyte adhesion deficiency	600065	1.061,00 €					
ITGB3	Thrombocytopenia, neonatal alloimmune	173470	1.127,00 €	445,00 €	1.422,00 €			x
ITGB4	Epidermolysis bullosa junctionalis with pyloric atresia	147557	1.977,00 €	842,00 €	2.669,00 €			x
ITGB6	Amelogenesis imperfecta type 1H	147558	1.127,00 €					

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ITK	Lymphoproliferative syndrome type 1	186973	1.127,00 €	328,00 €	1.305,00 €			
ITM2B	Dementia, familial, British type	603904	573,00 €					
ITPR1	Gillespie syndrome	147265	1.955,00 €					x
IVD	Isovaleric acidemia	607036	796,00 €	421,00 €	1.067,00 €			x
IYD	Thyroid dyshormonogenesis type 4	612025	737,00 €					
JAG1	Alagille syndrome type 1	601920	1.643,00 €	328,00 €	1.821,00 €			x
JAG2	Craniofacial and neuro-developmental abnormalities, JAG2 related	602570	1.643,00 €	548,00 €	2.041,00 €			x
JAGN1	Neutropenia, severe congenital type 6, autosomal recessive	616012	386,00 €					
JAK2	JAK2, selective sequencing of exons 12, 14 and 16	147796						
JAK3	SCID autosomal recessive T negative B positive type	600173	1.460,00 €					
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	606871	737,00 €					
JPH2	Cardiomyopathy, familial hypertrophic type 17	605267	573,00 €					
JPH3	Huntington disease-like type 2	605268				200,00 €		
JRK	Epilepsy, childhood absence, JRK related	603210	573,00 €					x
JUP	Arrhythmogenic right ventricular cardiomyopathy type 12	173325	1.193,00 €					
KANK1	Cerebral palsy type 2, spastic quadriplegic	607704	1.193,00 €	424,00 €	1.467,00 €			
KANSL1	Koolen syndrome	612452	1.127,00 €	328,00 €	1.305,00 €			
KARS	CMTRIB	601421	862,00 €					
KAT6A	Mental retardation, autosomal dominant type 32	601408	1.399,00 €					
KAT6B	Genitopatellar syndrome	605880	1.582,00 €	421,00 €	1.853,00 €			x
KBTBD13	Nemaline myopathy type 6	613727	468,00 €					
KCNA1	Episodic ataxia type 1	176260	468,00 €	328,00 €	646,00 €			x
KCNA2	Early infantile epileptic encephalopathy type 32	176262	468,00 €					
KCNA5	Atrial fibrillation type 7	176267	655,00 €					
KCNB1	Early infantile epileptic encephalopathy type 26	600397	655,00 €					
KCNC3	Spinocerebellar ataxia type 13, autosomal dominant	176264	737,00 €	390,00 €	977,00 €			x
KCND3	Spinocerebellar ataxia type 22, autosomal dominant	605411	796,00 €	437,00 €	1.083,00 €			x
KCNE1	Jervell and Lange-Nielsen syndrome type 2	176261	296,00 €	328,00 €	474,00 €			x
KCNE2	Atrial fibrillation type 4	603796	386,00 €	328,00 €	564,00 €			x
KCNE3	Brugada syndrome type 6	604433	296,00 €					
KCNH1	Temple-Baraitser syndrome	603305	928,00 €					
KCNH2	Long QT syndrome type 2	152427	1.193,00 €	328,00 €	1.371,00 €			x
KCNJ1	Bartter syndrome type 2	600359	573,00 €	343,00 €	766,00 €			x
KCNJ2	Short QT syndrome type 3	600681	491,00 €	328,00 €	669,00 €			x
KCNJ5	Hyperaldosteronism type 3	600734	491,00 €					
KCNJ10	SESAME syndrome	602208	374,00 €	343,00 €	567,00 €			x
KCNJ11	Diabetes mellitus, noninsulin-dependent	600937	468,00 €	296,00 €	614,00 €			x
KCNJ13	Leber congenital amaurosis type 16	603208	491,00 €					
KCNJ18	Thyrotoxic periodic paralysis type 2	613236	573,00 €	374,00 €	797,00 €			x
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia	600150	1.643,00 €					

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KCNQ1OT1	Beckwith-Wiedemann syndrome	604115		328,00 €				
KCNQ1	Atrial fibrillation type 3	607542	1.061,00 €	328,00 €	1.239,00 €			x
KCNQ2	Early infantile epileptic encephalopathy type 7	602235	1.260,00 €	328,00 €	1.438,00 €			x
KCNQ3	Seizures, benign neonatal, type 2	602232	1.127,00 €	328,00 €	1.305,00 €			x
KCNQ4	Deafness, autosomal dominant type 2A	603537	1.061,00 €	437,00 €	1.348,00 €			x
KCNT1	Early infantile epileptic encephalopathy type 14	608167	1.775,00 €	653,00 €	2.278,00 €			x
KCNV2	Retinal cone dystrophy type 3B	607604	573,00 €	343,00 €	766,00 €			x
KCTD3	Neurodevelopmental disorder, KCTD3 related	613272	1.326,00 €					
KCTD7	Progressive myoclonus epilepsy type 3	611725	491,00 €	437,00 €	778,00 €			x
KCTD17	DYT26, myoclonic	616386	819,00 €					
KDM5C	Mental retardation X-linked, syndromic, Claes-Jensen type	314690	1.582,00 €	548,00 €	1.980,00 €			
KDM6A	Kabuki syndrome type 2	300128	1.572,00 €	328,00 €	1.750,00 €			x
KDR	Hemangioma, capillary infantile, familial, susceptibility to	191306	1.572,00 €					
KEAP1	Goitre, multinodular	606016	655,00 €					
KEL	Hemolytic anemia, Kell-system related	613883	1.260,00 €					
KHDC3L	Hydatidiform mole, recurrent, type 2	611687	374,00 €	374,00 €	598,00 €			
KHK	Fructosuria essential	614058	655,00 €					
KIAA0196	Ritscher-Schinzel syndrome type 1	610657	1.643,00 €	611,00 €	2.104,00 €			x
KIAA0586	Joubert syndrome type 23	610178	1.825,00 €					
KIAA2022	Mental retardation, X-linked, nonsyndromic	300524	1.061,00 €					
KIF1A	HSN2C	601255	2.016,00 €	841,00 €	2.707,00 €			x
KIF1B	CMT2A1	605995	2.028,00 €	905,00 €	2.783,00 €			x
KIF1BP	Goldberg-Shprintzen megacolon syndrome	609367	729,00 €					
KIF1C	Spastic ataxia type 2, autosomal recessive	603060	1.460,00 €					
KIF2A	Neurodevelopmental malformation and microcephaly	602591	1.399,00 €					
KIF5A	SPG10	602821	1.764,00 €	611,00 €	2.225,00 €			x
KIF5C	Neurodevelopmental malformation and microcephaly	604593	1.764,00 €					
KIF7	Acrocallosal syndrome	611254	1.326,00 €	430,00 €	1.606,00 €			x
KIF11	Microcephaly with or without chorioretinopathy, Lymphedema, or Mental retardation, MCLMR	148760	1.338,00 €					
KIF21A	Fibrosis of extraocular muscles, congenital type 1	608283	1.927,00 €					
KIF23	Dyserythropoietic anemia, congenital, type 3	605064	1.278,00 €	484,00 €	1.612,00 €			x
KISS1R	Hypogonadotropic hypogonadism	604161	737,00 €	328,00 €	915,00 €			x
KISS1	Hypogonadotropic hypogonadism	603286	374,00 €	328,00 €	552,00 €			
KIT	Gastrointestinal stromal tumor, familial	164920	1.326,00 €	328,00 €	1.504,00 €			x
KLF1	Dyserythropoietic anemia, congenital, type 4	600599	468,00 €	374,00 €	692,00 €			x
KLF6	Gastric cancer, somatic	602053					1.900,00 €	
KLF8	Mental retardation non-syndromic	300286	655,00 €					
KLF11	Maturity-onset diabetes of the young type 7	603301	573,00 €	328,00 €	751,00 €			x
KLHL3	Pseudohypoaldosteronism type 2D	605775	1.061,00 €					x
KLHL7	Retinitis pigmentosa type 42, autosomal dominant	611119	729,00 €					

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KLK4	Amelogenesis imperfecta type 2A1	603767	491,00 €					
KMT2A	Wiedemann-Steiner syndrome	159555	2.262,00 €	758,00 €	2.870,00 €			x
KMT2C	Developmental delay, KMT2C related	606833	1.955,00 €	905,00 €	2.710,00 €			
KMT2D	Kabuki syndrome type 1	602113	2.106,00 €	328,00 €	2.284,00 €			x
KPTN	Mental retardation, autosomal recessive type 41	615620	862,00 €					
KRAS	Bladder cancer, somatic	190070					950,00 €	
KRIT1	Cerebral cavernous malformations type 1	604214	1.127,00 €	655,00 €	1.632,00 €			x
KRT1	Epidermolytic hyperkeratosis	139350	862,00 €					x
KRT2	Ichthyosis, bullous type	600194	729,00 €	421,00 €	1.000,00 €			x
KRT5	Epidermolysis bullosa simplex	148040	819,00 €	328,00 €	997,00 €			x
KRT6A	Pachyonychia congenita type 3	148041	737,00 €					x
KRT6B	Pachyonychia congenita type 4	148042	729,00 €					
KRT9	Epidermolytic palmoplantar keratoderma	607606	819,00 €	437,00 €	1.106,00 €			x
KRT10	Epidermolytic hyperkeratosis	148080	796,00 €	437,00 €	1.083,00 €			
KRT14	Dermatopathia pigmentosa reticularis	148066	862,00 €	437,00 €	1.149,00 €			x
KRT16	Pachyonychia congenita type 1	148067	737,00 €					
KRT17	Pachyonychia congenita type 2	148069	737,00 €					
KRT71	Hypotrichosis type 13	608245	729,00 €					
KRT74	Hypotrichosis type 3	608248	729,00 €					
KRT85	Ectodermal dysplasia type 4, hair/nail type	602767	729,00 €					
L1CAM	Hydrocephalus with aqueductal stenosis and congenital intestinal pseudoobstraction	308840	1.582,00 €	611,00 €	2.043,00 €			x
L2HGDH	L-2-hydroxyglutaric aciduria	609584	796,00 €	328,00 €	974,00 €			x
LAMA1	Poretti-Boltshauser syndrome	150320	1.955,00 €	905,00 €	2.710,00 €			
LAMA2	Muscular dystrophy type 1A	156225	2.282,00 €	655,00 €	2.787,00 €			x
LAMA3	Epidermolysis bullosa, generalized atrophic benign	600805	1.955,00 €	905,00 €	2.710,00 €			
LAMA5	Focal segmental glomerulosclerosis, LAMA5 related	601033	2.282,00 €					
LAMB1	Lissencephaly type 5	150240	1.927,00 €	716,00 €	2.493,00 €			
LAMB2	Nephrotic syndrome type 5	150325	1.764,00 €					x
LAMB3	Amelogenesis imperfecta type 1A	150310	1.460,00 €	484,00 €	1.794,00 €			x
LAMC1	Dandy-Walker malformation and occipital cephaloceles, LAMC1 related	150290	1.704,00 €					
LAMC2	Epidermolysis bullosa, junctional	150292	1.399,00 €					x
LAMC3	Cortical malformations, occipital	604349	1.825,00 €					
LAMP2	Danon disease	309060	928,00 €	421,00 €	1.199,00 €			x
LAMTOR2	Immunodeficiency due to defect in MAPBP-interacting protein	610389	468,00 €					
LARGE	Muscular dystrophy type 1D	603590	995,00 €	328,00 €	1.173,00 €			x
LARP7	Alazami syndrome	612026	862,00 €	445,00 €	1.157,00 €			
LBR	Greenberg skeletal dysplasia	600024	995,00 €					
LCA5	Leber congenital amaurosis type 5	611408	729,00 €	328,00 €	907,00 €			x
LCAT	Fish eye disease	606967	655,00 €	421,00 €	926,00 €			x
LCK	Immunodeficiency type 22	153390	729,00 €					

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LCT	Lactase deficiency, congenital	603202	1.582,00 €					
LDB3	Cardiomyopathy, dilated type 1C	605906	1.193,00 €					
LDHA	Glycogen storage disease type 11	150000	819,00 €	437,00 €	1.106,00 €			x
LDHB	Lactate dehydrogenase-B deficiency	150100	655,00 €					
LDLR	Hypercholesterolemia due to LDL-receptor-disorder autosomal dominant	606945	1.326,00 €	328,00 €	1.504,00 €			x
LDLRAP1	Hypercholesterolemia autosomal recessive	605747	729,00 €					
LEFTY2	Left-right axis malformations	601877	468,00 €					
LEMD3	Buschke-Ollendorff syndrome	607844	1.061,00 €	426,00 €	1.337,00 €			
LEP	Obesity due to leptin deficiency	164160	491,00 €	328,00 €	669,00 €			x
LFNG	Spondylocostal dysostosis, autosomal recessive type 3	602576	928,00 €	437,00 €	1.215,00 €			x
LGI1	Epilepsy, familial temporal lobe type 1	604619	819,00 €	328,00 €	997,00 €			x
LHB	Hypogonadotropic hypogonadism	152780	491,00 €					
LHCGR	Leydig cell hypoplasia type 1	152790	1.061,00 €	429,00 €	1.340,00 €			x
LHFPL5	Deafness, autosomal recessive type 67	609427	468,00 €					
LHX3	Pituitary hormone deficiency, combined type 3	600577	737,00 €	328,00 €	915,00 €			x
LHX4	Pituitary hormone deficiency, combined type 4	602146	655,00 €	328,00 €	833,00 €			x
LIAS	Pyruvate dehydrogenase lipoic acid synthetase deficiency	607031	729,00 €					
LIFR	Stuve-Wiedemann syndrome	151443	1.278,00 €	437,00 €	1.565,00 €			x
LIG4	LIG4 syndrome	601837	862,00 €					
LIM2	Cataract, cortical pulverulent, late-onset	154045	573,00 €					
LINS1	Mental retardation, autosomal recessive type 27	610350	729,00 €					
LIPA	Cholesteryl ester storage disease	613497	729,00 €	429,00 €	1.008,00 €			x
LIPH	Hypotrichosis type 7	607365	796,00 €					
LIPI	Hypertriglyceridemia, susceptibility to	609252	796,00 €					
LIPN	Ichthyosis, lamellar type 4	613924	819,00 €	421,00 €	1.090,00 €			x
LIPT1	Leigh syndrome due to pyruvate and alpha-ketoglutarate dehydrogenase deficiencies, LIPT1 related	610284	468,00 €					
LITAF	CMT1C	603795	491,00 €					
LMBRD1	Methylmalonic aciduria CblF type	612625	1.260,00 €					
LMNA	Cardiomyopathy, dilated type 1A	150330	995,00 €	328,00 €	1.173,00 €			x
LMNB1	Leukodystrophy demyelinating adult-onset, autosomal dominant	150340	862,00 €	328,00 €	1.040,00 €			x
LMX1B	Nail-Patella syndrome	602575	655,00 €	328,00 €	833,00 €			x
LONP1	CODAS syndrome	605490	1.326,00 €	421,00 €	1.597,00 €			x
LOR	Vohwinkel syndrome with ichthyosis	152445	374,00 €	343,00 €	567,00 €			x
LOXHD1	Deafness, autosomal recessive type 77	613072	1.931,00 €					
LOXL1	Exfoliation syndrome, susceptibility to	153456	796,00 €					
LPAR6	Hypotrichosis type 8	609239	468,00 €					
LPIN1	Myoglobinuria acute recurrent	605518	1.460,00 €	463,00 €	1.773,00 €			x
LPIN2	Majeed syndrome	605519	1.326,00 €					x
LPL	Hyperlipoproteinemia type 1	609708	729,00 €	328,00 €	907,00 €			x
LRAT	Retinitis pigmentosa juvenile	604863	374,00 €					

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LRBA	Immunodeficiency common variable type 8	606453	2.176,00 €	905,00 €	2.931,00 €			
LRIG2	Urofacial syndrome	608869	1.326,00 €					
LRP2	Donnai-Barrow syndrome	600073	1.955,00 €	905,00 €	2.710,00 €			
LRP5	Osteoporosis pseudoglioma syndrome	603506	1.521,00 €	328,00 €	1.699,00 €			x
LRPPRC	Leigh syndrome, French-Canadian type	607544	1.977,00 €					
LRRC6	Primary ciliary dyskinesia type 19	614930	928,00 €					
LRRC8A	Agammaglobulinemia type 5, autosomal recessive	608360	573,00 €					
LRRK2	PARK8 Parkinson	609007	2.028,00 €	328,00 €	2.206,00 €			x
LRSAM1	CMT2P	610933	1.582,00 €					
LRTOMT	Deafness, autosomal recessive type 63	612414	729,00 €					
LTBP2	Glaucoma, primary type 3D	602091	1.724,00 €					x
LTBP4	Cutis laxa type 1C, autosomal recessive	604710	1.673,00 €					
LYRM4	Combined oxidative phosphorylation deficiency type 19	613311	491,00 €					
LYST	Chediak-Higashi syndrome	606897	1.955,00 €					x
LZTFL1	Bardet-Biedl syndrome, LZTFL1 related	606568	819,00 €					
LZTR1	Noonan syndrome type 10	600574	1.338,00 €	328,00 €	1.516,00 €			
MACF1	Neurodevelopmental disorder, MACF1 related	608271	1.955,00 €	905,00 €	2.710,00 €			
MAF	Cataract, pulverulent or cerulean, with or without microcornea	177075	468,00 €	343,00 €	661,00 €			
MAFB	Multicentric carpotarsal osteolysis syndrome	608968	374,00 €					
MAGEL2	Schaaf-Yang syndrome	605283	819,00 €	328,00 €	997,00 €			
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia	300715	729,00 €					
MAK	Retinitis pigmentosa type 62, autosomal recessive	154235	1.061,00 €					
MALT1	Immunodeficiency type 12	604860	1.193,00 €					
MAMLD1	Hypospadias type 2, X-linked	300120	1.061,00 €	390,00 €	1.301,00 €			x
MAN1B1	Mental retardation, autosomal recessive type 15	604346	862,00 €					
MAN2B1	Mannosidosis-alpha	609458	1.326,00 €	505,00 €	1.681,00 €			x
MANBA	Mannosidosis-beta	609489	1.193,00 €	424,00 €	1.467,00 €			x
MANBAL	Mannosidosis, beta A, lysosomal-like		386,00 €					
MAOA	Brunner syndrome	309850	1.127,00 €					
MAP1A	Hearing loss, MAP1A related	600178	1.521,00 €					
MAP2K1	Cardiofaciocutaneous syndrome type 3	176872	796,00 €					
MAP2K2	Cardiofaciocutaneous syndrome type 4	601263	796,00 €	429,00 €	1.075,00 €			
MAPK10	Epileptic encephalopathy, Lennox-Gastaut type	602897	1.193,00 €					
MAPT	Dementia, frontotemporal	157140	1.193,00 €	328,00 €	1.371,00 €			x
MARS2	Combined oxidative phosphorylation deficiency type 25	609728	491,00 €					
MARVELD2	Deafness, autosomal recessive type 49	610572	737,00 €					
MASP1	3MC syndrome type 1	600521	1.278,00 €					
MASTL	Thrombocytopenia type 2	608221	1.061,00 €					
MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive	610550	819,00 €					
MAT2A	Aortic aneurysm, familial thoracic, MAT2A related	601468	819,00 €					

