Spinocerebellar Ataxia

Precision Panel

Overview

Spinocerebellar Ataxia (SCA) refers to a heterogeneous group of progressive neurodegenerative diseases of genetic origin. Currently, more than 30 types have been described, most of which are autosomal dominant. Classification is done according to the clinical manifestations or genetic nosology. The most common type is SCA3. Ataxia is defined as the absence of voluntary muscle coordination and loss of control of movement affecting gait stability, eye movement and speech. The clinical hallmark of SCA is loss of balance and coordination accompanied by slurred speech; onset is most often in adult life. The main disease mechanism of these SCAs include toxic RNA gain-of-function, mitochondrial dysfunction, channelopathies, autophagy and transcription dysregulation. The prognosis is dependent upon the individual and genetic properties, but most patients develop severe, irreversible disability, while retaining full mental capacity.

The Igenomix Spinocerebellar Ataxia Precision Panel can be used as a tool for an accurate diagnosis and differential diagnosis of loss of balance and coordination ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved, and their high or intermediate penetrance.

Indications

The Igenomix Spinocerebellar Ataxia Precision Panel is used for patients with a clinical suspicion or diagnosis presenting with or without the following symptoms:

- Broad based gait ataxia
- Tremor
- Limb ataxia
- Nystagmus
- Difficulty speaking
- Decreased coordination
- Dementia

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.

- Early initiation of treatment involving a multidisciplinary team in the form of orthopaedic care, use of special devices to assist with fine movements as well as speech therapy.
- Risk assessment of asymptomatic family members according to the mode of inheritance via genetic counselling.
- Improvement of delineation of genotype-phenotype correlation given the variability of severity and course of disease.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCB7	Anemia, Spinocerebellar Ataxia	X,XR,G	100	-
ABHD12	Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa, Cataract	AR	95.77	21 of 21
ABHD5	Chanarin-Dorfman Syndrome, Neutral Lipid Storage Disease, Ichthyosis	AR	99.98	37 of 37
ACADVL	Acyl-Coa Dehydrogenase Deficiency	AR	100	329 of 329
ACO2	Cerebellar-Retinal Degeneration, Optic Atrophy	AR	100	33 of 33
AFG3L2	Optic Atrophy, Spastic And Spinocerebellar Ataxia, Myoclonic Epilepsy, Neuropathy	AD,AR	99.74	42 of 42
AHI1	Joubert Syndrome, Ocular Defect, Retinitis Pigmentosa	AR	96.79	85 of 97
ALDH5A1	Succinic Semialdehyde Dehydrogenase Deficiency	AR	95.41	65 of 69
AMACR	Alpha-Methylacyl-Coa Racemase Deficiency, Bile Acid Synthesis Defect	AR	100	8 of 8
ANO10	Spinocerebellar And Cerebellar Ataxia	AR	100	28 of 28
AP1S2	Mental Retardation, Fried Syndrome, Dandy-Walker Malformation, Basal Ganglia Disease, Seizures, Intellectual Disability, Hypotonia	X,XR,G	84.15	-
APTX	Ataxia, Oculomotor Apraxia, Hypoalbuminemia	AR	100	44 of 44
ARL13B	Joubert Syndrome	AR	99.77	10 of 10
ARL6	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AD,AR,X,XR,G	100	17 of 21
ARSA	Metachromatic Leukodystrophy	AR	98	266 of 266
ATCAY	Cerebellar Ataxia	AR	99.98	3 of 3
ATG5	Spinocerebellar Ataxia	AR	99.83	1 of 3
ATM	Ataxia-Telangiectasia, Breast Cancer, Mantle Cell Lymphoma	AD,AR	99.93	1608 of 1632
ATN1	Congenital Hypotonia, Epilepsy, Developmental Delay, Digital Anomalies, Seizures, Dementia, Dentatorubral Pallidoluysian Atrophy	AD	99.86	11 of 11
ATP13A2	Kufor-Rakeb Syndrome, Spastic Paraplegia, Neuronal Ceroid Lipofuscinosis	AR	99.97	53 of 53
ATP1A3	Alternating Hemiplegia Of Childhood, Cerebellar Ataxia, Areflexia, Pes Cavus, Optic Atrophy, Sensorineuralhearing Loss, Dystonia, Parkinson Disease	AD	99.94	138 of 138
ATP2B3	Spinocerebellar And Cerebelar Ataxia	X,XR,G	100	-
ATP8A2	Cerebellar Ataxia, Mental Retardation, Dysequilibrium Syndrome	AR	99.99	20 of 20
ATXN1	Spinocerebellar Ataxia	AD	99.93	2 of 2
ATXN10	Spinocerebellar Ataxia	AD	99.77	-
ATXN2	Parkinson Disease, Spinocerebellar Ataxia, Amyotrophic Lateral Sclerosis	AD	91.78	9 of 10
ATXN3	Machado-Joseph Disease	AD	99.94	-
ATXN7	Spinocerebellar Ataxia, Retinal Degeneration, Macular Degeneration, Ophthalmoplegia	AD	94.99	-
ATXN8	Spinocerebellar Ataxia	AD	-	-

