Neurocognitive Evaluation Panel™

Comprehensive Neurocognitive Genetics Panel

Detect 64 risk factor genes associated with neurocognitive disorders

An accurate, timely diagnosis of a nonreversible decline in mental function is crucial to ensure a personalized and data-driven treatment plan is provided to the patient. Early-stage support can help avoid unnecessary struggles and expenses associated with missed treatment opportunities.

Lab offers our propriatery comprehensive neurocognitive evaluation panel, which examines 64 genes associated with an increased risk of developing neurocognitive disorders and detects both the diagnostic and risk factor genes for dementia, Alzheimer's disease, and Parkinson's disease. While no single test exists to diagnose dementia, DAP testing is an important component in the diagnostic process. Additionally, DAP testing rules out medical conditions that mimic the memory loss symptoms associated with dementia.

Parkinson's Disease

As a result of faulty genes being passed to a child by their parents, Parkinson's disease can run in families. Approximately 15% of people with Parkinson's have a family history of this disorder. Familial cases of Parkinson disease can be caused by mutations in the *LRRK2, PARK7, PINK1, PRKN, MAPT, GBA* or *SNCA* gene.

Alzheimer's Disease

One type of early-onset Alzheimer's disease is known as autosomal dominant Alzheimer's disease, or early-onset familial Alzheimer's disease (FAD), which affects approximately 3-5% of all people with Alzheimer's. What makes this type of early-onset Alzheimer's disease so unusual is that it is caused by a hereditary genetic mutation to one of three genes: *PSEN1*, *PSEN2* or APP. Genes that increase the risk of Alzheimer's disease are *APOE*, *TREM2* and *NOTCH3*.

Lewy-Body Dementia

Lewy body dementia, also known as dementia with Lewy bodies, is the second most common type of progressive dementia after Alzheimer's disease. Protein deposits, called Lewy bodies, develop in nerve cells in the brain regions involved in thinking, memory and movement (motor control). While much research is underway, it is widely accepted that familial cases of Lewy-Body Dementia can be caused by mutations in the *SNCA*, *APOE E4*, or *GBA* gene.

Frontotemporal Dementia

Frontotemporal Dementia (FTD) is the most common form of dementia for people under age 60. It represents a group of brain disorders caused by degeneration of the frontal and/or temporal lobes of the brain. FTD's estimated prevalence in the United States is around 60,000 cases, and many in the medical community remain unfamiliar with it. FTD is frequently misdiagnosed as Alzheimer's, depression, Parkinson's disease or a psychiatric condition. On average, it currently takes 3.6 years to get an accurate diagnosis. Up to 40% of patients have a positive family history with a diagnosis of dementia in at least one extra family member. Three major causal genes have been identified: MAPT, GRN and C9ORF72.

ALS

Approximately 5-10% of cases are due to genetic mutations and are inherited from a family member. If there are two or more family members with ALS, the disease is considered familial. There are an increasing number of gene mutations that have been identified both in familial and seemingly sporadic patients. The most commonly known ALS genes have been identified as: *C9orf72, FUS, TARDBP, and SOD1*.

Conditions

- Dementia
- Vascular dementia
- Cerebellar ataxia
- ALS
- Parkinson's
- · Alzheimer's
- Frontotemporal lobe dementia
- Lewy Body dementia

Symptoms along with Family History:

- Depression
- Anxiety
- Fatigue
- Insomnia
- Sleep disorder
- Tremor
- · Gait disturbance
- Repeated falls
- Amnesia
- Hallucinations
- Speech disturbance