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maternal UPD chr. 7	Silver-Russell syndrome			328,00 €				
maternal UPD chr. 1	Temple syndrome			328,00 €				
MATN3	Epiphyseal dysplasia, multiple, type 5	602109	819,00 €					
MATR3	Amyotrophic lateral sclerosis type 21	164015	1.193,00 €	421,00 €	1.464,00 €			
MAX	Pheochromocytoma type 9	154950	737,00 €	328,00 €	915,00 €			
MBD1	Autism, MBD1 related	156535	1.061,00 €					
MBD5	Mental retardation, autosomal dominant type 1	611472	1.127,00 €					
MBL2	Mannose-binding protein deficiency	154545	491,00 €	437,00 €	778,00 €			x
MBTPS2	Ichthyosis follicularis, atricia, and photophobia syndrome	300294	862,00 €					
MC1R	Melanoma, cutaneous malignant	155555	374,00 €	296,00 €	520,00 €			
MC2R	Glucocorticoid deficiency type 1	607397	374,00 €					
MC4R	Obesity	155541	491,00 €	328,00 €	669,00 €			x
MCCC1	CoA-3-methylcrontonyl carboxylase 1 deficiency	609010	1.260,00 €					
MCCC2	CoA-3-methylcrontonyl carboxylase 2 deficiency	609014	1.127,00 €					
MCEE	Methylmalonyl-CoA epimerase deficiency	608419	491,00 €					
MCM4	Immunodeficiency with natural killer cell deficiency	602638	1.061,00 €					
MCM6	Lactose intolerance, adult type	601806	1.193,00 €					
MCOLN1	Mucolipidosis type 4	605248	796,00 €	437,00 €	1.083,00 €			x
MCPH1	Microcephaly, autosomal recessive type 1	607117	1.260,00 €	328,00 €	1.438,00 €			x
MCTP2	Coarctation of the aorta		1.460,00 €					
MDM2	Accelerated tumor formation, susceptibility to	164785	862,00 €	328,00 €	1.040,00 €			
MECP2	Angelman-like syndrome	300005	468,00 €	328,00 €	646,00 €			x
MED12	FG syndrome type 1	300188	1.888,00 €	772,00 €	2.510,00 €			x
MED13L	Mental retardation and distinctive facial features with or without cardiac defects	608771	1.775,00 €					
MED23	Mental retardation, autosomal recessive type 18	605042	1.673,00 €					
MED25	CMT2B2	610197	1.061,00 €	421,00 €	1.332,00 €			x
MEF2C	Mental retardation, autosomal dominant type 20	600662	862,00 €	328,00 €	1.040,00 €			x
MEFV	Mediterranean fever	608107	862,00 €	328,00 €	1.040,00 €			x
MEGF8	Carpenter syndrome type 2	604267	1.888,00 €	721,00 €	2.459,00 €			
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	612453	1.582,00 €	548,00 €	1.980,00 €			
MEN1	Adrenal adenoma, somatic	613733					1.900,00 €	
MEOX1	Klippel-Feil syndrome type 2, autosomal dominant	600147	374,00 €	374,00 €	598,00 €			
MERTK	Retinitis pigmentosa type 38, autosomal recessive	604705	1.399,00 €	430,00 €	1.679,00 €			x
MESP2	Spondylocostal dysostosis, autosomal recessive type 2	605195	468,00 €	343,00 €	661,00 €			x
MET	Renal cell carcinoma, papillary type 1, familial	164860	1.399,00 €	328,00 €	1.577,00 €			
MFF	Mitochondrial encephalomyopathy	614785	491,00 €					
MFN2	CMT2A2	608507	1.061,00 €	328,00 €	1.239,00 €			x
MFRP	Microphthalmia, isolated type 5	606227	995,00 €					
MFSD8	Ceroid lipofuscinosis neuronal type 7	611124	862,00 €	426,00 €	1.138,00 €			x
MGAT2	Glycosylation disorde type 2A	602616	468,00 €					

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MGME1	Mitochondrial DNA depletion syndrome type 11	615076	491,00 €					
MGP	Keutel syndrome	154870	491,00 €					
MID1	Opitz G syndrome	300552	928,00 €	328,00 €	1.106,00 €			x
MID2	Mental retardation, X-linked type 101	300204	729,00 €	429,00 €	1.008,00 €			
MIR17HG	Feingold syndrome type 2	609415	468,00 €					
MIR96	Deafness, autosomal dominant type 50	611606	156,00 €					
MITF	Melanoma, cutaneous malignant	156845	1.061,00 €	328,00 €	1.239,00 €			x
MKKS	Bardet-Biedl syndrome type 6	604896	655,00 €	421,00 €	926,00 €			x
MKS1	Bardet-Biedl syndrome type 13	609883	1.127,00 €	424,00 €	1.401,00 €			
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	605908	862,00 €	328,00 €	1.040,00 €			x
MLH1	Colorectal cancer, hereditary nonpolyposis type 2	120436	1.260,00 €	328,00 €	1.438,00 €			
MLH3	Colorectal cancer, hereditary nonpolyposis type 7	604395	1.278,00 €					
MLPH	Griscelli syndrome type 3	606526	1.061,00 €					x
MLYCD	Malonyl-CoA decarboxylase deficiency	606761	655,00 €	328,00 €	833,00 €			x
MMAA	Methylmalonic aciduria CblA type	607481	655,00 €					
MMAB	Methylmalonic aciduria CblB type	607568	819,00 €					x
MMACHC	Methylmalonic aciduria CblC type	609831	468,00 €	437,00 €	755,00 €			x
MMADHC	Methylmalonic aciduria CblD type	611935	655,00 €	437,00 €	942,00 €			x
MMP1	Epidermolysis bullosa dystrophica, autosomal recessive, modifier of	120353	729,00 €					
MMP2	Multicentric osteolysis, nodulosis, and arthropathy	120360	995,00 €	426,00 €	1.271,00 €			x
MMP3	Coronary heart disease, susceptibility to, type 6	185250	819,00 €					
MMP9	Metaphyseal anadysplasia type 2	120361	928,00 €					
MMP13	Metaphyseal anadysplasia type 1	600108	862,00 €					
MMP14	Winchester Syndrome	600754	729,00 €	429,00 €	1.008,00 €			x
MMP20	Amelogenesis imperfecta type 2A2	604629	729,00 €					
MMP21	Heterotaxy, visceral type 7	608416	655,00 €					
MMR genes	MMR genes methylation analysis			328,00 €				
MN1	Meningioma, MN1 deficiency related	156100	862,00 €					
MNX1	Currarino syndrome	142994	737,00 €	374,00 €	961,00 €			x
MOCS1	Molybdenum cofactor deficiency type A	603707	729,00 €	429,00 €	1.008,00 €			x
MOCS2	Molybdenum cofactor deficiency type B	603708	737,00 €	437,00 €	1.024,00 €			
MOGS	Glycosylation disorder type 2B	601336	819,00 €					
MPC1	Mitochondrial pyruvate carrier deficiency	614738	491,00 €					
MPDU1	Glycosylation disorder type 1F	604041	468,00 €					
MPDZ	Hydrocephalus, nonsyndromic, autosomal recessive type 2	603785	2.059,00 €	905,00 €	2.814,00 €			
MPI	Glycosylation disorder type 1B	154550	737,00 €					x
MPL	MPL, selective sequencing of exon 10	159530						
MPLKIP	Trichothiodystrophy, nonphotosensitive type 1	609188	386,00 €	343,00 €	579,00 €			x
MPV17	Mitochondrial DNA depletion syndrome type 6	137960	491,00 €	328,00 €	669,00 €			x
MPZ	CMT1B	159440	655,00 €	328,00 €	833,00 €			x

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MR1	Major histocompatibility comples 1 deficiency	600764	573,00 €	437,00 €	860,00 €			x
MRAP	Glucocorticoid deficiency type 2	609196	468,00 €					
MRE11A	Ataxia telangiectasia like disorder	600814	1.326,00 €	437,00 €	1.613,00 €			x
MRPL3	Combined oxidative phosphorylation deficiency type 9	607118	729,00 €					
MRPL44	Combined oxidative phosphorylation deficiency type 16	611849	468,00 €					
MRPS16	Combined oxidative phosphorylation deficiency type 2	609204	468,00 €					
MRPS22	Combined oxidative phosphorylation deficiency type 5	605810	737,00 €					
MSH2	Colorectal cancer, hereditary nonpolyposis type 1	609309	1.193,00 €	328,00 €	1.371,00 €			
MSH3	Familial adenomatous polyposis type 4	600887	1.521,00 €	505,00 €	1.876,00 €			
MSH6	Colorectal cancer, hereditary nonpolyposis type 5	600678	1.127,00 €	328,00 €	1.305,00 €			
MSMO1	Microcephaly, MSMO1 related	607545	573,00 €					
MSRB3	Deafness, autosomal recessive type 74	613719	655,00 €					
MSTN	Muscle hypertrophy	601788	468,00 €					x
MSX1	Orofacial cleft type 5	142983	468,00 €	343,00 €	661,00 €			x
MSX2	Craniosynostosis type 2	123101	491,00 €	343,00 €	684,00 €			x
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma	156540	737,00 €	328,00 €	915,00 €			x
MT-ATP6	Leber optic atrophy	516060	296,00 €					x
MT-ATP8	Cardiomyopathy, apical hypertrophic, and neuropathy, MT-ATP8 related	516070	296,00 €					
MT-CO1	Cytochrome c oxidase 1 deficiency	516030	296,00 €					x
MT-CO2	Cytochrome c oxidase 2 deficiency	516040	296,00 €					
MT-CO3	Cytochrome c oxidase 3 deficiency	516050	296,00 €					x
MT-CYB	Leber optic atrophy	516020	296,00 €					x
MTFMT	Combined oxidative phosphorylation deficiency type 15	611766	819,00 €	421,00 €	1.090,00 €			
MTHFR	Homocystinuria	607093	729,00 €	328,00 €	907,00 €			x
MTM1	Myotubular myopathy X-linked	300415	1.061,00 €	328,00 €	1.239,00 €			x
MTMR2	CMT4B1	603557	1.061,00 €	328,00 €	1.239,00 €			x
MTMR14	Centronuclear myopathy type 1	611089	1.326,00 €					
MT-ND1	Leber optic atrophy	516000	296,00 €					x
MT-ND2	Leber optic atrophy	516001	296,00 €					x
MT-ND3	Leigh syndrome due to mitochondrial complex I deficiency	516002	296,00 €					x
MT-ND4L	Leber optic atrophy	516004	296,00 €					
MT-ND4	Leber optic atrophy	516003	296,00 €					x
MT-ND5	Leber optic atrophy	516005	296,00 €					x
MT-ND6	Leber optic atrophy	516006	296,00 €					x
MTO1	Combined oxidative phosphorylation deficiency type 10	614667	1.061,00 €	426,00 €	1.337,00 €			x
MTOR	Neurodevelopmental disorder, MTOR related	601231	2.067,00 €	905,00 €	2.822,00 €			
MTR	Methylcobalamin deficiency CblG type	156570	1.876,00 €					
MT-RNR1	Deafness, nonsyndromic, sensorineural, mitochondrial	561000	296,00 €					x
MT-RNR2	Chloramphenicol resistance, MT-RNR2 related	561010	296,00 €					
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	602568	1.127,00 €					

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MT-TA	Mitochondrial myopathy, MT-TA related	590000	296,00 €					
MT-TC	MELAS syndrome	590020	296,00 €					
MT-TD	Mitochondrial myopathy, isolated	590015	296,00 €					
MT-TE	Mitochondrial myopathy, infantile, transient, MT-TE related	590025	296,00 €					
MT-TF	MELAS syndrome	590070	296,00 €					
MT-TG	Cardiomyopathy, hypertrophic, MT-TG related	590035	296,00 €					
MT-TH	Cardiomyopathy, idiopathic dilated, mitochondrial, MT-TH related	590040	296,00 €					
MT-TI	Cardiomyopathy, fatal, MT-TI related	590045	296,00 €					
MT-TK	MERRF syndrome, MT-TK related	590060	296,00 €					
MT-TL1	MELAS syndrome, MT-TL1 related	590050	296,00 €					
MT-TL2	Encephalomyopathy, mitochondrial, MT-TL2 related	590055	296,00 €					
MT-TM	Mitochondrial myopathy, MT-TM related	590065	296,00 €					
MT-TN	Mitochondrial complex I deficiency, MT-TN related	590010	296,00 €					
MTTP	Abetalipoproteinemia	157147	1.326,00 €					
MT-TP	MERRF syndrome, MT-TP related	590075	296,00 €					
MT-TQ	Myopathy, MT-TQ related	590030	296,00 €					
MT-TR	Encephalomyopathy, mitochondrial, MT-TR related	590005	296,00 €					
MT-TS1	MERRF/MELAS overlap syndrome, MT-TS1 related	590080	296,00 €					
MT-TS2	MERRF/MELAS overlap syndrome, MT-TS2 related	590085	296,00 €					
MT-TT	Parkinson disease, susceptibility to, MT-TT related	590090	296,00 €					
MT-TV	Ataxia, progressive seizures, mental deterioration, and hearing loss, MT-TV related	590105	296,00 €					
MT-TW	Encephalopathy, mitochondrial, MT-TW related	590095	296,00 €					
MT-TY	Focal segmental glomerulosclerosis and dilated cardiomyopath, MT-TY related	590100	296,00 €					
MUSK	Myasthenic syndrome associated with acetylcholine receptor deficiency	601296	1.260,00 €	445,00 €	1.555,00 €			x
MUT	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	609058	995,00 €	426,00 €	1.271,00 €			x
MUTYH	Familial adenomatous polyposis type 2	604933	862,00 €	328,00 €	1.040,00 €			
MVK	Mevalonic aciduria	251170	729,00 €	429,00 €	1.008,00 €			x
MXRA5	Autism spectrum, MXRA5 related		1.399,00 €					
MYBPC1	Arthrogryposis, distal, type 1B	160794	1.724,00 €	632,00 €	2.206,00 €			
MYBPC3	Cardiomyopathy, dilated	600958	1.825,00 €	328,00 €	2.003,00 €			x
MYCN	Feingold syndrome	164840	573,00 €	328,00 €	751,00 €			x
MYD88	Macroglobulinemia, Waldenstrom, somatic	602170					1.900,00 €	
MYF6	Centronuclear myopathy type 3	159991	468,00 €					
MYH2	Inclusion body myopathy	160740	1.825,00 €					
MYH3	Arthrogryposis, distal, type 2A	160720	1.759,00 €	704,00 €	2.313,00 €			x
MYH6	Atrial septal defect type 3	160710	1.673,00 €					
MYH7B	Cardiomyopathy, left ventricular noncompaction, MYH7B related	609928	1.775,00 €					
MYH7	Cardiomyopathy, dilated type 1S	160760	1.759,00 €	328,00 €	1.937,00 €			x
MYH8	Arthrogryposis, distal, type 7	160741	1.977,00 €	842,00 €	2.669,00 €			x
MYH9	Deafness, autosomal dominant type 17	160775	2.028,00 €	328,00 €	2.206,00 €			x

