			Classification of rare metabolic diseases (Functional)	Source: www.orpha.net	State: 12.06.2018
ORPHA68367	10507	Group of phenomes	Rare inborn errors of metabolism		
ORPHA137	3553	Group of phenomes	Congenital disorder of glycosylation		
ORPHA309347	21377	Group of phenomes	Disorder of protein N-glycosylation		
ORPHA79318	11344	Disease	PMM2-CDG		
ORPHA79319	11345	Disease	MPI-CDG		
ORPHA79320	11346	Disease	ALG6-CDG		
ORPHA79321	11347	Disease	ALG3-CDG		
ORPHA79324	11350	Disease	ALG12-CDG		
ORPHA79325	11351	Disease	ALG8-CDG		
ORPHA79326	11352	Disease	ALG2-CDG		
ORPHA79327	11353	Disease	ALG1-CDG		
ORPHA79328	11354	Disease	ALG9-CDG		
ORPHA79329	11355	Disease	MGAT2-CDG		
ORPHA79330	11356	Disease	MOGS-CDG		
ORPHA86309	11726	Disease	DPAGT1-CDG		
ORPHA244310	19478	Disease	RFT1-CDG		
ORPHA280071	20425	Disease	ALG11-CDG		
ORPHA300536	21127	Disease	DDOST-CDG		
ORPHA314667	21512	Disease	TMEM165-CDG		
ORPHA319646	21686	Disease	PGM1-CDG		
ORPHA324422	21777	Disease	ALG13-CDG		
ORPHA353327	22137	Etiological subtype	Congenital myasthenic syndromes with glycosylation defect		
ORPHA370921	22500	Disease	STT3A-CDG		
ORPHA370924	22501	Disease	STT3B-CDG		
ORPHA370927	22502	Disease	SSR4-CDG		
ORPHA370943	22507	Disease	Autism spectrum disorder-epilepsy-arthrogryposis syndrome		
ORPHA397941	22709	Disease	MAN1B1-CDG		
ORPHA468699	24236	Disease	SLC39A8-CDG		
ORPHA309447	21378	Group of phenomes	Disorder of protein O-glycosylation		
ORPHA309450	21379	Group of phenomes	Disorder of O-xylosylglycan synthesis		
ORPHA321	3247	Disease	Multiple osteochondromas		
ORPHA2953	3480	Disease	Ehlers-Danlos syndrome, musculocontractural type		
ORPHA75496	11083	Disease	Ehlers-Danlos syndrome, progeroid type		
ORPHA93359	12290	Disease	Spondyloepimetaphyseal dysplasia with joint laxity		
ORPHA263463	20058	Disease	CHST3-related skeletal dysplasia		
ORPHA284139	20576	Malformation syndrome	Larsen-like syndrome, B3GAT3 type		
ORPHA363417	22298	Malformation syndrome	Temtamy preaxial brachydactyly syndrome		
ORPHA370930	22503	Disease	XYLT1-CDG		
ORPHA466926	24178	Disease	Seizures-scoliosis-macrocephaly syndrome		
ORPHA480682	25220	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2Z		
ORPHA309458	21380	Group of phenomes	Disorder of O-N-acetylgalactosaminylglycan synthesis		
ORPHA306661	21246	Clinical subtype	Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome		
ORPHA309463	21381	Group of phenomes	Disorder of O-xylosyl/N-acetylgalactosaminylglycan synthesis		
ORPHA3144	2813	Malformation syndrome	Schneckenbecken dysplasia		
ORPHA309469	21382	Group of phenomes	Disorder of O-mannosylglycan synthesis		
ORPHA272	8724	Disease	Congenital muscular dystrophy, Fukuyama type		
ORPHA899	8725	Disease	Walker-Warburg syndrome		
ORPHA588	8726	Disease	Muscle-eye-brain disease		
ORPHA34515	10337	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2I		
ORPHA86812	11732	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2K		
ORPHA206554	18521	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2M		
ORPHA206559	18522	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2N		

ORPHA206564	18523	Disease				Autosomal recessive limb-girdle muscular dystrophy type 2O
ORPHA352479	22066	Disease				Autosomal recessive limb-girdle muscular dystrophy type 2U
ORPHA363623	22329	Disease				Autosomal recessive limb-girdle muscular dystrophy type 2T
ORPHA370959	22509	Disease				Congenital muscular dystrophy with cerebellar involvement
ORPHA370968	22510	Disease				Congenital muscular dystrophy with intellectual disability
ORPHA370980	22511	Disease				Congenital muscular dystrophy without intellectual disability
ORPHA445110	23519	Disease				Limb-girdle muscular dystrophy due to POMK deficiency
ORPHA309505	21383	Group of phenomes		Disorder of fucoglycosan synthesis		
ORPHA709	968	Malformation syndrome				Peters plus syndrome
ORPHA2311	1042	Malformation syndrome				Autosomal recessive spondylocostal dysostosis
ORPHA79145	11171	Disease				Dowling-Degos disease
ORPHA309515	21384	Group of phenomes	Disorder of glycosphingolipid and glycosylphosphatidylinositol anchor glycosylation			
ORPHA447	21	Disease		Paroxysmal nocturnal hemoglobinuria		
ORPHA3474	3498	Malformation syndrome		CHIME syndrome		
ORPHA83639	11601	Disease		Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency		
ORPHA247262	19518	Disease		Hyperphosphatasia-intellectual disability syndrome		
ORPHA280633	20486	Malformation syndrome		Multiple congenital anomalies-hypotonia-seizures syndrome		
ORPHA300496	21120	Malformation syndrome		Multiple congenital anomalies-hypotonia-seizures syndrome type 2		
ORPHA369837	22433	Malformation syndrome		Intellectual disability-seizures-hypotonia-ophthalmologic-skeletal anomalies syndrome		
ORPHA370933	22504	Group of phenomes		GM3 synthase deficiency		
ORPHA171714	17933	Disease				Amish infantile epilepsy syndrome
ORPHA370938	22505	Disease				Salt-and-pepper syndrome
ORPHA401820	22835	Disease		Autosomal recessive spastic paraplegia type 67		
ORPHA488635	25411	Disease		Early-onset epilepsy-intellectual disability-brain anomalies syndrome		
ORPHA309526	21385	Group of phenomes	Disorder of multiple glycosylation			
ORPHA602	8729	Disease		GNE myopathy		
ORPHA79322	11348	Disease		DPM1-CDG		
ORPHA79323	11349	Disease		MPDU1-CDG		
ORPHA79332	11358	Disease		B4GALT1-CDG		
ORPHA91131	12108	Disease		DK1-CDG		
ORPHA98873	13890	Disease		Congenital dyserythropoietic anemia type II		
ORPHA99843	14416	Clinical subtype		Leukocyte adhesion deficiency type II		
ORPHA238459	19265	Disease		SLC35A1-CDG		
ORPHA263494	20063	Disease		DPM3-CDG		
ORPHA294049	20904	Disease		Reunion Island Larsen syndrome		
ORPHA309568	21386	Group of phenomes		Defect in conserved oligomeric Golgi complex		
ORPHA79333	11359	Disease				COG7-CDG
ORPHA95428	12587	Disease				COG8-CDG
ORPHA263487	20062	Disease				COG5-CDG
ORPHA263501	20064	Disease				COG4-CDG
ORPHA263508	20065	Disease				COG1-CDG
ORPHA435934	23320	Disease				COG2-CDG
ORPHA464443	24026	Disease				COG6-CGD
ORPHA309778	21387	Group of phenomes		Defect in V-ATPase		
ORPHA357058	22201	Disease				Autosomal recessive cutis laxa type 2A
ORPHA2834	2571	Clinical subtype				Wrinkly skin syndrome
ORPHA357074	22203	Clinical subtype				Autosomal recessive cutis laxa type 2, classic type
ORPHA324737	21803	Disease		SRD5A3-CDG		
ORPHA329178	21904	Disease		Congenital muscular dystrophy with intellectual disability and severe epilepsy		
ORPHA356961	22189	Disease		SLC35A2-CDG		
ORPHA443811	23468	Disease		PGM3-CDG		
ORPHA448010	23561	Disease		CAD-CDG		
ORPHA466703	24160	Disease		TMEM199-CDG		
ORPHA468684	24235	Disease		CCDC115-CDG		
ORPHA68366	10506	Group of phenomes	Lysosomal disease			

ORPHA763	571	Disease	Pycnodysostosis		
ORPHA216	650	Group of phenomes	Neuronal ceroid lipofuscinosis		
ORPHA1947	353	Disease		Progressive epilepsy-intellectual disability syndrome, Finnish type	
ORPHA79262	11288	Disease		Adult neuronal ceroid lipofuscinosis	
ORPHA228329	19105	Etiological subtype			CLN1 disease
ORPHA228337	19106	Etiological subtype			CLN10 disease
ORPHA228340	19107	Etiological subtype			CLN4A disease
ORPHA228343	19108	Etiological subtype			CLN4B disease
ORPHA228363	19114	Etiological subtype			CLN6 disease
ORPHA314629	21505	Etiological subtype			CLN11 disease
ORPHA352709	22104	Etiological subtype			CLN13 disease
ORPHA79263	11289	Disease		Infantile neuronal ceroid lipofuscinosis	
ORPHA228329	19105	Etiological subtype			CLN1 disease
ORPHA263516	20066	Clinical subtype			Progressive myoclonic epilepsy type 3
ORPHA79264	11290	Disease		Juvenile neuronal ceroid lipofuscinosis	
ORPHA228329	19105	Etiological subtype			CLN1 disease
ORPHA228337	19106	Etiological subtype			CLN10 disease
ORPHA228346	19109	Etiological subtype			CLN3 disease
ORPHA228349	19110	Etiological subtype			CLN2 disease
ORPHA228354	19111	Etiological subtype			CLN8 disease
ORPHA228357	19112	Etiological subtype			CLN9 disease
ORPHA168486	17765	Disease		Congenital neuronal ceroid lipofuscinosis	
ORPHA228337	19106	Etiological subtype			CLN10 disease
ORPHA168491	17766	Disease		Late infantile neuronal ceroid lipofuscinosis	
ORPHA228329	19105	Etiological subtype			CLN1 disease
ORPHA228337	19106	Etiological subtype			CLN10 disease
ORPHA228349	19110	Etiological subtype			CLN2 disease
ORPHA228354	19111	Etiological subtype			CLN8 disease
ORPHA228360	19113	Etiological subtype			CLN5 disease
ORPHA228363	19114	Etiological subtype			CLN6 disease
ORPHA228366	19115	Etiological subtype			CLN7 disease
ORPHA314632	21506	Disease		Parkinsonism due to ATP13A2 deficiency	
ORPHA35121	10372	Disease	Lysosomal acid phosphatase deficiency		
ORPHA79207	11233	Group of phenomes	Disorder of lysosomal amino acid transport		
ORPHA213	11	Disease		Cystinosis	
ORPHA411629	23023	Clinical subtype			Nephropathic infantile cystinosis
ORPHA411634	23024	Clinical subtype			Juvenile nephropathic cystinosis
ORPHA411641	23025	Clinical subtype			Ocular cystinosis
ORPHA834	578	Disease		Free sialic acid storage disease	
ORPHA309324	21372	Clinical subtype			Free sialic acid storage disease, infantile form
ORPHA309331	21373	Clinical subtype			Intermediate severe Salla disease
ORPHA309334	21374	Clinical subtype			Salla disease
ORPHA79213	11239	Group of phenomes	Mucopolysaccharidosis		
ORPHA583	24	Disease		Mucopolysaccharidosis type 6	
ORPHA276212	20356	Clinical subtype			Mucopolysaccharidosis type 6, rapidly progressing
ORPHA276223	20357	Clinical subtype			Mucopolysaccharidosis type 6, slowly progressing
ORPHA584	40	Disease		Mucopolysaccharidosis type 7	
ORPHA580	131	Disease		Mucopolysaccharidosis type 2	
ORPHA217085	18824	Clinical subtype			Mucopolysaccharidosis type 2, severe form
ORPHA217093	18825	Clinical subtype			Mucopolysaccharidosis type 2, attenuated form
ORPHA579	132	Disease		Mucopolysaccharidosis type 1	
ORPHA93473	12381	Clinical subtype			Hurler syndrome
ORPHA93474	12382	Clinical subtype			Scheie syndrome
ORPHA93476	12383	Clinical subtype			Hurler-Scheie syndrome
ORPHA581	653	Disease		Mucopolysaccharidosis type 3	

