Alzheimer Disease

Precision Panel

Overview

Alzheimer Disease (AD) is the most common cause of dementia and one of the leading causes of morbidity and mortality in the aging population. The main clinical characteristic of AD is dementia that typically begins with subtle and poorly recognized failure of memory also known as mild cognitive impairment, slowly becoming more severe and eventually incapacitating. Other findings include confusion, disorientation, language disturbances, agitation etc. 95% of all AD is late onset, typically over the age of 60-65, however, 5% is early onset. Approximately 25% of all AD is familial and 75% is nonfamilial. Familial forms of AD have a strong genetic predisposition. While the pathogenesis of AD remains unclear, all forms of AD appear to share overproduction and/or decreased clearance of amyloid beta peptides. The modes of inheritance of familial forms of AD are autosomal dominant and recessive patterns.

The Igenomix Alzheimer Disease Precision Panel can be used as a tool for an accurate diagnosis and differential diagnosis of dementia 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 Alzheimer Disease Precision Panel is used for patients with a clinical suspicion or diagnosis presenting with or without the following symptoms:

- Memory loss
- Family history of early-onset AD
- Confusion about the location of familiar places
- Taking longer to accomplish normal, daily tasks
- Trouble handling money and paying bills
- Compromised judgment
- Loss of spontaneity
- Mood and personality changes

Clinical Utility

The clinical utility of this panel is:

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

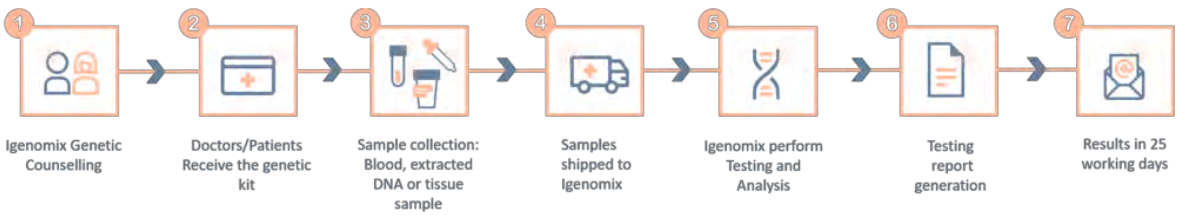
- Early initiation of treatment involving a multidisciplinary team in the form of medical care with antiparkinsonian agents, antiepileptic drugs, antidepressants, anxiolytics etc. Mental activities are recommended to potentiate cognitive retraining.
- Risk assessment of asymptomatic family members according 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 DISEASE	INHERITANCE	% GENE COVERAGE (20X)	HGMD**
A2M	Alzheimer Disease	AD	99.89	2 of 2
ABCA7	Alzheimer Disease, Early-Onset Autosomal Dominant Alzheimer Disease	AD	99.99	159 of 159
APOE	Alzheimer Disease, Sea-Blue Histiocyte Disease, Dysbetalipoproteinemia	AD,AR	99.53	65 of 68
APP	Alzheimer Disease, Cerebral Amyloid Angiopathy, Abeta Amyloidosis, Early-Onset Autosomal Dominant Alzheimer Disease	AD	100	69 of 77
CACNA1G	Spinocerebellar Ataxia	AD	99.52	16 of 16
GATA1	Down Syndrome Trisomy 21	X,XR,G	99.93	-
HFE	Alzheimer Disease, Hemochromatosis	AD,AR	100	55 of 57
MPO	Alzheimer Disease, Myeloperoxidase Deficiency	AD,AR	100	15 of 15
NOS3	Alzheimer Disease	AD,MU	99.98	13 of 13
PLAU	Alzheimer Disease	AD	100	5 of 5
PRNP	Creutzfeldt-Jakob Disease, Gerstmann-Straussler Disease, Spongiform Encephalopathy With Neuropsychiatric Features, Familial Alzheimer-Like Prion Disease	AD	100	69 of 69
PSEN1	Alzheimer Disease, Frontotemporal Dementia, Pick Disease Of Brain, Early-Onset Autosomal Dominant Alzheimer Disease	AD	100	326 of 332
PSEN2	Alzheimer Disease, Early-Onset Autosomal Dominant Alzheimer Disease	AD	100	72 of 72
SORL1	Early-Onset Autosomal Dominant Alzheimer Disease	-	100	155 of 155
TOMM40	Early-Onset Autosomal Dominant Alzheimer Disease	-	99.53	-
TREM2	Behavioral Variant Of Frontotemporal Dementia, Early-Onset Autosomal Dominant Alzheimer Disease	AD	100	55 of 55

*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.

References

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