

Neurology: Deletion/Duplication Panel

Test Code: MD280

Turnaround time: 2 weeks

CPT Codes: 81404 x1, 81405 x1, 81406 x1, 81228 x1, 81403 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 is important for prognosis, patient management, and development of therapeutic strategies.

References:

- Bras et al., (2012) *Nature Reviews Neuroscience*, 13:453-463.
- Nemeth et al., (2013) *Brain*, 136:3106-3118.

Genes

[ACTB](#), [ACTG1](#), [ADGRG1](#), [ADGRV1](#), [ADSL](#), [AHL1](#), [ALDH7A1](#), [AREGEE2](#), [ARHGEE9](#), [ARX](#), [ASPM](#), [ATP1A2](#), [ATP6AP2](#), [ATR](#), [ATRX](#), [BCKDK](#), [CACNB4](#), [CASK](#), [CC2D2A](#), [CDC6](#), [CDK5RAP2](#), [CDKL5](#), [CDT1](#), [CENPJ](#), [CEP135](#), [CEP152](#), [CEP290](#), [CEP41](#), [CEP63](#), [CHMP1A](#), [CHRNA2](#), [CHRNA4](#), [CHRN2](#), [CLN3](#), [CLN5](#), [CLN6](#), [CLN8](#), [CNTNAP2](#), [CPA6](#), [CSTB](#), [CTSD](#), [DCX](#), [DHCR7](#), [DISP1](#), [DNAJC5](#), [EFHC1](#), [EHMT1](#), [EOMES](#), [EPM2A](#), [EXOSC3](#), [EGF8](#), [FKRP](#), [FKTN](#), [FLNA](#), [FOLR1](#), [FOXG1](#), [FOXH1](#), [GABRA1](#), [GABRG2](#), [GAMT](#), [GATM](#), [GLI2](#), [GOSR2](#), [GRIN2A](#), [GRIN2B](#), [KCNJ10](#), [KCNMA1](#), [KCNQ2](#), [KCNQ3](#), [KCNT1](#), [KCTD7](#), [KIF7](#), [KIFBP](#), [KNL1](#), [LAMC3](#), [LARGE1](#), [LGI1](#), [LIAS](#), [MAGI2](#), [MAPK10](#), [MBD5](#), [MCPH1](#), [MECP2](#), [MEF2C](#), [MFSD8](#), [MKS1](#), [MYCN](#), [NDE1](#), [NHLRC1](#), [NIN](#), [NODAL](#), [NPHP1](#), [NRXN1](#), [OPHN1](#), [ORC4](#), [ORC6](#), [PAFAH1B1](#), [PCDH19](#), [PCNT](#), [PLCB1](#), [PNKP](#), [PNPO](#), [POC1A](#), [POLG](#), [POMGNT1](#), [POMT1](#), [POMT2](#), [PPT1](#), [POBP1](#), [PRICKLE1](#), [PTCH1](#), [RAB18](#), [RAB3GAP1](#), [RAB3GAP2](#), [RARS2](#), [RBBP8](#), [RELN](#), [RPGRI1](#), [RTTN](#), [SCARB2](#), [SCN1A](#), [SCN1B](#), [SCN2A](#), [SCN8A](#), [SCN9A](#), [SHH](#), [SIX3](#), [SLC19A3](#), [SLC25A19](#), [SLC25A22](#), [SLC2A1](#), [SLC9A6](#), [SPTAN1](#), [SRPX2](#), [ST3GAL3](#), [ST3GAL5](#), [STIL](#), [STXBP1](#), [SYN1](#), [TBC1D24](#), [TCF4](#), [TGIF1](#), [TMEM138](#), [TMEM216](#), [TMEM237](#), [TMEM67](#), [TPP1](#), [TSC1](#), [TSC2](#), [TSEN2](#), [TSEN34](#), [TSEN54](#), [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.

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: DNA, Isolated

Specimen Requirements:

Microtainer

3µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Type: Whole Blood (EDTA)

Specimen Requirements:

EDTA (Purple Top)

Infants and Young Children (2 years of age to 10 years old: 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

Related Tests

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