Asper Biogene LLC
Vaksali 17A, 50410 Tartu, Estonia
Phone +372 7307 295
info@asperbio.com
www.asperbio.com

DISEASES TESTED BY ATAXIA **NGS PANEL**

Gene	Phenotype/ Condition	Inheritance	Phenotype MIM number
ABCB7	Anemia, sideroblastic, with ataxia	XLR	301310
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract	AR	612674
ACO2	Infantile cerebellar-retinal degeneration	AR	614559
	Optic atrophy 9	AR	616289
AFG3L2	Spinocerebellar ataxia 28	AD	610246
	Spastic ataxia 5	AR	614487
	Optic atrophy 12	-	618977
AHI1	Joubert syndrome 3	AR	608629
ANO10	Spinocerebellar ataxia, autosomal recessive 10	AR	613728
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia (ataxia- oculomotor apraxia 1)	AR	208920
ATCAY	Ataxia, cerebellar, Cayman type	AR	601238
ATG5	Spinocerebellar ataxia, autosomal recessive 25	AR	617584
ATM	Ataxia-telangiectasia	AR	208900
ATP1A2	Migraine, familial hemiplegic, 2	AD	602481
	Migraine, familial basilar	AD	602481
ATP1A3	Alternating hemiplegia of childhood 2	AD	614820
	CAPOS syndrome	AD	601338
	Dystonia-12	AD	128235
ATP2B3	Spinocerebellar ataxia, 1	XLR	302500
ATP8A2	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4	AR	615268
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	AR	613227
CACNA1A	Epileptic encephalopathy, early infantile, 42	AD	617106
	Episodic ataxia, type 2	AD	108500
	Migraine, familial hemiplegic, 1	AD	141500
	Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia	AD	141500
CACNA1G	Spinocerebellar ataxia 42	AD	616795
	Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits	AD	618087
CACNB4	Episodic ataxia, type 5	AD	613855
	Epilepsy, idiopathic generalized, susceptibility to, 9	AD	607682
	Epilepsy, juvenile myoclonic, susceptibility to, 6	AD	607682
CAMTA1	Spastic ataxia with variable other neurological features	AD	
CAPN1	Spastic paraplegia 76	AR	616907
CASK	FG syndrome 4	-	300422
	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	XLD	300749
	Mental retardation, with or without nystagmus	-	300422

Asper Biogene LLC
Vaksali 17A, 50410 Tartu, Estonia
Phone +372 7307 295
info@asperbio.com
www.asperbio.com

CC2D2A	COACH syndrome	AR	216360
	Joubert syndrome 9	AR	612285
	Meckel syndrome 6	AR	612284
CCDC88C	Hydrocephalus, congenital, 1	AR	236600
	Spinocerebellar ataxia 40	AD	616053
CEP290	Joubert syndrome 5	AR	610188
	Leber congenital amaurosis 10	-	611755
	Meckel syndrome 4	AR	611134
	Senior-Loken syndrome 6	AR	610189
	Bardet-Biedl syndrome 14	AR	615991
CHP1	Spastic ataxia 9	AR	618438
CHRNA4	Epilepsy, nocturnal frontal lobe, 1	AD	600513
CLCN2	Leukoencephalopathy with ataxia	AR	615651
	Epilepsy, juvenile absence, susceptibility to, 2	AD	607628
	Hyperaldosteronism, familial, type II	AD	605635
	Epilepsy, juvenile myoclonic, susceptibility to, 8	AD	607628
	Epilepsy, idiopathic generalized, susceptibility to, 11	AD	607628
CLN5	Ceroid lipofuscinosis, neuronal, 5	AR	256731
COA7	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3	AR	618387
C9orf72	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1	AD	105550
COQ2	Coenzyme Q10 deficiency, primary, 1	AR	607426
COQ4	Coenzyme Q10 deficiency, primary, 7	AR	616276
COQ6	Coenzyme Q10 deficiency, primary, 6	AR	614650
COQ7	Coenzyme Q10 deficiency, primary, 8	AR	616733
COQ8A	Coenzyme Q10 deficiency, primary, 4 (spinocerebellar ataxia, autosomal recessive 9)	AR	612016
COQ9[1]	Coenzyme Q10 deficiency, primary, 5	AR	614654
CPLANE1	Orofaciodigital syndrome VI	AR	277170
	Joubert syndrome 17	AR	614615
CWF19L1	Spinocerebellar ataxia, autosomal recessive 17	AR	616127
CYP27A1	Cerebrotendinous xanthomatosis	AR	213700
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	AR	611105
DEPDC5	Epilepsy, familial focal, with variable foci 1	AD	604364
DNMT1	Cerebellar ataxia, deafness, and narcolepsy,	AD	604121
	Neuropathy, hereditary sensory, type IE	AD	614116
EEF2	Spinocerebellar ataxia 26	AD	609306
ELOVL4	Spinocerebellar ataxia 34	AD	133190
	Ichthyosis, spastic quadriplegia, and mental retardation	AR	614457

