In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic insertion is recommended. Analysis for the gene common in some Asian populations. For patients with 6 weeks molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype. Other regulatory element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of Next Generation Sequencing:

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 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 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, ADSS, AH1, ALDH5A1, ALDH7A1, ARFGEF2, ARHGEF9, ARX, ASPM, ATP1A2, ATP6AP2, ATR, ATRX, BCKDK, CACNB4, CASK, CC2D2A, CD6B, CD5SRAP2, CDL5, CDT1, CENPJ, CEP135, CEP152, CEP290, CEP41, CEP63, CHMP1A, CHRNA2, CHRNA4, CHRNA8, CLN3, CLN5, CLN8, CLN18, CNTNAP2, CPA6, CSTB, CTSD, CYPID2, DCX, DHCR7, DISP1, DNAJC5, EFCB1, EHM1, EOMES, EPM2A, EXOSC3, FGFB, FKBP, FKTN, FLNA, FOLR1, FOXL1, FOXH1, GABRA1, GABRG2, GAMT, GATM, GL2, GOSR2, GRIN2A, GRIN2B, KCNJ10, KCNMA1, KCNN2, KCNQ3, KCNT1, KCTD7, KIF18B, KIF24, KLI1, LAMC3, LARGE1, LGI1, LIAS, MAGI2, MAPK10, MBDS, MCPH1, MECP2, MEF2C, MFSDF8, MKS1, MYCEN, NDE1, NHLRC1, NIN, NODAL, NP4P1, NRPN1, OPHN1, ORC4, ORC6, PAFAH1B1, PCDH19, PCNT, PLOC1, PNKP, PONC1, POLG, POMGNT1, POMT1, POMT2, PPT1, PGBP1, PRICKLE1, PRRT2, PTCH1, Rab1B, Rab3GAP1, Rab3GAP2, RARS2, RBBP8, RELN, RPRGIF1, RTTN, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SHH, SIX3, SLC19A3, SLC25A19, SLC25A22, SLC2A1, SLC39A, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STIL, STXBP1, SYN1, TBC1D24, TCF4, TGF1, TMEM138, TMEM216, TMEM237, TMEM67, TTP1, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2B, TUBB3, UBE3A, VCP, VLDLR, VRK1, WDR62, ZEB2, ZIC2, ZNF335

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of neurological disorders.

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.
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