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MYH11	Aortic aneurysm, familial thoracic type 4	160745	2.028,00 €	905,00 €	2.783,00 €			
MYH14	Deafness, autosomal dominant type 4	608568	1.845,00 €					
MYL2	Cardiomyopathy, familial hypertrophic type 10	160781	491,00 €					x
MYL3	Cardiomyopathy, familial hypertrophic type 8	160790	573,00 €					
MYLK2	Cardiomyopathy, hypertrophic, midventricular, digenic	606566	1.127,00 €					
MYLK	Aortic aneurysm, familial thoracic type 7	600922	1.724,00 €					
MYO1A	Deafness, autosomal dominant type 48	601478	1.326,00 €					
MYO1E	Focal segmental glomerulosclerosis type 6	601479	1.764,00 €					
MYO3A	Deafness, autosomal recessive type 30	606808	1.775,00 €	737,00 €	2.362,00 €			x
MYO5A	Griscelli syndrome type 1	160777	2.262,00 €	704,00 €	2.816,00 €			x
MYO5B	Diarrhea type 2 with microvillus atrophy	606540	1.759,00 €	328,00 €	1.937,00 €			x
MYO6	Deafness, autosomal dominant type 22	600970	1.775,00 €					
MYO7A	Deafness, autosomal dominant type 11	276903	2.016,00 €	841,00 €	2.707,00 €			x
MYO15A	Deafness, autosomal recessive type 3	602666	2.282,00 €	905,00 €	3.037,00 €			
MYO16	Autism spectrum disorder, MYO16 related	615479	1.876,00 €	905,00 €	2.631,00 €			
MYO18B	Klippel-Feil syndrome type 4, autosomal dominant, with myopathy and facial dysmorphism	607295	2.145,00 €	905,00 €	2.900,00 €			
MYOC	Glaucoma, open angle type 1A	601652	655,00 €					
MYOT	Limb-girdle muscular dystrophy, autosomal dominant type 1A	604103	819,00 €	328,00 €	997,00 €			x
MYOZ2	Cardiomyopathy, familial hypertrophic type 16	605602	655,00 €					
MYPN	Cardiomyopathy, dilated type 1KK	608517	1.278,00 €					
NAA10	Microphthalmia, syndromic type 1	300013	737,00 €					
NAGA	Schindler disease	104170	819,00 €					x
NAGLU	Mucopolysaccharidosis type 3B	609701	819,00 €	421,00 €	1.090,00 €			x
NAGS	N-acetylglutamate synthase deficiency	608300	819,00 €	437,00 €	1.106,00 €			x
NAIP	Spinal muscular atrophy (SMA), NAIP related	600355	1.061,00 €					
NALCN	Neuroaxonal neurodegeneration, infantile, with facial dysmophism	611549	1.888,00 €	905,00 €	2.643,00 €			
NANOS1	Oligo-astheno-teratozoospermia	608226	729,00 €					
NARS2	Combined oxidative phosphorylation deficiency type 24	612803	928,00 €	437,00 €	1.215,00 €			
NBAS	Infantile liver failure syndrome type 2	608025	2.145,00 €					
NBN	Nijmegen breakage syndrome	602667	1.260,00 €					
NCAM1	Neurodevelopmental disorder, NCAM1 related	116930	1.521,00 €					
NCF1	Granulomatous disease, chronic, autosomal recessive, cytochrome b- positive, type 1	608512	862,00 €	429,00 €	1.141,00 €			x
NCF2	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 2	608515	995,00 €					
NCF4	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 3	601488	729,00 €					
NDE1	Lissencephaly type 4 with microcephaly	609449	737,00 €					
NDN	Prader-Willi syndrome	602117	468,00 €	328,00 €	646,00 €			x
NDP	Exudative vitreoretinopathy type 2	300658	491,00 €	374,00 €	715,00 €			x
NDRG1	CMT4D	605262	1.061,00 €					x
NDST1	Mental retardation, autosomal recessive type 46	600853	1.193,00 €					
NDUFA1	Mitochondrial complex I deficiency	300078	374,00 €					

Pricelist by gene

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NDUFA2	Leigh syndrome	602137	374,00 €					
NDUFA9	Leigh syndrome	603834	796,00 €					
NDUFA10	Leigh syndrome	603835	729,00 €					
NDUFA11	Mitochondrial complex I deficiency	612638	573,00 €					
NDUFA12	Leigh syndrome due to mitochondrial complex I deficiency	614530	468,00 €					
NDUFA13	Hurthle cell thyroid carcinoma, due to germline NDUFA13 mutation	609435	491,00 €					
NDUFAF1	Leigh syndrome	606934	374,00 €					
NDUFAF2	Leigh syndrome	609653	573,00 €	437,00 €	860,00 €			
NDUFAF3	Leigh syndrome	612911	468,00 €					
NDUFAF4	Mitochondrial complex I deficiency	611776	819,00 €					
NDUFAF5	Mitochondrial complex I deficiency	612360	796,00 €	429,00 €	1.075,00 €			x
NDUFAF6	Leigh syndrome	612392	819,00 €	421,00 €	1.090,00 €			
NDUFB3	Mitochondrial complex I deficiency	603839	374,00 €					
NDUFS1	Mitochondrial complex I deficiency	157655	1.338,00 €	430,00 €	1.618,00 €			x
NDUFS2	Mitochondrial complex I deficiency	602985	862,00 €					
NDUFS3	Leigh syndrome	603846	573,00 €					
NDUFS4	Leigh syndrome	602694	491,00 €					x
NDUFS6	Mitochondrial complex I deficiency	603848	468,00 €					
NDUFS7	Leigh syndrome	601825	737,00 €	437,00 €	1.024,00 €			x
NDUFS8	Leigh syndrome	602141	374,00 €					
NDUFV1	Mitochondrial complex I deficiency	161015	737,00 €					
NDUFV2	Mitochondrial complex I deficiency	600532	737,00 €	437,00 €	1.024,00 €			
NEB	Nemaline myopathy type 2, autosomal recessive	161650	1.955,00 €					x
NECAP1	Early infantile epileptic encephalopathy type 21	611623	737,00 €					
NEFH	Amyotrophic lateral sclerosis, susceptibility to	162230	862,00 €	437,00 €	1.149,00 €			x
NEFL	CMT1F	162280	573,00 €	328,00 €	751,00 €			x
NEK1	Short-rib thoracic dysplasia type 6 with or without polydactyly	604588	1.825,00 €					
NEK8	Nephronophthisis type 9	609799	995,00 €					
NEU1	Neuraminidase deficiency	608272	573,00 €	421,00 €	844,00 €			x
NEUROD1	Maturity-onset diabetes of the young type 6	601724	491,00 €	328,00 €	669,00 €			x
NEUROG3	Diarrhea type 4, malabsorptive, congenital	604882	386,00 €	343,00 €	579,00 €			x
NEXN	Cardiomyopathy, dilated type 1CC	613121	1.061,00 €					
NF1	Neurofibromatosis type 1	613113	2.225,00 €	655,00 €	2.730,00 €			x
NF2	Meningioma, NF2-related, somatic	607379					1.900,00 €	
NFIX	Sotos-like syndrome	164005	928,00 €	429,00 €	1.207,00 €			x
NFKB2	Immunodeficiency common variable type 10	164012	1.338,00 €					
NFU1	Multiple mitochondrial dysfunctions syndrome type 1	608100	737,00 €					
NGEF	Neurodevelopmental disorder, NGEF related	605991	995,00 €					
NGF	HSAN5	162030	386,00 €	374,00 €	610,00 €			x
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing ra	611290	655,00 €	437,00 €	942,00 €			x

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NHLRC1	Myoclonic epilepsy of Lafora	608072	386,00 €	328,00 €	564,00 €			x
NHP2	Dyskeratosis congenita, autosomal recessive type 2	606470	374,00 €					x
NHS	Cataract, X-linked	300457	1.399,00 €	421,00 €	1.670,00 €			x
NIN	Seckel syndrome type 7	608684	1.825,00 €					
NIPA1	SPG6	608145	573,00 €	328,00 €	751,00 €			x
NIPAL4	Ichthyosiform erythroderma, congenital, nonbullous type 1	609383	737,00 €					
NIPBL	Cornelia de Lange syndrome type 1	608667	2.145,00 €	655,00 €	2.650,00 €			x
NKX2-1	Chorea, hereditary benign	600635	468,00 €	328,00 €	646,00 €			x
NKX2-2	Maturity-onset diabetes of the young, NKX2-2 related	604612	491,00 €					
NKX2-5	Atrial septal defect with atrioventricular conduction defects	600584	655,00 €	328,00 €	833,00 €			x
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia	602183	573,00 €	343,00 €	766,00 €			x
NLGN3	Asperger syndrome susceptibility X-linked type 2	300336	796,00 €	437,00 €	1.083,00 €			
NLGN4X	Autism susceptibility, X-linked type 2	300427	796,00 €	328,00 €	974,00 €			
NLRP1	Corneal intraepithelial dyskeratosis and ectodermal dysplasia	606636	1.460,00 €	424,00 €	1.734,00 €			x
NLRP3	Muckle-wells syndrome	606416	995,00 €	429,00 €	1.274,00 €			x
NLRP7	Hydatidiform mole	609661	796,00 €	429,00 €	1.075,00 €			
NLRP12	Cold autoinflammatory syndrome type 2	609648	928,00 €					
NME8	Primary ciliary dyskinesia type 6	607421	1.326,00 €					
NMNAT1	Leber congenital amaurosis type 9	608700	491,00 €	437,00 €	778,00 €			
NOD2	Blau syndrome	605956	1.326,00 €	421,00 €	1.597,00 €			x
NODAL	Heterotaxy, visceral type 5	601265	468,00 €	328,00 €	646,00 €			
NOL3	Myoclonus, familial cortical	605235	468,00 €					
NOP10	Dyskeratosis congenita, autosomal recessive type 1	606471	386,00 €					
NOP56	Spinocerebellar ataxia type 36, autosomal dominant	614154				200,00 €		
NOTCH1	Aortic valve disease type 1	190198	1.876,00 €	716,00 €	2.442,00 €			x
NOTCH2	Alagille syndrome type 2	600275	1.977,00 €	716,00 €	2.543,00 €			x
NOTCH3	CADASIL	600276	1.876,00 €	328,00 €	2.054,00 €			
NOTCH4	Schizophrenia, NOTCH4 related	164951	1.673,00 €					
NPC1	Niemann-Pick disease type C1	607623	1.643,00 €	328,00 €	1.821,00 €			x
NPC2	Niemann-Pick disease type C2	601015	491,00 €	328,00 €	669,00 €			x
NPHP1	Joubert syndrome type 4	607100	1.278,00 €	328,00 €	1.456,00 €			x
NPHP3	Nephronophthisis type 3	608002	1.704,00 €					
NPHP4	Nephronophthisis type 4	607215	1.825,00 €	632,00 €	2.307,00 €			x
NPHS1	Nephrosis, Finnish type	602716	1.582,00 €	611,00 €	2.043,00 €			x
NPHS2	Nephrotic syndrome	604766	737,00 €	437,00 €	1.024,00 €			x
NPM1	NPM1, selective sequencing of exon 11	164040						
NPPA	Atrial fibrillation type 6	108780	374,00 €					
NPR2	Acromesomelic dysplasia, Maroteaux type	108961	1.278,00 €	463,00 €	1.591,00 €			x
NR0B1	Adrenal hypoplasia	300473	491,00 €	328,00 €	669,00 €			x
NR1H4	Cholestasis, infantile, NR1H4 related	603826	796,00 €					

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NR2E1	Polymicrogyria bilateral occipital	603849	928,00 €					
NR2E3	Retinitis pigmentosa type 37, autosomal dominant/recessive	604485	737,00 €					
NR3C1	Glucocorticoid resistance, generalized	138040	862,00 €	421,00 €	1.133,00 €			
NR3C2	Hypertension early onset	600983	928,00 €	421,00 €	1.199,00 €			x
NR5A1	SPGF8	184757	737,00 €	328,00 €	915,00 €			x
NRAS	Colorectal cancer, hereditary	164790	468,00 €					
NRG1	Hirschsprung disease	142445	995,00 €					
NRL	Retinitis pigmentosa type 27, autosomal dominant	162080	386,00 €					
NRTN	Hirschsprung disease	602018	468,00 €	328,00 €	646,00 €			
NRXN1	Pitt-Hopkins syndrome	600565	1.825,00 €	328,00 €	2.003,00 €			x
NSD1	Beckwith-Wiedemann syndrome	606681	1.775,00 €	328,00 €	1.953,00 €			x
NSDHL	CHILD syndrome	300275	737,00 €	421,00 €	1.008,00 €			x
NSMF	Hypogonadotropic hypogonadism	608137	995,00 €	424,00 €	1.269,00 €			x
NSUN2	Mental retardation, autosomal recessive type 5	610916	1.326,00 €	430,00 €	1.606,00 €			x
NT5C2	SPG45	600417	995,00 €					
NTF4	Glaucoma, open angle type 1F	162662	386,00 €					
NTHL1	Familial adenomatous polyposis type 3	602656	491,00 €	421,00 €	762,00 €			
NTNG1	Autism, NTNG1 related	608818	729,00 €	328,00 €	907,00 €			x
NTRK1	HSAN4	191315	1.193,00 €	424,00 €	1.467,00 €			x
NUBPL	Leigh syndrome	613621	796,00 €					
NXF5	Mental retardation non-syndromic	300319	1.260,00 €					
NYX	Night blindness, congenital stationary type 1A	300278	491,00 €					
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia	613349	796,00 €	429,00 €	1.075,00 €			x
OBSL1	Three M syndrome type 2	610991	1.704,00 €	442,00 €	1.996,00 €			x
OCA2	Albinism, oculocutaneous type 2	611409	1.460,00 €	328,00 €	1.638,00 €			x
OCLN	Band-like calcification with simplified gyration and polymicrogyria	602876	729,00 €	421,00 €	1.000,00 €			
OCRL	Dent disease type 2	300535	1.460,00 €	505,00 €	1.815,00 €			x
OFD1	Joubert syndrome type 10	300170	1.521,00 €	484,00 €	1.855,00 €			x
OGDH	Alpha-ketoglutarate dehydrogenase deficiency	613022	1.399,00 €					
OPA1	Optic atrophy type 1	605290	1.764,00 €	328,00 €	1.942,00 €			x
OPA3	3-methylglutaconic aciduria type 3	606580	468,00 €	343,00 €	661,00 €			x
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	300127	1.521,00 €	527,00 €	1.898,00 €			x
OPLAH	5-oxoprolinase deficiency	614243	1.127,00 €					
OPTN	Amyotrophic lateral sclerosis type 12	602432	928,00 €	424,00 €	1.202,00 €			x
OR13H1	Autism, OR13H1 related		374,00 €					
ORAI1	Immunodeficiency type 9	610277	386,00 €					
ORC1	Meier-Gorlin syndrome type 1	601902	1.127,00 €	424,00 €	1.401,00 €			x
OSMR	Amyloidosis, primary localized cutaneous, type 1	601743	1.326,00 €					
OSTM1	Osteopetrosis, autosomal recessive type 5	607649	1.127,00 €					
OTC	Ornithine transcarbamoylase deficiency	300461	862,00 €	328,00 €	1.040,00 €			x

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OTOA	Deafness, autosomal recessive type 22	607038	1.825,00 €					
OTOF	Deafness, autosomal recessive type 9	603681	1.931,00 €	807,00 €	2.588,00 €			x
OTX2	Microphthalmia syndromic type 5	600037	468,00 €	390,00 €	708,00 €			x
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency	601424	1.193,00 €	424,00 €	1.467,00 €			x
OXTR	Autism, OXTR related	167055	468,00 €					
P2RX2	Progressive hearing loss	600844	862,00 €					
P2RY12	Bleeding disorder, platelet-type 8	600515	374,00 €					
P3H1	Osteogenesis imperfecta type 8	610339	1.061,00 €					
PABPN1	Muscular dystrophy, oculopharyngeal	602279	655,00 €	437,00 €	942,00 €	200,00 €		
PACS1	Mental retardation, autosomal dominant type 17	607492	1.278,00 €					
PAFAH1B1	Lissencephaly type 1	601545	729,00 €	328,00 €	907,00 €			x
PAH	Phenylketonuria	612349	928,00 €	328,00 €	1.106,00 €			x
PAK3	Mental retardation, X-linked type 30	300142	1.193,00 €	328,00 €	1.371,00 €			
PALB2	Breast cancer, susceptibility to	610355	1.193,00 €	328,00 €	1.371,00 €			x
PANK2	Pantothenate kinase-associated neurodegeneration	606157	819,00 €	328,00 €	997,00 €			x
PARK2	PARK2 Parkinson	602544	862,00 €	328,00 €	1.040,00 €			x
PARK7	PARK7 Parkinson	602533	655,00 €	328,00 €	833,00 €			x
PARN	Dyskeratosis congenita, autosomal recessive type 6	604212	1.460,00 €					
paternal UPD chr. 14	Kagami-Ogata syndrome			328,00 €				
PAX2	Focal segmental glomerulosclerosis type 7	167409	862,00 €	429,00 €	1.141,00 €			x
PAX3	Waardenburg syndrome type 1	606597	796,00 €	328,00 €	974,00 €			x
PAX4	Maturity-onset diabetes of the young type 9	167413	729,00 €	328,00 €	907,00 €			x
PAX5	Leukemia, acute lymphoblastic, susceptibility to	167414					1.900,00 €	
PAX6	Aniridia	607108	1.061,00 €	328,00 €	1.239,00 €			x
PAX8	Hypothyroidism congenital nongoitrous type 2, familial	167415	928,00 €	328,00 €	1.106,00 €			
PAX9	Tooth agenesis, selective type 3	167416	573,00 €	390,00 €	813,00 €			x
PC	Leigh syndrome due to pyruvate carboxylase deficiency	608786	1.278,00 €	484,00 €	1.612,00 €			x
PCBD1	Hyperphenylalaninemia, BH4 deficient, type D	126090	374,00 €					
PCCA	Propionic acidemia	232000	1.521,00 €	328,00 €	1.699,00 €			x
PCCB	Propionic acidemia	232050	1.193,00 €	424,00 €	1.467,00 €			x
PCDH11X	Dyslexia	300246	1.193,00 €					
PCDH15	Deafness, autosomal recessive type 23	605514	2.059,00 €	328,00 €	2.237,00 €			
PCDH19	Early infantile epileptic encephalopathy type 9	300460	862,00 €	328,00 €	1.040,00 €			x
PCK1	Phosphoenolpyruvate carboxykinase deficiency, cytosolic	614168	729,00 €					
PCK2	Phosphoenolpyruvate carboxykinase deficiency, mitochondrial	614095	796,00 €					
PCNT	Microcephalic osteodysplastic primordial dwarfism type 2	605925	2.145,00 €	807,00 €	2.802,00 €			x
PCSK1	Obesity with impaired prohormone processing	162150	1.127,00 €					
PCSK9	Hypercholesterolemia autosomal dominant type 3	607786	862,00 €	421,00 €	1.133,00 €			x
PDCD10	Cerebral cavernous malformations type 3	609118	655,00 €	328,00 €	833,00 €			x
PDE4D	Acrodysostosis 2	600129	1.521,00 €	445,00 €	1.816,00 €			x

