Peters Plus Syndrome: B3GLCT Gene Deletion/Duplication

Test Code: DB3QA
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.

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

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

* Preferred specimen type: Whole Blood

Type: Whole Blood

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
Infants (2 years): 3-5 ml
Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

Type: Saliva

Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.
Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection 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.