64 Genes Tested in Our Propiratry Pannel

Gene	Gene Symbol	Gene ID	Associated Diseases (OMIM)
APOE	APOE (apolipoprotein E)	348	Alzheimer Disease; Sea-blue histiocyte disease; Macular Degeneration, Age-Related, early-onset familial Alzheimer disease-3; Lipoprotein glomerulopathy
PRNP	PRNP (prion protein)	5621	Creutzfeldt-Jakob disease; Gerstmann-Straussler disease; Kuru, susceptibility to; Insomnia, fatal familial; Hun- tington disease-like 1; Prion disease with protracted course
GCH1	GCH1 (GTP cyclohydrolase 1)	2643	Parkinson disease
PSEN1	PSEN1 (presenilin 1)	5663	Pick disease; Dementia, frontotemporal; early-onset familial Alzheimer disease-3; dilated cardiomyopathy-1U; Acne inversa, familial
LRRK2	LRRK2 (leucine rich repeat kinase 2)	120892	Parkinson disease, Dementia
APP	APP (amyloid beta precursor protein)	351	Alzheimer Disease; Cerebral Amyloid Angiopathy, App-Related
GRN	GRN (granulin precursor)	2896	frontotemporal lobar degeneration with ubiquitin-positive inclusions; neuronal ceroid lipofuscinosis type 11
MAPT	MAPT (microtubule associated protein tau)	4137	Susceptibility to late-onset Parkinson disease; Pick disease; Dementia, frontotemporal
NOTCH3	NOTCH3 (notch receptor 3)	4854	CADASIL; Lateral meningocele syndrome
POLG	POLG (DNA polymerase gamma, catalytic subunit)	5428	progressive external ophthalmoplegia
PSEN2	PSEN2 (presenilin 2)	5664	Alzheimer disease, type 4; dilated cardiomyopathy-1V
TH ATP13A2	TH (tyrosine hydroxylase) ATP13A2 (ATPase cation transporting 13A2)	7054 23400	Segawa Syndrome, Autosomal Recessive and Dystonia Parkinson disease, Dementia
ATP1A3	ATP1A3 (ATPase Na+/K+ transporting subunit alpha 3)	478	rapid-onset dystonia-parkinsonism, cerebellar ataxia
C9orf72	C9orf72 (C9orf72-SMCR8 complex subunit)	203228	frontotemporal dementia and/or amyotrophic lateral sclerosis
CSF1R	CSF1R (colony stimulating factor 1 receptor)	1436	hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia
DCTN1	DCTN1 (dynactin subunit 1)	1639	amyotrophic lateral sclerosis ; Perry syndrome; Neuronopathy, Distal Hereditary Motor, Type Viib
DNMT1	DNMT1 (DNA methyltransferase 1)	1786	autosomal dominant cerebellar ataxia, deafness, and narcolepsy
EIF4G1	EIF4G1 (eukaryotic translation initiation factor 4 gamma 1)	1981	Cerebellar ataxia
FBX07	FBX07 (F-box protein 7)	25793	Parkinson Disease, Autosomal Recessive Early-Onset and Early-Onset Parkinson's Disease
GBA	GBA (glucosylceramidase beta)	2629	Dementia, Parkinson disease
HTRA2	HTRA2 (HtrA serine peptidase 2)	27429	Parkinson's disease
PARK7 PINK1	PARKT (Parkinsonism associated deglycase)	11315	Parkinson's disease Parkinson disease
PLA2G6	PINK1 (PTEN induced kinase 1) PLA2G6 (phospholipase A2 group VI)	65018 8398	Neurodegeneration
PRKN	PRKN (parkin RBR E3 ubiquitin protein ligase)	5071	Parkinson disease
PRKRA	PRKRA (protein activator of interferon induced protein kinase EIF2AK2)	8575	Dystonia
SLC6A3	SLC6A3 (solute carrier family 6 member 3)	6531	Dopamine transporter deficiency syndrome
SNCA	SNCA (synuclein alpha)	6622	Lewy body dementia; Parkinson disease
SNCB	SNCB (synuclein beta)	6620	Parkinson disease, dementia, DLB
TAF1	TAF1 (TATA-box binding protein associated factor 1)	6872	Mental Retardation, X-Linked, Syndromic 33 and Dystonia
TREM2	TREM2 (triggering receptor expressed on myeloid cells 2)	54209	polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy type 2