Asper Biogene LLC
Vaksali 17A, 50410 Tartu, Estonia
Phone +372 7307 295
info@asperbio.com
www.asperbio.com

ELOVL4	Stargardt disease 3	AD	600110
ELOVL5	Spinocerebellar ataxia 38	AD	615957
FAT2	Spinocerebellar ataxia 45	AD	617769
FGF12	Epileptic encephalopathy, early infantile, 47	AD	617166
FGF14	Spinocerebellar ataxia 27	AD	609307
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa	AR	609033
FXN	Friedreich ataxia	AR	229300
	Friedreich ataxia with retained reflexes	AR	229300
GDAP2	Spinocerebellar ataxia, autosomal recessive 27	AR	618369
GSS	Glutathione synthetase deficiency	AR	266130
	Hemolytic anemia due to glutathione synthetase deficiency	AR	231900
GOSR2	Epilepsy, progressive myoclonic 6	AR	614018
GRID2	Spinocerebellar ataxia, autosomal recessive 18	AR	616204
GRM1	Spinocerebellar ataxia, autosomal recessive 13	AR	614831
	Spinocerebellar ataxia 44	AD	617691
GSX2	Diencephalic-mesencephalic junction dysplasia syndrome 2	AR	618646
IFRD1	Spinocerebellar ataxia 18	AD	607458
ITPR1	Spinocerebellar ataxia 29, congenital nonprogressive	AD	117360
	Gillespie syndrome	AD, AR	206700
	Spinocerebellar ataxia 15	AD	606658
KCNA1	Episodic ataxia 1/myokymia syndrome	AD	160120
KCNC3	Spinocerebellar ataxia 13	AD	605259
KCND3	Spinocerebellar ataxia 19	AD	607346
	Brugada syndrome 9	AD	616399
KCNJ10	Enlarged vestibular aqueduct, digenic	AR	612780
	SESAME syndrome	AR	600791
KCNMA1	Epilepsy, idiopathic generalized, susceptibility to, 16	AD	618596
	Cerebellar atrophy, developmental delay, and seizures	AR	617643
	Liang-Wang syndrome	AD	618729
	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy	AD	609446
KCNQ2	Myokymia; Seizures, benign neonatal, 1	AD	121200
	Epileptic encephalopathy, early infantile, 7	AD	613720
KIF1C	Spastic ataxia 2	AR	611302
LAMA1	Poretti-Boltshauser syndrome	AR	615960
MAN2B1	Mannosidosis, alpha-, types I and II	AR	248500
MARS2	Spastic ataxia 3	AR	611390
	Combined oxidative phosphorylation deficiency 25	AR	616430
MME	Charcot-Marie-Tooth disease, axonal, type 2T	AD, AR	617017
	Spinocerebellar ataxia 43	AD	617018
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities	AR	617282

Asper Biogene LLC
Vaksali 17A, 50410 Tartu, Estonia
Phone +372 7307 295
info@asperbio.com
www.asperbio.com

