Amyotrophic Lateral Sclerosis

Precision Panel

Overview

Amyotrophic Lateral Sclerosis (ALS) is the most common degenerative disease of the motor neuron system. It is characterized by the progressive loss of motor neurons in the brain and spinal cord, leading to paralysis. It begins insidiously with focal weakness but spreads ceaselessly to involve most muscles, including the diaphragm. The diagnosis of ALS rests on a history of painless progressive weakness coupled with examination findings of upper and lower motor dysfunction. Typically, death is due to respiratory paralysis and it occurs within 3 to 5 years of diagnosis. Many different genes and pathophysiological processes contribute to the disease, more so, there are two types of ALS differentiated by genetics: familial and sporadic. Although ALS is incurable and fatal, with median survival of 3 years, early treatment can lengthen and substantially increase quality of life for patients.

The Igenomix Amyotrophic Lateral Sclerosis Precision Panel can serve as an accurate and directed diagnostic tool 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.

Indications

The Igenomix Amyotrophic Lateral Sclerosis Precision Panel is indicated in patients with a clinical suspicion or diagnosis presenting with the following manifestations:

- Reduced finger dexterity and cramping
- Tripping, stumbling or awkwardness when running
- Wrist drop and foot drop
- Slurred speech and hoarseness
- Depression
- Impaired executive function
- Aspiration or choking
- Muscle atrophy
- Spasticity
- Muscle cramps
- Voice changes

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form of medical care with riluzole, enteral nutrition, non-invasive ventilation and preventive measures of complications.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

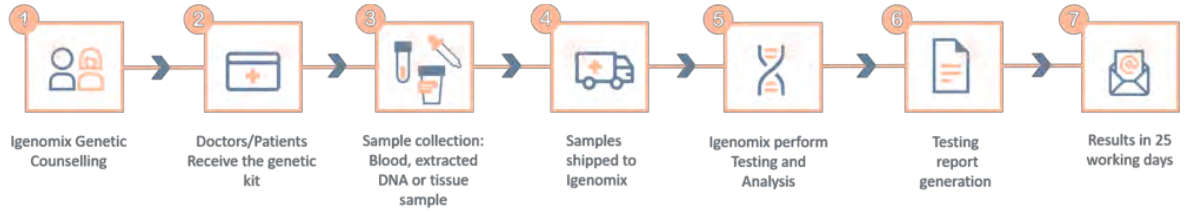
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ALS2	Amyotrophic Lateral Sclerosis, Primary Lateral Sclerosis, Spastic Paralysis	AR	99.09	83 of 83
ANG	Amyotrophic Lateral Sclerosis	AD	-	-
ANXA11	Amyotrophic Lateral Sclerosis	AD	99.91	15 of 15
ATL1	Neuropathy, Spastic Paraplegia	AD	100	93 of 93
ATXN2	Parkinson Disease, Spinocerebellar Ataxia, Amyotrophic Lateral Sclerosis	AD	91.78	9 of 10
BSCL2	Encephalopathy, Lipodystrophy, Neuronopathy, Spastic Paraplegia, Neurodegenerative Syndrome	AD,AR	99.83	60 of 61
C9ORF72	Amyotrophic Lateral Sclerosis, Frontotemporal And Semantic Dementia, Huntington Disease, Aphasia	AD	-	-
CCNF	Amyotrophic Lateral Sclerosis	-	99.11	22 of 23
CFAP410	Retinal Dystrophy, Spondylometaphyseal Dysplasia, Amyotrophic Lateral Sclerosis, Cone Rod Dystrophy	AR	-	-
CHCHD10	Frontotemporal Dementia, Myopathy, Spinal Muscular Atrophy, Amyotrophic Lateral Sclerosis	AD	95.3	22 of 30
CHMP2B	Amyotrophic Lateral Sclerosis, Frontotemporal And Semantic Dementia, Aphasia	AD	99.99	22 of 22
DAO	Amyotrophic Lateral Sclerosis	-	100	12 of 12
DCTN1	Amyotrophic Lateral Sclerosis, Neuronopathy, Parkinsonism, Perry Syndrome	AD,AR	100	56 of 56
EPHA4	Amyotrophic Lateral Sclerosis	-	99.98	5 of 5
ERBB4	Amyotrophic Lateral Sclerosis	AD	100	13 of 14
FIG4	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease, Cleidocranial Dysplasia, Polymicrogyria, Yunis-Varon Syndrome	AD,AR	99.92	72 of 72
FUS	Amyotrophic Lateral Sclerosis, Frontotemporal Dementia, Myxoid Cell Liposarcoma	AD	100	117 of 117
GBE1	Glycogen Storage Disease, Polyglucosan Body Disease	AR	99.95	71 of 74
GLE1	Congenital Arthrogryposis, Anterior Horn Cell Disease, Lethal Congenital Contracture Syndrome, Amyotrophic Lateral Sclerosis	AR	100	17 of 17
GLT8D1	Amyotrophic Lateral Sclerosis	-	100	7 of 7
GRN	Ceroid Lipofuscinosis, Progressive Non-Fluent Aphasia, Frontotemporal And Semantic Dementia	AD,AR	100	220 of 229
HEXA	Tay-Sachs Disease	AR	100	205 of 206
HNRNPA1	Amyotrophic Lateral Sclerosis, Inclusion Body Myopathy, Paget Disease, Frontotemporal Dementia	AD	99.98	13 of 13
HNRNPA2B1	Inclusion Body Myopathy, Paget Disease, Frontotemporal Dementia	-	99.98	5 of 6
HSPD1	Leukodystrophy, Spastic Paraplegiainant	AD,AR	100	7 of 7
KIF5A	Amyotrophic Lateral Sclerosis, Myoclonus, Spastic Paraplegia	AD	100	85 of 85