ATXN8OS	Parkinson Disease, Spinocerebellar Ataxia	AD	-	-
B9D1	Joubert Syndrome, Meckel Syndrome	AR	90.23	11 of 11
BBS1	Bardet-Biedl Syndrome	AR	100	102 of 105
BBS12	Bardet-Biedl Syndrome	AR	99.78	61 of 61
BEAN1	Spinocerebellar Ataxia	AD	97.28	1 of 1
BSCL2	Encephalopathy, Neuronopathy, Spastic Paraplegia, Berardinelli-Seip Lipodystrophy, Motor Neuropathy, Neurodegenerative Syndrome	AD,AR	99.83	60 of 61
BTD	Biotinidase Deficiency	AR	100	261 of 262
C12ORF65	Oxidative Phosphorylation Deficiency, Spastic Paraplegia	AR	-	-
C19ORF12	Neurodegeneration, Brain Iron Accumulation, Spastic Paraplegia	AD,AR	-	-
CA8	Cerebellar Ataxia, Mental Retardation, Dysequilibrium Syndrome	AR	100	4 of 4
CACNA1A	Epileptic Encephalopathy, Migraine, Paroxysmal Torticollis, Spinocerebellar And Paroxisomal Ataxia	AD	96.13	249 of 266
CACNA1G	Spinocerebellar Ataxia	AD	99.52	16 of 16
CACNB4	Epilepsy, Ataxia	AD	99.87	5 of 5
CAMTA1	Cerebellar Ataxia, Mental Retardation	AD	99.8	6 of 6
CASK	Anemia, Fg Syndrome, Mental Retardation, Microcephaly, Cerebellar Hypoplasia, Epileptic Encephalopathy	X,XR,XD,G	99.98	-
CC2D2A	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.43	98 of 100
CCDC88C	Hydrocephalus, Spinocerebellar Ataxia	AD,AR	99.44	13 of 14
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Amaurosis, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47	293 of 327
CEP41	Joubert Syndrome	AR	100	17 of 17
CHMP1A	Pontocerebellar Hypoplasia	AR	100	4 of 4
CLCN2	Epilepsy, Seizures, Hyperaldosteronism, Leukoencephalopathy, Ataxia	AD,AR	100	39 of 39
CLN5	Ceroid Lipofuscinosis	AR	99.56	52 of 55
CLN6	Ceroid Lipofuscinosis	AR	99.94	98 of 99
CLPP	Perrault Syndrome	AR	99.91	11 of 11
COA7	Spinocerebellar Ataxia, Axonal Neuropathy	AR	99.99	6 of 6
COASY	Neurodegeneration, Brain Iron Accumulation, Pontocerebellar Hypoplasia	AR	99.5	5 of 5
COQ2	Coenzyme Q10 Deficiency, Multiple System Atrophy, Leigh Syndrome, Nephrotic Syndrome	AD,AR	99.61	37 of 38
COQ8A	Coenzyme Q10 Deficiency, Ataxia	AR	100	-
COQ9	Coenzyme Q10 Deficiency	AR	99.87	6 of 6
COX20	Mitochondrial Complex Iv Deficiency	AR,MI	99.88	5 of 5
CP	Aceruloplasminemia	AR	99.91	58 of 59
CPLANE1	Joubert Syndrome, Varadi-Papp Syndrome, Joubert Syndrome, Monomelic Amyotrophy, Orofaciodigital Syndrome	AR	-	-
CSPP1	Joubert Syndrome, Jeune Asphyxiating Thoracic Dystrophy, Meckel Syndrome	AR	98.32	29 of 30
CWF19L1	Spinocerebellar Ataxia	AR	100	6 of 6
CYP27A1	Cerebrotendinous Xanthomatosis	AR	100	118 of 118
DAB1	Spinocerebellar Ataxia	AD	99.98	-
DARS2	Leukoencephalopathy, Brainstem, Spinal Cord Involvement	AR	100	65 of 65
DLAT	Pyruvate Dehydrogenase E2 Deficiency	AR	99.99	5 of 5
DNAJC19	3-Methylglutaconic Aciduria, Dilated Cardiomyopathy, Ataxia	AR	100	6 of 6
DNAJC5	Ceroid Lipofuscinosis	AD	100	2 of 2
DNMT1	Cerebellar Ataxia, Deafness, Narcolepsy, Neuropathy	AD	97.87	30 of 30