TYROBP	TYROBP (transmembrane immune signaling adaptor TYROBP)	7305	polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy type 1
UCHL1	UCHL1 (ubiquitin C-terminal hydrolase L1)	7345	Autosomal Recessive and Parkinson Disease
VPS35 ALS2	VPS35 (VPS35 retromer complex component) ALS2 (alsin Rho guanine nucleotide exchange factor ALS2)	55737	Parkinson disease
ATP7B	ATP7B (ATPase copper transporting beta)	57679 540	amyotrophic lateral sclerosis; Spastic paralysis, infantile onset ascending Wilson disease
ERBB4	ERBB4 (erb-b2 receptor tyrosine kinase 4)	2066	Amyotrophic lateral sclerosis
FUS	FUS (FUS RNA binding protein)	2521	amyotrophic lateral sclerosis ; Tremor, hereditary essential,
HNRNPA1	HNRNPA1 (heterogeneous nuclear ribonucleoprotein A1)	3178	Amyotrophic lateral sclerosis
ІТМ2В	ITM2B (integral membrane protein 2B)	9445	Cerebral amyloid angiopathy, itm2b-related; Cerebral amyloid angiopathy, itm2b-related,
MATR3	MATR3 (matrin 3)	9782	Myopathy, Distal,
NPC1	NPC1 (NPC intracellular cholesterol transporter 1)	4864	Niemann-Pick disease type C/D
CHCHD10	CHCHD10	615903	Frontotemporal dementia and/or amyotrophic lateral sclerosis
OPTN	OPTN (optineurin)	10133	Adult-onset primary open angle glaucoma; Glaucoma, normal tension, susceptibility to; amyotrophic lateral sclerosis
SETX	SETX (senataxin)	23064	amyotrophic lateral sclerosis; autosomal recessive spinocerebellar ataxia
SIGMAR1 SOD1	SIGMARI (sigma non-opioid intracellular receptor 1) SOD1 (superoxide dismutase 1)	10280	distal spinal muscular atrophy type 2; amyotrophic lateral sclerosis amyotrophic lateral sclerosis
SORL1	SORL1 (sortilin related receptor 1)	6653	Alzheimer disease
SPG11	SPG11 (SPG11 vesicle trafficking associated, spatacsin)	80208	Amyotrophic lateral sclerosis, juvenile; spastic paraplegia type; Charcot-Marie-Tooth disease, axonal, type 2X
SQSTM1	SQSTM1 (sequestosome 1)	8878	Frontotemporal dementia and/or amyotrophic lateral sclerosis; Neurodegeneration with ataxia, dystonia, and gaze
TARDBP	TARDBP (TAR DNA binding protein)	23435	palsy, childhood-onset amyotrophic lateral sclerosis
TBK1		29110	Frontotemporal dementia and/or amyotrophic lateral sclerosis type 4; Encephalopathy, acute, infection-induced
	THRAM (*ubulin aloha 4a)		(herpes-specific), susceptibility to,
TUBA4A UBQLN2	TUBA4A (tubulin alpha 4a) UBQLN2 (ubiquilin 2)	7277 29978	amyotrophic lateral sclerosis amyotrophic lateral sclerosis
			amyotrophic lateral scienosis inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia; amyotrophic lat-
VCP	VCP (valosin containing protein)	7415	eral sclerosis; Charcot-Marie-Tooth disease type 2Y
CHMP2B	CHMP28 (charged multivesicular body protein 28)	25978	Dementia, familial, nonspecific; amyotrophic lateral sclerosis
HNRNPA2B1 VPS13C	HNRNPA2B1 (heterogeneous nuclear ribonucleoprotein A2/B1) VPS13C (vacuolar protein sorting 13 homolog C)	3181 54832	Frontotemporal dementia, ALS Parkinson disease, dementia, frontotemporal dementia
ATXN2	VPSI3C (vacuolar protein sorting 13 homolog C) Ataxin 2	601517	Parkinson disease, dementia, frontotemporal dementia Susceptibility to late-onset Parkinson disease; spinocerebellar ataxia 2
ATM	ATM serine/threonine kinase	472	Susceptibility to data: and SCHIZOPHRENIA
	ATP binding cassette subfamily A member 7)	10347	Alzheimer disease
ABCA7			
ABCA7 PLCG2	PLCG2 (phospholipase C gamma 2)"	5336	Alzheimer Disease, Late Onset