ORPHA79269	11295	Etiological subtype				Sanfilippo syndrome type A
ORPHA79270	11296	Etiological subtype				Sanfilippo syndrome type B
ORPHA79271	11297	Etiological subtype				Sanfilippo syndrome type C
ORPHA79272	11298	Etiological subtype				Sanfilippo syndrome type D
ORPHA582	872	Disease		Mucopolysaccharidosis type 4		
ORPHA309297	21369	Clinical subtype				Mucopolysaccharidosis type 4A
ORPHA309310	21370	Clinical subtype				Mucopolysaccharidosis type 4B
ORPHA67041	10901	Disease		Hyaluronidase deficiency		
ORPHA79225	11251	Group of phenomes	Sphingolipidosis			
ORPHA585	6	Disease		Multiple sulfatase deficiency		
ORPHA333	12	Disease		Farber disease		
ORPHA487	22	Disease		Krabbe disease		
ORPHA206436	18495	Clinical subtype				Infantile Krabbe disease
ORPHA206443	18496	Clinical subtype				Late-infantile/juvenile Krabbe disease
ORPHA206448	18497	Clinical subtype				Adult Krabbe disease
ORPHA324	94	Disease		Fabry disease		
ORPHA512	112	Disease		Metachromatic leukodystrophy		
ORPHA309256	21362	Clinical subtype				Metachromatic leukodystrophy, late infantile form
ORPHA309263	21363	Clinical subtype				Metachromatic leukodystrophy, juvenile form
ORPHA309271	21364	Clinical subtype				Metachromatic leukodystrophy, adult form
ORPHA355	644	Disease		Gaucher disease		
ORPHA2072	1989	Clinical subtype				Gaucher disease-ophthalmoplegia-cardiovascular calcification syndrome
ORPHA77259	11102	Clinical subtype				Gaucher disease type 1
ORPHA77260	11103	Clinical subtype				Gaucher disease type 2
ORPHA77261	11104	Clinical subtype				Gaucher disease type 3
ORPHA85212	11662	Clinical subtype				Fetal Gaucher disease
ORPHA309252	21361	Clinical subtype				Atypical Gaucher disease due to saposin C deficiency
ORPHA77292	11105	Disease		Niemann-Pick disease type A		
ORPHA77293	11106	Disease		Niemann-Pick disease type B		
ORPHA79204	11230	Group of phenomes		Lipid storage disease		
ORPHA646	853	Disease				Niemann-Pick disease type C
ORPHA216972	18801	Clinical subtype				Niemann-Pick disease type C, severe perinatal form
ORPHA216975	18802	Clinical subtype				Niemann-Pick disease type C, severe early infantile neurologic onset
ORPHA216978	18803	Clinical subtype				Niemann-Pick disease type C, late infantile neurologic onset
ORPHA216981	18804	Clinical subtype				Niemann-Pick disease type C, juvenile neurologic onset
ORPHA216986	18805	Clinical subtype				Niemann-Pick disease type C, adult neurologic onset
ORPHA275761	20326	Disease				Lysosomal acid lipase deficiency
ORPHA75233	11067	Clinical subtype				Wolman disease
ORPHA75234	11068	Clinical subtype				Cholesteryl ester storage disease
ORPHA99022	14039	Disease		Niemann-Pick disease type E		
ORPHA139406	16887	Disease		Encephalopathy due to prosaposin deficiency		
ORPHA309144	21350	Group of phenomes		Gangliosidosis		
ORPHA354	643	Disease				GM1 gangliosidosis
ORPHA79255	11281	Clinical subtype				GM1 gangliosidosis type 1
ORPHA79256	11282	Clinical subtype				GM1 gangliosidosis type 2
ORPHA79257	11283	Clinical subtype				GM1 gangliosidosis type 3
ORPHA309152	21352	Group of phenomes				GM2 gangliosidosis
ORPHA796	38	Disease				Sandhoff disease
ORPHA309155	21353	Clinical subtype				Sandhoff disease, infantile form
ORPHA309162	21354	Clinical subtype				Sandhoff disease, juvenile form
ORPHA309169	21355	Clinical subtype				Sandhoff disease, adult form
ORPHA845	888	Disease				Tay-Sachs disease
ORPHA309178	21356	Clinical subtype				Tay-Sachs disease, B variant, infantile form
ORPHA309185	21357	Clinical subtype				Tay-Sachs disease, B variant, juvenile form
ORPHA309192	21358	Clinical subtype				Tay-Sachs disease, B variant, adult form

ORPHA309239	21359	Clinical subtype				Tay-Sachs disease, B1 variant
ORPHA309246	21360	Disease				GM2 gangliosidosis, AB variant
ORPHA306511	21221	Disease		Autosomal recessive spastic paraplegia type 48		
ORPHA309279	21365	Group of phenomes		Glycoproteinosis		
ORPHA79212	11238	Group of phenomes			Mucolipidosis	
ORPHA576	27	Disease				Mucolipidosis type II
ORPHA577	28	Disease				Mucolipidosis type III
ORPHA423461	23158	Clinical subtype				Mucolipidosis type III alpha/beta
ORPHA423470	23159	Clinical subtype				Mucolipidosis type III gamma
ORPHA578	29	Disease				Mucolipidosis type IV
ORPHA79215	11241	Group of phenomes			Oligosaccharidosis	
ORPHA61	3	Disease				Alpha-mannosidosis
ORPHA309282	21366	Clinical subtype				Alpha-mannosidosis, infantile form
ORPHA309288	21367	Clinical subtype				Alpha-mannosidosis, adult form
ORPHA93	5	Disease				Aspartylglucosaminuria
ORPHA118	7	Disease				Beta-mannosidosis
ORPHA349	13	Disease				Fucosidosis
ORPHA351	498	Disease				Galactosialidosis
ORPHA3137	673	Disease				Alpha-N-acetylgalactosaminidase deficiency
ORPHA79279	11305	Clinical subtype				Alpha-N-acetylgalactosaminidase deficiency type 1
ORPHA79280	11306	Clinical subtype				Alpha-N-acetylgalactosaminidase deficiency type 2
ORPHA79281	11307	Clinical subtype				Alpha-N-acetylgalactosaminidase deficiency type 3
ORPHA309294	21368	Group of phenomes				Sialidosis
ORPHA812	26	Disease				Sialidosis type 1
ORPHA87876	11801	Disease				Sialidosis type 2
ORPHA93399	12321	Clinical subtype				Juvenile sialidosis type 2
ORPHA93400	12322	Clinical subtype				Congenital sialidosis type 2
ORPHA309319	21371	Group of phenomes		Disorder of sialic acid metabolism		
ORPHA3166	766	Disease			Sialuria	
ORPHA309337	21375	Group of phenomes		Lysosomal glycogen storage disease		
ORPHA365	14	Disease			Glycogen storage disease due to acid maltase deficiency	
ORPHA308552	21321	Clinical subtype				Glycogen storage disease due to acid maltase deficiency, infantile onset
ORPHA420429	23106	Clinical subtype				Glycogen storage disease due to acid maltase deficiency, late-onset
ORPHA34587	10348	Disease			Glycogen storage disease due to LAMP-2 deficiency	
ORPHA68373	10513	Group of phenomes	Peroxisomal disease			
ORPHA79189	11215	Group of phenomes		Peroxisome biogenesis disorder		
ORPHA912	225	Disease			Zellweger syndrome	
ORPHA44	410	Disease			Neonatal adrenoleukodystrophy	
ORPHA772	5016	Disease			Infantile Refsum disease	
ORPHA309810	21391	Group of phenomes		Disorder of peroxisomal alpha-, beta- and omega-oxidation		
ORPHA773	381	Disease			Refsum disease	
ORPHA926	794	Disease			Acatalasemia	
ORPHA35706	10395	Disease			Glutaric acidemia type 3	
ORPHA79095	11137	Disease			Congenital bile acid synthesis defect type 4	
ORPHA79188	11214	Group of phenomes			Peroxisomal beta-oxidation disorder	
ORPHA2971	567	Disease				Peroxisomal acyl-CoA oxidase deficiency
ORPHA43	761	Disease				X-linked adrenoleukodystrophy
ORPHA139396	16884	Clinical subtype				X-linked cerebral adrenoleukodystrophy
ORPHA139399	16885	Clinical subtype				Adrenomyeloneuropathy
ORPHA300	3578	Disease				Bifunctional enzyme deficiency
ORPHA163684	17522	Disease				Leukoencephalopathy-dystonia-motor neuropathy syndrome
ORPHA93598	12431	Clinical subtype			Primary hyperoxaluria type 1	
ORPHA369942	22451	Disease		CADDS		
ORPHA3276	23428	Group of phenomes		Disorder of plasmalogens biosynthesis		
ORPHA177	3567	Disease			Rhizomelic chondrodysplasia punctata	

ORPHA309789	21388	Etiological subtype	Rhizomelic chondrodysplasia punctata type 1
ORPHA309796	21389	Etiological subtype	Rhizomelic chondrodysplasia punctata type 2
ORPHA309803	21390	Etiological subtype	Rhizomelic chondrodysplasia punctata type 3
ORPHA468717	24237	Etiological subtype	Rhizomelic chondrodysplasia punctata type 5
ORPHA438178	23367	Disease	Severe intellectual disability-epilepsy-cataract syndrome due to fatty acyl-CoA reductase 1 deficiency
ORPHA79062	11124	Group of phenomes	Disorder of amino acid and other organic acid metabolism
ORPHA79166	11192	Group of phenomes	Disorder of amino acid absorption and transport
ORPHA534	123	Malformation syndrome	Oculocerebrorenal syndrome of Lowe
ORPHA214	202	Disease	Cystinuria
ORPHA93612	12445	Etiological subtype	Cystinuria type A
ORPHA93613	12446	Etiological subtype	Cystinuria type B
ORPHA2195	3356	Disease	Dicarboxylic aminoaciduria
ORPHA470	3366	Disease	Lysinuric protein intolerance
ORPHA1032	3367	Disease	Hyperdibasic aminoaciduria type 1
ORPHA94086	12555	Disease	Blue diaper syndrome
ORPHA238517	19271	Group of phenomes	Hypotonia-cystinuria type 1 syndrome
ORPHA163690	17524	Disease	Hypotonia-cystinuria syndrome
ORPHA163693	17525	Disease	2p21 microdeletion syndrome
ORPHA238523	19272	Disease	Atypical hypotonia-cystinuria syndrome
ORPHA308451	21314	Group of phenomes	Disorder of neutral amino acid transport
ORPHA2116	502	Disease	Hartnup disease
ORPHA42062	10466	Disease	Iminoglycinuria
ORPHA363429	22302	Disease	Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome
ORPHA324262	21758	Clinical subtype	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency
ORPHA363432	22303	Clinical subtype	Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency
ORPHA79167	11193	Group of phenomes	Disorder of urea cycle metabolism and ammonia detoxification
ORPHA664	168	Disease	Ornithine transcarbamylase deficiency
ORPHA90	417	Disease	Argininemia
ORPHA23	459	Disease	Argininosuccinic aciduria
ORPHA147	461	Disease	Carbamoyl-phosphate synthetase 1 deficiency
ORPHA187	762	Group of phenomes	Citrullinemia
ORPHA247525	19524	Disease	Citrullinemia type I
ORPHA247546	19525	Clinical subtype	Acute neonatal citrullinemia type I
ORPHA247573	19526	Clinical subtype	Adult-onset citrullinemia type I
ORPHA247582	19527	Group of phenomes	Citrin deficiency
ORPHA247585	19528	Disease	Citrullinemia type II
ORPHA247598	19529	Disease	Neonatal intrahepatic cholestasis due to citrin deficiency
ORPHA415	770	Disease	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
ORPHA927	3370	Disease	Hyperammonemia due to N-acetylglutamate synthase deficiency
ORPHA35878	10403	Disease	Hyperinsulinism-hyperammonemia syndrome
ORPHA401948	22857	Disease	Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency
ORPHA79173	11199	Group of phenomes	Disorder of methionine cycle and sulfur amino acid metabolism
ORPHA394	173	Disease	Classic homocystinuria
ORPHA833	468	Disease	Encephalopathy due to sulfite oxidase deficiency
ORPHA99731	14304	Clinical subtype	Isolated sulfite oxidase deficiency
ORPHA99732	14305	Clinical subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency
ORPHA308386	21306	Etiological subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A
ORPHA308393	21307	Etiological subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type B
ORPHA308400	21308	Etiological subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type C
ORPHA1035	1342	Biological anomaly	Beta-mercaptolactate cysteine disulfiduria
ORPHA212	3365	Disease	Cystathioninuria
ORPHA622	3369	Disease	Homocystinuria without methylmalonic aciduria
ORPHA2169	2063	Clinical subtype	Methylcobalamin deficiency type cblE
ORPHA2170	3351	Clinical subtype	Methylcobalamin deficiency type cblG
ORPHA308380	21305	Clinical subtype	Methylcobalamin deficiency type cblDv1

ORPHA88618	11805	Disease	Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency			
ORPHA168598	17780	Disease	Brain demyelination due to methionine adenosyltransferase deficiency			
ORPHA289290	20707	Disease	Hypermethioninemia encephalopathy due to adenosine kinase deficiency			
ORPHA289891	20772	Disease	Hypermethioninemia due to glycine N-methyltransferase deficiency			
ORPHA79181	11207	Group of phenomes	Disorder of histidine metabolism			
ORPHA2157	3355	Disease	Histidinemia			
ORPHA210128	18685	Disease	Urocanic aciduria			
ORPHA79185	11211	Group of phenomes	Disorder of ornithine or proline metabolism			
ORPHA289866	20769	Group of phenomes	Disorder of proline metabolism			
ORPHA419	3729	Disease	Hyperprolinemia type 1			
ORPHA79101	11143	Disease	Hyperprolinemia type 2			
ORPHA289869	20770	Group of phenomes	Disorder of ornithine metabolism			
ORPHA414	3349	Disease	Gyrate atrophy of choroid and retina			
ORPHA79187	11213	Group of phenomes	Disorder of peptide metabolism			
ORPHA1361	1569	Disease	Carnosinemia			
ORPHA742	1727	Disease	Prolidase deficiency			
ORPHA2168	3357	Disease	Homocarnosinosis			
ORPHA79190	11216	Group of phenomes	Disorder of phenylalanin or tyrosine metabolism			
ORPHA284814	20625	Group of phenomes	Disorder of phenylalanine metabolism			
ORPHA716	611	Disease	Phenylketonuria			
ORPHA79253	11279	Clinical subtype	Mild phenylketonuria			
ORPHA79254	11280	Clinical subtype	Classic phenylketonuria			
ORPHA79651	11543	Clinical subtype	Mild hyperphenylalaninemia			
ORPHA293284	20847	Clinical subtype	Tetrahydrobiopterin-responsive hyperphenylalaninemia/phenylketonuria			
ORPHA2209	1889	Malformation syndrome	Maternal phenylketonuria			
ORPHA284818	20626	Group of phenomes	Disorder of tyrosine metabolism			
ORPHA56	411	Disease	Alkaptonuria			
ORPHA2118	503	Disease	Hawkinsinuria			
ORPHA3402	3372	Disease	Transient tyrosinemia of the newborn			
ORPHA882	3494	Disease	Tyrosinemia type 1			
ORPHA28378	8772	Disease	Tyrosinemia type 2			
ORPHA69723	10935	Disease	Tyrosinemia type 3			
ORPHA79194	11220	Group of phenomes	Disorder of serine or glycine metabolism			
ORPHA3129	773	Disease	Sarcosinemia			
ORPHA407	3556	Disease	Glycine encephalopathy			
ORPHA289857	20766	Clinical subtype	Neonatal glycine encephalopathy			
ORPHA289860	20767	Clinical subtype	Infantile glycine encephalopathy			
ORPHA289863	20768	Clinical subtype	Atypical glycine encephalopathy			
ORPHA35705	10394	Group of phenomes	Neurometabolic disorder due to serine deficiency			
ORPHA79350	11376	Disease	3-phosphoserine phosphatase deficiency			
ORPHA284417	20609	Disease	Phosphoserine aminotransferase deficiency			
ORPHA422519	23137	Group of phenomes	3-Phosphoglycerate dehydrogenase deficiency			
ORPHA2671	2439	Malformation syndrome	Neu-Laxova syndrome			
ORPHA79351	11377	Disease	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form			
ORPHA447997	23560	Disease	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome			
ORPHA243343	19468	Disease	Dimethylglycine dehydrogenase deficiency			
ORPHA79196	11222	Group of phenomes	Disorder of the gamma-glutamyl cycle			
ORPHA32	711	Disease	Glutathione synthetase deficiency			
ORPHA289846	20764	Clinical subtype	Glutathione synthetase deficiency with 5-oxoprolinuria			
ORPHA289849	20765	Clinical subtype	Glutathione synthetase deficiency without 5-oxoprolinuria			
ORPHA33572	10327	Disease	5-oxoprolinase deficiency			
ORPHA33573	10328	Disease	Gamma-glutamyl transpeptidase deficiency			
ORPHA33574	10329	Disease	Glutamate-cysteine ligase deficiency			
ORPHA79197	11223	Group of phenomes	Disorder of branched-chain amino acid metabolism			
ORPHA511	708	Disease	Maple syrup urine disease			