EEF2	Spinocerebellar Ataxia	AD	94.75	4 of 4
EIF2B1	Leukoencephalopathy, Vanishing White Matter	AR	100	9 of 9
EIF2B2	Leukoencephalopathy, Vanishing White Matter	AR	100	30 of 30
EIF2B3	Leukoencephalopathy, Vanishing White Matter	AR	97.55	26 of 26
EIF2B4	Leukoencephalopathy, Vanishing White Matter	AR	100	31 of 31
EIF2B5	Leukoencephalopathy, Vanishing White Matter	AR	100	99 of 99
ELOVL4	Erythrokeratodermia, Ichthyosis, Spastic Quadriplegia, Mental Retardation, Stargardt Disease, Spinocerebellar Ataxia	AD,AR	100	16 of 17
ELOVL5	Spinocerebellar Ataxia	AD	99.47	6 of 6
EXOSC3	Pontocerebellar Hypoplasia	AR	100	19 of 20
FA2H	Spastic Paraplegia, Fatty Acid Hydroxylase-Associated Neurodegeneration	AR	88.77	60 of 62
FAT2	Spinocerebellar Ataxia	AD	100	6 of 6
FBXL4	Mitochondrial Dna Depletion Syndrome	AR	99.26	46 of 51
FGF14	Spinocerebellar Ataxia	AD	99.91	7 of 8
FLVCR1	Posterior Column Ataxia, Retinitis Pigmentosa	AR	99.96	26 of 26
FTL	Basal Ganglia Disease, L-Ferritin Deficiency, Hyperferritinemia, Neuroferritinopathy	AD,AR	100	21 of 63
FXN	Friedreich Ataxia	AR	99.93	52 of 52
GALC	Krabbe Disease	AR	99.38	252 of 254
GBA	Dementia, Gaucher Disease, Parkinson Disease, Ophthalmoplegia, Cardiovascular Calcification	AD,AR	100	469 of 471
GBA2	Spastic Paraplegia, Cerebellar Ataxia	AR	99.96	25 of 25
GDAP2	Spinocerebellar Ataxia	AR	99.96	3 of 3
GFAP	Alexander Disease	AD	99.98	143 of 143
GJB1	Charcot-Marie-Tooth Disease, Cerebellar Ataxia	X,XR,XD,G	100	-
GJC2	Leukodystrophy, Lymphedema, Spastic Paraplegia, Milroy Disease	AD,AR	95.37	52 of 63
GOSR2	Epilepsy	AR	88.39	6 of 6
GRID2	Spinocerebellar Ataxia	AR	99.95	16 of 21
GRM1	Spinocerebellar Ataxia	AD,AR	99.88	20 of 21
GSS	Glutathione Synthetase Deficiency, Hemolytic Anemia	AR	100	36 of 38
HEPACAM	Megalencephalic Leukoencephalopathy, Subcortical Cysts, Mental Retardation	AD,AR	97.87	30 of 30
HEXB	Sandhoff Disease	AR	99.92	109 of 115
HIBCH	3-Hydroxyisobutyryl-Coa Hydrolase Deficiency, Neurodegeneration	AR	96.47	27 of 27
IFRD1	Spinocerebellar Ataxia		100	2 of 2
INPP5E	Joubert Syndrome, Mental Retardation, Truncal Obesity, Retinal Dystrophy	AR	99.89	56 of 56
ITM2B	Dementia, Retinal Dystrophy	AD	92	8 of 8
ITPR1	Aniridia, Cerebellar Ataxia, Mental Deficiency, Spinocerebellar Ataxia	AD,AR	99.87	93 of 97
KCNA1	Ataxia, Epileptic Encephalopathy, Continuous Muscle Fiber Activity, Paroxysmal Kinesigenic Dyskinesia	AD	100	49 of 49
KCNC3	Spinocerebellar Ataxia	AD	79.25	12 of 13
KCND3	Brugada Syndrome, Spinocerebellar Ataxia	AD	100	32 of 32
KCNJ10	Enlarged Vestibular Aqueduct, Pendred Syndrome, Seizures, Sensorineural Deafness, Ataxia, Mental Retardation, East Syndrome	AR	93.53	27 of 32
KIF1A	Mental Retardation, Neuropathy, Spastic Paraplegia, Hereditary Sensory And Autonomic Neuropathy, Peho Syndrome	AD,AR	100	76 of 76
KIF1C	Ataxia, Spastic Paraplegia	AR	99.74	18 of 19
KIF5A	Amyotrophic Lateral Sclerosis, Myoclonus, Spastic Paraplegia	AD	100	85 of 85

