In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic
81405, 81406, 81407 are common in some Asian populations. For patients with
6 weeks
insertion is available as a
136,3106-3118. Insertion is recommended. Analysis for the
insertion is recommended. Analysis for the
Next Generation Sequencing:
detection. Large deletions/duplications will not be detected by this analysis. Results of
molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype.
variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown
significance. Variants of unknown significance may require further studies of the patient and/or family members.
Disorders that affect the nervous system include a large group of conditions with genetic and phenotypic heterogeneity. As a group, neurological disorders often have overlapping clinical features, such as intellectual disability, seizures, microcephaly, and motor disability. Other characteristics may include brain malformations (lissencephaly, molar tooth sign), vision loss, speech difficulties, and respiratory failure. This wide phenotypic spectrum can make diagnosis challenging, but obtaining a specific diagnosis is important for prognosis, patient management, and development of therapeutic strategies.
Note: This test does not detect the retrotransposon insertion in the 3' UTR of the FKTN gene common in some Asian populations. For patients with suspected Fukuyama congenital muscular dystrophy, testing for the FKTN insertion is recommended. Analysis for the FKTN insertion is available as a separate assay.
References:

Genes
ACTB, ACTG1, ADGRG1, ADSL, AH1, ALDH5A1, ALDH7A1, ARFGEF2, ARHGEF9, ARX, ASPM, ATP1A2, ATP6AP2, ATR, ATRX, BCKDK, CACNB4, CASK, CC2D2A, CDC6, CDK5RAP2, CDKL5, CDT1, CENPJ, CEP135, CEP152, CEP290, CEP41, CEP63, CHMP1A, CHRNA2, CHRNA4, CHRNA8, CLN3, CLN5, CLN8, CLN9B, CNTNAP2, CPA7, CUB, CTSQ, CYD5, CYP27A1, DCX, DHCRI7, DISP1, DNAJC5, EFHC1, EHT1, EOMES, EPM2A, EXOSC3, FGFB, FKBP, FKTN, FLNA, FOLR1, FOXL1, FOXH1, GABRA1, GABRG2, GMAT, Gatm, GLI2, GOSR2, GRIN2A, GRIN2B, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCTD7, KIF18B, KIF7, KN1, LMHC3, LARGE1, LGI1, LIAA, MAGI2, MAPK10, MBD5, MCH1, MECP2, MEF2C, MFS8, MKS1, MYCN, NDE1, NELRC1, NIN, NODAL, NPH1P1, NRXN1, OPHN1, ORC1, ORC6, PAFAHIB1, PCDH19, PCNT, PLCB1, PNKP, PNPO, POC1A, POLG, POMIFNT1, POMT1, POMT2, PPT1, POGP1, PRICKLE1, PRRT2, PTCH1, RAB18, RAB3GAP1, RAB3GAP2, RARS2, RBSP8, RELN, RPRGIF1P1, RTTN, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SHH, SIX3, SLC19A3, SLC25A19, SLC25A22, SLC2A1, SLC3A2, SPTAN1, SRPX2, ST3GA3L, ST3GAL5, STIL, STXB1, SYN1, TBC1D24, TCF4, TGF1, TMEM138, TMEM216, TMEM237, TMEM67, TPP1, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2B, TUBB3, UBE3A, VCP, VDLR, VRK1, WDR62, ZEB2, ZIC2, ZNF335

Indications
This test is indicated for:
- Confirmation of a clinical diagnosis of neurological disorders.

Methodology
Next Generation Sequencing: In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

Detection
Next Generation Sequencing: Clinical Sensitivity: Unknown. Pathogenic variants in the promoter region, some pathogenic variants in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype.

Analytical Sensitivity: ~99%.

Specimen Requirements
Submit only 1 of the following specimen types

Type: Whole Blood
Specimen Requirements:
In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml.

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 60 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**

- Brain Malformations Panel
- Seizure Disorders Panel
- Neurology: Deletion/Duplication Panel