Parkinson and Early Onset Parkinson Disease

Precision Panel

Overview

Parkinson Disease (PD) is one of the second-most common neurodegenerative disorder affecting 2-3% of the population >65 years of age. It has traditionally been considered a motor system disorders, now recognized to be a complex condition with diverse clinical features. It is characterized by neuronal dopaminergic loss in the substantia nigra, a region in the brain in charge of gross motor movements. The clinical hallmarks of Parkinson disease include slowing of movements (bradykinesia), pin-wheel rigidity, resting tremor and postural instability. The etiology of Parkinson Disease is still unclear, but it is hypothesized to be a combination of genetic and environmental factors. Although incidence increases rapidly over the age of 60 it can also have an early onset. Generally, these patients have more involuntary movements and a poorer prognosis.

The Igenomix Parkinson and Early Onset Parkinson Disease Precision Panel can serve as an accurate and directed diagnostic tool as well as for a differential diagnosis of resting tremor 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 Parkinson and Early Onset Parkinson Disease Precision Panel is indicated in patients with a clinical suspicion or diagnosis presenting with the following manifestations:

- Resting tremor
- Family history of early onset PD
- Slowing of movements (bradykinesia)
- Rigidity
- Postural instability
- Cognitive dysfunction
- Psychosis
- Mood disorders
- Sleep disturbances
- Fatigue
- Pain and sensory disturbances

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 treatment to increase dopamine, nonpharmacologic treatment including education, support and neuroprotective benefits of exercise and nutrition as well of surgical care if needed.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

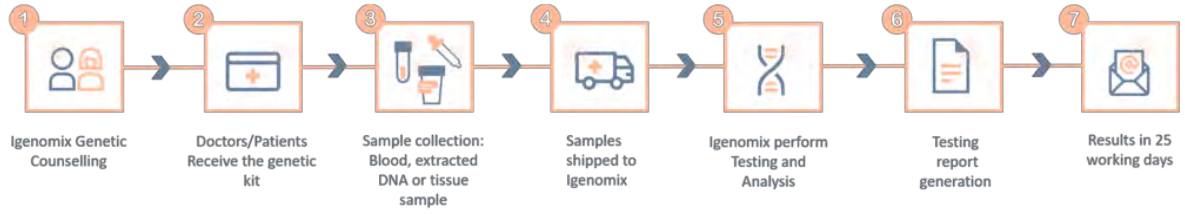
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ATP13A2	Kufor-Rakeb Syndrome, Spastic Paraplegia, Ceroid Lipofuscinosis	AR	99.97	53 of 53
ATP1A3	Hemiplegia Of Childhood, Cerebellar Ataxia, Pes Cavus, Optic Atrophy, Sensorineuralhearing Loss, Dystonia, Parkinson Disease	AD	99.94	138 of 138
ATP7B	Wilson Disease	AR	99.97	989 of 1000
CHCHD2	Parkinson Disease	AD	100	14 of 14
CSF1R	Brain Abnormalities, Neurodegeneration, Dysosteosclerosis, Gliosis	AD,AR	100	122 of 124
DCTN1	Amyotrophic Lateral Sclerosis, Neuronopathy, Parkinson Disease, Alveolar Hypoventilation, Mental Depression, Perry Syndrome	AD,AR	100	56 of 56
DNAJC6	Parkinson Disease	AR	99.86	13 of 14
EIF4G1	Parkinson Disease	AD	99.92	23 of 23
FBXO7	Parkinsonian-Pyramidal Syndrome	AR	100	19 of 20
GBA	Dementia, Gaucher Disease, Parkinson Disease	AD,AR	100	469 of 471
GCH1	Dystonia, Gtp Cyclohydrolase I Deficiency	AD,AR	99.41	225 of 244
GIGYF2	Parkinson Disease	AD	99.88	49 of 49
HTRA2	3-Methylglutaconic Aciduria, Parkinson Disease	AD,AR	99.81	18 of 18
LRRK2	Parkinson Disease	AD	100	154 of 155
MAPT	Frontotemporal Dementia, Parkinson Disease, Pick Disease Of Brain, Supranuclear Palsy, Aphasia, Gait Freezing	AD,AR	97.65	110 of 111
PARK7	Parkinson Disease	AR	100	29 of 31
PDE8B	Adrenocortical Disease, Striatal Degeneration	AD	99.98	10 of 10
PDGFB	Basal Ganglia Calcification, Meningioma, Striopallidodentate Calcinosis, Dermatofibrosarcoma Protuberans	AD	100	22 of 22
PDGFRB	Basal Ganglia Calcification, Kosaki Overgrowth Syndrome, Myeloproliferative Disorder, Myofibromatosis, Premature Aging Syndrome	AD	99.64	28 of 28
PINK1	Parkinson Disease	AR	92.42	124 of 127
PLA2G6	Neuroaxonal Dystrophy, Neurodegeneration, Brain Iron Accumulation, Parkinson Disease	AR	99.94	190 of 191
PODXL	Parkinson Disease	-	99.77	11 of 11
PRKN	Lung Cancer, Parkinson Disease, Lung And Ovarian Cancer	AD,AR	100	-
PRKRA	Dystonia	AR	100	9 of 9
RAB39B	Mental Retardation, Parkinson Disease	X,XR,G	100	-

SLC20A2	Basal Ganglia Calcification, Striopallidodentate Calcinosis	AD	99.96	123 of 127
SLC39A14	Hypermanganesemia, Dystonia, Hyperostosis Cranialis Interna, Parkinson Disease	AD,AR	100	9 of 9
SLC6A3	Dystonia, Parkinson Disease	AR	100	31 of 31
SNCA	Dementia, Parkinson Disease, Parkinsonian-Pyramidal Syndrome	AD	100	11 of 12
SPR	Dystonia, Sepiapterin Reductase Deficiency	AD,AR	99.89	27 of 27
SYNJ1	Epileptic Encephalopathy, Parkinson Disease	AR	99.81	30 of 32
TAF1	Dystonia, Mental Retardation, Parkinson Disease	X,XR,G	99.74	-
TH	Segawa Syndrome, Dystonia	AR	100	71 of 71
TMEM230	Parkinson Disease, Mitochondrial Complex Iv Deficiency	-	96.41	7 of 7
UCHL1	Neurodegeneration, Optic Atrophy, Parkinson Disease	AD,AR	100	5 of 5
VAC14	Striatonigral Degeneration, Yunis-Varon Syndrome	AR	100	11 of 11
VPS13A	Choreoacanthocytosis	AR	99.37	120 of 122
VPS13C	Parkinson Disease	AR	99.72	10 of 12
VPS35	Parkinson Disease	AD	98	24 of 24
XPR1	Basal Ganglia Calcification, Striopallidodentate Calcinosis	AD	99.88	14 of 14

*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



Contact us

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.

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