ORPHA268145	20168	Clinical subtype							Classic maple syrup urine disease
ORPHA268162	20169	Clinical subtype							Intermediate maple syrup urine disease
ORPHA268173	20170	Clinical subtype							Intermittent maple syrup urine disease
ORPHA268184	20171	Clinical subtype							Thiamine-responsive maple syrup urine disease
ORPHA289307	20709	Disease						Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency	
ORPHA308410	21310	Disease						Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency	
ORPHA401948	22857	Disease						Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency	
ORPHA289829	20761	Group of phenomes					Disorder of tryptophan metabolism		
ORPHA2224	2100	Disease						Hypertryptophanemia	
ORPHA79155	11181	Disease						Encephalopathy due to hydroxykynureninuria	
ORPHA289832	20762	Group of phenomes					Disorder of lysine and hydroxylysine metabolism		
ORPHA2203	3353	Disease						Hyperlysinemia	
ORPHA3124	3354	Disease						Saccharopinuria	
ORPHA79154	11180	Disease						2-aminoadipic 2-oxoadipic aciduria	
ORPHA79156	11182	Disease						Seizures-intellectual disability due to hydroxylysinuria syndrome	
ORPHA289841	20763	Group of phenomes					Disorder of glutamine metabolism		
ORPHA71278	11000	Disease						Congenital brain dysgenesis due to glutamine synthetase deficiency	
ORPHA289899	20774	Group of phenomes					Organic aciduria		
ORPHA79158	11184	Group of phenomes						Cerebral organic aciduria	
ORPHA19	3362	Group of phenomes							2-hydroxyglutaric aciduria
ORPHA79314	11340	Disease							L-2-hydroxyglutaric aciduria
ORPHA79315	11341	Disease							D-2-hydroxyglutaric aciduria
ORPHA356978	22190	Disease							D,L-2-hydroxyglutaric aciduria
ORPHA25	3564	Disease							Glutaryl-CoA dehydrogenase deficiency
ORPHA308448	21313	Group of phenomes							Aminoacylase deficiency
ORPHA141	8	Disease							Canavan disease
ORPHA314911	21537	Clinical subtype							Severe Canavan disease
ORPHA314918	21538	Clinical subtype							Mild Canavan disease
ORPHA137754	16711	Disease							Neurological conditions associated with aminoacylase 1 deficiency
ORPHA391417	22621	Disease							HSD10 disease
ORPHA85295	11685	Clinical subtype							HSD10 disease, atypical type
ORPHA391428	22622	Clinical subtype							HSD10 disease, infantile type
ORPHA391457	22623	Clinical subtype							HSD10 disease, neonatal type
ORPHA79163	11189	Group of phenomes						Classic organic aciduria	
ORPHA33	399	Disease							Isovaleric acidemia
ORPHA148	462	Group of phenomes							Multiple carboxylase deficiency
ORPHA79241	11267	Disease							Biotinidase deficiency
ORPHA79242	11268	Disease							Holocarboxylase synthetase deficiency
ORPHA26	710	Disease							Methylmalonic acidemia with homocystinuria
ORPHA79282	11308	Clinical subtype							Methylmalonic acidemia with homocystinuria, type cblC
ORPHA79283	11309	Clinical subtype							Methylmalonic acidemia with homocystinuria, type cblD
ORPHA79284	11310	Clinical subtype							Methylmalonic acidemia with homocystinuria type cblF
ORPHA369955	22453	Clinical subtype							Methylmalonic acidemia with homocystinuria, type cblJ
ORPHA369962	22454	Clinical subtype							Methylmalonic acidemia with homocystinuria, type cblX
ORPHA134	713	Disease							Beta-ketothiolase deficiency
ORPHA939	1260	Disease							3-hydroxyisobutyric aciduria
ORPHA20	3296	Disease							3-hydroxy-3-methylglutaric aciduria
ORPHA6	3297	Disease							3-methylcrotonyl-CoA carboxylase deficiency
ORPHA35	3557	Disease							Propionic acidemia
ORPHA79157	11183	Disease							2-methylbutyryl-CoA dehydrogenase deficiency
ORPHA79159	11185	Disease							Isobutyryl-CoA dehydrogenase deficiency
ORPHA88639	11817	Disease							Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency
ORPHA289504	20731	Disease							Combined malonic and methylmalonic acidemia
ORPHA289902	20775	Group of phenomes							3-methylglutaconic aciduria
ORPHA111	1059	Disease							Barth syndrome

ORPHA66634	10892	Disease							Dilated cardiomyopathy with ataxia
ORPHA67046	10906	Disease							3-methylglutaconic aciduria type 1
ORPHA67047	10907	Disease							3-methylglutaconic aciduria type 3
ORPHA67048	10908	Disease							3-methylglutaconic aciduria type 4
ORPHA352328	22056	Disease							MEGDEL syndrome
ORPHA445038	23516	Disease							3-methylglutaconic aciduria type 7
ORPHA505208	26323	Disease							3-methylglutaconic aciduria type 8
ORPHA505216	26324	Disease							3-methylglutaconic aciduria type 9
ORPHA293355	20854	Group of phenomes						Methylmalonic acidemia without homocystinuria	
ORPHA27	1263	Disease							Vitamin B12-unresponsive methylmalonic acidemia
ORPHA79312	11338	Clinical subtype							Vitamin B12-unresponsive methylmalonic acidemia type mut-
ORPHA289916	20777	Clinical subtype							Vitamin B12-unresponsive methylmalonic acidemia type mut0
ORPHA28	3260	Disease							Vitamin B12-responsive methylmalonic acidemia
ORPHA79310	11336	Clinical subtype							Vitamin B12-responsive methylmalonic acidemia type cblA
ORPHA79311	11337	Clinical subtype							Vitamin B12-responsive methylmalonic acidemia type cblB
ORPHA308442	21312	Clinical subtype							Vitamin B12-responsive methylmalonic acidemia, type cblDv2
ORPHA308425	21311	Disease							Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency
ORPHA308407	21309	Group of phenomes			Disorder of beta and omega amino acid metabolism				
ORPHA2066	3577	Disease				Gamma-aminobutyric acid transaminase deficiency			
ORPHA391381	22614	Group of phenomes			Disorder of asparagine metabolism				
ORPHA391376	22613	Disease				Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome			
ORPHA468726	24238	Disease			Severe primary trimethylaminuria				
ORPHA79161	11187	Group of phenomes		Disorder of carbohydrate metabolism					
ORPHA79177	11203	Group of phenomes			Gluconeogenesis disorder				
ORPHA348	676	Disease				Fructose-1,6-bisphosphatase deficiency			
ORPHA2880	3374	Disease				Phosphoenolpyruvate carboxykinase deficiency			
ORPHA3008	8026	Disease				Pyruvate carboxylase deficiency			
ORPHA353308	22134	Clinical subtype					Pyruvate carboxylase deficiency, infantile type		
ORPHA353314	22135	Clinical subtype					Pyruvate carboxylase deficiency, severe neonatal type		
ORPHA353320	22136	Clinical subtype					Pyruvate carboxylase deficiency, benign type		
ORPHA401948	22857	Disease				Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency			
ORPHA79179	11205	Group of phenomes			Disorder of glycerol metabolism				
ORPHA308993	21332	Group of phenomes				Glycerol kinase deficiency			
ORPHA408	463	Disease					Isolated glycerol kinase deficiency		
ORPHA284408	20606	Clinical subtype						Glycerol kinase deficiency, infantile form	
ORPHA284411	20607	Clinical subtype						Glycerol kinase deficiency, juvenile form	
ORPHA284414	20608	Clinical subtype						Glycerol kinase deficiency, adult form	
ORPHA261476	19879	Disease					Xp21 microdeletion syndrome		
ORPHA79201	11227	Group of phenomes			Glycogen storage disease				
ORPHA365	14	Disease				Glycogen storage disease due to acid maltase deficiency			
ORPHA308552	21321	Clinical subtype					Glycogen storage disease due to acid maltase deficiency, infantile onset		
ORPHA420429	23106	Clinical subtype					Glycogen storage disease due to acid maltase deficiency, late-onset		
ORPHA366	15	Disease				Glycogen storage disease due to glycogen debranching enzyme deficiency			
ORPHA367	16	Disease				Glycogen storage disease due to glycogen branching enzyme deficiency			
ORPHA206583	18528	Clinical subtype					Adult polyglucosan body disease		
ORPHA308621	21325	Clinical subtype					Glycogen storage disease due to glycogen branching enzyme deficiency, progressive hepatic form		
ORPHA308638	21326	Clinical subtype					Glycogen storage disease due to glycogen branching enzyme deficiency, non progressive hepatic form		
ORPHA308655	21327	Clinical subtype					Glycogen storage disease due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form		
ORPHA308670	21328	Clinical subtype					Glycogen storage disease due to glycogen branching enzyme deficiency, congenital neuromuscular form		
ORPHA308684	21329	Clinical subtype					Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form		
ORPHA308698	21330	Clinical subtype					Glycogen storage disease due to glycogen branching enzyme deficiency, childhood neuromuscular form		
ORPHA308712	21331	Clinical subtype					Glycogen storage disease due to glycogen branching enzyme deficiency, adult neuromuscular form		
ORPHA368	17	Disease				Glycogen storage disease due to muscle glycogen phosphorylase deficiency			
ORPHA369	18	Disease				Glycogen storage disease due to liver glycogen phosphorylase deficiency			
ORPHA371	19	Disease				Glycogen storage disease due to muscle phosphofructokinase deficiency			

ORPHA370	357	Group of phenomes	Glycogen storage disease due to phosphorylase kinase deficiency
ORPHA715	677	Disease	Glycogen storage disease due to muscle phosphorylase kinase deficiency
ORPHA79240	11266	Disease	Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency
ORPHA264580	20103	Disease	Glycogen storage disease due to liver phosphorylase kinase deficiency
ORPHA364	645	Disease	Glycogen storage disease due to glucose-6-phosphatase deficiency
ORPHA79258	11284	Clinical subtype	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ia
ORPHA79259	11285	Clinical subtype	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib
ORPHA57	738	Disease	Glycogen storage disease due to aldolase A deficiency
ORPHA713	739	Disease	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency
ORPHA2088	3513	Disease	Glycogen storage disease due to GLUT2 deficiency
ORPHA2364	3572	Disease	Glycogen storage disease due to lactate dehydrogenase deficiency
ORPHA284426	20610	Clinical subtype	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency
ORPHA284435	20611	Clinical subtype	Glycogen storage disease due to lactate dehydrogenase H-subunit deficiency
ORPHA34587	10348	Disease	Glycogen storage disease due to LAMP-2 deficiency
ORPHA97234	12860	Disease	Glycogen storage disease due to phosphoglycerate mutase deficiency
ORPHA99849	14422	Disease	Glycogen storage disease due to muscle beta-enolase deficiency
ORPHA263297	20039	Disease	Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency
ORPHA308520	21320	Group of phenomes	Glycogen storage disease due to glycogen synthase deficiency
ORPHA2089	3271	Disease	Glycogen storage disease due to hepatic glycogen synthase deficiency
ORPHA137625	16694	Disease	Glycogen storage disease due to muscle and heart glycogen synthase deficiency
ORPHA397937	22708	Disease	Polyglucosan body myopathy type 1
ORPHA439854	23393	Disease	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease
ORPHA456369	23680	Disease	Polyglucosan body myopathy type 2
ORPHA308459	21315	Group of phenomes	Disorder of glycolysis
ORPHA868	325	Disease	Triose phosphate-isomerase deficiency
ORPHA712	3304	Disease	Hemolytic anemia due to glucophosphate isomerase deficiency
ORPHA79299	11325	Disease	Hyperinsulinism due to glucokinase deficiency
ORPHA308463	21316	Group of phenomes	Disorder of fructose metabolism
ORPHA469	517	Disease	Hereditary fructose intolerance
ORPHA348	676	Disease	Fructose-1,6-bisphosphatase deficiency
ORPHA2056	3313	Disease	Essential fructosuria
ORPHA308467	21317	Group of phenomes	Disorder of galactose metabolism
ORPHA352	355	Group of phenomes	Galactosemia
ORPHA79237	11263	Disease	Galactokinase deficiency
ORPHA79238	11264	Disease	Galactose epimerase deficiency
ORPHA308473	21318	Clinical subtype	Erythrocyte galactose epimerase deficiency
ORPHA308487	21319	Clinical subtype	Generalized galactose epimerase deficiency
ORPHA79239	11265	Disease	Classic galactosemia
ORPHA308998	21333	Group of phenomes	Disorder of glyoxylate metabolism
ORPHA941	3360	Disease	D-glyceric aciduria
ORPHA416	3529	Disease	Primary hyperoxaluria
ORPHA93598	12431	Clinical subtype	Primary hyperoxaluria type 1
ORPHA93599	12432	Clinical subtype	Primary hyperoxaluria type 2
ORPHA93600	12433	Clinical subtype	Primary hyperoxaluria type 3
ORPHA309001	21334	Group of phenomes	Disorder of carbohydrate absorption and transport
ORPHA469	517	Disease	Hereditary fructose intolerance
ORPHA35122	10373	Disease	Congenital sucrase-isomaltase deficiency
ORPHA306436	21213	Clinical subtype	Congenital sucrase-isomaltase deficiency with starch intolerance
ORPHA306446	21214	Clinical subtype	Congenital sucrase-isomaltase deficiency with minimal starch tolerance
ORPHA306462	21215	Clinical subtype	Congenital sucrase-isomaltase deficiency without starch intolerance
ORPHA306474	21216	Clinical subtype	Congenital sucrase-isomaltase deficiency with starch and lactose intolerance
ORPHA306486	21217	Clinical subtype	Congenital sucrase-isomaltase deficiency without sucrose intolerance
ORPHA53690	10735	Disease	Congenital lactase deficiency
ORPHA79178	11204	Group of phenomes	Glucose transport disorder
ORPHA2088	3513	Disease	Glycogen storage disease due to GLUT2 deficiency