MAPT	Frontotemporal And Semantic Dementia, Parkinson Disease, Supranuclear Palsy, Aphasia, Gait Freezing Syndrome	AD,AR	97.65	110 of 111
MATR3	Amyotrophic Lateral Sclerosis, Vocal Cord And Pharyngeal Distal Myopathy	AD	99.98	21 of 21
NEFH	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease	AD,AR	97.5	28 of 31
NEK1	Amyotrophic Lateral Sclerosis, Short Rib-Polydactyly Syndrome, Orofaciodigital Syndrome	AD,AR,MU,D	99.83	73 of 74
OPTN	Amyotrophic Lateral Sclerosis, Glaucoma	AD,AR	97.02	68 of 68
PFN1	Amyotrophic Lateral Sclerosis	AD	80.25	8 of 8
PON1	Amyotrophic Lateral Sclerosis	-	100	8 of 8
PON2	Amyotrophic Lateral Sclerosis	-	99.98	3 of 3
PON3	Amyotrophic Lateral Sclerosis	-	100	3 of 3
PPARGC1A	Amyotrophic Lateral Sclerosis	-	99.88	6 of 6
PRF1	Aplastic Anemia, Hemophagocytic Lymphohistiocytosis, Lymphoma Non-Hodgkin	AR	99.99	196 of 196
PRPH	Amyotrophic Lateral Sclerosis	AD,AR	100	10 of 10
PSEN1	Acne Inversa, Alzheimer Disease, Cardiomyopathy, Frontotemporal And Semantic Dementia, Pick Disease Of Brain	AD	100	326 of 332
REEP1	Neuronopathy, Spastic Paraplegia	AD	100	62 of 62
SETX	Amyotrophic Lateral Sclerosis, Spinocerebellar Ataxia	AD,AR	99.71	219 of 227
SIGMAR1	Amyotrophic Lateral Sclerosis, Spinal Muscular Atrophy	AR	100	20 of 20
SLC52A2	Brown-Vialetto-Van Laere Syndrome, Spinocerebellar Ataxia-Blindness-Deafness Syndrome	AR	100	31 of 32
SLC52A3	Bulbar Palsy	AR	100	43 of 43
SOD1	Amyotrophic Lateral Sclerosis, Spastic Tetraplegia And Axial Hypotonia	AD,AR	100	217 of 221
SPART	Spastic Paraplegia	AR	99.9	-
SPAST	Spastic Paraplegia	AD	99.98	616 of 655
SPG11	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease, Spastic Paraplegia	AR	99.93	289 of 297
SQSTM1	Frontotemporal And Semantic Dementia, Amyotrophic Lateral Sclerosis, Myopathy, Neurodegeneration, Dystonia, Gaze Palsy, Paget Disease Of Bone	AD,AR	99.25	105 of 107
TAF15	Chondrosarcoma, Amyotrophic Lateral Sclerosis	-	95.53	13 of 13
TARDBP	Amyotrophic Lateral Sclerosis, Frontotemporal Dementia	AD	90.84	71 of 74
TBK1	Encephalopathy, Frontotemporal Dementia, Amyotrophic Lateral Sclerosis, Herpes Simplex Virus Encephalitis	AD	99.91	141 of 142
TIA1	Welander Distal Myopathy	AD,AR	100	13 of 13
TREM2	Polycystic Lipomembranous Osteodysplasia, Sclerosing Leukoencephalopathy, Amyotrophic Lateral Sclerosis, Frontotemporal And Semantic Dementia, Alzheimer Disease, Nasu-Hakola Disease	AD	100	55 of 55
TRPM7	Amyotrophic Lateral Sclerosis, Parkinsonism	AD	99.59	6 of 6
TUBA4A	Amyotrophic Lateral Sclerosis, Frontotemporal Dementia	AD	99.97	13 of 13
UBQLN2	Amyotrophic Lateral Sclerosis, Frontotemporal Dementia	X,XD,G	99.78	-
UNC13A	Amyotrophic Lateral Sclerosis	-	99.41	12 of 12
VAPB	Amyotrophic Lateral Sclerosis, Spinal Muscular Atrophy	AD	100	9 of 9
VCP	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease, Inclusion Body Myopathy, Paget Disease, Frontotemporal And Semantic Dementia, Spastic Paraplegia	AD	100	68 of 69
WASHC5	Dandy-Walker-Like Malformation, Spastic Paraplegia, 3c Syndrome	AD,AR	99.99	-

*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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