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Full name:	Neurodegeneration (99 genes) - IPG
Abbreviation:	Neurodegeneration
Type of panel:	Custom panel
Version number:	V4
Laboratory:	Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
Created:	26 Nov 2019 - 14:48
Changed:	01 Dec 2023 - 12:58

Related Diseases

- ATP13A2-related juvenile neuronal ceroid lipofuscinosis
- Adult-onset distal myopathy due to VCP mutation
- Amyotrophic lateral sclerosis
- Amyotrophic lateral sclerosis type 4
- Autosomal dominant Charcot-Marie-Tooth disease type 2 due to KIF5A mutation
- Autosomal dominant adult-onset proximal spinal muscular atrophy
- Autosomal dominant spastic paraplegia type 10
- Autosomal recessive spastic paraplegia type 11
- Behavioral variant of frontotemporal dementia
- Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy
- Classic progressive supranuclear palsy syndrome
- Distal hereditary motor neuropathy type 7
- Distal hereditary motor neuropathy, Jerash type
- Early-onset autosomal dominant Alzheimer disease
- Familial Alzheimer-like prion disease
- Fatal familial insomnia
- Fetal Gaucher disease
- Frontotemporal dementia with motor neuron disease
- Gaucher disease type 1

- Gaucher disease type 2
- Gaucher disease type 3
- Gaucher disease-ophthalmoplegia-cardiovascular calcification syndrome
- Gerstmann-Straussler-Scheinker syndrome
- Hereditary late-onset Parkinson disease
- Huntington disease-like 1
- Huntington disease-like syndrome due to C9ORF72 expansions
- Inclusion body myopathy with Paget disease of bone and frontotemporal dementia
- Inherited Creutzfeldt-Jakob disease
- Juvenile amyotrophic lateral sclerosis
- Kufor-Rakeb syndrome
- Kuru
- Parkinson-dementia complex of Guam
- Parkinsonian-pyramidal syndrome
- Perry syndrome
- Progressive non-fluent aphasia
- Progressive supranuclear palsy-corticobasal syndrome
- Progressive supranuclear palsy-parkinsonism syndrome
- Progressive supranuclear palsy-progressive non-fluent aphasia syndrome
- Progressive supranuclear palsy-pure akinesia with gait freezing syndrome
- Semantic dementia
- Spastic paraplegia-Paget disease of bone syndrome
- Spinocerebellar ataxia with axonal neuropathy type 2
- Young-onset Parkinson disease

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
AARS2	100.00	1	NM_020745.4
ABAT	100.00	1	NM_020686.6
ABCB7	100.00	1	NM_001271696.3
ABCD1	100.00	1	NM_000033.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ADPRS	100.00	1	NM_017825.3
AFG3L2	100.00	1	NM_006796.3
ANG	100.00	1	NM_001145.4
ANXA11	100.00	1	NM_145868.2
AP5Z1	100.00	1	NM_014855.3
APOE	100.00	1	NM_000041.4
APP	100.00	1	NM_000484.4
ARSA	100.00	1	NM_000487.6
ATP13A2	100.00	1	NM_022089.4
ATP1A3	100.00	1	NM_152296.5
ATP6AP2	100.00	1	NM_005765.3
C19ORF12	100.00	1	NM_031448.6
C9ORF72	100.00	1	NM_018325.5
CCNF	100.00	1	NM_001761.3
CHCHD10	100.00	1	NM_213720.3
CHCHD2	100.00	1	NM_016139.4
CHMP2B	100.00	1	NM_014043.4
CLN3	100.00	1	NM_001042432.2
CLN5	100.00	1	NM_006493.4
CLN6	100.00	1	NM_017882.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
CLN8	100.00	1	NM_018941.4
COA7	100.00	1	NM_023077.3
COASY	100.00	1	NM_025233.7
CRAT	100.00	1	NM_000755.5
CSF1R	100.00	1	NM_005211.3
CTSD	100.00	1	NM_001909.5
CTSF	100.00	1	NM_003793.4
DCTN1	100.00	1	NM_004082.4
DNAJC13	100.00	1	NM_015268.4
DNAJC5	100.00	1	NM_025219.3
DNAJC6	100.00	1	NM_001256864.2
EIF4G1	100.00	1	NM_198241.3
ERBB4	100.00	1	NM_005235.3
FA2H	100.00	1	NM_024306.5
FBXO7	100.00	1	NM_012179.4
FIG4	100.00	1	NM_014845.6
FTL	100.00	1	NM_000146.4
FUS	100.00	1	NM_004960.4
FXN	100.00	1	NM_000144.5
GBA1	100.00	1	NM_000157.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
GCH1	100.00	1	NM_000161.3
GIGYF2	100.00	1	NM_001103146.3
GLUD2	100.00	1	NM_012084.4
GRID2	100.00	1	NM_001510.4
GRN	100.00	1	NM_002087.3
IREB2	100.00	1	NM_004136.4
ITM2B	100.00	1	NM_021999.5
KIF5A	100.00	1	NM_004984.4
KLC4	100.00	1	NM_201521.3
LRRK2	100.00	1	NM_198578.4
MAPT	100.00	1	NM_001123066.3
MATR3	100.00	1	NM_018834.6
MFSD8	100.00	1	NM_001371596.2
NEFH	100.00	1	NM_021076.4
NEK1	100.00	1	NM_001199397.3
NOTCH3	100.00	1	NM_000435.3
NPC1	100.00	1	NM_000271.5
NPC2	100.00	1	NM_006432.5
OPTN	100.00	1	NM_001008212.2
PANK2	100.00	1	NM_153638.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
PARK7	100.00	1	NM_007262.5
PFN1	100.00	1	NM_005022.4
PGAP1	100.00	1	NM_024989.4
PINK1	100.00	1	NM_032409.3
PLA2G6	100.00	1	NM_003560.4
PODXL	94.00	1	NM_001018111.3
POLG	100.00	1	NM_001126131.2
PPT1	100.00	1	NM_000310.4
PRKN	100.00	1	NM_004562.3
PRNP	100.00	1	NM_000311.5
PRPH	100.00	1	NM_006262.4
PSEN1	100.00	1	NM_000021.4
PSEN2	100.00	1	NM_000447.3
RAB18	100.00	1	NM_021252.5
REPS1	100.00	1	NM_001286611.1
SETX	100.00	1	NM_015046.7
SIGMAR1	100.00	1	NM_005866.4
SLC6A3	100.00	1	NM_001044.5
SNCA	100.00	1	NM_000345.4
SOD1	100.00	1	NM_000454.5

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
SPG11	100.00	1	NM_025137.4
SPG21	100.00	1	NM_016630.7
SQSTM1	100.00	1	NM_003900.5
SYNJ1	100.00	1	NM_003895.3
TARDBP	100.00	1	NM_007375.4
TBK1	100.00	1	NM_013254.4
TUBA4A	100.00	1	NM_006000.3
UBQLN2	100.00	1	NM_013444.3
UBTF	100.00	1	NM_014233.4
UCHL1	100.00	1	NM_004181.5
VAPB	100.00	1	NM_004738.5
VCP	100.00	1	NM_007126.5
VPS13C	100.00	1	NM_020821.3
VPS35	100.00	1	NM_018206.6
WDR45	100.00	1	NM_001029896.2