ORPHA35710	10398	Disease					Glucose-galactose malabsorption
ORPHA69076	10912	Disease					Familial renal glucosuria
ORPHA71277	10999	Disease					Encephalopathy due to GLUT1 deficiency
ORPHA103907	14983	Disease			Chronic diarrhea due to glucoamylase deficiency		
ORPHA103909	14985	Disease			Trehalase deficiency		
ORPHA165991	17595	Disease			Exercise-induced hyperinsulinism		
ORPHA247794	19548	Disease			Juvenile cataract-microcornea-renal glucosuria syndrome		
ORPHA440701	23410	Group of phenomes		Disorders of pentose/polyol metabolism			
ORPHA2843	3363	Disease			Pentosuria		
ORPHA79186	11212	Group of phenomes			Disorder of pentose phosphate metabolism		
ORPHA101028	14739	Disease					Transaldolase deficiency
ORPHA440706	23411	Disease					Ribose-5-P isomerase deficiency
ORPHA440713	23412	Disease					Isolated sedoheptulokinase deficiency
ORPHA488618	25408	Malformation syndrome					Transketolase deficiency
ORPHA79200	11226	Group of phenomes	Disorder of energy metabolism				
ORPHA68380	10520	Group of phenomes	Mitochondrial disease				
ORPHA223713	18973	Group of phenomes			Mitochondrial oxidative phosphorylation disorder		
ORPHA2443	277	Group of phenomes					Mitochondrial oxidative phosphorylation disorder due to nuclear DNA anomalies
ORPHA506	532	Group of phenomes					Leigh syndrome
ORPHA70472	10945	Disease					Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type
ORPHA70474	10947	Disease					Leigh syndrome with cardiomyopathy
ORPHA255241	19814	Disease					Leigh syndrome with leukodystrophy
ORPHA255249	19815	Disease					Leigh syndrome with nephrotic syndrome
ORPHA1561	3379	Disease					Fatal infantile cytochrome C oxidase deficiency
ORPHA35656	10380	Group of phenomes					Coenzyme Q10 deficiency
ORPHA1168	1427	Disease					Ataxia-oculomotor apraxia type 1
ORPHA139485	16908	Disease					Autosomal recessive ataxia due to ubiquinone deficiency
ORPHA254898	19797	Disease					Deafness-encephaloneuropathy-obesity-valvulopathy syndrome
ORPHA255249	19815	Disease					Leigh syndrome with nephrotic syndrome
ORPHA280406	20470	Disease					Familial steroid-resistant nephrotic syndrome with sensorineural deafness
ORPHA319678	21692	Disease					Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome
ORPHA457185	23703	Disease					Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome
ORPHA35696	10386	Group of phenomes					Mitochondrial disorder due to a defect in mitochondrial protein synthesis
ORPHA2598	1213	Disease					Mitochondrial myopathy and sideroblastic anemia
ORPHA2855	2589	Disease					Perrault syndrome
ORPHA99013	14030	Disease					Spastic paraplegia type 7
ORPHA101109	14820	Disease					Spinocerebellar ataxia type 28
ORPHA137681	16706	Disease					Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1
ORPHA137898	16739	Disease					Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome
ORPHA137908	16742	Disease					Hypotonia with lactic acidemia and hyperammonemia
ORPHA166073	17610	Malformation syndrome					Pontocerebellar hypoplasia type 6
ORPHA168566	17773	Disease					Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3
ORPHA217371	18849	Disease					Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins
ORPHA238329	19262	Disease					Severe X-linked mitochondrial encephalomyopathy
ORPHA254343	19738	Disease					Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome
ORPHA254920	19801	Disease					Combined oxidative phosphorylation defect type 2
ORPHA254925	19802	Disease					Combined oxidative phosphorylation defect type 4
ORPHA314051	21475	Disease					Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome
ORPHA314637	21507	Disease					Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency
ORPHA319504	21663	Disease					Combined oxidative phosphorylation defect type 8
ORPHA319509	21664	Disease					Combined oxidative phosphorylation defect type 9
ORPHA319514	21665	Disease					Combined oxidative phosphorylation defect type 13
ORPHA319519	21666	Disease					Combined oxidative phosphorylation defect type 14
ORPHA319524	21667	Disease					Combined oxidative phosphorylation defect type 15
ORPHA324535	21783	Disease					Combined oxidative phosphorylation defect type 11

ORPHA352563	22078	Disease	Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency
ORPHA363694	22340	Disease	Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome
ORPHA369913	22447	Disease	Combined oxidative phosphorylation defect type 17
ORPHA420728	23121	Disease	Combined oxidative phosphorylation defect type 20
ORPHA420733	23122	Disease	Combined oxidative phosphorylation defect type 21
ORPHA436174	23332	Disease	Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome
ORPHA444013	23474	Disease	Combined oxidative phosphorylation defect type 23
ORPHA444458	23489	Disease	Combined oxidative phosphorylation defect type 24
ORPHA447954	23553	Disease	Combined oxidative phosphorylation defect type 25
ORPHA457223	23709	Disease	Syndromic sensorineural deafness due to combined oxidative phosphorylation defect
ORPHA466722	24162	Disease	Autosomal recessive spastic paraplegia type 77
ORPHA466784	24168	Disease	Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect
ORPHA477684	25139	Disease	Combined oxidative phosphorylation defect type 26
ORPHA477774	25150	Disease	Combined oxidative phosphorylation defect type 27
ORPHA478042	25167	Disease	Combined oxidative phosphorylation defect type 30
ORPHA497623	25864	Group of phenomes	C12ORF65-related combined oxidative phosphorylation defect
ORPHA254930	19803	Disease	Combined oxidative phosphorylation defect type 7
ORPHA320375	21716	Disease	Autosomal recessive spastic paraplegia type 55
ORPHA199337	18404	Disease	Pancreatic insufficiency-anemia-hyperostosis syndrome
ORPHA254822	19780	Group of phenomes	Mitochondrial oxidative phosphorylation disorder with no known mechanism
ORPHA50812	10651	Disease	Zellweger-like syndrome without peroxisomal anomalies
ORPHA67036	10897	Disease	Autosomal dominant optic atrophy and cataract
ORPHA98673	13690	Disease	Autosomal dominant optic atrophy, classic form
ORPHA166105	17619	Disease	FASTKD2-related infantile mitochondrial encephalomyopathy
ORPHA227976	19058	Disease	Autosomal recessive optic atrophy, OPA7 type
ORPHA250932	19594	Disease	Autosomal dominant optic atrophy and peripheral neuropathy
ORPHA330050	21964	Disease	Lethal encephalopathy due to mitochondrial and peroxisomal fission defect
ORPHA391348	22609	Disease	Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome
ORPHA436271	23337	Disease	Non-progressive predominantly posterior cavitating leukoencephalopathy with peripheral neuropathy
ORPHA457050	23691	Disease	Autosomal dominant mitochondrial myopathy with exercise intolerance
ORPHA485421	25311	Malformation syndrome	Leigh-like basal ganglia disease-optic atrophy-peripheral neuropathy syndrome
ORPHA309136	21348	Group of phenomes	Mitochondrial disorder due to a defect in assembly or maturation of the respiratory chain complexes
ORPHA1194	5015	Disease	TMEM70-related mitochondrial encephalo-cardio-myopathy
ORPHA123	8566	Disease	Björnstad syndrome
ORPHA53693	10737	Disease	GRACILE syndrome
ORPHA254843	19786	Group of phenomes	Exercise intolerance with lactic acidosis
ORPHA43115	10579	Disease	Hereditary myopathy with lactic acidosis due to ISCU deficiency
ORPHA99901	14474	Disease	Acyl-CoA dehydrogenase 9 deficiency
ORPHA254902	19798	Disease	Renal tubulopathy-encephalopathy-liver failure syndrome
ORPHA289527	20735	Disease	Fatal infantile hypertrophic cardiomyopathy due to mitochondrial complex I deficiency
ORPHA289573	20743	Group of phenomes	Multiple mitochondrial dysfunctions syndrome
ORPHA363424	22301	Disease	Multiple mitochondrial dysfunctions syndrome type 3
ORPHA401869	22845	Disease	Multiple mitochondrial dysfunctions syndrome type 1
ORPHA401874	22846	Disease	Multiple mitochondrial dysfunctions syndrome type 2
ORPHA457406	23727	Disease	Multiple mitochondrial dysfunctions syndrome type 4
ORPHA352328	22056	Disease	MEGDEL syndrome
ORPHA397593	22684	Disease	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency
ORPHA330054	21965	Disease	Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome
ORPHA352456	22064	Group of phenomes	Mitochondrial DNA maintenance syndrome
ORPHA298	8030	Disease	Mitochondrial neurogastrointestinal encephalomyopathy
ORPHA35698	10388	Group of phenomes	Mitochondrial DNA depletion syndrome
ORPHA254803	19777	Group of phenomes	Mitochondrial DNA depletion syndrome, encephalomyopathic form
ORPHA1933	491	Disease	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria
ORPHA17	776	Disease	Fatal infantile lactic acidosis with methylmalonic aciduria
ORPHA255235	19813	Disease	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy

ORPHA369897	22445	Disease	Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies
ORPHA254871	19792	Group of phenomes	Mitochondrial DNA depletion syndrome, hepatocerebral form
ORPHA1186	1443	Disease	Infantile onset spinocerebellar ataxia
ORPHA726	1730	Disease	Alpers-Huttenlocher syndrome
ORPHA255229	19812	Disease	Navajo neurohepatopathy
ORPHA279934	20419	Disease	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency
ORPHA363534	22319	Disease	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form
ORPHA254875	19793	Disease	Mitochondrial DNA depletion syndrome, myopathic form
ORPHA313772	21449	Disease	Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome
ORPHA254807	19778	Group of phenomes	Multiple mitochondrial DNA deletion syndrome
ORPHA1215	1461	Disease	Autosomal dominant optic atrophy plus syndrome
ORPHA254818	19779	Group of phenomes	Ataxia neuropathy spectrum
ORPHA70595	10966	Disease	Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome
ORPHA94125	12565	Disease	Recessive mitochondrial ataxia syndrome
ORPHA254881	19794	Disease	Spinocerebellar ataxia with epilepsy
ORPHA254886	19795	Disease	Autosomal recessive progressive external ophthalmoplegia
ORPHA254892	19796	Disease	Autosomal dominant progressive external ophthalmoplegia
ORPHA329314	21925	Disease	Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency
ORPHA352470	22065	Disease	Mitochondrial DNA deletion syndrome with progressive myopathy
ORPHA352447	22063	Disease	Progressive external ophthalmoplegia-myopathy-emaciation syndrome
ORPHA391351	22610	Disease	SURF1-related Charcot-Marie-Tooth disease type 4
ORPHA435998	23324	Disease	Autosomal recessive intermediate Charcot-Marie-Tooth disease type D
ORPHA478049	25168	Disease	Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome
ORPHA254758	19772	Group of phenomes	Mitochondrial oxidative phosphorylation disorder due to mitochondrial DNA anomalies
ORPHA254767	19773	Group of phenomes	Mitochondrial oxidative phosphorylation disorder due to a large-scale single deletion of mitochondrial DNA
ORPHA480	61	Disease	Kearns-Sayre syndrome
ORPHA699	193	Disease	Pearson syndrome
ORPHA663	3558	Disease	Maternally-inherited progressive external ophthalmoplegia
ORPHA1670	3613	Disease	Chronic diarrhea with villous atrophy
ORPHA329336	21930	Disease	Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy
ORPHA254776	19774	Group of phenomes	Mitochondrial oxidative phosphorylation disorder due to a point mutation of mitochondrial DNA
ORPHA550	63	Disease	MELAS
ORPHA551	64	Disease	MERRF
ORPHA104	167	Disease	Leber hereditary optic neuropathy
ORPHA644	182	Disease	NARP syndrome
ORPHA225	7037	Disease	Maternally-inherited diabetes and deafness
ORPHA90641	12050	Etiological subtype	Mitochondrial non-syndromic sensorineural deafness
ORPHA99718	14291	Disease	Leber plus disease
ORPHA168609	17783	Etiological subtype	Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure
ORPHA254788	19775	Group of phenomes	Maternally-inherited mitochondrial myopathy
ORPHA2596	3612	Disease	Myopathy and diabetes mellitus
ORPHA254854	19789	Disease	Pure mitochondrial myopathy
ORPHA254857	19790	Disease	Lethal infantile mitochondrial myopathy
ORPHA254864	19791	Disease	Mitochondrial myopathy with reversible cytochrome C oxidase deficiency
ORPHA254851	19788	Disease	Maternally-inherited mitochondrial dystonia
ORPHA255210	19809	Disease	Maternally-inherited Leigh syndrome
ORPHA324525	21781	Disease	Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation
ORPHA397750	22699	Disease	Periodic paralysis with later-onset distal motor neuropathy
ORPHA254793	19776	Group of phenomes	Mitochondrial oxidative phosphorylation disorder due to a duplication of mitochondrial DNA
ORPHA3390	600	Disease	Proximal tubulopathy-diabetes mellitus-cerebellar ataxia syndrome
ORPHA254846	19787	Group of phenomes	Isolated oxidative phosphorylation complex disorder
ORPHA2609	369	Disease	Isolated complex I deficiency
ORPHA3208	3377	Disease	Isolated succinate-CoQ reductase deficiency
ORPHA1460	3378	Disease	Isolated complex III deficiency
ORPHA254905	19799	Disease	Isolated cytochrome C oxidase deficiency