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PDE6A	Retinitis pigmentosa type 43, autosomal recessive	180071	1.460,00 €					
PDE6B	Night blindness, congenital stationary, autosomal dominant type 2	180072	1.521,00 €					
PDE6C	Cone-rod dystrophy type 4	600827	1.399,00 €					
PDE6D	Joubert syndrome type 22	602676	491,00 €					
PDE6G	Retinitis pigmentosa type 57, autosomal recessive	180073	374,00 €					
PDE6H	Achromatopsia type 6	601190	468,00 €					
PDE8B	Striatal degeneration	603390	1.521,00 €					
PDGFB	Basal ganglia calcification type 5, idiopathic	190040	655,00 €	437,00 €	942,00 €			x
PDGFRA	Gastrointestinal stromal tumor, somatic	173490					1.900,00 €	
PDGFRB	Basal ganglia calcification type 4	173410	1.399,00 €	484,00 €	1.733,00 €			x
PDHA1	Leigh syndrome, X-linked	300502	862,00 €	328,00 €	1.040,00 €			x
PDHB	Pyruvate dehydrogenase E1-beta deficiency	179060	819,00 €	429,00 €	1.098,00 €			x
PDHX	Lacticacidemia due to PDX1 deficiency	608769	796,00 €	429,00 €	1.075,00 €			
PDP1	Pyruvate dehydrogenase phosphatase deficiency	605993	491,00 €					
PDSS1	Coenzyme Q10 deficiency type 2	607429	729,00 €	421,00 €	1.000,00 €			x
PDSS2	Coenzyme Q10 deficiency type 3	610564	737,00 €	437,00 €	1.024,00 €			x
PDX1	Maturity-onset diabetes of the young type 4	600733	468,00 €	328,00 €	646,00 €			x
PDYN	Spinocerebellar ataxia type 23, autosomal dominant	131340	374,00 €	328,00 €	552,00 €			x
PDZD7	Usher syndrome type 2C	612971	1.061,00 €					
PEAR1	Platelet aggregation disorder	610278	1.521,00 €					
PECAM1	Immunological disorder, PECAM1 related	173445	1.061,00 €					
PEPD	Prolidase deficiency	613230	1.061,00 €					
PET100	Mitochondrial complex IV deficiency	614770	374,00 €					
PEX1	Heimler syndrome type 1	602136	1.643,00 €	505,00 €	1.998,00 €			x
PEX2	Zellweger syndrome	170993	468,00 €					x
PEX3	Peroxisome biogenesis disorder type 10A	603164	796,00 €					x
PEX5	Peroxisome biogenesis disorder type 2A	600414	1.061,00 €	424,00 €	1.335,00 €			x
PEX6	Zellweger syndrome	601498	1.193,00 €	424,00 €	1.467,00 €			
PEX7	Refsum disease	601757	729,00 €	429,00 €	1.008,00 €			x
PEX10	Zellweger syndrome	602859	655,00 €					x
PEX11B	Peroxisome biogenesis disorder 14B	603867	573,00 €	328,00 €	751,00 €			
PEX12	Zellweger syndrome	601758	468,00 €	374,00 €	692,00 €			x
PEX13	Zellweger syndrome	601789	491,00 €	437,00 €	778,00 €			
PEX14	Zellweger syndrome	601791	729,00 €					
PEX16	Zellweger syndrome	603360	819,00 €					
PEX19	Zellweger syndrome	600279	655,00 €	437,00 €	942,00 €			x
PEX26	Zellweger syndrome	608666	491,00 €					x
PFKM	Glycogen storage disease type 7	610681	1.338,00 €	527,00 €	1.715,00 €			x
PFN1	Amyotrophic lateral sclerosis type 18	176610	374,00 €					
PGAM2	Glycogen storage disease type 10	612931	468,00 €	374,00 €	692,00 €			x

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PGAP1	Mental retardation, autosomal recessive type 42	611655	1.704,00 €					
PGAP2	Hyperphosphatasia with mental retardation syndrome type 3	615187	729,00 €					
PGAP3	Hyperphosphatasia with mental retardation syndrome type 4	611801	737,00 €					
PGK1	Phosphoglycerate kinase 1 deficiency	311800	862,00 €					
PGM1	Glycogen storage disease type 14	171900	995,00 €	421,00 €	1.266,00 €			
PHC1	Microcephaly, autosomal recessive type 11	602978	1.193,00 €					
PHEX	Hypophosphatemic rickets, X-linked	300550	1.521,00 €	328,00 €	1.699,00 €			x
PHF6	Borjeson-Forssman-Lehmann syndrome	300414	819,00 €	429,00 €	1.098,00 €			x
PHF8	Mental retardation, X-linked, Siderius type	300560	1.704,00 €					
PHGDH	Phosphoglycerate dehydrogenase deficiency	606879	796,00 €	421,00 €	1.067,00 €			x
PHKA1	Muscle glycogenosis	311870	1.622,00 €	674,00 €	2.146,00 €			
PHKA2	Glycogen storage disease type 9A	300798	1.673,00 €	695,00 €	2.218,00 €			x
PHKB	Glycogen storage disease type 9B	172490	1.622,00 €	653,00 €	2.125,00 €			x
PHKG2	Glycogen storage disease type 9C	172471	796,00 €					x
PHOX2A	Central hypoventilation syndrome, congenital	602753	491,00 €	374,00 €	715,00 €			x
PHOX2B	Central hypoventilation syndrome with or without Hirschsprung disease	603851	468,00 €	374,00 €	692,00 €	200,00 €		x
PHYH	Refsum disease	602026	819,00 €	421,00 €	1.090,00 €			x
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	600286	2.145,00 €					
PICALM	Immunological disorder, PICALM related	603025	1.338,00 €					
PIEZO1	Dehydrated hereditary stomatocytosis	611184	2.106,00 €	796,00 €	2.752,00 €			
PIEZO2	Arthrogryposis, distal, type 3	613629	2.184,00 €	811,00 €	2.845,00 €			
PIGA	Early infantile epileptic encephalopathy type 20	311770	655,00 €	421,00 €	926,00 €			
PIGL	CHIME syndrome	605947	655,00 €					
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome type 1	606097	1.704,00 €	653,00 €	2.207,00 €			x
PIGO	Hyperphosphatasia with mental retardation syndrome type 2	614730	928,00 €					
PIGQ	Neurodevelopmental disorder, PIGQ related	605754	796,00 €					
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome type 3	610272	796,00 €					
PIGV	Hyperphosphatasia with mental retardation syndrome type 1	610274	573,00 €					
PIGW	Hyperphosphatasia with mental retardation syndrome type 5	610275	468,00 €					
PIK3CA	Cowden syndrome type 5	171834	1.326,00 €	442,00 €	1.618,00 €			
PIK3CD	Immunodeficiency type 14	602839	1.326,00 €					
PIK3R1	Agammaglobulinemia type 7, autosomal recessive	171833	1.260,00 €					
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	603157	995,00 €	424,00 €	1.269,00 €			x
PIK3R5	Ataxia-oculomotor apraxia type 3	611317	1.127,00 €	430,00 €	1.407,00 €			x
PINK1	PARK6 Parkinson	608309	819,00 €	328,00 €	997,00 €			
PITPNM3	Cone-rod dystrophy type 5	608921	1.399,00 €					
PITX1	Club foot	602149	491,00 €	374,00 €	715,00 €			x
PITX2	Axenfeld-Rieger syndrome type 1	601542	573,00 €	328,00 €	751,00 €			x
PITX3	Anterior segment mesenchymal dysgenesis	602669	491,00 €					
PKD1	Polycystic kidney disease type 1, autosomal dominant	601313	2.176,00 €	655,00 €	2.681,00 €			x

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PKD2	Polycystic kidney disease type 2, autosomal dominant	173910	1.127,00 €	328,00 €	1.305,00 €			x
PKHD1L1	Autism, PKHD1L1 related	607843	1.955,00 €	905,00 €	2.710,00 €			
PKHD1	Polycystic kidney and hepatic disease	606702	1.955,00 €	655,00 €	2.460,00 €			x
PKLR	Adenosine triphosphate, elevated, of erythrocytes	609712	796,00 €	328,00 €	974,00 €			x
PKP1	Ectodermal dysplasia/skin fragility syndrome	601975	995,00 €					
PKP2	Arrhythmogenic right ventricular cardiomyopathy type 9	602861	1.127,00 €	328,00 €	1.305,00 €			x
PLA2G5	Fleck retina, familial benign	601192	491,00 €					
PLA2G6	Infantile neuroaxonal dystrophy type 1	603604	1.127,00 €	328,00 €	1.305,00 €			x
PLCB1	Early infantile epileptic encephalopathy type 12	607120	1.673,00 €	674,00 €	2.197,00 €			x
PLCB4	Auriculocondylar syndrome type 2	600810	1.876,00 €	758,00 €	2.484,00 €			x
PLCE1	Nephrotic syndrome type 3	608414	1.825,00 €	695,00 €	2.370,00 €			
PLEC	Epidermolysis bullosa simplex with muscular dystrophy	601282	1.955,00 €					x
PLEKHG4	Spinocerebellar ataxia type 4, autosomal dominant	609526	1.260,00 €	484,00 €	1.594,00 €			x
PLEKHG5	Spinal muscular atrophy distal, autosomal recessive type 4	611101	1.338,00 €	463,00 €	1.651,00 €			x
PLEKHM1	Osteopetrosis, autosomal recessive type 6	611466	928,00 €					
PLG	Plasminogen deficiency type 1	173350	1.326,00 €					
PLN	Cardiomyopathy, dilated type 1P	172405	296,00 €					x
PLOD1	Ehlers-Danlos syndrome type 6	153454	1.326,00 €	328,00 €	1.504,00 €			x
PLOD2	Osteogenesis imperfecta with congenital joint contractures	601865	1.278,00 €					
PLP1	Pelizaeus-Merzbacher disease	300401	737,00 €	328,00 €	915,00 €			x
PLS3	Bone mineral density QTL18, osteoporosis	300131	1.193,00 €	328,00 €	1.371,00 €			
PLXNB3	Adrenoleukodystrophy, x-linked	300214	2.028,00 €					
PMM2	Glycosylation disorder type 1A	601785	819,00 €	437,00 €	1.106,00 €			
PMP22	CMT1A	601097	468,00 €	328,00 €	646,00 €			x
PMS1	Nonpolyposis hereditary colon cancer, PMS1 related	600258	1.193,00 €	426,00 €	1.469,00 €			x
PMS2	Colorectal cancer, hereditary nonpolyposis type 4	600259	995,00 €	328,00 €	1.173,00 €			
PNKD	Paroxysmal nonkinesigenic dyskinesia	609023	796,00 €	429,00 €	1.075,00 €			x
PNKP	Ataxia-oculomotor apraxia type 4	605610	796,00 €	424,00 €	1.070,00 €			x
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency	164050	491,00 €					
PNPLA1	Ichthyosis congenital, autosomal recessive, PNPLA1 related	612121	819,00 €					
PNPLA2	Neutral lipid storage disease with myopathy	609059	737,00 €					
PNPLA6	SPG39	603197	1.622,00 €	716,00 €	2.188,00 €			x
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	603287	655,00 €	437,00 €	942,00 €			x
PNPT1	Combined oxidative phosphorylation deficiency type 13	610316	1.521,00 €					
POC1B	Cone-rod dystrophy type 20	614784	862,00 €					
POGZ	Mental retardation, autosomal dominant type 37	614787	1.521,00 €					
POLD1	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome	174761	1.127,00 €					
POLE	FILS syndrome	174762	2.059,00 €	905,00 €	2.814,00 €			
POLG2	Progressive external ophthalmoplegia with mitochondrial deletions type 4	604983	737,00 €	328,00 €	915,00 €			x
POLG	Mitochondrial DNA depletion syndrome type 4A	174763	1.521,00 €	328,00 €	1.699,00 €			x

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POLH	Xeroderma pigmentosum, variant type	603968	928,00 €					x
POLR1C	Treacher Collins syndrome type 3	610060	655,00 €					
POLR1D	Treacher Collins syndrome type 2	613715	468,00 €	343,00 €	661,00 €			x
POLR3A	Leukodystrophy hypomyelinating type 7	614258	1.572,00 €	653,00 €	2.075,00 €			x
POLR3B	Leukodystrophy hypomyelinating type 8	614366	1.704,00 €	590,00 €	2.144,00 €			x
POMC	Obesity with adrenal insufficiency and red hair	176830	374,00 €	328,00 €	552,00 €			x
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A3	606822	1.061,00 €	328,00 €	1.239,00 €			x
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A8	614828	468,00 €					
POMK	Limb-girdle muscular dystrophy, autosomal recessive type 12C	615247	386,00 €					
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	613386	573,00 €					
POMT1	Limb-girdle muscular dystrophy, autosomal recessive type 2K	607423	1.193,00 €	328,00 €	1.371,00 €			x
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A2	607439	1.278,00 €	328,00 €	1.456,00 €			x
POR	Adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	124015	1.061,00 €	328,00 €	1.239,00 €			x
PORCN	Focal dermal hypoplasia	300651	729,00 €	445,00 €	1.024,00 €			x
POT1	Melanoma, cutaneous malignant, familial type 10, susceptibility to	606478	1.326,00 €					
POU1F1	Pituitary hormone deficiency type 1	173110	573,00 €	328,00 €	751,00 €			x
POU3F4	Deafness, X-linked type 2	300039	468,00 €	296,00 €	614,00 €			x
POU4F3	Deafness, autosomal dominant type 15	602460	468,00 €					
PPARG	Carotid intimal medial thickness type 1	601487	655,00 €	328,00 €	833,00 €			
PPIB	Osteogenesis imperfecta type 9	123841	491,00 €					
PPM1K	Maple syrup urine disease, mild variant	611065	573,00 €					
PPOX	Porphyria variegata	600923	862,00 €	328,00 €	1.040,00 €			
PPP1R8	Cardiac defects, PPP1R8 related	602636	737,00 €					
PPP2R2B	Spinocerebellar ataxia type 12, autosomal dominant	604325				200,00 €		x
PPT1	Ceroid lipofuscinosis neuronal type 1	600722	819,00 €	421,00 €	1.090,00 €			x
PQBP1	Renpenning syndrome	300463	491,00 €	328,00 €	669,00 €			x
PRCD	Retinitis pigmentosa type 36, autosomal recessive	610598	374,00 €					
PRDM12	HSAN8	616458	573,00 €					
PRDM16	Cardiomyopathy, dilated type 1LL	605557	1.193,00 €					
PREPL	Cystinuria	609557	1.127,00 €	328,00 €	1.305,00 €			x
PRF1	Aplastic anemia	170280	491,00 €	328,00 €	669,00 €			x
PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	604283	1.260,00 €	426,00 €	1.536,00 €			
PRICKLE1	Progressive myoclonus epilepsy type 1A	608500	729,00 €	437,00 €	1.016,00 €			x
PRICKLE2	Epilepsy, progressive myoclonic type 5	608501	819,00 €					
PRKAG2	Cardiomyopathy, familial hypertrophic type 6	602743	1.127,00 €	424,00 €	1.401,00 €			x
PRKAR1A	Acrodysostosis type 1, with or without hormone resistance	188830	796,00 €	429,00 €	1.075,00 €			x
PRKCD	Autoimmune lymphoproliferative syndrome type 3	176977	1.260,00 €					
PRKCG	Spinocerebellar ataxia type 14, autosomal dominant	176980	1.260,00 €	430,00 €	1.540,00 €			x
PRKCSH	Polycystic liver disease	177060	995,00 €					
PRKDC	Immunodeficiency type 26, with or without neurologic abnormalities	600899	1.955,00 €	905,00 €	2.710,00 €			x

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PRKG1	Aortic aneurysm, familial thoracic type 8	176894	1.326,00 €					
PRKRA	DYT16	603424	655,00 €	328,00 €	833,00 €			x
PRNP	Creutzfeldt-Jakob disease	176640	386,00 €	343,00 €	579,00 €	200,00 €		
PROC	Protein C Deficiency, AD	612283	737,00 €	328,00 €	915,00 €			x
PRODH	Hyperprolinemia type 1	606810	1.061,00 €					
PROK2	Kallmann syndrome type 4	607002	468,00 €	328,00 €	646,00 €			x
PROKR2	Hypogonadotropic hypogonadism type 3 with or without anosmia	607123	468,00 €	328,00 €	646,00 €			x
PROM1	Cone-rod dystrophy type 12	604365	1.704,00 €	569,00 €	2.123,00 €			x
PROP1	Pituitary hormone deficiency type 2	601538	374,00 €	328,00 €	552,00 €			x
PROS1	Protein S Deficiency, autosomal dominant	176880	1.061,00 €	328,00 €	1.239,00 €			x
PRPF3	Retinitis pigmentosa type 18, autosomal dominant	607301	1.061,00 €					x
PRPF6	Retinitis pigmentosa type 60	613979	1.643,00 €					
PRPF8	Retinitis pigmentosa type 13, autosomal dominant	607300	1.876,00 €	905,00 €	2.631,00 €			x
PRPF31	Retinitis pigmentosa type 11, autosomal dominant	606419	796,00 €	328,00 €	974,00 €			x
PRPH2	Choroidal dystrophy, central areolar type 2	179605	468,00 €	328,00 €	646,00 €			x
PRPS1	Arts syndrome	311850	655,00 €	437,00 €	942,00 €			x
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis	614386	468,00 €	374,00 €	692,00 €			x
PRSS1	Pancreatitis	276000	573,00 €	328,00 €	751,00 €			x
PRSS2	Pancreatitis, chronic, protection against	601564	491,00 €					
PRSS56	Microphthalmia, isolated type 6	613858	995,00 €					
PRX	CMT4F	605725	995,00 €	328,00 €	1.173,00 €			x
PSAP	Gaucher disease, atypical	176801	1.061,00 €	445,00 €	1.356,00 €			x
PSAT1	Phosphoserine aminotransferase deficiency	610936	819,00 €					x
PSEN1	Acne inversa familial type 3	104311	928,00 €	328,00 €	1.106,00 €			x
PSEN2	Alzheimer disease type 4	600759	729,00 €	328,00 €	907,00 €			x
PSMB8	Autoinflammation, lipodystroph and dermatosis syndrome	177046	655,00 €	421,00 €	926,00 €			x
PSPH	Phosphoserine phosphatase deficiency	172480	573,00 €					
PSTPIP1	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	606347	1.061,00 €					
PTCH1	Basal cell nevus syndrome	601309	1.764,00 €	328,00 €	1.942,00 €			x
PTCH2	Basal cell nevus syndrome due to germline PTCH2 mutation	603673	1.460,00 €					
PTCHD1	Autism susceptibility, X-linked type 4	300828	655,00 €					
PTDSS1	Lenz-Majewski hyperostotic dwarfism	612792	995,00 €					
PTEN	Cowden syndrome type 1	601728	796,00 €	328,00 €	974,00 €			x
PTF1A	Pancreatic agenesis type 2	607194	374,00 €	343,00 €	567,00 €			
PTH1R	Chondrodysplasia, Blomstrand type	168468	1.061,00 €	424,00 €	1.335,00 €			x
PTH	Hypoparathyroidism	168450	374,00 €					
PTPN11	Leukemia, juvenile myelomonocytic	176876					1.900,00 €	
PTPN14	Choanal atresia and lymphedema	603155	1.399,00 €					
PTPN23	Ciliogenesis related disorder	606584	1.521,00 €					
PTPRC	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	151460	1.724,00 €	695,00 €	2.269,00 €			