MTCL1	Early-onset ataxia	AR	-
MTPAP	Spastic ataxia 4	AR	613672
NKX6-2	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy	AR	617560
NOP56	Spinocerebellar ataxia 36	AD	614153
NPC1	Niemann-Pick disease, type C1	AR	257220
	Niemann-Pick disease, type D	AR	257220
NPC2	Niemann-pick disease, type C2	AR	607625
OFD1	Orofaciodigital syndrome I	XLD	311200
	Simpson-Golabi-Behmel syndrome, type 2	XLR	300209
	Retinitis pigmentosa 23	XLR	300424
	Joubert syndrome 10	XLR	300804
OPHN1	Mental retardation, with cerebellar hypoplasia and distinctive facial appearance	XLR	300486
PAX6	Coloboma of optic nerve	AD	120430
	Coloboma, ocular	AD	120200
	Morning glory disc anomaly	AD	120430
	Aniridia	AD	106210
	Anterior segment dysgenesis 5, multiple subtypes	AD	604229
	Cataract with late-onset corneal dystrophy	AD	106210
	Foveal hypoplasia 1	AD	136520
	Keratitis	AD	148190
	Optic nerve hypoplasia	AD	165550
PCDH12	Diencephalic-mesencephalic junction dysplasia syndrome 1	AR	251280
PDSS1	Coenzyme Q10 deficiency, primary, 2	AR	614651
PDSS2	Coenzyme Q10 deficiency, primary, 3	AR	614652
PDYN	Spinocerebellar ataxia 23	AD	610245
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR	215100
	Peroxisome biogenesis disorder 9B	AR	614879
PHYH	Refsum disease	AR	266500
PIK3R5	Ataxia-oculomotor apraxia 3	AR	615217
PLD3	Spinocerebellar ataxia 46	AD	617770
PMPCA	Spinocerebellar ataxia, autosomal recessive 2	AR	213200
PNKD	Paroxysmal nonkinesigenic dyskinesia 1	AD	118800
PNKP	Microcephaly, seizures, and developmental delay	AR	613402
	Charcot-Marie-Tooth disease, type 2B2	AR	605589
	Ataxia-oculomotor apraxia 4	AR	616267
PNPLA6	Spastic paraplegia 39	AR	612020
	Laurence-Moon syndrome	AR	245800
	Oliver-McFarlane syndrome	AR	275400
	Boucher-Neuhauser syndrome	AR	215470
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type)	AR	203700
	Mitochondrial DNA depletion syndrome 4B (MNGIE type)	AR	613662

Asper Biogene LLC
Vaksali 17A, 50410 Tartu, Estonia
Phone +372 7307 295
info@asperbio.com
www.asperbio.com

POLG	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)	AR	607459
	Progressive external ophthalmoplegia, autosomal dominant 1	AD	157640
	Progressive external ophthalmoplegia, autosomal recessive 1	AR	258450
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogo nadism	AR	607694
	Wiedemann-Rautenstrauch syndrome	AR	264090
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	AR	614381
PRKCG	Spinocerebellar ataxia 14	AD	605361
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis	AD	602066
	Episodic kinesigenic dyskinesia 1	AD	128200
	Seizures, benign familial infantile, 2	AD	605751
PUM1	Spinocerebellar ataxia 47	AD	617931
RUBCN	Spinocerebellar ataxia, autosomal recessive 15	AR	615705
PTF1A	Pancreatic and cerebellar agenesis	AR	609069
	Pancreatic agenesis 2	AR	615935
RELN	Lissencephaly 2 (Norman-Roberts type)	AR	257320
	Epilepsy, familial temporal lobe, 7	AD	616436
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism (Gordon Holmes syndrome)	AR	212840
SACS	Spastic ataxia, Charlevoix-Saguenay type	AR	270550
SAMD9L	Ataxia-pancytopenia syndrome	AD	159550
SCN1A	Migraine, familial hemiplegic, 3	AD	609634
SCN2A	Seizures, benign familial infantile, 3	AD	607745
	Epileptic encephalopathy, early infantile, 11	AD	613721
	Episodic ataxia, type 9	AD	618924
SCN8A	Myoclonus, familial, 2	AD	618364
	Cognitive impairment with or without cerebellar ataxia	AD	614306
	Epileptic encephalopathy, early infantile, 13	AD	614558
	Seizures, benign familial infantile, 5	AD	617080
SCYL1	Spinocerebellar ataxia, autosomal recessive 21	AR	616719
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	AR	614739
SETX	Spinocerebellar ataxia with axonal neuropathy 2 (ataxia-oculomotor apraxia 2)	AR	606002
	Amyotrophic lateral sclerosis 4, juvenile	AD	602433
SIL1	Marinesco-Sjogren syndrome	AR	248800
SLC1A3	Episodic ataxia, type 6	AD	612656
SLC2A1	GLUT1 deficiency syndrome 1, infantile onset, severe	AD, AR	606777
	GLUT1 deficiency syndrome 2, childhood onset	AD	612126
	Stomatin-deficient cryohydrocytosis with neurologic defects	AD	608885