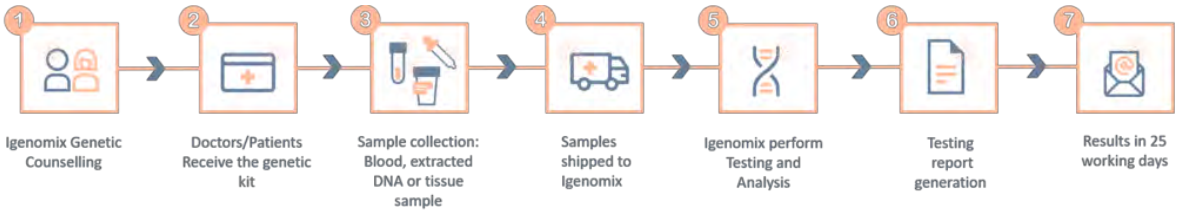
KIF7	Acrocallosal Syndrome, Hydrolethalus Syndrome, Macrocephaly, Multiple Epiphyseal Dysplasia, Orofaciodigital Syndrome	AR	94.91	47 of 50
LAMA1	Poretti-Boltshauser Syndrome, Ataxia, Intellectual Disability, Oculomotor Apraxia	AR	100	43 of 43
LMNB1	Leukodystrophy	AD	99.66	4 of 4
LRPPRC	Leigh Syndrome	AR	98.94	18 of 18
MARS2	Ataxia, Oxidative Phosphorylation Deficiency, Leukoencephalopathy	AR	99.94	3 of 3
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
MLC1	Megalencephalic Leukoencephalopathy	AR	100	104 of 106
MME	Charcot-Marie-Tooth Disease, Spinocerebellar Ataxia, Congenital Membranous Nephropathy, Fetomaternal Anti-Neutral Endopeptidase Alloimmunization	AD,AR	100	33 of 33
MRE11	Ataxia-Telangiectasia-Like Disorder, Breast And Ovarian Cancer Syndrome	AR	99.95	-
MTFMT	Oxidative Phosphorylation Deficiency, Mitochondrial Complex I Deficiency, Leigh Syndrome, Leukodystrophy	AR	99.52	18 of 18
MTPAP	Spastic Ataxia, Optic Atrophy, Dysarthria	AR	99.99	2 of 2
MTTP	Abdominal Obesity-Metabolic Syndrome, Abetalipoproteinemia	AD,AR	100	69 of 71
NDUFAF6	Fanconi Renotubular Syndrome, Mitochondrial Complex I Deficiency, Leigh Syndrome, Leukodystrophy	AR	99.4	12 of 13
NDUFS1	Mitochondrial Complex I Deficiency, Leigh Syndrome, Leukodystrophy	AR	99.98	30 of 30
NDUFS2	Mitochondrial Complex I Deficiency, Leber Optic Neuropathy, Leigh Syndrome, Cardiomyopathy, Leukodystrophy	AR	100	26 of 26
NDUFS4	Mitochondrial Complex I Deficiency, Leigh Syndrome, Leukodystrophy	AR,X,XD,MI,G	100	15 of 15
NDUFS7	Mitochondrial Complex I Deficiency, Leigh Syndrome, Leukodystrophy	AR	88	6 of 7
NDUFV1	Mitochondrial Complex I Deficiency, Leigh Syndrome, Leukodystrophy	AR	100	36 of 36