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PTPRQ	Deafness, autosomal recessive type 84	603317	2.145,00 €	905,00 €	2.900,00 €			x
PTRF	Lipodystrophy generalized type 4	603198	468,00 €	343,00 €	661,00 €			x
PTS	Hyperphenylalaninemia, BH4-deficient, type A	612719	819,00 €	421,00 €	1.090,00 €			x
PURA	Mental retardation, autosomal dominant type 31	600473	386,00 €					
PUS1	Mitochondrial myopathy and sideroblastic anemia type 1	608109	573,00 €					
PVRL1	Orofacial cleft type 7	600644	819,00 €					
PXDN	Corneal opacification and other ocular anomalies	605158	1.643,00 €					
PYCR1	Cutis laxa type 2B, autosomal recessive	179035	737,00 €					
PYGL	Glycogen storage disease type 6B	613741	1.278,00 €	437,00 €	1.565,00 €			x
PYGM	Glycogen storage disease type 5	608455	1.326,00 €	437,00 €	1.613,00 €			x
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	603727	1.278,00 €					
QDPR	Hyperphenylalaninemia, BH4 deficient, type C	612676	737,00 €					x
RAB3GAP1	Warburg micro syndrome type 1	602536	1.460,00 €	527,00 €	1.837,00 €			x
RAB3GAP2	Martsolf syndrome	609275	1.775,00 €	905,00 €	2.530,00 €			
RAB7A	CMT2B	602298	491,00 €	328,00 €	669,00 €			x
RAB18	Warburg micro syndrome 3	602207	655,00 €					
RAB23	Carpenter syndrome	606144	573,00 €	437,00 €	860,00 €			
RAB27A	Griscelli syndrome type 2	603868	491,00 €	437,00 €	778,00 €			x
RAB39B	Mental retardation, X-linked type 72	300774	374,00 €					
RAB40AL	Mental retardation, X-linked	300405	374,00 €					
RABGGTA	Autism spectrum disorder	601905	862,00 €					
RAC2	Neutrophil immunodeficiency syndrome	602049	655,00 €					
RAD21	Cornelia de Lange syndrome type 4	606462	1.061,00 €	437,00 €	1.348,00 €			
RAD50	Hereditary breast and ovarian cancer syndrome, RAD50 related	604040	1.582,00 €	328,00 €	1.760,00 €			
RAD51C	Breast-ovarian cancer	602774	819,00 €	328,00 €	997,00 €			
RAD51D	Breast-ovarian cancer, familial, susceptibility to, type 4	602954	796,00 €	328,00 €	974,00 €			
RAD51	Mirror movements type 2	179617	796,00 €	429,00 €	1.075,00 €			
RAF1	Noonan syndrome type 5	164760	1.061,00 €	424,00 €	1.335,00 €			x
RAG1	Severe combined immunodeficiency, B cell-negative	179615	819,00 €					
RAG2	Combined cellular and humoral immune defects with granulomas	179616	491,00 €	328,00 €	669,00 €			x
RAI1	Smith-Magenis syndrome	607642	1.193,00 €	328,00 €	1.371,00 €			x
RANBP2	Encephalopathy acute necrotizing type 1	601181	1.931,00 €	611,00 €	2.392,00 €			x
RAPSN	Fetal akinesia deformation sequence	601592	655,00 €	437,00 €	942,00 €			x
RARS2	Pontocerebellar hypoplasia type 6	611524	1.278,00 €					x
RARS	Leukodystrophy hypomyelinating type 9	107820	1.061,00 €					
RASA1	Capillary malformation-arteriovenous malformation	139150	1.643,00 €	328,00 €	1.821,00 €			x
RAX2	Cone-rod dystrophy type 11	610362	374,00 €					
RAX	Microphthalmia, isolated type 3	601881	491,00 €					
RB1	Hereditary Retinoblastoma	614041	1.673,00 €	328,00 €	1.851,00 €			x
RBBP8	Jawad syndrome	604124	1.278,00 €					x

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RBCK1	Polyglucosan body myopathy type 1 with or without immunodeficiency	610924	862,00 €					
RBM8A	Thromocytopenia-Absent-Radius-Syndrome	605313	491,00 €	421,00 €	762,00 €			x
RBM20	Cardiomyopathy, dilated type 1DD	613171	1.260,00 €					
RBP3	Retinitis pigmentosa type 66, autosomal recessive	180290	862,00 €					
RBPJ	Adams-Oliver syndrome type 3	147183	995,00 €					
RD3	Leber congenital amaurosis type 12	180040	386,00 €					
RDH5	Fundus albipunctatus	601617	491,00 €					
RDH12	Retinitis pigmentosa type 53, autosomal recessive	608830	737,00 €	328,00 €	915,00 €			x
RDX	Deafness, autosomal recessive type 24	179410	1.193,00 €					
RECQL4	Baller-Gerold syndrome	603780	1.193,00 €	442,00 €	1.485,00 €			
RECQL	Breast cancer, susceptibility to	600537	928,00 €					
REEP1	SPG31	609139	737,00 €	328,00 €	915,00 €			x
REEP2	SPG72	609347	655,00 €					
RELN	Epilepsy, familial temporal lobe type 7	600514	1.955,00 €					x
REN	Renal tubular dysgenesis	179820	729,00 €	429,00 €	1.008,00 €			x
RET	Central hypoventilation syndrome, congenital	164761	1.338,00 €	328,00 €	1.516,00 €			x
RFT1	Glycosylation disorder type 1N	611908	928,00 €					
RFX6	Maturity-onset diabetes of the young, RFX6 related	612659	1.278,00 €	430,00 €	1.558,00 €			x
RFXANK	Bare lymphocyte syndrome, type 2	603200	737,00 €	429,00 €	1.016,00 €			x
RGR	Retinitis pigmentosa type 44, autosomal dominant/recessive	600342	655,00 €					
RGS9BP	Bradyopsia	607814	655,00 €					
RGS9	Bradyopsia	604067	1.460,00 €					
RHBDF2	Tylosis with esophageal cancer	614404	1.260,00 €					
RHO	Night blindness type 1, congenital stationary, autosomal dominant	180380	491,00 €	328,00 €	669,00 €			x
RIMS1	Cone-rod dystrophy type 7	606629	2.028,00 €	905,00 €	2.783,00 €			
RIN2	Macrocephaly, alopecia, cutis laxa, and scoliosis	610222	1.127,00 €					
RINT1	Breast cancer, RINT1 related	610089	862,00 €					
RIPK4	Popliteal pterygium syndrome, lethal type	605706	796,00 €					x
RIT1	Noonan syndrome type 8	609591	491,00 €					x
RLBP1	Bothnia retinal dystrophy	180090	573,00 €	421,00 €	844,00 €			
RMND1	Combined oxidative phosphorylation deficiency type 11	614917	862,00 €					
RMRP	Cartilage-hair hypoplasia	157660	296,00 €	296,00 €	442,00 €			x
RNASEH2A	Aicardi-Goutieres syndrome type 4	606034	655,00 €	328,00 €	833,00 €			x
RNASEH2B	Aicardi-Goutieres syndrome type 2	610326	796,00 €	328,00 €	974,00 €			x
RNASEH2C	Aicardi-Goutieres syndrome type 3	610330	374,00 €	328,00 €	552,00 €			x
RNASEL	Prostate cancer, hereditary type 1	180435	819,00 €					
RNASET2	Leukoencephalopathy, cystic without megalencephaly	612944	819,00 €	421,00 €	1.090,00 €			x
RNF39	Hippocampal longterm potentiation, RFN39 related	607524	573,00 €					
RNF128	Autism, RNF128 related	300439	737,00 €					
RNF213	Moyamoya disease type 2, susceptibility to	613768	1.955,00 €	905,00 €	2.710,00 €			

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RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism type 1	601428	296,00 €					
ROBO2	Vesicoureteral reflux type 2	602431	1.704,00 €	590,00 €	2.144,00 €			x
ROBO3	Gaze palsy, horizontal, with progressive scoliosis	608630	1.582,00 €					
ROGDI	Kohlschutter Tonz syndrome	614574	655,00 €	429,00 €	934,00 €			x
ROM1	Retinitis pigmentosa type 7	180721	573,00 €					
ROR2	Brachydactyly type B1	602337	862,00 €	328,00 €	1.040,00 €			x
RP1L1	Occult macular dystrophy	608581	1.521,00 €	437,00 €	1.808,00 €			x
RP1	Retinitis pigmentosa type 1, autosomal dominant	603937	1.775,00 €	328,00 €	1.953,00 €			x
RP2	Retinitis pigmentosa type 2 X-linked	300757	573,00 €	328,00 €	751,00 €			x
RP9	Retinitis pigmentosa type 9, autosomal dominant	607331	491,00 €					
RPE65	Retinitis pigmentosa type 20, autosomal recessive	180069	796,00 €	328,00 €	974,00 €			x
RPGR	Retinitis pigmentosa type 3 X-linked	312610	1.521,00 €	328,00 €	1.699,00 €			x
RPGRIP1L	COACH syndrome	610937	1.704,00 €					x
RPGRIP1	Cone-rod dystrophy type 13	605446	1.582,00 €	328,00 €	1.760,00 €			x
RPL5	Diamond-Blackfan anemia type 6	603634	819,00 €	328,00 €	997,00 €			x
RPL10	Autism susceptibility, X-linked type 5	312173	573,00 €					
RPL11	Diamond-Blackfan anemia type 7	604175	655,00 €	328,00 €	833,00 €			x
RPL15	Diamond-Blackfan anemia type 12	604174	468,00 €	437,00 €	755,00 €			x
RPL21	Hypotrichosis type 12	603636	491,00 €					
RPL26	Diamond-Blackfan anemia type 11	603704	374,00 €	437,00 €	661,00 €			x
RPL35A	Diamond-Blackfan anemia type 5	180468	468,00 €	328,00 €	646,00 €			x
RPS6KA3	Coffin-Lowry syndrome	300075	1.521,00 €	328,00 €	1.699,00 €			x
RPS7	Diamond-Blackfan anemia type 8	603658	737,00 €					
RPS10	Diamond-Blackfan anemia type 9	603632	491,00 €					
RPS17	Diamond-Blackfan anemia type 4	180472	573,00 €	328,00 €	751,00 €			x
RPS19	Diamond-Blackfan anemia type 1	603474	737,00 €	328,00 €	915,00 €			x
RPS24	Diamond-blackfan anemia type 3	602412	737,00 €					x
RPS26	Diamond-Blackfan anemia type 10	603701	468,00 €	328,00 €	646,00 €			x
RPS28	Diamond Blackfan anemia type 15 with mandibulofacial dysostosis	603685	374,00 €	437,00 €	661,00 €			x
RPS29	Diamond-Blackfan anemia type 13	603633	374,00 €	374,00 €	598,00 €			x
RRM1	Autism, RRM1 related		1.278,00 €					
RRM2B	Mitochondrial DNA depletion syndrome 8B, MNGIE type	604712	729,00 €	328,00 €	907,00 €			x
RS1	Retinoschisis	300839	573,00 €	421,00 €	844,00 €			
RSPH1	Primary ciliary dyskinesia type 24	609314	819,00 €					
RSPH4A	Primary ciliary dyskinesia type 11	612647	655,00 €					
RSPH9	Primary ciliary dyskinesia type 12	612648	655,00 €					
RTEL1	Dyskeratosis congenita, autosomal recessive type 5	608833	1.775,00 €					x
RTN2	SPG12	603183	796,00 €	429,00 €	1.075,00 €			x
RTN3	Alzheimers disease, RTN3 related	604249	729,00 €					
RTTN	Microcephaly, short stature, and polymicrogyria with seizures	610436	2.106,00 €					

Pricelist by gene

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RUBCN	Salih ataxia	613516	1.338,00 €					
RUNX1	Leukemia, acute myeloid	151385					1.900,00 €	
RUNX2	Cleidocranial dysplasia	600211	819,00 €	328,00 €	997,00 €			x
RXFP2	Cryptorchidism	606655	1.278,00 €					
RYR1	Central core disease	180901	1.955,00 €	655,00 €	2.460,00 €			x
RYR2	Arrhythmogenic right ventricular dysplasia type 2	180902	1.955,00 €					x
SACS	Spastic ataxia Charlevoix-Saguenay type	604490	1.802,00 €	328,00 €	1.980,00 €			x
SAG	Oguchi disease	181031	1.061,00 €					
SALL1	Townes-Brocks syndrome	602218	862,00 €	374,00 €	1.086,00 €			x
SALL4	Duane Retraction syndrome	607343	729,00 €	437,00 €	1.016,00 €			x
SAMHD1	Aicardi-Goutieres syndrome type 5	606754	1.061,00 €	328,00 €	1.239,00 €			x
SAR1B	Chylomicron retention disease	607690	573,00 €					
SARDH	Sarcosinemia	604455	1.338,00 €					
SATB2	Glass syndrome	612313	862,00 €					
SBDS	Aplastic anemia, SBDS related	607444	491,00 €	390,00 €	731,00 €			x
SBF2	CMT4B2	607697	1.973,00 €	328,00 €	2.151,00 €			x
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure	602257	862,00 €	421,00 €	1.133,00 €			x
SCARF2	Van den Ende-Gupta syndrome	613619	928,00 €					
SCN1A	Early infantile epileptic encephalopathy type 6	182389	1.572,00 €	328,00 €	1.750,00 €			x
SCN1B	Brugada syndrome type 5	600235	573,00 €	374,00 €	797,00 €			x
SCN2A	Convulsions, benign familial infantile, 3	182390	1.673,00 €	569,00 €	2.092,00 €			x
SCN3A	Epilepsy, focal, SCN3A related	182391	1.825,00 €					
SCN3B	Brugada syndrome type 7	608214	491,00 €					
SCN4A	Hyperkalemic periodic paralysis	603967	1.764,00 €	328,00 €	1.942,00 €			x
SCN4B	Long QT syndrome type 10	608256	491,00 €					
SCN5A	Atrial fibrillation type 10	600163	1.673,00 €	328,00 €	1.851,00 €			x
SCN8A	Early infantile epileptic encephalopathy type 13	600702	1.825,00 €					
SCN9A	Dravet syndrome, modifier of	603415	1.572,00 €	569,00 €	1.991,00 €			x
SCN10A	Episodic pain syndrome type 2, familial	604427	1.622,00 €					
SCN11A	Episodic pain syndrome type 3, familial	604385	1.572,00 €					
SCNN1A	Bronchiectasis with or without elevated sweat chloride type 2	600228	862,00 €	421,00 €	1.133,00 €			x
SCNN1B	Liddle syndrome	600760	796,00 €	426,00 €	1.072,00 €			x
SCNN1G	Liddle syndrome	600761	796,00 €	426,00 €	1.072,00 €			x
SCO1	Hepatic failure, early onset, and neurologic disorder	603644	737,00 €					
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency	604272	468,00 €	343,00 €	661,00 €			
SCP2	Leukoencephalopathy with dystonia and motor neuropathy	184755	1.193,00 €					
SDCCAG8	Senior-Loken syndrome type 7	613524	1.338,00 €					
SDHA	Cardiomyopathy, dilated type 1GG	600857	1.061,00 €	328,00 €	1.239,00 €			x
SDHAF1	Mitochondrial complex II deficiency	612848	374,00 €	328,00 €	552,00 €			
SDHAF2	Pheochromocytoma type 5	613019	468,00 €	328,00 €	646,00 €			x

