Macrocephaly and Overgrowth Syndromes: Deletion/Duplication Panel

Test Code: MD260  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1, 81405 x1

**Condition Description**

Macrocephaly is defined as a head circumference, which is 2 standard deviations larger than the average when matched for age and sex. It refers to an abnormally large head inclusive of the scalp, cranial bone, and intracranial contents. Macrocephaly can arise due to a true enlargement of the brain (megalencephaly) or other conditions such as hydrocephalus and be either syndromic or non-syndromic. The genetic subtypes of macrocephaly include familial forms of macrocephaly, autism, syndromic associations such as PTEN hamartoma syndrome, Noonan syndrome, Sotos syndrome, and metabolic disorders such as glutaric aciduria type 1 and D-2-hydroxyglutaric aciduria.

Reference:


**Genes**

- CUL4B, EZH2, GLI3, GPC3, MED12, NFIX, NSD1, PHF6, PTCH1, PTEN, UPF3B

**Indications**

This test is indicated for:

- Patients with macrocephaly.

**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™ 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**

- Macrocephaly: Sequencing Panel