Brain Malformations: Deletion/Duplication Panel

**Test Code:** MD250  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81403 x1, 81404 x1

### Condition Description

Many genes are associated with Mendelian forms of malformations of brain development. Genes involved in the malformations in the brain show locus heterogeneity and have been typically identified in small kindred sizes. Therefore, the diagnostic classifications can be difficult and may not reflect the molecular pathogenesis. Pathogenic variants have been detected in more than 51 genes and are known to cause lissencephaly, cerebellar hypoplasia disorders, and Joubert syndrome. These disorders can be inherited in an autosomal dominant, autosomal recessive, or X-linked manner.

**References:**

**Genes**

ACTB, ACTG1, ADGRG1, AHI1, ARFGEF2, ARX, CASK, CC2D2A, CEP290, CEP41, CHMP1A, DCX, EOMES, EXOSC3, FKRP, FKTN, FLNA, KIF1B, KIF7, LAMC3, LARGE1, MKS1, NPHP1, OPHN1, PFAH1B1, POMGNT1, POMT1, POMT2, POBP1, RAB3GAP1, RAB3GAP2, RARS2, RELN, RPGRIP1L, RTTN, SPPX2, TMEM138, TMEM216, TMEM237, TMEM67, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2B, TUBB3, VLDLR, VRK1, WDR62

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of a brain malformation.

### 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

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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: Sequencing Panel