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SDHB	Paragangliomas type 4	185470	737,00 €	328,00 €	915,00 €			
SDHC	Pheochromocytoma type 3	602413	573,00 €	328,00 €	751,00 €			x
SDHD	Carcinoid tumors, intestinal	602690	468,00 €	328,00 €	646,00 €			x
SEC23B	Anemia dyserythropoietic type 2	610512	1.278,00 €	437,00 €	1.565,00 €			x
SECISBP2	Thyroid hormone metabolism abnormal	607693	1.193,00 €					
SELENON	Myopathy with fiber-type disproportion	606210	995,00 €	426,00 €	1.271,00 €			x
SEMA3A	Kallmann syndrome, SEMA3A related	603961	1.193,00 €	424,00 €	1.467,00 €			x
SEMA4A	Retinitis pigmentosa type 35, autosomal dominant/recessive	607292	1.127,00 €					
SEMA4C	Retinitis pigmentosa SEMA4C related	604462	1.193,00 €					
SEPSECS	Pontocerebellar hypoplasia type 2D	613009	819,00 €					
SEPT9	Amyotrophy hereditary neuralgic	604061	1.127,00 €	328,00 €	1.305,00 €			x
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	614725	1.127,00 €	424,00 €	1.401,00 €			
SERHL2	Serine hydrolase deficiency, SERHL2 related		862,00 €					
SERPINA1	Antitrypsin-alpha-1 deficiency	107400	573,00 €	437,00 €	860,00 €			x
SERPINB6	Deafness, autosomal recessive type 91	173321	819,00 €					
SERPINC1	Antithrombin III deficiency	107300	819,00 €	328,00 €	997,00 €			x
SERPINE1	Plasminogen activator inhibitor type 1	173360	655,00 €					x
SERPINF1	Osteogenesis imperfecta type 6	172860	655,00 €					
SERPING1	Angioedema, hereditary	606860	819,00 €	328,00 €	997,00 €			x
SERPINH1	Osteogenesis imperfecta type 10	613848	573,00 €					
SERPINI1	Encephalopathy, familial, with neuroserpin inclusion bodies	602445	737,00 €					x
SETBP1	Schinzel-Giedion midface retraction syndrome	611060	1.127,00 €					
SETD2	Autism, SETD2 related	612778	1.876,00 €					
SETD5	Mental retardation, autosomal dominant type 23	615743	1.521,00 €					
SETX	Amyotrophic lateral sclerosis type 4	608465	1.825,00 €	328,00 €	2.003,00 €			x
SF3B4	Acrofacial dysostosis 1, Nager type	605593	655,00 €	421,00 €	926,00 €			x
SFTPA1	Pulmonary fibrosis, idiopathic	178630	573,00 €					
SFTPA2	Pulmonary fibrosis, idiopathic	178642	491,00 €					
SFTPB	Surfactant metabolism dysfunction type 1	178640	729,00 €	421,00 €	1.000,00 €			x
SFTPC	Surfactant metabolism dysfunction type 2	178620	491,00 €	421,00 €	762,00 €			x
SFTPD	Surfactant metabolism dysfunction	178635	655,00 €					
SFXN4	Combined oxidative phosphorylation deficiency type 18	615564	995,00 €					
SGCA	Limb-girdle muscular dystrophy, autosomal recessive type 2D	600119	819,00 €	328,00 €	997,00 €			x
SGCB	Limb-girdle muscular dystrophy, autosomal recessive type 2E	600900	573,00 €	328,00 €	751,00 €			x
SGCD	Cardiomyopathy, dilated type 1L	601411	737,00 €	328,00 €	915,00 €			x
SGCE	DYT11	604149	928,00 €	328,00 €	1.106,00 €			x
SGCG	Limb-girdle muscular dystrophy, autosomal recessive type 2C	608896	655,00 €	328,00 €	833,00 €			x
SGSH	Mucopolysaccharidosis type 3A	605270	819,00 €	437,00 €	1.106,00 €			x
SH2D1A	Lymphoproliferative syndrome, X-linked type 1	300490	468,00 €	328,00 €	646,00 €			
SH3PXD2B	Frank-ter Haar syndrome	613293	1.127,00 €					

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SH3TC2	CMT4C	608206	1.338,00 €	328,00 €	1.516,00 €			x
SHANK2	Autism susceptibility, X-linked type 17	603290	1.521,00 €	328,00 €	1.699,00 €			
SHANK3	Phelan-McDermid syndrome	606230	1.704,00 €	328,00 €	1.882,00 €			
SHBG	Androgen-binding protein deficiency	182205	573,00 €					
SHH	Holoprosencephaly type 3	600725	796,00 €	328,00 €	974,00 €			x
SHOC2	Noonan syndrom like	602775	737,00 €	421,00 €	1.008,00 €			
SHOX	Short stature syndrome	312865	655,00 €	328,00 €	833,00 €			x
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome	300579	1.399,00 €					
SI	Sucrase-isomaltase deficiency	609845	1.888,00 €	905,00 €	2.643,00 €			
SIGMAR1	Amyotrophic lateral sclerosis type 16	601978	491,00 €					
SIK1	Early infantile epileptic encephalopathy type 30	605705	995,00 €					
SIL1	Cataract, congenital, associated with Marinesco-Sjogren Syndrome	608005	819,00 €	429,00 €	1.098,00 €			x
SIX1	Deafness, autosomal dominant type 23	601205	374,00 €	343,00 €	567,00 €			x
SIX3	Holoprosencephaly type 2	603714	491,00 €	328,00 €	669,00 €			x
SIX5	Branchiootorenal syndrome type 2	600963	737,00 €	374,00 €	961,00 €			x
SIX6	Microphthalmia syndromic type 6	606326	374,00 €					
SKI	Shprintzen-Goldberg syndrome	164780	737,00 €	437,00 €	1.024,00 €			x
SKIV2L	Trichohepatoenteric syndrome type 2	600478	1.338,00 €	590,00 €	1.778,00 €			x
SLC1A3	Episodic ataxia type 6	600111	819,00 €	429,00 €	1.098,00 €			x
SLC1A4	Developmental delay and microcephaly, SLC1A4 related	600229	819,00 €					
SLC2A1	DYT8	138140	819,00 €	328,00 €	997,00 €			x
SLC2A2	Fanconi-Bickel syndrome	138160	796,00 €	429,00 €	1.075,00 €			x
SLC2A5	Fructose uptake deficiency, SLC2A5 related	138230	796,00 €					
SLC2A9	Hypouricemia, renal type 2	606142	928,00 €					
SLC2A10	Arterial Tortuosity Syndrome	606145	729,00 €	390,00 €	969,00 €			x
SLC3A1	Cystinuria	104614	862,00 €	328,00 €	1.040,00 €			x
SLC4A1	Ovalocytosis	109270	1.260,00 €					
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities	603345	1.582,00 €	527,00 €	1.959,00 €			x
SLC4A5	Renal tubular acidosis, SLC4A5 related	606757	1.643,00 €	548,00 €	2.041,00 €			x
SLC4A11	Corneal endothelial dystrophy type 2	610206	1.193,00 €					
SLC5A1	Glucose/Galactose malabsorption	182380	1.061,00 €	445,00 €	1.356,00 €			x
SLC5A2	Renal glucosuria	182381	928,00 €	437,00 €	1.215,00 €			x
SLC5A5	Thyroid dyshormonogenesis type 1	601843	1.061,00 €					
SLC6A2	Orthostatic intolerance	163970	1.127,00 €					
SLC6A3	Parkinsonism-Dystonia, infantile	126455	995,00 €	445,00 €	1.290,00 €			x
SLC6A5	Hyperekplexia	604159	1.193,00 €	328,00 €	1.371,00 €			x
SLC6A8	Creatine deficiency syndrome X-linked	300036	819,00 €	328,00 €	997,00 €			x
SLC6A14	Cystic fibrosis, SLC6A14 related	300444	995,00 €	437,00 €	1.282,00 €			
SLC6A19	Hartnup disorder	608893	796,00 €	421,00 €	1.067,00 €			x
SLC7A5	Phenylketonuria modifier, SLC7A5 related	600182	862,00 €					

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SLC7A7	LYSINURIC PROTEIN INTOLERANCE	603593	796,00 €	429,00 €	1.075,00 €			x
SLC7A9	Cystinuria	604144	862,00 €	328,00 €	1.040,00 €			x
SLC9A3R1	Nephrolithiasis/osteoporosis, hypophosphatemic, type 2	604990	655,00 €					
SLC9A6	Mental retardation, X-linked syndromic, Christianson type	300231	1.127,00 €					
SLC10A2	Bile acid malabsorption, primary	601295	655,00 €					
SLC12A1	Bartter syndrome type 1	600839	1.764,00 €	569,00 €	2.183,00 €			x
SLC12A2	Bartter syndrome	600840	1.775,00 €					x
SLC12A3	Bartter syndrome	600968	1.825,00 €	328,00 €	2.003,00 €			x
SLC12A5	Bartter syndrome	606726	1.704,00 €					
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	604878	1.643,00 €	527,00 €	2.020,00 €			x
SLC12A7	Bartter syndrome	604879	1.572,00 €					
SLC13A5	Early infantile epileptic encephalopathy type 25	608305	862,00 €	421,00 €	1.133,00 €			
SLC16A1	Erythrocyte lactate transporter defect	600682	655,00 €	390,00 €	895,00 €			x
SLC16A2	Allan-Herndon-Dudley syndrome	300095	655,00 €	421,00 €	926,00 €			x
SLC17A5	Sialuria, finish type	604322	796,00 €	429,00 €	1.075,00 €			x
SLC17A8	Deafness, autosomal dominant type 25	607557	995,00 €					
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	603941	655,00 €					
SLC19A3	Encephalopathy thiamine-responsive	606152	655,00 €	421,00 €	926,00 €			x
SLC20A2	Basal ganglia calcification type 1, ideopathic	158378	796,00 €	429,00 €	1.075,00 €			
SLC22A5	Carnitine deficiency	603377	862,00 €	328,00 €	1.040,00 €			x
SLC22A9	Autism, SLC22A9 related	607579	819,00 €					
SLC22A12	Hypouricemia, renal type 1	607096	796,00 €					
SLC24A4	Amelogenesis imperfecta type 2A5	609840	1.338,00 €					
SLC24A5	Albinism, oculocutaneous nonsyndromic	609802	737,00 €					x
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria	190315	737,00 €	437,00 €	1.024,00 €			
SLC25A4	Progressive external ophthalmoplegia with mitochondrial deletions type 2	103220	491,00 €	328,00 €	669,00 €			x
SLC25A13	Citrin deficiency	603859	1.193,00 €	421,00 €	1.464,00 €			x
SLC25A15	Hyperornithinemia- Hyperammonemia - Homocitrullinuria syndrome	603861	573,00 €	437,00 €	860,00 €			
SLC25A19	Microcephaly, Amish type	606521	573,00 €					
SLC25A20	Carnitine-acylcarnitine translocase deficiency	613698	819,00 €	421,00 €	1.090,00 €			x
SLC25A22	Early infantile epileptic encephalopathy type 3	609302	655,00 €					
SLC25A38	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive	610819	655,00 €					
SLC26A1	Renal dysfunction due to SLC26A1 deficiency	610130	573,00 €					
SLC26A2	Achondrogenesis type 1B	606718	655,00 €	374,00 €	879,00 €			x
SLC26A3	Diarrhea type 1, secretory chloride, congenital	126650	1.326,00 €	328,00 €	1.504,00 €			x
SLC26A4	Pendred syndrome	605646	1.326,00 €	328,00 €	1.504,00 €			x
SLC26A5	Deafness, autosomal recessive type 61	604943	1.582,00 €					
SLC26A6	Hyperoxaluria, SLC26A6 related	610068	1.278,00 €					
SLC27A4	Ichthyosis prematurity syndrome	604194	729,00 €					
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome	612373	573,00 €					

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SLC30A10	Hypermanganesemia with dystonia, polycythemia and cirrhosis	611146	573,00 €	437,00 €	860,00 €			x
SLC33A1	SPG42	603690	655,00 €	421,00 €	926,00 €			x
SLC34A1	Fanconi renotubular syndrome type 2	182309	729,00 €					x
SLC34A3	Hypophosphatemic rickets with hypercalciuria	609826	862,00 €	426,00 €	1.138,00 €			x
SLC35A1	Glycosylation disorder type 2F	605634	655,00 €					
SLC35A2	Glycosylation disorder type 2M	314375	819,00 €					
SLC35A3	Arthrogryposis, mental retardation, and seizures	605632	737,00 €					
SLC35C1	Glycosylation disorde type 2C	605881	491,00 €					
SLC35D1	Schneckenbecken dysplasia	610804	796,00 €					
SLC37A4	Glycogen storage disease type 1B	602671	737,00 €	429,00 €	1.016,00 €			x
SLC39A4	Acrodermatitis enteropathica	607059	729,00 €	421,00 €	1.000,00 €			x
SLC39A13	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like	608735	729,00 €					
SLC40A1	Hemochromatosis type 4	604653	819,00 €	328,00 €	997,00 €			x
SLC45A2	Albinism, oculocutaneous type 4	606202	655,00 €	437,00 €	942,00 €			x
SLC46A1	Folate malabsorption, hereditary	611672	573,00 €					
SLC52A1	Riboflavin deficiency	607883	491,00 €					
SLC52A2	Brown-Vialetto-Van Laere syndrome type 2	607882	573,00 €					
SLC52A3	Brown-Vialetto-Van Laere syndrome 1	613350	573,00 €	390,00 €	813,00 €			x
SLCO1B1	Hyperbilirubinemia, Rotor type	604843	1.061,00 €	445,00 €	1.356,00 €			x
SLCO1B3	Hyperbilirubinemia, Rotor type	605495	995,00 €	424,00 €	1.269,00 €			x
SLCO2A1	Hypertrophic osteoarthropathy type 2	601460	1.061,00 €					
SLITRK1	Tourette syndrome	609678	737,00 €	296,00 €	883,00 €			x
SLMAP	Brugada syndrome type 9	602701	1.460,00 €					
SLURP1	Mal de Meleda	606119	374,00 €					
SLX4	Fanconi anemia type P	613278	1.338,00 €					x
SMAD2	Thoracic aortic aneurysm dissection	601366	729,00 €					
SMAD3	Loeys-Dietz syndrome type 1C	603109	729,00 €					
SMAD4	Juvenile polyposis syndrome	600993	729,00 €	328,00 €	907,00 €			x
SMAD6	Aortic valve disease type 2	602931	737,00 €	437,00 €	1.024,00 €			x
SMARCA1	Mental retardation X-linked, SMARCA1 related	300012	1.643,00 €					
SMARCA2	Nicolaides Baraitser syndrome	600014	1.927,00 €	716,00 €	2.493,00 €			x
SMARCA4	Mental retardation, autosomal dominant type 16	603254	1.876,00 €	758,00 €	2.484,00 €			
SMARCAL1	Schimke immunoosseous dysplasia	606622	1.260,00 €					
SMARCB1	Mental retardation, autosomal dominant type 15	601607	819,00 €	328,00 €	997,00 €			x
SMARCE1	Coffin-Siris syndrome, SMARCE1 related	603111	729,00 €	429,00 €	1.008,00 €			
SMC1A	Cornelia de Lange syndrome type 2	300040	1.460,00 €	527,00 €	1.837,00 €			x
SMC3	Cornelia de Lange syndrome type 3	606062	1.764,00 €	611,00 €	2.225,00 €			x
SMN1	Spinal muscular atrophy type 1	600354	819,00 €	655,00 €	1.324,00 €			x
SMN2	Spinal muscular atrophy type 3, modifier of	601627		655,00 €				x
SMPD1	Niemann-Pick disease type A/B	607608	655,00 €	328,00 €	833,00 €			x

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SMPX	Deafness, X-linked type 4	300226	374,00 €					
SMS	Mental retardation, X-linked, Snyder-Robinson type	300105	796,00 €					
SNAI2	Piebaldism	602150	468,00 €	328,00 €	646,00 €			x
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, palmoplantar keratoderma syndrome	604202	573,00 €					
SNCA	Dementia, Lewy body	163890	573,00 €	328,00 €	751,00 €			
SNRNP200	Retinitis pigmentosa type 33, autosomal dominant	601664	1.931,00 €					
SNRPE	Hypotrichosis type 11	128260	573,00 €					
SNRPN	Prader-Willi syndrome	182279	573,00 €	328,00 €	751,00 €			x
SNTA1	Long QT syndrome type 12	601017	737,00 €					
SNX10	Osteopetrosis of infancy, malignant	614780	573,00 €	437,00 €	860,00 €			x
SNX14	Cerebellar ataxia, SNX14 related	616105	1.643,00 €					
SOAT1	Atherosclerosis, SOAT1 related	102642	1.127,00 €					
SOD1	Amyotrophic lateral sclerosis type 1	147450	729,00 €	390,00 €	969,00 €			x
SOD2	Microvascular complications of diabetes type 6, susceptibility to	147460	573,00 €					
SOHLH1	Oogenesis dysfunction	610224	819,00 €					
SORD	Cataract, congenital	182500	819,00 €					
SORL1	Alzheimers disease, early onset, autosomal dominant	602005	2.102,00 €	905,00 €	2.857,00 €			
SOS1	Noonan syndrome type 4	182530	1.582,00 €					x
SOST	Craniodiaphyseal dysplasia, autosomal dominant	605740	386,00 €					
SOX2	Lung cancer, SOX2 related, somatic	184429					1.900,00 €	
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency	313430	468,00 €	296,00 €	614,00 €			x
SOX9	Campomelic dysplasia	608160	573,00 €	328,00 €	751,00 €			x
SOX10	Peripheral demyelinating neuropathy Waardenburg syndrome and Hirschsprung disease	602229	573,00 €	437,00 €	860,00 €			x
SOX11	Mental retardation, autosomal dominant type 27	600898	374,00 €					
SOX17	Vesicoureteral reflux type 3	610928	573,00 €	343,00 €	766,00 €			x
SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome	601618	468,00 €					
SP7	Osteogenesis imperfecta type 12	606633	491,00 €					x
SP110	Hepatic venoocclusive disease with immunodeficiency	604457	1.643,00 €					
SPAG1	Primary ciliary dyskinesia type 28	603395	1.193,00 €					
SPAST	SPG4	604277	1.193,00 €	328,00 €	1.371,00 €			x
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome	613940	1.260,00 €					
SPATA7	Leber congenital amaurosis type 3	609868	928,00 €					
SPATA16	SPGF6	609856	796,00 €	429,00 €	1.075,00 €			
SPG7	SPG7	602783	1.260,00 €	328,00 €	1.438,00 €			x
SPG11	SPG11	610844	1.931,00 €	328,00 €	2.109,00 €			x
SPG20	SPG20	607111	796,00 €	421,00 €	1.067,00 €			x
SPG21	SPG21	608181	737,00 €	421,00 €	1.008,00 €			
SPINK1	Pancreatitis	167790	468,00 €	328,00 €	646,00 €			x
SPINK5	Netherton syndrome	605010	1.724,00 €	716,00 €	2.290,00 €			x
SPR	Dystonia, DOPA-responsive, autosomanl recessive	182125	374,00 €	374,00 €	598,00 €			x