ORPHA254913	19800	Disease				Isolated ATP synthase deficiency
ORPHA254827	19781	Group of phenomes		Mitochondrial membrane transport disorder		
ORPHA254830	19782	Group of phenomes			Mitochondrial substrate carrier disorder	
ORPHA1369	433	Disease				Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome
ORPHA1935	890	Clinical syndrome				Early myoclonic encephalopathy
ORPHA91130	12107	Disease				Cardiomyopathy-hypotonia-lactic acidosis syndrome
ORPHA255132	19805	Disease				Adult-onset autosomal recessive sideroblastic anemia
ORPHA353217	22119	Disease				Epileptic encephalopathy with global cerebral demyelination
ORPHA466784	24168	Disease				Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect
ORPHA254834	19783	Group of phenomes			Mitochondrial protein import disorder	
ORPHA52368	10691	Disease				Mohr-Tranebjaerg syndrome
ORPHA254837	19784	Group of phenomes		Unspecified mitochondrial disorder		
ORPHA51188	10672	Disease			Ethylmalonic encephalopathy	
ORPHA401854	22841	Group of phenomes		Lipoic acid biosynthesis defect		
ORPHA2394	5520	Clinical subtype			Pyruvate dehydrogenase E3 deficiency	
ORPHA289573	20743	Group of phenomes			Multiple mitochondrial dysfunctions syndrome	
ORPHA363424	22301	Disease				Multiple mitochondrial dysfunctions syndrome type 3
ORPHA401869	22845	Disease				Multiple mitochondrial dysfunctions syndrome type 1
ORPHA401874	22846	Disease				Multiple mitochondrial dysfunctions syndrome type 2
ORPHA457406	23727	Disease				Multiple mitochondrial dysfunctions syndrome type 4
ORPHA401859	22842	Disease			Lipoic acid synthetase deficiency	
ORPHA401862	22843	Disease			Lipoyl transferase 1 deficiency	
ORPHA401866	22844	Disease			Childhood-onset spasticity with hyperglycinemia	
ORPHA79172	11198	Group of phenomes	Creatine deficiency syndrome			
ORPHA382	1726	Disease		Guanidinoacetate methyltransferase deficiency		
ORPHA35704	10393	Disease		L-Arginine:glycine amidinotransferase deficiency		
ORPHA52503	10699	Disease		X-linked creatine transporter deficiency		
ORPHA79174	11200	Group of phenomes	Disorder of fatty acid oxidation and ketone body metabolism			
ORPHA79183	11209	Group of phenomes		Disorder of ketolysis		
ORPHA134	713	Disease			Beta-ketothiolase deficiency	
ORPHA832	3298	Disease			Succinyl-CoA:3-ketoacid CoA transferase deficiency	
ORPHA309115	21343	Group of phenomes		Disorder of fatty acid oxidation and ketogenesis		
ORPHA746	3294	Disease			Mitochondrial trifunctional protein deficiency	
ORPHA20	3296	Disease			3-hydroxy-3-methylglutaric aciduria	
ORPHA35701	10391	Disease			3-hydroxy-3-methylglutaryl-CoA synthase deficiency	
ORPHA309120	21344	Group of phenomes			Acyl-CoA dehydrogenase deficiency	
ORPHA42	3570	Disease				Medium chain acyl-CoA dehydrogenase deficiency
ORPHA26791	8766	Disease				Multiple acyl-CoA dehydrogenase deficiency
ORPHA394529	22659	Clinical subtype				Multiple acyl-CoA dehydrogenase deficiency, severe neonatal type
ORPHA394532	22660	Clinical subtype				Multiple acyl-CoA dehydrogenase deficiency, mild type
ORPHA26792	8767	Disease				Short chain acyl-CoA dehydrogenase deficiency
ORPHA26793	8768	Disease				Very long chain acyl-CoA dehydrogenase deficiency
ORPHA329942	21949	Disease				Transient neonatal multiple acyl-CoA dehydrogenase deficiency
ORPHA309127	21345	Group of phenomes			3-hydroxyacyl-CoA dehydrogenase deficiency	
ORPHA5	3555	Disease				Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
ORPHA71212	10987	Disease				Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency
ORPHA309130	21346	Group of phenomes		Disorder of carnitine cycle and carnitine transport		
ORPHA157	901	Disease			Carnitine palmitoyltransferase II deficiency	
ORPHA228302	19099	Clinical subtype				Carnitine palmitoyl transferase II deficiency, myopathic form
ORPHA228305	19100	Clinical subtype				Carnitine palmitoyl transferase II deficiency, severe infantile form
ORPHA228308	19101	Clinical subtype				Carnitine palmitoyl transferase II deficiency, neonatal form
ORPHA156	1215	Disease			Carnitine palmitoyl transferase 1A deficiency	
ORPHA158	3316	Disease			Systemic primary carnitine deficiency	
ORPHA159	3343	Disease			Carnitine-acylcarnitine translocase deficiency	
ORPHA309133	21347	Group of phenomes		Metabolic disease due to other fatty acid oxidation disorder		

ORPHA943	3295	Disease	Malonic aciduria
ORPHA99900	14473	Disease	Long chain acyl-CoA dehydrogenase deficiency
ORPHA438072	23360	Group of phenomes	Disorder of keton body transport
ORPHA438075	23361	Disease	Ketoacidosis due to monocarboxylate transporter-1 deficiency
ORPHA254746	19770	Group of phenomes	Pyruvate metabolism disorder
ORPHA765	467	Disease	Pyruvate dehydrogenase deficiency
ORPHA2394	5520	Clinical subtype	Pyruvate dehydrogenase E3 deficiency
ORPHA79243	11269	Clinical subtype	Pyruvate dehydrogenase E1-alpha deficiency
ORPHA79244	11270	Clinical subtype	Pyruvate dehydrogenase E2 deficiency
ORPHA79246	11272	Clinical subtype	Pyruvate dehydrogenase phosphatase deficiency
ORPHA255138	19806	Clinical subtype	Pyruvate dehydrogenase E1-beta deficiency
ORPHA255182	19807	Clinical subtype	Pyruvate dehydrogenase E3-binding protein deficiency
ORPHA766	3257	Disease	Hemolytic anemia due to red cell pyruvate kinase deficiency
ORPHA447784	23543	Disease	Mitochondrial pyruvate carrier deficiency
ORPHA254749	19771	Group of phenomes	Tricarboxylic acid cycle disorder
ORPHA31	1259	Disease	Oxoglutaricaciduria
ORPHA24	3376	Disease	Fumaric aciduria
ORPHA313850	21458	Disease	Infantile cerebellar-retinal degeneration
ORPHA79214	11240	Group of phenomes	Disorder of biogenic amine metabolism and transport
ORPHA79169	11195	Group of phenomes	Disorder of neurotransmitter metabolism and transport
ORPHA3057	2747	Disease	Monoamine oxidase A deficiency
ORPHA309819	21394	Group of phenomes	Disorder of pterin metabolism
ORPHA70594	10965	Disease	Dopa-responsive dystonia due to sepiapterin reductase deficiency
ORPHA98808	13825	Disease	Autosomal dominant dopa-responsive dystonia
ORPHA101150	14826	Disease	Autosomal recessive dopa-responsive dystonia
ORPHA238583	19279	Disease	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency
ORPHA226	457	Clinical subtype	Dihydropteridine reductase deficiency
ORPHA13	771	Clinical subtype	6-pyruvoyl-tetrahydropterin synthase deficiency
ORPHA2102	787	Clinical subtype	GTP cyclohydrolase I deficiency
ORPHA1578	3391	Clinical subtype	Pterin-4 alpha-carbinolamine dehydratase deficiency
ORPHA309830	21397	Group of phenomes	Disorder of catecholamine synthesis
ORPHA230	8743	Disease	Dopamine beta-hydroxylase deficiency
ORPHA35708	10397	Disease	Aromatic L-amino acid decarboxylase deficiency
ORPHA352649	22091	Disease	Brain dopamine-serotonin vesicular transport disease
ORPHA79175	11201	Group of phenomes	Disorder of gamma-aminobutyric acid metabolism
ORPHA22	402	Disease	Succinic semialdehyde dehydrogenase deficiency
ORPHA2066	3577	Disease	Gamma-aminobutyric acid transaminase deficiency
ORPHA79192	11218	Group of phenomes	Disorder of pyridoxine metabolism
ORPHA3006	880	Disease	Pyridoxine-dependent epilepsy
ORPHA79096	11138	Disease	Pyridoxal phosphate-responsive seizures
ORPHA79219	11245	Group of phenomes	Metabolic disease involving other neurotransmitter deficiency
ORPHA3197	2862	Disease	Hereditary hyperekplexia
ORPHA132	3704	Disease	Butyrylcholinesterase deficiency
ORPHA79097	11139	Disease	Folinic acid-responsive seizures
ORPHA79224	11250	Group of phenomes	Disorder of purine or pyrimidine metabolism
ORPHA79191	11217	Group of phenomes	Disorder of purine metabolism
ORPHA45	458	Disease	Adenosine monophosphate deaminase deficiency
ORPHA760	671	Disease	Purine nucleoside phosphorylase deficiency
ORPHA3467	704	Disease	Hereditary xanthinuria
ORPHA93601	12434	Etiological subtype	Xanthinuria type I
ORPHA93602	12435	Etiological subtype	Xanthinuria type II
ORPHA46	763	Disease	Adenylosuccinate lyase deficiency
ORPHA976	775	Disease	Adenine phosphoribosyltransferase deficiency
ORPHA1187	1444	Disease	Lethal ataxia with deafness and optic atrophy
ORPHA3222	2879	Disease	Phosphoribosylpyrophosphate synthetase superactivity

ORPHA411536	23018	Clinical subtype						Mild phosphoribosylpyrophosphate synthetase superactivity
ORPHA411543	23019	Clinical subtype						Severe phosphoribosylpyrophosphate synthetase superactivity
ORPHA206428	18494	Group of phenomes			Hypoxanthine-guanine phosphoribosyltransferase deficiency			
ORPHA510	197	Disease						Lesch-Nyhan syndrome
ORPHA79233	11259	Disease						Hypoxanthine guanine phosphoribosyltransferase partial deficiency
ORPHA209886	18663	Disease			Familial juvenile hyperuricemic nephropathy type 1			
ORPHA250977	19596	Disease			AICA-ribosiduria			
ORPHA279934	20419	Disease			Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency			
ORPHA423479	23160	Disease			X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome			
ORPHA457375	23724	Disease			ITPA-related encephalopathy			
ORPHA79193	11219	Group of phenomes		Disorder of pyrimidine metabolism				
ORPHA30	404	Disease			Hereditary orotic aciduria			
ORPHA1675	774	Disease			Dihydropyrimidine dehydrogenase deficiency			
ORPHA298	8030	Disease			Mitochondrial neurogastrointestinal encephalomyopathy			
ORPHA35120	10371	Disease			Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency			
ORPHA38874	10451	Disease			Dihydropyrimidinuria			
ORPHA65287	10869	Disease			Beta-ureidopropionase deficiency			
ORPHA254875	19793	Disease			Mitochondrial DNA depletion syndrome, myopathic form			
ORPHA309147	21351	Disease			Hyper-beta-alaninemia			
ORPHA448010	23561	Disease			CAD-CDG			
ORPHA91088	12103	Group of phenomes	Other metabolic disease					
ORPHA60	194	Disease		Alpha-1-antitrypsin deficiency				
ORPHA714	3306	Disease		Hemolytic anemia due to diphosphoglycerate mutase deficiency				
ORPHA79345	11371	Malformation syndrome		Brachytelephalangic chondrodysplasia punctata				
ORPHA79507	11533	Disease		Hypotonia-failure to thrive-microcephaly syndrome				
ORPHA99845	14418	Disease		Genetic recurrent myoglobinuria				
ORPHA99846	14419	Disease		Autosomal dominant myoglobinuria				
ORPHA404454	22923	Disease		Alacrimia-choreoathetosis-liver dysfunction syndrome				
ORPHA309005	21335	Group of phenomes	Disorder of lipid metabolism					
ORPHA79226	11252	Group of phenomes		Sterol metabolism disorder				
ORPHA79168	11194	Group of phenomes			Disorder of bile acid synthesis			
ORPHA84065	11606	Disease						Idiopathic malabsorption due to bile acid synthesis defects
ORPHA163631	17508	Group of phenomes						Bile acid synthesis defect with cholestasis and malabsorption
ORPHA909	605	Disease						Cerebrotendinous xanthomatosis
ORPHA238475	19267	Disease						Familial hypercholanemia
ORPHA276066	20346	Disease						Bile acid CoA ligase deficiency and defective amidation
ORPHA485631	25322	Group of phenomes						Congenital bile acid synthesis defect
ORPHA79095	11137	Disease						Congenital bile acid synthesis defect type 4
ORPHA79301	11327	Disease						Congenital bile acid synthesis defect type 1
ORPHA79302	11328	Disease						Congenital bile acid synthesis defect type 3
ORPHA79303	11329	Disease						Congenital bile acid synthesis defect type 2
ORPHA209902	18665	Disease						Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency
ORPHA79195	11221	Group of phenomes			Sterol biosynthesis disorder			
ORPHA1426	1632	Disease						Greenberg dysplasia
ORPHA139	2136	Disease						CHILD syndrome
ORPHA818	3574	Malformation syndrome						Smith-Lemli-Opitz syndrome
ORPHA35107	10370	Disease						Desmosterolosis
ORPHA35173	10376	Disease						X-linked dominant chondrodysplasia punctata
ORPHA46059	10592	Disease						Lathosterolosis
ORPHA251383	19639	Malformation syndrome						CK syndrome
ORPHA309025	21338	Group of phenomes						Mevalonate kinase deficiency
ORPHA29	403	Disease						Mevalonic aciduria
ORPHA343	3276	Disease						Hyperimmunoglobulinemia D with periodic fever
ORPHA401973	22861	Malformation syndrome						MEND syndrome
ORPHA488168	25388	Malformation syndrome						Microcephaly-congenital cataract-psoriasiform dermatitis syndrome