NOP56	Spinocerebellar Ataxia	AD	99.41	-
NPC1	Niemann-Pick Disease	AR	97	503 of 505
NPC2	Niemann-Pick Disease	AR	100	27 of 27
NPHP1	Joubert Syndrome, Nephronophthisis, Senior-Loken Syndrome, Bardet-Biedl Syndrome	AR	100	58 of 59
NUBPL	Mitochondrial Complex I Deficiency	AR	95.2	13 of 13
OFD1	Joubert Syndrome, Orofaciodigital Syndrome, Retinitis Pigmentosa, Simpson-Golabi-Behmel Syndrome, Ciliary Dyskinesia	X,XR,XD,G	98.09	-
OPA1	Behr Syndrome, Mitochondrial Dna Depletion Syndrome, Optic Atrophy, Ophthalmoplegia, Myopathy, Ataxia, Neuropathy	AD,AR	99.98	397 of 402
OPA3	3-Methylglutaconic Aciduria, Optic Atrophy	AD,AR	100	18 of 18
OPHN1	Mental Retardation, Cerebellar Hypoplasia	X,XR,G	100	-
PANK2	Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa, Pallidal Degeneration, Neurodegeneration, Brain Iron Accumulation, Pantothenate Kinase-Associated Neurodegeneration	AR	98.92	177 of 182
PAX6	Aniridia, Anterior Segment Dysgenesis, Coloboma Of Optic Nerve, Hypoplasia, Keratitis, Optic Nerve Hypoplasia, Wilms Tumor, Chromosome 11p13 Deletion Syndrome, Morning Glory Disc Anomaly, Peters Anomaly, Wagr Syndrome	AD	100	460 of 485
PDHX	Pyruvate Dehydrogenase E3-Binding Protein Deficiency	AR	97.21	18 of 20
PDSS1	Coenzyme Q10 Deficiency	AR	97.34	5 of 5
PDSS2	Coenzyme Q10 Deficiency, Leigh Syndrome, Nephrotic Syndrome	AR	99.99	6 of 6
PDYN	Spinocerebellar Ataxia	AD	100	13 of 13
PEX10	Ataxia, Refsum Disease, Adrenoleukodystrophy, Zellweger Syndrome	AR	99.76	29 of 32
PEX2	Refsum Disease, Adrenoleukodystrophy, Zellweger Syndrome	AR	99.89	17 of 17
PEX6	Heimler Syndrome, Spinocerebellar Ataxia, Blindness, Deafness, Refsum Disease, Adrenoleukodystrophy, Zellweger Syndrome	AD,AR	99.94	105 of 108