Pricelist by gene

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Phone: +49 (0)381 203 652 222
E-mail: testing@centogene.com

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SPRED1	Legius syndrome	609291	737,00 €	328,00 €	915,00 €			x
SPTA1	Spherocytosis type 3	182860	2.145,00 €	905,00 €	2.900,00 €			
SPTAN1	Early infantile epileptic encephalopathy type 5	182810	2.067,00 €					
SPTB	Anemia, neonatal hemolytic, fatal and near-fatal	182870	1.927,00 €					
SPTBN2	Spinocerebellar ataxia type 5, autosomal dominant	604985	1.977,00 €	779,00 €	2.606,00 €			x
SPTBN5	Neuronal migration disorder	605916	1.955,00 €					
SPTLC1	HSAN1	605712	1.127,00 €	328,00 €	1.305,00 €			x
SQSTM1	Paget disease of bone	601530	737,00 €					
SRCAP	Floating-Harbor syndrome	611421	2.028,00 €	716,00 €	2.594,00 €			x
SRD5A2	Prostate cancer	607306	468,00 €	328,00 €	646,00 €			x
SRD5A3	Congenital disorder of glycosylation, type Iq	611715	491,00 €					
SRGAP1	Thyroid cancer type 2, nonmedullary, susceptibility to	606523	1.399,00 €					
SRGAP2	Neuronal migration disorder	606524	1.764,00 €					
SRP72	Bone marrow failure syndrome type 1	602122	1.260,00 €	430,00 €	1.540,00 €			x
SRPX2	Rolandic epilepsy, mental retardation, and speech dyspraxia	300642	729,00 €					x
SRY	46,XX sex reversal type 1	480000	374,00 €	328,00 €	552,00 €			
SS18L1	Amyotrophic lateral sclerosis, CREST related	606472	928,00 €					
SSR4	Glycosylation disorder x-linked	300090	655,00 €					
ST3GAL3	Early infantile epileptic encephalopathy type 15	606494	862,00 €					
ST3GAL5	Amish infantile epilepsy syndrome	604402	737,00 €					
ST6GAL2	Beta-Galactosamide alpha-2,6-Sialyltransferase 2 deficiency	608472	655,00 €					x
ST14	Ichthyosis, congenital, autosomal recessive, type 11	606797	928,00 €					
STAC3	Native American myopathy	615521	862,00 €					
STAG1	Prostate cancer	604358	1.825,00 €	905,00 €	2.580,00 €			
STAMBP	Microcephaly-capillary malformation syndrome	606247	729,00 €					
STAR	Lipoid congenital adrenal hyperplasia	600617	655,00 €	437,00 €	942,00 €			x
STAT1	Atypical Mycobacterial infection	600555	1.399,00 €					
STAT3	Hyper-IgE recurrent infection syndrome	102582	1.260,00 €	328,00 €	1.438,00 €			x
STAT5B	Growth hormone insensitivity with immunodeficiency	604260	1.260,00 €	328,00 €	1.438,00 €			
STIL	Microcephaly, autosomal recessive type 7	181590	1.278,00 €					x
STIM1	Immunodeficiency type 10	605921	928,00 €					
STK11	Melanoma, malignant, somatic	602216					1.900,00 €	
STRA6	Microphthalmia syndromic type 9	610745	1.399,00 €	430,00 €	1.679,00 €			x
STRC	Deafness and male infertility	606440	2.028,00 €	611,00 €	2.489,00 €			x
STS	Ichthyosis, X-linked	300747	729,00 €	328,00 €	907,00 €			x
STT3A	Congenital disorder of glycosylation, type Iw	601134	1.127,00 €					
STX11	Hemophagocytic lymphohistiocytosis type 4	605014	374,00 €	328,00 €	552,00 €			x
STXBP1	Early infantile epileptic encephalopathy type 4	602926	1.278,00 €	437,00 €	1.565,00 €			x
STXBP2	Hemophagocytic lymphohistiocytosis type 5	601717	995,00 €					
SUCLA2	Mitochondrial DNA depletion syndrome	603921	796,00 €	328,00 €	974,00 €			x

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SUCLG1	Mitochondrial DNA depletion syndrome, encephalomyopathic type with methylmalonic aciduria	611224	819,00 €	328,00 €	997,00 €			x
SUFU	Basal cell nevus syndrome	607035	928,00 €	421,00 €	1.199,00 €			x
SUGCT	Glutaric aciduria type 3	609187	1.061,00 €					
SULT2A1	Polycystic ovary syndrome type 1	125263	573,00 €					x
SULT2B1	Polycystic ovary syndrome type 1	604125	655,00 €					
SUMF1	Sulfatase deficiency	607939	819,00 €	421,00 €	1.090,00 €			x
SUMO1	Orofacial cleft type 10	601912	491,00 €					
SUN1	Deafness, autosomal recessive	607723	1.338,00 €					
SUOX	Sulfite oxidase deficiency	606887	655,00 €	421,00 €	926,00 €			x
SURF1	Leigh syndrome due to COX deficiency	185620	729,00 €	421,00 €	1.000,00 €			x
SYCP3	SPGF4	604759	737,00 €					
SYN1	Epilepsy, X-linked, with learning disabilities and behavior disorders	313440	995,00 €	426,00 €	1.271,00 €			x
SYNE1	Emery-Dreifuss muscular dystrophy type 4	608441	1.955,00 €	905,00 €				x
SYNE2	Emery-Dreifuss muscular dystrophy type 5	608442	1.955,00 €	905,00 €	2.710,00 €			x
SYNE4	Deafness, autosomal recessive type 76	615535	819,00 €					
SYNGAP1	Mental retardation, autosomal dominant type 5	603384	1.278,00 €					
SYNJ1	PARK20 Parkinson	604297	1.775,00 €	674,00 €	2.299,00 €			x
SYP	Mental retardation, X-linked type 96	313475	573,00 €					
TAB2	Congenital heart defects multiple types	605101	729,00 €					
TAC3	Hypogonadotropic hypogonadism type 10 with or without anosmia	162330	737,00 €					
TACO1	Leigh syndrome due to the mitochondrial complex IV deficiency	612958	655,00 €					
TACR3	Hypogonadotropic hypogonadism type 11 with or without anosmia	162332	573,00 €	390,00 €	813,00 €			x
TAF1	DYT3	313650	1.927,00 €	821,00 €	2.598,00 €			
TALDO1	Transaldolase deficiency	602063	737,00 €	437,00 €	1.024,00 €			x
TANC1	Psychomotor retardation	611397	1.643,00 €					
TANC2	TANC2 related brain disorders	615047	1.582,00 €					
TARDBP	Amyotrophic lateral sclerosis type 10	605078	819,00 €	421,00 €	1.090,00 €			x
TARS2	Combined oxidative phosphorylation deficiency type 21	612805	1.260,00 €					
TAT	Tyrosinemia type 2	613018	729,00 €	421,00 €	1.000,00 €			
TAZ	Barth syndrome	300394	655,00 €	429,00 €	934,00 €			x
TBC1D24	Deafness, autosomal dominant type 65	613577	819,00 €	437,00 €	1.106,00 €			x
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome	604934	1.193,00 €	421,00 €	1.464,00 €			x
TBP	Spinocerebellar ataxia type 17, autosomal dominant	600075				200,00 €		x
TBR1	Intellectual disability, TBR1 related	604616	737,00 €					
TBX1	DiGeorge syndrome	602054	995,00 €	655,00 €	1.500,00 €			x
TBX3	Ulnar-Mammary syndrome	601621	729,00 €	437,00 €	1.016,00 €			x
TBX5	Holt-Oram syndrome	601620	729,00 €	328,00 €	907,00 €			x
TBX6	Spondylocostal dysostosis type 5	602427	655,00 €					
TBX15	Cousin syndrome	604127	819,00 €					
TBX19	Adrenocorticotropic hormone deficiency	604614	737,00 €					

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TBX20	Atrial septal defect type 4	606061	737,00 €	437,00 €	1.024,00 €			
TBXAS1	Ghosal hematodiaphyseal syndrome	274180	1.061,00 €					
TCAP	Cardiomyopathy, dilated type 1N	604488	386,00 €	343,00 €	579,00 €			x
TCF4	Pitt-Hopkins syndrome	602272	1.582,00 €	328,00 €	1.760,00 €			x
TCF12	Craniosynostosis type 3	600480	1.278,00 €					
TCIRG1	Osteopetrosis, autosomal recessive type 1	604592	1.338,00 €					x
TCN2	Transcobalamin II deficiency	613441	729,00 €					x
TCOF1	Treacher Collins syndrome type 1	606847	1.764,00 €	328,00 €	1.942,00 €			x
TCTN1	Joubert syndrome type 13	609863	1.193,00 €					
TCTN2	Joubert syndrome type 24	613846	1.260,00 €					
TCTN3	Joubert syndrome type 18	613847	928,00 €	437,00 €	1.215,00 €			
TDP1	Spinocerebellar ataxia with axonal neuropathy, autosomal recessive	607198	1.127,00 €	424,00 €	1.401,00 €			x
TDRD7	Cataract, autosomal recessive congenital type 4	611258	1.338,00 €					
TEAD1	Sveinsson choreoretinal atrophy	189967	796,00 €					
TECPR2	SPG49	615000	1.326,00 €					
TECR	Mental retardation, autosomal recessive type 14	610057	737,00 €					
TECTA	Deafness, autosomal dominant type 12	602574	1.572,00 €					x
TENM1	Colobomatous microphthalmia	610083	1.802,00 €	674,00 €	2.326,00 €			
TENM3	Microphthalmia, isolated with coloboma type 9	610083	1.825,00 €					
TERC	Aplastic anemia	602322	296,00 €	328,00 €	474,00 €			x
TERT	Dyskeratosis congenita, autosomal recessive type 4/ autosomal dominant type 2	187270	1.260,00 €	328,00 €	1.438,00 €			x
TET2	Myelodysplastic syndrome, somatic	612839					1.900,00 €	
TFAP2A	Branchiooculofacial syndrome	107580	862,00 €					
TFG	Hereditary motor and sensory neuropathy, Okinawa type	602498	655,00 €					
TFR2	Hemochromatosis type 3	604720	1.061,00 €	328,00 €	1.239,00 €			x
TG	Thyroid dyshormonogenesis type 3	188450	2.106,00 €	905,00 €	2.861,00 €			
TGFB1	Camurati-Engelmann disease	190180	737,00 €					
TGFB2	Loeys-Dietz syndrome type 4	190220	737,00 €	437,00 €	1.024,00 €			x
TGFB3	Arrhythmogenic right ventricular cardiomyopathy type 1	190230	796,00 €	328,00 €	974,00 €			
TGFBI	Corneal dystrophy, epithelial basement membrane	601692	1.127,00 €					
TGFBR1	Aortic aneurysm, familial thoracic type 5	190181	819,00 €	328,00 €	997,00 €			x
TGFBR2	Aortic aneurysm, familial thoracic type 3	190182	862,00 €	328,00 €	1.040,00 €			x
TGIF1	Holoprosencephaly type 4	602630	468,00 €	328,00 €	646,00 €			
TGM1	Ichthyosis, congenital, autosomal recessive type 1	190195	1.127,00 €	445,00 €	1.422,00 €			x
TGM5	Peeling skin syndrome type 2	603805	928,00 €	426,00 €	1.204,00 €			x
TGM6	Spinocerebellar ataxia type 35, autosomal dominant	613900	995,00 €					
TH	Segawa syndrome, autosomal recessive	191290	796,00 €	437,00 €	1.083,00 €			x
THAP1	DYT6	609520	374,00 €	328,00 €	552,00 €			x
THBD	Hemolytic uremic syndrome	188040	573,00 €					x
THRA	Hypothyroidism congenital nongoitrous type 6	190120	819,00 €	429,00 €	1.098,00 €			x

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THRB	Thyroid hormone resistance	190160	819,00 €	421,00 €	1.090,00 €			x
TIMM8A	Dystonia-deafness syndrome	300356	374,00 €	343,00 €	567,00 €			x
TIMM21	Mitochondrial respiratory chain disease, TIMM21 related	615180	573,00 €					
TIMP1	Bicuspid aortic valve	305370	491,00 €					
TIMP3	Sorsby fundus dystrophy	188826	573,00 €					
TINF2	Revesz syndrome	604319	737,00 €					x
TJP1	TJP1 deficiency	601009	1.622,00 €					
TJP2	Hypercholanemia	607709	1.521,00 €					
TK2	Mitochondrial DNA depletion syndrome	188250	862,00 €	328,00 €	1.040,00 €			x
TLR3	Herpes simplex encephalitis type 2, susceptibility to	603029	737,00 €					
TMC1	Deafness, autosomal dominant type 36	606706	1.399,00 €	505,00 €	1.754,00 €			x
TMEM5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A10	605862	655,00 €					
TMEM38B	Osteogenesis imperfecta type 14	611236	573,00 €	421,00 €	844,00 €			x
TMEM43	Arrhythmogenic right ventricular cardiomyopathy type 5	612048	862,00 €					
TMEM67	COACH syndrome	609884	1.704,00 €					
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	612418	491,00 €	374,00 €	715,00 €			x
TMEM126A	Optic atrophy type 7	612988	468,00 €					x
TMEM127	Pheochromocytoma type 8	613403	468,00 €					
TMEM138	Joubert syndrome type 16	614459	468,00 €					
TMEM165	Glycosylation disorder type 2K	614726	655,00 €	421,00 €	926,00 €			x
TMEM173	Vasculopathy, infantile-onset, TMEM173/STING related	612374	573,00 €	437,00 €	860,00 €			x
TMEM216	Joubert syndrome type 2	613277	491,00 €					x
TMEM231	Joubert syndrome type 20	614949	729,00 €					
TMEM237	Joubert syndrome type 14	614423	995,00 €					
TMEM240	Spinocerebellar ataxia type 21, autosomal dominant	616101	374,00 €					
TMIE	Deafness, autosomal recessive type 6	607237	468,00 €					
TMPO	Cardiomyopathy, dilated type 1T	188380	928,00 €					
TMPRSS3	Deafness, autosomal recessive type 8/10	605511	1.127,00 €					
TMPRSS6	Iron-refractory iron deficiency anemia	609862	1.326,00 €	430,00 €	1.606,00 €			
TMPRSS15	Enterokinase deficiency	606635	1.582,00 €					
TNFAIP3	Rheumatoid arthritis, TNFAIP3 related	191163	729,00 €					
TNFRSF1A	Periodic fever autosomal dominant	191190	819,00 €	429,00 €	1.098,00 €			x
TNFRSF11A	Osteolysis, familial expansile	603499	928,00 €					
TNFRSF11B	Paget disease, juvenile	602643	491,00 €					
TNFRSF13B	Immunodeficiency common variable type 2	604907	573,00 €	390,00 €	813,00 €			
TNFRSF13C	Immunodeficiency common variable type 4	606269	374,00 €					
TNFSF11	Osteopetrosis, autosomal recessive type 2	602642	573,00 €					
TNNC1	Cardiomyopathy, dilated type 1Z	191040	491,00 €					
TNNI2	Arthrogryposis, distal, type 2B	191043	655,00 €					x
TNNI3	Cardiomyopathy, dilated type 2A	191044	737,00 €					

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TNNT1	Nemaline myopathy type 5	191041	862,00 €	437,00 €	1.149,00 €			x
TNNT2	Cardiomyopathy, dilated type 1D	191045	1.061,00 €	328,00 €	1.239,00 €			x
TNNT3	Arthrogryposis, distal, type 2B	600692	1.061,00 €					
TNXB	Ehlers-Danlos syndrome type 3	600985	2.016,00 €	328,00 €	2.194,00 €			x
TOPORS	Retinitis pigmentosa type 31, autosomal dominant	609507	796,00 €					
TOR1A	DYT1	605204	491,00 €	328,00 €	669,00 €			x
TP53	Basal cell carcinoma type 7, susceptibility to, somatic	191170					1.900,00 €	
TP63	ADULT syndrome, split hand-foot malformation	603273	1.061,00 €	424,00 €	1.335,00 €			x
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency	190450	491,00 €					
TPK1	Thiamine metabolism dysfunction syndrome type 5	606370	737,00 €					
TPM1	Cardiomyopathy, dilated type 1Y	191010	995,00 €					
TPM2	Arthrogryposis, distal, type 1A	190990	819,00 €	421,00 €	1.090,00 €			x
TPM3	Nemaline myopathy type 1	191030	928,00 €	429,00 €	1.207,00 €			x
TPMT	TPMT deficiency	187680	737,00 €	328,00 €	915,00 €			
TPO	Thyroid dyshormonogenesis type 2A	606765	1.193,00 €	328,00 €	1.371,00 €			
TPP1	Ceroid lipofuscinosis neuronal type 2	607998	862,00 €	426,00 €	1.138,00 €			x
TPRN	Deafness, autosomal recessive type 79	613354	491,00 €					
TRAPPC9	Mental retardation, autosomal dominant type 13	611966	1.521,00 €					
TRAPPC11	Limb-girdle muscular dystrophy, autosomal recessive type 2S	614138	1.825,00 €					
TRDN	Ventricular tachycardia, catecholaminergic polymorphic type 5	603283	1.888,00 €	905,00 €	2.643,00 €			
TREM2	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	605086	491,00 €	390,00 €	731,00 €			x
TREX1	Aicardi-Goutieres syndrome type 1	606609	374,00 €	328,00 €	552,00 €			x
TRHR	Hypothyroidism, isolated, TRHR related	188545	374,00 €					
TRIM32	Bardet-Biedl syndrome type 11	602290	819,00 €					x
TRIM37	Mulibrey nanism	605073	1.582,00 €	527,00 €	1.959,00 €			x
TRIOBP	Deafness, autosomal recessive type 28	609761	1.673,00 €					
TRIP11	Achondrogenesis type 1A	604505	1.622,00 €	442,00 €	1.914,00 €			x
TRMT5	Combined oxidative phosphorylation deficiency type 26	611023	655,00 €					
TRMU	Liver failure transient infantile	610230	796,00 €					x
TRNT1	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	612907	737,00 €					
TRPC6	Focal segmental glomerulosclerosis type 2	603652	995,00 €					
TRPM1	Night blindness, congenital stationar type 1C	603576	1.764,00 €	569,00 €	2.183,00 €			
TRPM4	Progressive familial heart block	606936	1.704,00 €					
TRPM6	Hypomagnesemia type 1	607009	1.888,00 €	821,00 €	2.559,00 €			x
TRPS1	Langer-Giedion syndrome	604386	995,00 €	328,00 €	1.173,00 €			x
TRPV3	Olmsted syndrome	607066	1.193,00 €					
TRPV4	CMT2C	605427	1.127,00 €	424,00 €	1.401,00 €			x
TSC1	Bladder cancer, TSC1-related, somatic	605284					1.900,00 €	
TSC2	Lymphangioleiomyomatosis, somatic	191092					1.900,00 €	
TSEN2	Pontocerebellar hypoplasia type 2B	608753	862,00 €					x