ORPHA101953	14889	Group of phenomes	Rare dyslipidemia				
ORPHA309028	21339	Group of phenomes	Disorder of lipid absorption and transport				
ORPHA352301	22047	Group of phenomes	Disorder of phospholipids, sphingolipids and fatty acids biosynthesis				
ORPHA352306	22049	Group of phenomes		Disorder of phospholipids, sphingolipids and fatty acids biosynthesis with central nervous system predominant involvement			
ORPHA816	586	Disease			Sjögren-Larsson syndrome		
ORPHA139480	16907	Disease			Autosomal recessive spastic paraplegia type 39		
ORPHA157850	17156	Disease			Pantothenate kinase-associated neurodegeneration		
ORPHA329303	21923	Group of phenomes			PLA2G6-associated neurodegeneration		
ORPHA35069	10365	Disease				Infantile neuroaxonal dystrophy	
ORPHA199351	18408	Disease				Adult-onset dystonia-parkinsonism	
ORPHA329308	21924	Disease			Fatty acid hydroxylase-associated neurodegeneration		
ORPHA352328	22056	Disease			MEGDEL syndrome		
ORPHA352333	22057	Disease			Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome		
ORPHA370933	22504	Group of phenomes			GM3 synthase deficiency		
ORPHA171714	17933	Disease				Amish infantile epilepsy syndrome	
ORPHA370938	22505	Disease				Salt-and-pepper syndrome	
ORPHA423296	23152	Disease			Spinocerebellar ataxia type 38		
ORPHA424027	23187	Disease			Progressive myoclonic epilepsy type 8		
ORPHA431361	23249	Disease			Progressive encephalopathy with leukodystrophy due to DECR deficiency		
ORPHA506353	26450	Disease			Autosomal recessive complex spastic paraplegia due to Kennedy pathway dysfunction		
ORPHA352309	22050	Group of phenomes		Disorder of phospholipids, sphingolipids and fatty acids biosynthesis with peripheral nerves predominant involvement			
ORPHA36386	10423	Disease			Hereditary sensory and autonomic neuropathy type 1		
ORPHA171848	17940	Disease			Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome		
ORPHA352312	22051	Group of phenomes		Disorder of phospholipids, sphingolipids and fatty acids biosynthesis with skeletal muscle predominant involvement			
ORPHA1369	433	Disease			Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome		
ORPHA111	1059	Disease			Barth syndrome		
ORPHA165	2137	Group of phenomes			Neutral lipid storage disease		
ORPHA98907	13924	Disease				Dorfman-Chanarin disease	
ORPHA98908	13925	Disease				Neutral lipid storage myopathy	
ORPHA99845	14418	Disease			Genetic recurrent myoglobinuria		
ORPHA280671	20495	Disease			Megaconial congenital muscular dystrophy		
ORPHA506334	26449	Disease		Familial steroid-resistant nephrotic syndrome with adrenal insufficiency			
ORPHA309340	21376	Group of phenomes	Disorder of lysosomal-related organelles				
ORPHA309813	21392	Group of phenomes	Disorder of porphyrin and haem metabolism				
ORPHA738	657	Group of phenomes	Porphyria				
ORPHA79277	11303	Disease		Congenital erythropoietic porphyria			
ORPHA79278	11304	Disease		Autosomal erythropoietic protoporphyria			
ORPHA95157	12578	Group of phenomes		Acute hepatic porphyria			
ORPHA79273	11299	Disease			Hereditary coproporphyria		
ORPHA79276	11302	Disease			Acute intermittent porphyria		
ORPHA79473	11499	Disease			Porphyria variegata		
ORPHA100924	14678	Disease			Porphyria due to ALA dehydratase deficiency		
ORPHA95161	12581	Group of phenomes		Chronic hepatic porphyria			
ORPHA95159	12580	Disease			Hepatoerythropoietic porphyria		
ORPHA101330	14841	Disease			Porphyria cutanea tarda		
ORPHA443057	23434	Clinical subtype				Sporadic porphyria cutanea tarda	
ORPHA443062	23435	Clinical subtype				Familial porphyria cutanea tarda	
ORPHA280379	20463	Disease		Erythropoietic uroporphyria associated with myeloid malignancy			
ORPHA443197	23451	Disease		X-linked erythropoietic protoporphyria			
ORPHA75563	11087	Disease	X-linked sideroblastic anemia				
ORPHA309816	21393	Group of phenomes	Disorder of bilirubin metabolism and excretion				
ORPHA205	242	Disease		Crigler-Najjar syndrome			
ORPHA79234	11260	Clinical subtype			Crigler-Najjar syndrome type 1		
ORPHA79235	11261	Clinical subtype			Crigler-Najjar syndrome type 2		
ORPHA234	805	Disease		Dubin-Johnson syndrome			

ORPHA3111	882	Disease	Rotor syndrome
ORPHA172	1073	Disease	Progressive familial intrahepatic cholestasis
ORPHA79304	11330	Clinical subtype	Progressive familial intrahepatic cholestasis type 2
ORPHA79305	11331	Clinical subtype	Progressive familial intrahepatic cholestasis type 3
ORPHA79306	11332	Clinical subtype	Progressive familial intrahepatic cholestasis type 1
ORPHA480476	25205	Clinical subtype	Progressive familial intrahepatic cholestasis type 5
ORPHA480483	25206	Clinical subtype	Progressive familial intrahepatic cholestasis type 4
ORPHA480491	25207	Clinical subtype	MYO5B-related progressive familial intrahepatic cholestasis
ORPHA65682	10872	Disease	Benign recurrent intrahepatic cholestasis
ORPHA99960	14533	Clinical subtype	Benign recurrent intrahepatic cholestasis type 1
ORPHA99961	14534	Clinical subtype	Benign recurrent intrahepatic cholestasis type 2
ORPHA415286	23081	Clinical syndrome	Kernicterus spectrum disorder
ORPHA309824	21395	Group of phenomes	Disorder of metabolite absorption and transport
ORPHA309827	21396	Group of phenomes	Disorder of vitamin and non-protein cofactor absorption and transport
ORPHA79171	11197	Group of phenomes	Disorder of cobalamin metabolism and transport
ORPHA26	710	Disease	Methylmalonic acidemia with homocystinuria
ORPHA79282	11308	Clinical subtype	Methylmalonic acidemia with homocystinuria, type cblC
ORPHA79283	11309	Clinical subtype	Methylmalonic acidemia with homocystinuria, type cblD
ORPHA79284	11310	Clinical subtype	Methylmalonic acidemia with homocystinuria type cblF
ORPHA369955	22453	Clinical subtype	Methylmalonic acidemia with homocystinuria, type cblJ
ORPHA369962	22454	Clinical subtype	Methylmalonic acidemia with homocystinuria, type cblX
ORPHA859	1729	Disease	Transcobalamin deficiency
ORPHA28	3260	Disease	Vitamin B12-responsive methylmalonic acidemia
ORPHA79310	11336	Clinical subtype	Vitamin B12-responsive methylmalonic acidemia type cblA
ORPHA79311	11337	Clinical subtype	Vitamin B12-responsive methylmalonic acidemia type cblB
ORPHA308442	21312	Clinical subtype	Vitamin B12-responsive methylmalonic acidemia, type cblDv2
ORPHA2967	3358	Disease	Transcobalamin I deficiency
ORPHA332	3359	Disease	Congenital intrinsic factor deficiency
ORPHA622	3369	Disease	Homocystinuria without methylmalonic aciduria
ORPHA2169	2063	Clinical subtype	Methylcobalamin deficiency type cblE
ORPHA2170	3351	Clinical subtype	Methylcobalamin deficiency type cblG
ORPHA308380	21305	Clinical subtype	Methylcobalamin deficiency type cblDv1
ORPHA35858	10402	Disease	Gräsbeck-Imerslund disease
ORPHA280183	20433	Biological anomaly	Methylmalonic aciduria due to transcobalamin receptor defect
ORPHA285657	20670	Group of phenomes	Disorder of folate metabolism and transport
ORPHA395	465	Disease	Homocystinuria due to methylene tetrahydrofolate reductase deficiency
ORPHA51208	10673	Disease	Formiminoglutamic aciduria
ORPHA90045	11940	Disease	Hereditary folate malabsorption
ORPHA217382	18851	Disease	Neurodegenerative syndrome due to cerebral folate transport deficiency
ORPHA319651	21687	Disease	Constitutional megaloblastic anemia with severe neurologic disease
ORPHA298644	21080	Group of phenomes	Disorder of thiamine metabolism and transport
ORPHA49827	10643	Disease	Thiamine-responsive megaloblastic anemia syndrome
ORPHA99742	14315	Malformation syndrome	Amish lethal microcephaly
ORPHA199348	18407	Disease	Thiamine-responsive encephalopathy
ORPHA217396	18854	Disease	Progressive polyneuropathy with bilateral striatal necrosis
ORPHA293955	20894	Disease	Childhood encephalopathy due to thiamine pyrophosphokinase deficiency
ORPHA309833	21398	Group of phenomes	Disorder of other vitamins and cofactors metabolism and transport
ORPHA96	149	Disease	Ataxia with vitamin E deficiency
ORPHA79096	11138	Disease	Pyridoxal phosphate-responsive seizures
ORPHA79241	11267	Disease	Biotinidase deficiency
ORPHA98434	13451	Disease	Hereditary combined deficiency of vitamin K-dependent clotting factors
ORPHA99732	14305	Clinical subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency
ORPHA308386	21306	Etiological subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A
ORPHA308393	21307	Etiological subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type B
ORPHA308400	21308	Etiological subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type C

ORPHA199285	18389	Disease							Hereditary hypercarotenemia and vitamin A deficiency
ORPHA352718	22106	Disease							Progressive retinal dystrophy due to retinol transport defect
ORPHA411712	23032	Disease							Maternal riboflavin deficiency
ORPHA309836	21399	Group of phenomes			Disorder of mineral absorption and transport				
ORPHA309839	21400	Group of phenomes				Disorder of copper metabolism			
ORPHA905	134	Disease							Wilson disease
ORPHA565	278	Disease							Menkes disease
ORPHA1551	1723	Disease							Familial benign copper deficiency
ORPHA198	7035	Disease							Occipital horn syndrome
ORPHA171851	17941	Disease							MEDNIK syndrome
ORPHA309842	21401	Group of phenomes				Disorder of iron metabolism and transport			
ORPHA220489	18915	Group of phenomes							Rare hereditary hemochromatosis
ORPHA79230	11256	Disease							Hemochromatosis type 2
ORPHA139491	16909	Disease							Hemochromatosis type 4
ORPHA225123	18976	Disease							Hemochromatosis type 3
ORPHA447792	23545	Disease							Hemochromatosis type 5
ORPHA465508	24056	Disease							Symptomatic form of hemochromatosis type 1
ORPHA309845	21402	Group of phenomes				Disorder of zinc metabolism and transport			
ORPHA309848	21403	Group of phenomes				Disorder of magnesium transport			
ORPHA34526	10344	Group of phenomes							Familial primary hypomagnesemia
ORPHA306516	21222	Group of phenomes							Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis
ORPHA2196	2078	Disease							Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement
ORPHA31043	9284	Disease							Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement
ORPHA306519	21223	Group of phenomes							Familial primary hypomagnesemia with hypocalcuria
ORPHA34528	10346	Disease							Autosomal dominant primary hypomagnesemia with hypocalciuria
ORPHA306522	21224	Group of phenomes							Familial primary hypomagnesemia with normocalcuria
ORPHA30924	9282	Disease							Primary hypomagnesemia with secondary hypocalcemia
ORPHA34527	10345	Disease							Familial primary hypomagnesemia with normocalciuria and normocalcemia
ORPHA199326	18401	Disease							Isolated autosomal dominant hypomagnesemia, Glaudemans type
ORPHA309851	21404	Group of phenomes				Disorder of manganese transport			
ORPHA309854	21405	Disease							Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome

