Neurology: Deletion/Duplication Panel

Test Code: MD280
Turnaround time: 2 weeks
CPT Codes: 81228 x1, 81403 x1, 81404 x1, 81405 x1, 81406 x1

Condition Description

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 in the context of the patient's clinical and/or biochemical phenotype may be crucial for prognosis, patient management, and development of therapeutic strategies.

References:

Genes

ACTB, ACTG1, ADGRG1, ADGRV1, ADSL, AH1, 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, CLN6, CLN8, CLN10, CNTNAP2, CPAP, CSTB, CTSD, DCX, DHCR7, DISP1, DNAJC5, EFHC1, EHMT1, EOMES, EPM2A, EXOSC3, FGF8, FKRP, FKTN, FLNA, FOLR1, FOXG1, FOXH1, GABRA1, GABRG2, GAMT, GATM, GLI2, GOSP2, GRIN2A, GRIN2B, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KIF7, KNL1, LAMC3, LARGE1, LGI1, LIAS, MAGI2, MAPK10, MBDS, MCHP1, MECP2, MEF2C, MFSD8, MKS1, MYCN, NDE1, NLRRC1, NIN, NODAL, NPHP1, NRXN1, OPHN1, ORC4, ORC6, PAFAH1B1, PCDH19, PCNT, PLCB1, PNNP, PNO, POA1, POLG, POMGNT1, POMT1, POMT2, PPT1, PRICKLE1, PTC1, RAB18, RAB3GAP1, RAB3GAP2, RARS2, RBBP8, RELN, RGRIPI1L, RTTN, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SHH, SIX3, SLC19A3, SLC25A19, SLC25A22, SLC2A1, SLC9A6, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STIL, STXB1, SYN1, TBC1D24, TCF4, TGF1, TMEM138, TMEM216, TMEM237, TMEM67, TPP1, TSC1, TSC2, TSEN2, TSEN34, TSEN35, TUBA1A, TUBA8, TUBB2B, TUBB3, UBE3A, VLDLR, VRK1, WDR62, ZEB2, ZIC2, ZNF335

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of neurological disorders.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region. Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

Detection

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

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: 10 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

- Neurology: Sequencing Panel
- Brain Malformations: Sequencing Panel
- Epilepsy and Seizure Disorders: Sequencing Panel