PEX7	Refsum Disease, Rhizomelic Chondrodysplasia Punctata, Refsum Disease, Zellweger Syndrome	AR	99.21	47 of 53
PHYH	Refsum Disease	AR	100	34 of 34
PIK3R5	Ataxia, Oculomotor Apraxia, Spinocerebellar Ataxia	AR	100	2 of 2
PLA2G6	Neuroaxonal Dystrophy, Neurodegeneration, Brain Iron Accumulation, Parkinson Disease	AR	99.94	190 of 191
PLD3	Spinocerebellar Ataxia	AD	99.83	22 of 22
PLEKHG4	Spinocerebellar Ataxia		99.97	2 of 2
PLP1	Pelizaeus-Merzbacher Disease, Spastic Paraplegia	X,XR,G	100	-
PMPCA	Spinocerebellar Ataxia, Cerebelloparenchymal Disorder	AR	99.91	9 of 9
PNKD	Paroxysmal Non-Kinesigenic Dyskinesia	AD	99.98	6 of 6
PNKP	Ataxia, Oculomotor Apraxia, Charcot-Marie-Tooth Disease, Epileptic Encephalopathy	AR	100	36 of 36
PNPLA6	Boucher-Neuhauser Syndrome, Laurence-Moon Syndrome, Oliver-Mcfarlane Syndrome, Spastic Paraplegia, Hypogonadism, Choroidal Dystrophy, Cerebellar Ataxia	AR	100	65 of 65
POLG	Mitochondrial Dna Depletion Syndrome, Ophthalmoplegia, Sensory Ataxic Neuropathy, Dysarthria, Ophthalmoparesis, Alpers-Huttenlocher Syndrome, Neurogastrointestinal Encephalomyopathy, Mitochondrial Ataxia	AD,AR	99.92	325 of 326
POLR3A	Leukodystrophy, Progeroid Syndrome, Hypomyelination, Hypogonadotropic Hypogonadism, Hypodontia, Tremor, Ataxia, Central Hypomyelination, Wiedemann-Rautenstrauch Syndrome	AR	100	122 of 122
POLR3B	Leukodystrophy, Hypomyelination, Hypogonadotropic Hypogonadism, Hypodontia	AR	100	61 of 61
PPP2R2B	Spinocerebellar Ataxia	AD	99.97	2 of 2
PRICKLE1	Epilepsy, Unverricht-Lundborg Disease	AR	98.41	23 of 23
PRKCG	Spinocerebellar Ataxia	AD	100	52 of 52
PRRT2	Convulsions, Kinesigenic Dyskinesia, Hemiplegic Migraine, Choreoathetosis	AD	99.93	111 of 111
PUM1	Spinocerebellar Ataxia	AD	99.98	8 of 8
RARS2	Pontocerebellar Hypoplasia	AR	99.98	39 of 40
RPGRIP1L	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.96	52 of 52
RRM2B	Mitochondrial Dna Depletion Syndrome, Ophthalmoplegia, Mitochondrial Myopathy, Kearns-Sayre Syndrome, Neurogastrointestinal Encephalomyopathy	AD,AR	92.38	46 of 46
RUBCN	Spinocerebellar Ataxia, Epilepsy, Intellectual Disability	AR	99.96	-
SACS	Spastic Ataxia	AR	99.91	291 of 292
SCN2A	Epileptic Encephalopathy, Episodic Ataxia, Seizures, Dravet Syndrome, West Syndrome	AD	100	351 of 351
SCYL1	Spinocerebellar Ataxia, Acute Infantile Liver Failure, Peripheral Sensory Motor Neuropathy	AR	99.98	13 of 13
SETX	Amyotrophic Lateral Sclerosis, Spinocerebellar Ataxia	AD,AR	99.71	219 of 227
SIL1	Marinesco-Sjogren Syndrome	AR	100	47 of 48
SLC16A2	Allan-Herndon-Dudley Syndrome	X,XR,G	99.94	-
SLC17A5	Sialuria	AR	99.91	49 of 49
SLC1A3	Episodic Ataxia	AD	100	13 of 13
SLC20A2	Basal Ganglia Calcification, Bilateral Striopallidodentate Calcinosis	AD	99.96	123 of 127
SLC25A46	Neuropathy	AR	99.79	16 of 17
SLC2A1	Choreoathetosis, Epilepsy, Glucose Transport Defect, Cryohydrocytosis, Ataxia, Dyskinesia	AD,AR	99.99	301 of 304
SLC52A2	Brown-Vialetto-Van Laere Syndrome, Spinocerebellar Ataxia, Blindness, Deafness	AR	100	31 of 32
SLC52A3	Bulbar Palsy, Deafness	AR	100	43 of 43
SLC9A1	Lichtenstein-Knorr Syndrome	AR	99.2	6 of 6

