Peters Plus Syndrome: B3GLCT Gene Deletion/Duplication

Test Code: DB3GA
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Peters plus syndrome is characterized by developmental delay/intellectual disability, disproportionate short stature, cleft lip and/or palate, and anterior chamber eye anomalies with the most common being Peters' anomaly. Typical facial features include a cupid's bow-shaped upper lip, narrow palpebral fissures, a prominent forehead, and a long philtrum. Mutations in the B3GLCT gene (13q12.3) cause the autosomal recessive Peters plus syndrome. Many individuals with Peters plus syndrome have the common c660+1G>A mutation in the B3GLCT gene. Sequencing analysis is recommended before deletion/duplication analysis.

References:
- GeneReviews
- OMIM #610308: B3GLCT gene
- OMIM #261540: Peters plus syndrome

Genes
B3GLCT

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of Peters plus syndrome in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of Peters plus syndrome in whom sequence analysis was negative.

Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Detection

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 (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 24 hours of collection. Do not refrigerate or freeze.

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.

Related Tests
• Sequence analysis for the B3GLCT gene is also available.

• Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.

• Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.