Asper Biogene LLC
Vaksali 17A, 50410 Tartu, Estonia
Phone +372 7307 295
info@asperbio.com
www.asperbio.com

SLC2A1	Epilepsy, idiopathic generalized, susceptibility to, 12	AD	614847
	Dystonia 9	AD	601042
SLC9A1	Lichtenstein-Knorr syndrome	AR	616291
SLC9A6	Mental retardation, syndromic, Christianson type	XLD	300243
SLC16A2	Allan-Herndon-Dudley syndrome	XL	300523
SLC25A46	Neuropathy, hereditary motor and sensory, type VIB	AR	616505
SLC52A2	Brown-Vialetto-Van Laere syndrome 2	AR	614707
SNX14	Spinocerebellar ataxia, autosomal recessive 20	AR	616354
SPG7	Spastic paraplegia 7	AR	607259
SPTBN2	Spinocerebellar ataxia, autosomal recessive 14	AR	615386
	Spinocerebellar ataxia 5	AD	600224
SQSTM1	Myopathy, distal, with rimmed vacuoles	AD	617158
	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	AR	617145
	Paget disease of bone 3	AD	167250
	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3	AD	616437
STUB1	Spinocerebellar ataxia 48	AD	618093
	Spinocerebellar ataxia, autosomal recessive 16	AR	615768
SYNE1	Emery-Dreifuss muscular dystrophy 4, autosomal dominant	AD	612998
	Arthrogryposis multiplex congenita 3, myogenic type	AR	618484
	Spinocerebellar ataxia, autosomal recessive 8	AR	610743
SYT14	Spinocerebellar ataxia, autosomal recessive 11	AR	614229
TDP1	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1	AR	607250
TDP2	Spinocerebellar ataxia, autosomal recessive 23	AR	616949
TGM6	Spinocerebellar ataxia 35	AD	613908
THG1L	Spinocerebellar ataxia, autosomal recessive 28	AR	618800
TMEM231	Joubert syndrome 20	AR	614970
	Meckel syndrome 11	AR	615397
TMEM240	Spinocerebellar ataxia 21	AD	607454
TTBK2	Spinocerebellar ataxia 11	AD	604432
TPP1	Ceroid lipofuscinosis, neuronal, 2	AR	204500
	Spinocerebellar ataxia, autosomal recessive 7	AR	609270
TRPC3	Spinocerebellar ataxia 41	AD	616410
TSFM	Combined oxidative phosphorylation deficiency 3	AR	610505
TTC19	Mitochondrial complex III deficiency, nuclear type 2	AR	615157
TTPA	Ataxia with isolated vitamin E deficiency	AR	277460
TUBB4A	Dystonia 4, torsion	AD	128101
	Leukodystrophy, hypomyelinating, 6	AD	612438
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	AR	271245

Asper Biogene LLC
Vaksali 17A, 50410 Tartu, Estonia
Phone +372 7307 295
info@asperbio.com
www.asperbio.com