SLC9A6	Mental Retardation, Christianson Syndrome	X,XD,G	98.87	-
SNX14	Spinocerebellar Ataxia, Intellectual Disability, Macrocephaly, Cerebellar Hypotrophy	AR	99.8	18 of 18
SPG11	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease, Spastic Paraplegia	AR	99.93	289 of 297
SPG7	Spastic Paraplegia, Primary Lateral Sclerosis	AD,AR	99.94	125 of 126
SPR	Dystonia,Sepiapterin Reductase Deficiency	AD,AR	99.89	27 of 27
SPTBN2	Spinocerebellar Ataxia	AD,AR	99.92	39 of 39
STUB1	Spinocerebellar Ataxia	AD,AR	99.93	36 of 36
SYNE1	Arthrogryposis Multiplex Congenita, Emery-Dreifuss Muscular Dystrophy, Spinocerebellar Ataxia	AD,AR	99.99	193 of 193
SYT14	Spinocerebellar Ataxia, Psychomotor Delay	AR	100	3 of 3
TBP	Parkinson Disease, Spinocerebellar Ataxia	AD	99.9	1 of 1
TCTN2	Joubert Syndrome, Meckel Syndrome	AR	100	14 of 14
TDP1	Spinocerebellar Ataxia, Axonal Neuropathy	AR	99.47	5 of 5
TDP2	Spinocerebellar Ataxia, Epilepsy, Intellectual Disability	AR	99.93	8 of 8
TGM6	Spinocerebellar Ataxia	AD	100	26 of 26
THG1L	Spinocerebellar Ataxia	AR	100	7 of 7
TMEM216	Joubert Syndrome, Meckel Syndrome, Orofaciodigital Syndrome	AR	98.74	8 of 8
TMEM237	Joubert Syndrome, Oculorenal Defect	AR	100	11 of 11
TMEM240	Spinocerebellar Ataxia	AD	99.1	9 of 9
TMEM67	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Nephronophthisis, Rhyns Syndrome	AR	96.93	177 of 179
TPP1	Ceroid Lipofuscinosis, Spinocerebellar Ataxia	AR	100	147 of 147
TRPC3	Spinocerebellar Ataxia	AD	98.78	2 of 2
TSEN2	Pontocerebellar Hypoplasia	AR	95.47	4 of 5
TSEN34	Pontocerebellar Hypoplasia	AR	100	1 of 1
TSEN54	Encephalopathy, Olivopontocerebellar Hypoplasia	AR	96.94	20 of 22
TTBK2	Spinocerebellar Ataxia	AD	99.31	10 of 10
TTC19	Mitochondrial Complex Iii Deficiency	AR	95.3	10 of 12
TTPA	Vitamin E Deficiency, Ataxia	AR	99.31	30 of 30
TUBB4A	Dystonia Musculorum Deformans, Leukodystrophy	AD	89.81	44 of 44
TWNK	Spinocerebellar Ataxia, Perrault Syndrome, Ophthalmoplegia, Mitochondrial Dna Deletions, Sensory Ataxic Neuropathy, Dysarthria, Ophthalmoparesis	AD,AR	-	-
UBA5	Epileptic Encephalopathy, Spinocerebellar Ataxia	AR	99.98	19 of 19
VAMP1	Ataxia, Myasthenic Syndrome	AD,AR	99.51	8 of 8
VLDLR	Cerebellar Hypoplasia, Mental Retardation, Dysequilibrium Syndrome	AR	100	20 of 20
VPS13D	Spinocerebellar Ataxia	AR	99.97	19 of 19
VRK1	Pontocerebellar Hypoplasia	AR	99.64	15 of 15
VWA3B	Spinocerebellar Ataxia	AR	99.95	4 of 4
WDR81	Cerebellar Hypoplasia, Mental Retardation, Hydrocephalus, Dysequilibrium Syndrome	AR	99.94	19 of 19
WFS1	Cataract, Deafness, Diabetes Mellitus, Wolfram Syndrome	AD,AR	99.97	390 of 395
WWOX	Epileptic Encephalopathy, Esophageal Cancer, Spinocerebellar Ataxia, Partial Gonadal Dysgenesis, Squamous Cell Carcinoma Of The Esophagus	AR	99.94	44 of 44
XRCC1	Spinocerebellar Ataxia	AR	94.43	3 of 3
ZFYVE26	Spastic Paraplegia	AR	99.95	48 of 48

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



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Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

References

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