Classification of rare neurologic diseases (Functional)			Source: www.orpha.net	State: 12.06.2018
ORPHA98006	13024	Group of phenomes	Rare neurologic disease	
ORPHA68385	10525	Group of phenomes	Neurometabolic disease	
ORPHA61	3	Disease	Alpha-mannosidosis	
ORPHA309282	21366	Clinical subtype	Alpha-mannosidosis, infantile form	
ORPHA309288	21367	Clinical subtype	Alpha-mannosidosis, adult form	
ORPHA93	5	Disease	Aspartylglucosaminuria	
ORPHA585	6	Disease	Multiple sulfatase deficiency	
ORPHA141	8	Disease	Canavan disease	
ORPHA314911	21537	Clinical subtype	Severe Canavan disease	
ORPHA314918	21538	Clinical subtype	Mild Canavan disease	
ORPHA333	12	Disease	Farber disease	
ORPHA487	22	Disease	Krabbe disease	
ORPHA206436	18495	Clinical subtype	Infantile Krabbe disease	
ORPHA206443	18496	Clinical subtype	Late-infantile/juvenile Krabbe disease	
ORPHA206448	18497	Clinical subtype	Adult Krabbe disease	
ORPHA812	26	Disease	Sialidosis type 1	
ORPHA95	45	Disease	Friedreich ataxia	
ORPHA480	61	Disease	Kearns-Sayre syndrome	
ORPHA550	63	Disease	MELAS	
ORPHA551	64	Disease	MERRF	
ORPHA512	112	Disease	Metachromatic leukodystrophy	
ORPHA309256	21362	Clinical subtype	Metachromatic leukodystrophy, late infantile form	
ORPHA309263	21363	Clinical subtype	Metachromatic leukodystrophy, juvenile form	
ORPHA309271	21364	Clinical subtype	Metachromatic leukodystrophy, adult form	
ORPHA580	131	Disease	Mucopolysaccharidosis type 2	
ORPHA217085	18824	Clinical subtype	Mucopolysaccharidosis type 2, severe form	
ORPHA217093	18825	Clinical subtype	Mucopolysaccharidosis type 2, attenuated form	
ORPHA579	132	Disease	Mucopolysaccharidosis type 1	
ORPHA93473	12381	Clinical subtype	Hurler syndrome	
ORPHA93474	12382	Clinical subtype	Scheie syndrome	
ORPHA93476	12383	Clinical subtype	Hurler-Scheie syndrome	
ORPHA905	134	Disease	Wilson disease	
ORPHA96	149	Disease	Ataxia with vitamin E deficiency	
ORPHA394	173	Disease	Classic homocystinuria	
ORPHA644	182	Disease	NARP syndrome	
ORPHA14	252	Disease	Abetalipoproteinemia	
ORPHA565	278	Disease	Menkes disease	
ORPHA868	325	Disease	Triose phosphate-isomerase deficiency	
ORPHA385	358	Group of phenomes	Neurodegeneration with brain iron accumulation	
ORPHA3464	3045	Disease	Woodhouse-Sakati syndrome	
ORPHA48818	10633	Disease	Aceruloplasminemia	
ORPHA157846	17155	Disease	Neuroferritinopathy	
ORPHA157850	17156	Disease	Pantothenate kinase-associated neurodegeneration	
ORPHA216866	18796	Clinical subtype	Classic pantothenate kinase-associated neurodegeneration	
ORPHA216873	18797	Clinical subtype	Atypical pantothenate kinase-associated neurodegeneration	

ORPHA289560	20741	Disease	Mitochondrial membrane protein-associated neurodegeneration
ORPHA306674	21249	Disease	Kufor-Rakeb syndrome
ORPHA329284	21922	Disease	Beta-propeller protein-associated neurodegeneration
ORPHA329303	21923	Group of phenomes	PLA2G6-associated neurodegeneration
ORPHA35069	10365	Disease	Infantile neuroaxonal dystrophy
ORPHA199351	18408	Disease	Adult-onset dystonia-parkinsonism
ORPHA329308	21924	Disease	Fatty acid hydroxylase-associated neurodegeneration
ORPHA397725	22696	Disease	COASY protein-associated neurodegeneration
ORPHA496756	25847	Disease	Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome
ORPHA2609	369	Disease	Isolated complex I deficiency
ORPHA773	381	Disease	Refsum disease
ORPHA22	402	Disease	Succinic semialdehyde dehydrogenase deficiency
ORPHA833	468	Disease	Encephalopathy due to sulfite oxidase deficiency
ORPHA99731	14304	Clinical subtype	Isolated sulfite oxidase deficiency
ORPHA99732	14305	Clinical subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency
ORPHA308386	21306	Etiological subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A
ORPHA308393	21307	Etiological subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type B
ORPHA308400	21308	Etiological subtype	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type C
ORPHA255	484	Group of phenomes	Dopa-responsive dystonia
ORPHA70594	10965	Disease	Dopa-responsive dystonia due to sepiapterin reductase deficiency
ORPHA98808	13825	Disease	Autosomal dominant dopa-responsive dystonia
ORPHA101150	14826	Disease	Autosomal recessive dopa-responsive dystonia
ORPHA351	498	Disease	Galactosialidosis
ORPHA2116	502	Disease	Hartnup disease
ORPHA506	532	Group of phenomes	Leigh syndrome
ORPHA70472	10945	Disease	Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type
ORPHA70474	10947	Disease	Leigh syndrome with cardiomyopathy
ORPHA255241	19814	Disease	Leigh syndrome with leukodystrophy
ORPHA255249	19815	Disease	Leigh syndrome with nephrotic syndrome
ORPHA834	578	Disease	Free sialic acid storage disease
ORPHA309324	21372	Clinical subtype	Free sialic acid storage disease, infantile form
ORPHA309331	21373	Clinical subtype	Intermediate severe Salla disease
ORPHA309334	21374	Clinical subtype	Salla disease
ORPHA816	586	Disease	Sjögren-Larsson syndrome
ORPHA909	605	Disease	Cerebrotendinous xanthomatosis
ORPHA716	611	Disease	Phenylketonuria
ORPHA79253	11279	Clinical subtype	Mild phenylketonuria
ORPHA79254	11280	Clinical subtype	Classic phenylketonuria
ORPHA79651	11543	Clinical subtype	Mild hyperphenylalaninemia
ORPHA293284	20847	Clinical subtype	Tetrahydrobiopterin-responsive hyperphenylalaninemia/phenylketonuria
ORPHA354	643	Disease	GM1 gangliosidosis
ORPHA79255	11281	Clinical subtype	GM1 gangliosidosis type 1
ORPHA79256	11282	Clinical subtype	GM1 gangliosidosis type 2
ORPHA79257	11283	Clinical subtype	GM1 gangliosidosis type 3
ORPHA216	650	Group of phenomes	Neuronal ceroid lipofuscinosis
ORPHA1947	353	Disease	Progressive epilepsy-intellectual disability syndrome, Finnish type

ORPHA79262	11288	Disease	Adult neuronal ceroid lipofuscinosis
ORPHA228329	19105	Etiological subtype	CLN1 disease
ORPHA228337	19106	Etiological subtype	CLN10 disease
ORPHA228340	19107	Etiological subtype	CLN4A disease
ORPHA228343	19108	Etiological subtype	CLN4B disease
ORPHA228363	19114	Etiological subtype	CLN6 disease
ORPHA314629	21505	Etiological subtype	CLN11 disease
ORPHA352709	22104	Etiological subtype	CLN13 disease
ORPHA79263	11289	Disease	Infantile neuronal ceroid lipofuscinosis
ORPHA228329	19105	Etiological subtype	CLN1 disease
ORPHA263516	20066	Clinical subtype	Progressive myoclonic epilepsy type 3
ORPHA79264	11290	Disease	Juvenile neuronal ceroid lipofuscinosis
ORPHA228329	19105	Etiological subtype	CLN1 disease
ORPHA228337	19106	Etiological subtype	CLN10 disease
ORPHA228346	19109	Etiological subtype	CLN3 disease
ORPHA228349	19110	Etiological subtype	CLN2 disease
ORPHA228354	19111	Etiological subtype	CLN8 disease
ORPHA228357	19112	Etiological subtype	CLN9 disease
ORPHA168486	17765	Disease	Congenital neuronal ceroid lipofuscinosis
ORPHA228337	19106	Etiological subtype	CLN10 disease
ORPHA168491	17766	Disease	Late infantile neuronal ceroid lipofuscinosis
ORPHA228329	19105	Etiological subtype	CLN1 disease
ORPHA228337	19106	Etiological subtype	CLN10 disease
ORPHA228349	19110	Etiological subtype	CLN2 disease
ORPHA228354	19111	Etiological subtype	CLN8 disease
ORPHA228360	19113	Etiological subtype	CLN5 disease
ORPHA228363	19114	Etiological subtype	CLN6 disease
ORPHA228366	19115	Etiological subtype	CLN7 disease
ORPHA314632	21506	Disease	Parkinsonism due to ATP13A2 deficiency
ORPHA581	653	Disease	Mucopolysaccharidosis type 3
ORPHA79269	11295	Etiological subtype	Sanfilippo syndrome type A
ORPHA79270	11296	Etiological subtype	Sanfilippo syndrome type B
ORPHA79271	11297	Etiological subtype	Sanfilippo syndrome type C
ORPHA79272	11298	Etiological subtype	Sanfilippo syndrome type D
ORPHA3137	673	Disease	Alpha-N-acetylgalactosaminidase deficiency
ORPHA79279	11305	Clinical subtype	Alpha-N-acetylgalactosaminidase deficiency type 1
ORPHA79280	11306	Clinical subtype	Alpha-N-acetylgalactosaminidase deficiency type 2
ORPHA79281	11307	Clinical subtype	Alpha-N-acetylgalactosaminidase deficiency type 3
ORPHA43	761	Disease	X-linked adrenoleukodystrophy
ORPHA139396	16884	Clinical subtype	X-linked cerebral adrenoleukodystrophy
ORPHA139399	16885	Clinical subtype	Adrenomyeloneuropathy
ORPHA646	853	Disease	Niemann-Pick disease type C
ORPHA216972	18801	Clinical subtype	Niemann-Pick disease type C, severe perinatal form
ORPHA216975	18802	Clinical subtype	Niemann-Pick disease type C, severe early infantile neurologic onset
ORPHA216978	18803	Clinical subtype	Niemann-Pick disease type C, late infantile neurologic onset
ORPHA216981	18804	Clinical subtype	Niemann-Pick disease type C, juvenile neurologic onset

ORPHA216986	18805	Clinical subtype	Niemann-Pick disease type C, adult neurologic onset
ORPHA3006	880	Disease	Pyridoxine-dependent epilepsy
ORPHA31	1259	Disease	Oxoglutaricaciduria
ORPHA382	1726	Disease	Guanidinoacetate methyltransferase deficiency
ORPHA742	1727	Disease	Prolidase deficiency
ORPHA726	1730	Disease	Alpers-Huttenlocher syndrome
ORPHA2388	2219	Disease	Choreoacanthocytosis
ORPHA2962	2673	Disease	De Barsy syndrome
ORPHA35664	10381	Etiological subtype	ALDH18A1-related De Barsy syndrome
ORPHA293633	20864	Etiological subtype	PYCR1-related De Barsy syndrome
ORPHA3057	2747	Disease	Monoamine oxidase A deficiency
ORPHA3197	2862	Disease	Hereditary hyperekplexia
ORPHA19	3362	Group of phenomes	2-hydroxyglutaric aciduria
ORPHA79314	11340	Disease	L-2-hydroxyglutaric aciduria
ORPHA79315	11341	Disease	D-2-hydroxyglutaric aciduria
ORPHA356978	22190	Disease	D,L-2-hydroxyglutaric aciduria
ORPHA622	3369	Disease	Homocystinuria without methylmalonic aciduria
ORPHA2169	2063	Clinical subtype	Methylcobalamin deficiency type cblE
ORPHA2170	3351	Clinical subtype	Methylcobalamin deficiency type cblG
ORPHA308380	21305	Clinical subtype	Methylcobalamin deficiency type cblDv1
ORPHA24	3376	Disease	Fumaric aciduria
ORPHA407	3556	Disease	Glycine encephalopathy
ORPHA289857	20766	Clinical subtype	Neonatal glycine encephalopathy
ORPHA289860	20767	Clinical subtype	Infantile glycine encephalopathy
ORPHA289863	20768	Clinical subtype	Atypical glycine encephalopathy
ORPHA663	3558	Disease	Maternally-inherited progressive external ophthalmoplegia
ORPHA25	3564	Disease	Glutaryl-CoA dehydrogenase deficiency
ORPHA177	3567	Disease	Rhizomelic chondrodysplasia punctata
ORPHA309789	21388	Etiological subtype	Rhizomelic chondrodysplasia punctata type 1
ORPHA309796	21389	Etiological subtype	Rhizomelic chondrodysplasia punctata type 2
ORPHA309803	21390	Etiological subtype	Rhizomelic chondrodysplasia punctata type 3
ORPHA468717	24237	Etiological subtype	Rhizomelic chondrodysplasia punctata type 5
ORPHA2066	3577	Disease	Gamma-aminobutyric acid transaminase deficiency
ORPHA419	3729	Disease	Hyperprolinemia type 1
ORPHA230	8743	Disease	Dopamine beta-hydroxylase deficiency
ORPHA31150	9288	Disease	Tangier disease
ORPHA35656	10380	Group of phenomes	Coenzyme Q10 deficiency
ORPHA1168	1427	Disease	Ataxia-oculomotor apraxia type 1
ORPHA139485	16908	Disease	Autosomal recessive ataxia due to ubiquinone deficiency
ORPHA254898	19797	Disease	Deafness-encephaloneuropathy-obesity-valvulopathy syndrome
ORPHA255249	19815	Disease	Leigh syndrome with nephrotic syndrome
ORPHA280406	20470	Disease	Familial steroid-resistant nephrotic syndrome with sensorineural deafness
ORPHA319678	21692	Disease	Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome
ORPHA457185	23703	Disease	Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome
ORPHA35704	10393	Disease	L-Arginine:glycine amidinotransferase deficiency
ORPHA35705	10394	Group of phenomes	Neurometabolic disorder due to serine deficiency