SLC2A1	Progressive external ophthalmoplegia with mitochondrial DNA deletions, 3	AD	609286
	Perrault syndrome 5	AR	616138
TXN2	Combined oxidative phosphorylation deficiency 29	AR	616811
UBA5	Epileptic encephalopathy, early infantile, 44	AR	617132
	Spinocerebellar ataxia, autosomal recessive 24	AR	617133
UBR4	Episodic ataxia, type 8	AD	616055
VAMP1	Myasthenic syndrome, congenital, 25	AR	618323
	Spastic ataxia 1	AD	108600
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	AR	224050
VPS13D	Spinocerebellar ataxia, autosomal recessive 4	AR	607317
VWA3B	Spinocerebellar ataxia, autosomal recessive 22	AR	616948
WDR73	Galloway-Mowat syndrome 1 (spinocerebellar ataxia, autosomal recessive 5)	AR	251300
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2	AR	610185
WFS1	Wolfram-like syndrome	AD	614296
	Deafness, 6/14/38	AD	600965
	Cataract 41	AD	116400
	Diabetes mellitus, noninsulin-dependent, association with	AD	125853
	Wolfram syndrome 1	AR	222300
WWOX	Epileptic encephalopathy, early infantile, 28	AR	616211
	Esophageal squamous cell carcinoma, somatic	-	133239
	Spinocerebellar ataxia, autosomal recessive 12	AR	614322
XRCC1	Spinocerebellar ataxia, autosomal recessive 26	AR	617633

DISEASES TESTED BY **REPEAT EXPANSION PANEL**

Gene	Phenotype/ Condition	Inheritance	Phenotype MIM number
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies	AD	618494
	Dentatorubral-pallidoluysian atrophy	AD	125370
ATXN1	Spinocerebellar ataxia 1	AD	164400
ATXN2	Spinocerebellar ataxia 2	AD	183090
ATXN3	Machado-Joseph disease (spinocerebellar ataxia 3)	AD	109150
ATXN7	Spinocerebellar ataxia 7	AD	164500
ATXN8	Spinocerebellar ataxia 8	AD	608768
ATXN10	Spinocerebellar ataxia 10	AD	603516
BEAN1	Spinocerebellar ataxia 31	AD	117210
CACNA1A	Spinocerebellar ataxia 6	AD	183086
FXN	Friedreich ataxia; Friedreich ataxia with retained reflexes	AR	229300
NOP56	Spinocerebellar ataxia 36	AD	614153
PPP2R2B	Spinocerebellar ataxia 12	AD	604326
TBP	Spinocerebellar ataxia 17	AD	607136

Asper Biogene LLC
Vaksali 17A, 50410 Tartu, Estonia
Phone +372 7307 295
info@asperbio.com
www.asperbio.com

DISEASES TESTED BY **REPEAT EXPANSION ANALYSIS**

Gene	Phenotype/ Condition	Inheritance	Phenotype MIM number
FMR1	Fragile X tremor/ataxia syndrome	XLD	300623
	Fragile X syndrome	XLD	300624
	Premature ovarian failure 1	XL	311360

DISEASES TESTED BY **MITOCHONDRIAL GENOME SEQUENCING**

Gene	Phenotype/ Condition	Inheritance	Phenotype MIM number
MT-TK *MTT-L1* *MT-TH* *MT-TS1* *MT-TS2* *MT-TF* *MT-ND5*	Myoclonic Epilepsy associated with Ragged-Red Fibers (MERRF)	mitochondrial	545000
MT-ATP6	Neuropahty, Ataxia, and Retinitis Pigmetosa (NARP)	mitochondrial	551500
mtDNA deletions	Kearns-Sayre syndrome	mitochondrial	530000

DISEASES TESTED BY **CHROMOSOMAL MICROARRAY ANALYSIS**

Gene	Phenotype/ Condition	Inheritance	Phenotype MIM number
CAMTA1	Cerebellar ataxia, and intellectual disability (CANPMR)	AD	614756