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TSEN34	Pontocerebellar hypoplasia type 2C	608754	491,00 €					
TSEN54	Pontocerebellar hypoplasia type 2A	608755	862,00 €	429,00 €	1.141,00 €			x
TSFM	Combined oxidative phosphorylation deficiency type 3	604723	729,00 €					
TSHB	Hypothyroidism congenital nongoitrous type 4	188540	386,00 €					
TSHR	Hypothyroidism congenital nongoitrous type 1	603372	1.193,00 €	328,00 €	1.371,00 €			x
TSPAN7	Mental retardation, X-linked type 58	300096	655,00 €	328,00 €	833,00 €			x
TSPAN12	Exudative vitreoretinopathy type 5	613138	737,00 €					
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome	604714	374,00 €					
TSR2	Diamond-Blackfan anemia type 14 with mandibulofacial dysostosis	300945	468,00 €	390,00 €	708,00 €			x
TTBK2	Spinocerebellar ataxia type 11, autosomal dominant	611695	1.278,00 €	445,00 €	1.573,00 €			x
TTC7A	Intestinal atresia, multiple	609332	1.399,00 €	442,00 €	1.691,00 €			x
TTC8	Bardet-Biedl syndrome type 8	608132	1.193,00 €					
TTC19	Mitochondrial complex III deficiency, nuclear type 2	613814	796,00 €	429,00 €	1.075,00 €			
TTC21B	Nephronophthisis type 12	612014	1.572,00 €	611,00 €	2.033,00 €			x
TTC37	Trichohepatoenteric syndrome type 1	614589	1.927,00 €	738,00 €	2.515,00 €			x
TTI2	Mental retardation, autosomal recessive type 39	614426	737,00 €					
TTN	Cardiomyopathy, dilated type 1G	188840	2.225,00 €	905,00 €	2.980,00 €			
TTPA	Vitamin E familial deficiency	600415	491,00 €	390,00 €	731,00 €			x
TTR	Amyloidosis	176300	491,00 €	437,00 €	778,00 €			
TUBA1A	Lissencephaly type 3	602529	655,00 €					
TUBA8	Polymicrogyria with optic nerve hypoplasia	605742	573,00 €	390,00 €	813,00 €			x
TUBB2A	Cortical dysplasia, complex, with other brain malformations, type 5	615101	655,00 €	437,00 €	942,00 €			
TUBB2B	Microcephaly, TUBB2B related	612850	491,00 €					
TUBB3	Cortical dysplasia, complex, with other brain malformations, type 1	602661	573,00 €					
TUBB4A	DYT4	602662	655,00 €	390,00 €	895,00 €			x
TUBB	Neurodevelopmental disorder, TUBB related	191130	655,00 €					
TUBG1	Neurodevelopmental malformation and microcephaly	191135	862,00 €					
TUBGCP6	Microcephaly and chorioretinopathy with or without mental retardation	610053	1.673,00 €	527,00 €	2.050,00 €			x
TUFM	Combined oxidative phosphorylation deficiency type 4	602389	862,00 €					
TUFT1	Tuftelin deficiency	600087	995,00 €					
TULP1	Retinitis pigmentosa type 14, autosomal recessive	602280	862,00 €	445,00 €	1.157,00 €			x
TUSC3	Mental retardation, autosomal recessive type 7	601385	796,00 €	429,00 €	1.075,00 €			
TWIST1	Craniosynostosis type 1	601622	386,00 €	328,00 €	564,00 €			x
TWNK	Mitochondrial DNA depletion syndrome type 7	606075	737,00 €	328,00 €	915,00 €			x
TYK2	Immunodeficiency type 35	176941	1.460,00 €	527,00 €	1.837,00 €			x
TYMP	Mitochondrial neurogastrointestinal encephalopathy syndrome without leukoencephalopathy	131222	819,00 €	429,00 €	1.098,00 €			x
TYR	Albinism, oculocutaneous type 1A	606933	655,00 €	328,00 €	833,00 €			x
TYROBP	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	604142	468,00 €	390,00 €	708,00 €			x
TYRP1	Albinism, oculocutaneous type 3	115501	655,00 €	437,00 €	942,00 €			x
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type	312180	655,00 €					

Pricelist by gene

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UBE3A	Angelman syndrome	601623	1.061,00 €	328,00 €	1.239,00 €			x
UBE3B	Blepharophimosis-ptosis-intellectual disability syndrome	608047	1.704,00 €					
UBQLN2	Amyotrophic lateral sclerosis, x-linked juvenile and adult-onset ALS	300264	491,00 €					x
UBR1	Johanson Blizzard syndrome	605981	2.059,00 €	807,00 €	2.716,00 €			x
UCHL1	PARK5 Parkinson	191342	655,00 €	328,00 €	833,00 €			x
UCP2	Hyperinsulinism, UCP2 related	601693	491,00 €					
UGT1A1	Crigler-Najjar syndrome, type 1	191740	737,00 €	390,00 €	977,00 €			x
ULK2	Smith-Magenis syndrome, ULK2 related	613653	1.704,00 €					
UMOD	Glomerulocystic kidney disease with hyperuricemia and isosthenuria	191845	862,00 €	421,00 €	1.133,00 €			x
UMPS	Orotic aciduria	613891	655,00 €	421,00 €	926,00 €			x
UNC13B	Autism, UNC13B related	605836	1.802,00 €					
UNC13D	Hemophagocytic lymphohistiocytosis type 3	608897	1.278,00 €	328,00 €	1.456,00 €			x
UNC45B	Cataract type 43	611220	1.326,00 €					
UNC119	Cone-rod dystrophy	604011	573,00 €					
UNG	Immunodeficiency type 5, with hyper IgM	191525	737,00 €					
UPB1	Beta-ureidopropionase deficiency	606673	729,00 €	429,00 €	1.008,00 €			
UPD chr. 6	6q24-related transient neonatal diabetes mellitus type 1			328,00 €				
UPF3B	Mental retardation, X-linked type 14	300298	729,00 €	429,00 €	1.008,00 €			x
UQCC2	Mitochondrial complex III deficiency, nuclear type 7	614461	468,00 €					
UQCRB	Mitochondrial complex III deficiency	191330	819,00 €					
UQCRC2	Mitochondrial complex III deficiency	191329	995,00 €	437,00 €	1.282,00 €			x
UQCRQ	Mitochondrial complex III deficiency	612080	374,00 €					
UROC1	Urocanase deficiency	613012	1.399,00 €					
UROD	Porphyria cutanea tarda	613521	737,00 €	328,00 €	915,00 €			x
UROS	Porphyria congenital erythropoietic	606938	819,00 €	328,00 €	997,00 €			x
USB1	Poikiloderma with neutropenia	613276	737,00 €	437,00 €	1.024,00 €			
USF1	Hyperlipidemia, familial combined, susceptibility to	191523	737,00 €					
USH1C	Deafness, autosomal recessive type 18	605242	1.643,00 €	569,00 €	2.062,00 €			x
USH1G	Usher syndrome type 1G	607696	573,00 €					
USH2A	Retinitis pigmentosa type 39	608400	1.955,00 €	655,00 €	2.460,00 €			x
USP8	SPG59, USP8 related	603158	1.326,00 €					
USP9X	Mental retardation, X-linked type 99	300072	1.931,00 €	905,00 €	2.686,00 €			
UVSSA	UV-sensitive syndrome type 3	614632	928,00 €					
VAMP1	Spastic ataxia type 1, autosomal dominant	185880	573,00 €	390,00 €	813,00 €			x
VAPB	Amyotrophic lateral sclerosis type 8	605704	573,00 €					x
VARS2	Combined oxidative phosphorylation deficiency type 20	612802	1.582,00 €					
VAX1	Microphthalmia, syndromic type 11	604294	573,00 €					
VAX2	Microphthalmia, VAX2 related	604295	374,00 €					
VCAN	Wagner syndrome	118661	1.775,00 €					
VCL	Cardiomyopathy, dilated type 1W	193065	1.582,00 €					

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VCP	Amyotrophic lateral sclerosis type 14	601023	1.127,00 €					x
VDAC1	Encephalopathy mitochondrial	604492	655,00 €					x
VDR	Rickets, vitamin D-resistant, type 2A	601769	729,00 €	429,00 €	1.008,00 €			x
VEGFA	Microvascular complications of diabetes type 1	192240	819,00 €					
VHL	Hemangioblastoma, cerebellar, somatic	608537					1.900,00 €	
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis type 2	613401	1.326,00 €	437,00 €	1.613,00 €			x
VKORC1	Coumarin resistance	608547	573,00 €					
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion type 1	192977	1.260,00 €	328,00 €	1.438,00 €			x
VPS13A	Choreoacanthocytosis	605978	1.955,00 €					
VPS13B	Cohen syndrome	607817	1.955,00 €	655,00 €	2.460,00 €			
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis type 1	608552	1.326,00 €					
VPS35	PARK17 Parkinson	601501	1.193,00 €	424,00 €	1.467,00 €			x
VPS37A	SPG53	609927	796,00 €					
VPS45	Neutropenia, severe congenital type 5, autosomal recessive	610035	1.127,00 €					
VPS53	Pontocerebellar hypoplasia type 2E	615850	1.399,00 €					
VPS54	Amyotrophic lateral sclerosis, VPS54 related	614633	1.521,00 €					
VRK1	Pontocerebellar hypoplasia type 1A	602168	862,00 €	426,00 €	1.138,00 €			x
VSX1	Corneal dystrophy, posterior polymorphous, type 1	605020	655,00 €					
VSX2	Microphthalmia, isolated type 2	142993	491,00 €	390,00 €	731,00 €			x
VWF	von Willebrand disease	613160	2.028,00 €	655,00 €	2.533,00 €			
WAS	Thrombocytopenia, X-linked, intermittent	300392	928,00 €	421,00 €	1.199,00 €			x
WDPCP	Bardet-Biedl syndrome type 15	613580	1.338,00 €					
WDR11	Hypogonadtropic hypogonadism type 14	606417	1.764,00 €	611,00 €	2.225,00 €			x
WDR19	Cranioectodermal dysplasia type 4	608151	1.927,00 €					
WDR27	WDR27-related brain disorders		1.582,00 €					
WDR34	Short-rib thoracic dysplasia type 11 with or without polydactyly	613363	796,00 €					
WDR35	Cranioectodermal dysplasia type 2	613602	1.825,00 €					
WDR36	Glaucoma, open angle type 1G	609669	1.460,00 €					x
WDR45	Neurodegeneration with brain iron accululation type 5	300526	928,00 €	421,00 €	1.199,00 €			
WDR48	SPG60, WDR48 related	612167	1.260,00 €					
WDR60	Short-rib thoracic dysplasia type 8 with or without polydactyly	615462	1.582,00 €					
WDR62	Microcephaly with cortical malformations, autosomal recessive type 2	613583	1.775,00 €	674,00 €	2.299,00 €			x
WDR72	Amelogenesis imperfecta type 2A3	613214	1.399,00 €					
WDR73	Galloway-Mowat syndrome	616144	737,00 €					
WDR81	Cerebellar ataxia with mental retardation and dysequilibrium syndrome type 2	614218	1.338,00 €					
WFS1	Cataract type 41	606201	796,00 €	328,00 €	974,00 €			x
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood	603400	573,00 €					x
WNK1	HSAN2A	605232	1.802,00 €	590,00 €	2.242,00 €			x
WNK4	Pseudohypoaldosteronism, type 2B	601844	1.399,00 €					
WNT1	Osteogenesis imperfecta type 15	164820	491,00 €					

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WNT3	Tetraamelia, autosomal recessive	165330	491,00 €					
WNT4	SERKAL syndrome	603490	491,00 €	328,00 €	669,00 €			x
WNT5A	Robinow syndrome, autosomal dominant type 1	164975	819,00 €	390,00 €	1.059,00 €			x
WNT7A	Fibular aplasia or hypoplasia, femoral bowing and poly-, syn-, and oligodactyly	601570	468,00 €	437,00 €	755,00 €			x
WNT10A	Odontoonychodermal dysplasia	606268	491,00 €	437,00 €	778,00 €			x
WNT10B	Split-hand/foot malformation type 6	601906	491,00 €	390,00 €	731,00 €			x
WRN	Werner syndrome	604611	1.927,00 €	905,00 €	2.682,00 €			
WT1	Mesothelioma, somatic	607102					1.900,00 €	
WWOX	Early infantile epileptic encephalopathy type 28	605131	729,00 €	328,00 €	907,00 €			x
XDH	Xanthinuria type 1	607633	1.876,00 €					x
XIAP	Lymphoproliferative syndrome, X-linked type 2	300079	737,00 €	328,00 €	915,00 €			
XK	McLeod syndrome with or without chronic granulomatous disease	314850	491,00 €	374,00 €	715,00 €			x
XPA	Xeroderma pigmentosum, group A	611153	573,00 €					
XPC	Xeroderma pigmentosum, group C	613208	1.326,00 €					
XPNPEP3	Nephronophthisis-like nephropathy type 1	613553	729,00 €					
XPR1	Basal ganglia calcification type 6, idiopathic	605237	1.061,00 €	445,00 €	1.356,00 €			
XRCC2	Fanconi anemia, XRCCR2 related	600375	468,00 €	374,00 €	692,00 €			x
XRCC3	Breast cancer, susceptibility to	600675	729,00 €					
XRCC4	Short stature, microcephaly, and endocrine dysfunction	194363	655,00 €	437,00 €	942,00 €			
XYLT1	Desbuquois dysplasia type 2	608124	862,00 €					
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia type 2	610957	737,00 €					
YARS	DI-CMTC	603623	928,00 €	426,00 €	1.204,00 €			x
YWHAE	Miller Dieker lissencephaly syndrome	605066	819,00 €	328,00 €	997,00 €			
ZAP70	Selective T-cell defect	176947	862,00 €					
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome type 2	614064	819,00 €					
ZC4H2	Wieacker-Wolff syndrome	300897	491,00 €					
ZCCHC12	Mental retardation non-syndromic	300701	468,00 €					
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type	300646	819,00 €					
ZDHHC15	Mental retardation, X-linked type 91	300576	862,00 €	421,00 €	1.133,00 €			x
ZDHHC17	Huntington disease, ZDHHC17 related	607799	1.193,00 €					
ZEB2	Central hypoventilation syndrome, congenital	605802	862,00 €	328,00 €	1.040,00 €			x
ZFHX4	Ptosis, congenital	606940	1.622,00 €					
ZFP57	Maturity-onset diabetes of the young, ZFP57 related	612192	655,00 €	328,00 €	833,00 €			x
ZFPM2	Diaphragmatic hernia type 3	603693	928,00 €					
ZFR	SPG71, ZFR related	615635	1.326,00 €					
ZFYVE26	SPG15	612012	1.931,00 €	721,00 €	2.502,00 €			x
ZFYVE27	SPG33	610243	796,00 €	421,00 €	1.067,00 €			x
ZIC1	Craniosynostosis type 6	600470	573,00 €	328,00 €	751,00 €			x
ZIC2	Holoprosencephaly type 5	603073	737,00 €	328,00 €	915,00 €			x
ZIC3	Heterotaxy, visceral type 1	300265	491,00 €	328,00 €	669,00 €			x

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ZIC5	ZIC5 related brain disorders		491,00 €					
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy	606480	729,00 €	328,00 €	907,00 €			x
ZNF41	Mental retardation, X-linked type 89	314995	862,00 €					
ZNF81	Mental retardation, X-linked type 45	314998	819,00 €					
ZNF311	Neurodevelopmental disorder, ZNF311 related		796,00 €					
ZNF423	Nephronophthisis type 14	604557	1.193,00 €					
ZNF469	Brittle cornea syndrome	612078	2.141,00 €					
ZNF513	Retinitis pigmentosa type 58, autosomal recessive	613598	655,00 €					
ZNF674	Mental retardation, X-linked type 92	300573	737,00 €					
ZNF711	Mental retardation, X-linked type 97	314990	729,00 €					
ZNF778	Autism, ZNF778 related		796,00 €					
ZNF783	Prostate cancer		819,00 €					
ZP1	Oocyte maturation defect	195000	928,00 €					
ZSWIM6	Acromelic frontonasal dysostosis	615951	1.193,00 €	437,00 €	1.480,00 €			x