ORPHA79350	11376	Disease	3-phosphoserine phosphatase deficiency
ORPHA284417	20609	Disease	Phosphoserine aminotransferase deficiency
ORPHA422519	23137	Group of phenomes	3-Phosphoglycerate dehydrogenase deficiency
ORPHA2671	2439	Malformation syndrome	Neu-Laxova syndrome
ORPHA79351	11377	Disease	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form
ORPHA447997	23560	Disease	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome
ORPHA35708	10397	Disease	Aromatic L-amino acid decarboxylase deficiency
ORPHA52503	10699	Disease	X-linked creatine transporter deficiency
ORPHA59306	10791	Disease	McLeod neuroacanthocytosis syndrome
ORPHA65284	10866	Disease	Biotin-thiamine-responsive basal ganglia disease
ORPHA65287	10869	Disease	Beta-ureidopropionase deficiency
ORPHA71212	10987	Disease	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency
ORPHA71277	10999	Disease	Encephalopathy due to GLUT1 deficiency
ORPHA71278	11000	Disease	Congenital brain dysgenesis due to glutamine synthetase deficiency
ORPHA77260	11103	Clinical subtype	Gaucher disease type 2
ORPHA77261	11104	Clinical subtype	Gaucher disease type 3
ORPHA77292	11105	Disease	Niemann-Pick disease type A
ORPHA79096	11138	Disease	Pyridoxal phosphate-responsive seizures
ORPHA79097	11139	Disease	Folinic acid-responsive seizures
ORPHA79101	11143	Disease	Hyperprolinemia type 2
ORPHA79155	11181	Disease	Encephalopathy due to hydroxykynureninuria
ORPHA79157	11183	Disease	2-methylbutyryl-CoA dehydrogenase deficiency
ORPHA79189	11215	Group of phenomes	Peroxisome biogenesis disorder
ORPHA912	225	Disease	Zellweger syndrome
ORPHA44	410	Disease	Neonatal adrenoleukodystrophy
ORPHA772	5016	Disease	Infantile Refsum disease
ORPHA88639	11817	Disease	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency
ORPHA137754	16711	Disease	Neurological conditions associated with aminoacylase 1 deficiency
ORPHA139406	16887	Disease	Encephalopathy due to prosaposin deficiency
ORPHA166105	17619	Disease	FASTKD2-related infantile mitochondrial encephalomyopathy
ORPHA168598	17780	Disease	Brain demyelination due to methionine adenosyltransferase deficiency
ORPHA206428	18494	Group of phenomes	Hypoxanthine-guanine phosphoribosyltransferase deficiency
ORPHA510	197	Disease	Lesch-Nyhan syndrome
ORPHA79233	11259	Disease	Hypoxanthine guanine phosphoribosyltransferase partial deficiency
ORPHA210128	18685	Disease	Urocanic aciduria
ORPHA238329	19262	Disease	Severe X-linked mitochondrial encephalomyopathy
ORPHA238583	19279	Disease	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency
ORPHA226	457	Clinical subtype	Dihydropteridine reductase deficiency
ORPHA13	771	Clinical subtype	6-pyruvoyl-tetrahydropterin synthase deficiency
ORPHA2102	787	Clinical subtype	GTP cyclohydrolase I deficiency
ORPHA1578	3391	Clinical subtype	Pterin-4 alpha-carbinolamine dehydratase deficiency
ORPHA254803	19777	Group of phenomes	Mitochondrial DNA depletion syndrome, encephalomyopathic form
ORPHA1933	491	Disease	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria
ORPHA17	776	Disease	Fatal infantile lactic acidosis with methylmalonic aciduria
ORPHA255235	19813	Disease	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy
ORPHA369897	22445	Disease	Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies

ORPHA254875	19793	Disease	Mitochondrial DNA depletion syndrome, myopathic form
ORPHA254886	19795	Disease	Autosomal recessive progressive external ophthalmoplegia
ORPHA254892	19796	Disease	Autosomal dominant progressive external ophthalmoplegia
ORPHA255210	19809	Disease	Maternally-inherited Leigh syndrome
ORPHA289290	20707	Disease	Hypermethioninemia encephalopathy due to adenosine kinase deficiency
ORPHA289573	20743	Group of phenomes	Multiple mitochondrial dysfunctions syndrome
ORPHA363424	22301	Disease	Multiple mitochondrial dysfunctions syndrome type 3
ORPHA401869	22845	Disease	Multiple mitochondrial dysfunctions syndrome type 1
ORPHA401874	22846	Disease	Multiple mitochondrial dysfunctions syndrome type 2
ORPHA457406	23727	Disease	Multiple mitochondrial dysfunctions syndrome type 4
ORPHA289877	20771	Particular clinical situation in a disease or syndrc	Transient hyperammonemia of the newborn
ORPHA300313	21109	Disease	Congenital cataract-hearing loss-severe developmental delay syndrome
ORPHA306511	21221	Disease	Autosomal recessive spastic paraplegia type 48
ORPHA309152	21352	Group of phenomes	GM2 gangliosidosis
ORPHA796	38	Disease	Sandhoff disease
ORPHA845	888	Disease	Tay-Sachs disease
ORPHA309246	21360	Disease	GM2 gangliosidosis, AB variant
ORPHA313850	21458	Disease	Infantile cerebellar-retinal degeneration
ORPHA324535	21783	Disease	Combined oxidative phosphorylation defect type 11
ORPHA329336	21930	Disease	Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy
ORPHA330050	21964	Disease	Lethal encephalopathy due to mitochondrial and peroxisomal fission defect
ORPHA352328	22056	Disease	MEGDEL syndrome
ORPHA352333	22057	Disease	Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome
ORPHA352447	22063	Disease	Progressive external ophthalmoplegia-myopathy-emaciation syndrome
ORPHA369942	22451	Disease	CADDS
ORPHA371047	22516	Group of phenomes	Congenital disorder of glycosylation with neurological involvement
ORPHA2953	3480	Disease	Ehlers-Danlos syndrome, musculocontractural type
ORPHA602	8729	Disease	GNE myopathy
ORPHA34515	10337	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2I
ORPHA79324	11350	Disease	ALG12-CDG
ORPHA79332	11358	Disease	B4GALT1-CDG
ORPHA206554	18521	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2M
ORPHA206564	18523	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2O
ORPHA263494	20063	Disease	DPM3-CDG
ORPHA263516	20066	Clinical subtype	Progressive myoclonic epilepsy type 3
ORPHA300536	21127	Disease	DDOST-CDG
ORPHA314667	21512	Disease	TMEM165-CDG
ORPHA353327	22137	Etiological subtype	Congenital myasthenic syndromes with glycosylation defect
ORPHA370980	22511	Disease	Congenital muscular dystrophy without intellectual disability
ORPHA371054	22517	Group of phenomes	X-linked congenital disorder of glycosylation with intellectual disability as a major feature
ORPHA356961	22189	Disease	SLC35A2-CDG
ORPHA371064	22518	Group of phenomes	Non-X-linked congenital disorder of glycosylation with intellectual disability as a major feature
ORPHA709	968	Malformation syndrome	Peters plus syndrome
ORPHA3474	3498	Malformation syndrome	CHIME syndrome
ORPHA272	8724	Disease	Congenital muscular dystrophy, Fukuyama type
ORPHA899	8725	Disease	Walker-Warburg syndrome

ORPHA588	8726	Disease	Muscle-eye-brain disease
ORPHA79318	11344	Disease	PMM2-CDG
ORPHA79321	11347	Disease	ALG3-CDG
ORPHA79323	11349	Disease	MPDU1-CDG
ORPHA79325	11351	Disease	ALG8-CDG
ORPHA79326	11352	Disease	ALG2-CDG
ORPHA79327	11353	Disease	ALG1-CDG
ORPHA79329	11355	Disease	MGAT2-CDG
ORPHA86309	11726	Disease	DPAGT1-CDG
ORPHA86812	11732	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2K
ORPHA95428	12587	Disease	COG8-CDG
ORPHA99843	14416	Clinical subtype	Leukocyte adhesion deficiency type II
ORPHA206559	18522	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2N
ORPHA238459	19265	Disease	SLC35A1-CDG
ORPHA244310	19478	Disease	RFT1-CDG
ORPHA263487	20062	Disease	COG5-CDG
ORPHA263501	20064	Disease	COG4-CDG
ORPHA263508	20065	Disease	COG1-CDG
ORPHA280071	20425	Disease	ALG11-CDG
ORPHA280333	20455	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2P
ORPHA280633	20486	Malformation syndrome	Multiple congenital anomalies-hypotonia-seizures syndrome
ORPHA324737	21803	Disease	SRD5A3-CDG
ORPHA329178	21904	Disease	Congenital muscular dystrophy with intellectual disability and severe epilepsy
ORPHA352479	22066	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2U
ORPHA357058	22201	Disease	Autosomal recessive cutis laxa type 2A
ORPHA2834	2571	Clinical subtype	Wrinkly skin syndrome
ORPHA357074	22203	Clinical subtype	Autosomal recessive cutis laxa type 2, classic type
ORPHA363417	22298	Malformation syndrome	Temtamy preaxial brachydactyly syndrome
ORPHA363623	22329	Disease	Autosomal recessive limb-girdle muscular dystrophy type 2T
ORPHA370921	22500	Disease	STT3A-CDG
ORPHA370924	22501	Disease	STT3B-CDG
ORPHA370930	22503	Disease	XYLT1-CDG
ORPHA370933	22504	Group of phenomes	GM3 synthase deficiency
ORPHA171714	17933	Disease	Amish infantile epilepsy syndrome
ORPHA370938	22505	Disease	Salt-and-pepper syndrome
ORPHA370943	22507	Disease	Autism spectrum disorder-epilepsy-arthrogryposis syndrome
ORPHA370959	22509	Disease	Congenital muscular dystrophy with cerebellar involvement
ORPHA370968	22510	Disease	Congenital muscular dystrophy with intellectual disability
ORPHA397941	22709	Disease	MAN1B1-CDG
ORPHA371071	22519	Group of phenomes	Congenital disorder of glycosylation with epilepsy as a major feature
ORPHA3474	3498	Malformation syndrome	CHIME syndrome
ORPHA272	8724	Disease	Congenital muscular dystrophy, Fukuyama type
ORPHA899	8725	Disease	Walker-Warburg syndrome
ORPHA588	8726	Disease	Muscle-eye-brain disease
ORPHA79318	11344	Disease	PMM2-CDG
ORPHA79320	11346	Disease	ALG6-CDG

ORPHA79321	11347	Disease	ALG3-CDG
ORPHA79322	11348	Disease	DPM1-CDG
ORPHA79323	11349	Disease	MPDU1-CDG
ORPHA79326	11352	Disease	ALG2-CDG
ORPHA79327	11353	Disease	ALG1-CDG
ORPHA79328	11354	Disease	ALG9-CDG
ORPHA79330	11356	Disease	MOGS-CDG
ORPHA79333	11359	Disease	COG7-CDG
ORPHA83639	11601	Disease	Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency
ORPHA86309	11726	Disease	DPAGT1-CDG
ORPHA95428	12587	Disease	COG8-CDG
ORPHA99843	14416	Clinical subtype	Leukocyte adhesion deficiency type II
ORPHA238459	19265	Disease	SLC35A1-CDG
ORPHA244310	19478	Disease	RFT1-CDG
ORPHA263501	20064	Disease	COG4-CDG
ORPHA280071	20425	Disease	ALG11-CDG
ORPHA280633	20486	Malformation syndrome	Multiple congenital anomalies-hypotonia-seizures syndrome
ORPHA300496	21120	Malformation syndrome	Multiple congenital anomalies-hypotonia-seizures syndrome type 2
ORPHA324422	21777	Disease	ALG13-CDG
ORPHA329178	21904	Disease	Congenital muscular dystrophy with intellectual disability and severe epilepsy
ORPHA356961	22189	Disease	SLC35A2-CDG
ORPHA357058	22201	Disease	Autosomal recessive cutis laxa type 2A
ORPHA2834	2571	Clinical subtype	Wrinkly skin syndrome
ORPHA357074	22203	Clinical subtype	Autosomal recessive cutis laxa type 2, classic type
ORPHA370921	22500	Disease	STT3A-CDG
ORPHA370924	22501	Disease	STT3B-CDG
ORPHA370927	22502	Disease	SSR4-CDG
ORPHA370933	22504	Group of phenomes	GM3 synthase deficiency
ORPHA171714	17933	Disease	Amish infantile epilepsy syndrome
ORPHA370938	22505	Disease	Salt-and-pepper syndrome
ORPHA370943	22507	Disease	Autism spectrum disorder-epilepsy-arthrogryposis syndrome
ORPHA466926	24178	Disease	Seizures-scoliosis-macrocephaly syndrome
ORPHA488635	25411	Disease	Early-onset epilepsy-intellectual disability-brain anomalies syndrome
ORPHA435934	23320	Disease	COG2-CDG
ORPHA464443	24026	Disease	COG6-CGD
ORPHA391348	22609	Disease	Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome
ORPHA391376	22613	Disease	Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome
ORPHA391417	22621	Disease	HSD10 disease
ORPHA85295	11685	Clinical subtype	HSD10 disease, atypical type
ORPHA391428	22622	Clinical subtype	HSD10 disease, infantile type
ORPHA391457	22623	Clinical subtype	HSD10 disease, neonatal type
ORPHA397937	22708	Disease	Polyglucosan body myopathy type 1
ORPHA401859	22842	Disease	Lipoic acid synthetase deficiency
ORPHA401866	22844	Disease	Childhood-onset spasticity with hyperglycinemia
ORPHA415286	23081	Clinical syndrome	Kernicterus spectrum disorder
ORPHA420728	23121	Disease	Combined oxidative phosphorylation defect type 20

ORPHA420733	23122	Disease	Combined oxidative phosphorylation defect type 21
ORPHA423479	23160	Disease	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome
ORPHA431361	23249	Disease	Progressive encephalopathy with leukodystrophy due to DECR deficiency
ORPHA436174	23332	Disease	Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome
ORPHA436271	23337	Disease	Non-progressive predominantly posterior cavitating leukoencephalopathy with peripheral neuropathy
ORPHA438178	23367	Disease	Severe intellectual disability-epilepsy-cataract syndrome due to fatty acyl-CoA reductase 1 deficiency
ORPHA444013	23474	Disease	Combined oxidative phosphorylation defect type 23
ORPHA445038	23516	Disease	3-methylglutaconic aciduria type 7
ORPHA445110	23519	Disease	Limb-girdle muscular dystrophy due to POMK deficiency
ORPHA447784	23543	Disease	Mitochondrial pyruvate carrier deficiency
ORPHA447954	23553	Disease	Combined oxidative phosphorylation defect type 25
ORPHA456369	23680	Disease	Polyglucosan body myopathy type 2
ORPHA457050	23691	Disease	Autosomal dominant mitochondrial myopathy with exercise intolerance
ORPHA457375	23724	Disease	ITPA-related encephalopathy
ORPHA477774	25150	Disease	Combined oxidative phosphorylation defect type 27
ORPHA485421	25311	Malformation syndrome	Leigh-like basal ganglia disease-optic atrophy-peripheral neuropathy syndrome
ORPHA497623	25864	Group of phenomes	C12ORF65-related combined oxidative phosphorylation defect
ORPHA254930	19803	Disease	Combined oxidative phosphorylation defect type 7
ORPHA320375	21716	Disease	Autosomal recessive spastic paraplegia type 55
ORPHA505208	26323	Disease	3-methylglutaconic aciduria type 8
ORPHA505216	26324	Disease	3-methylglutaconic aciduria type 9