Uniparental Disomy of Chromosome 14 (UPD14): Methylation Analysis

Test Code: OO
Turnaround time: 3 weeks
CPT Codes: 81402 x1

Condition Description

The clinical phenotypes for maternal and paternal UPD14 are caused by dysregulation of maternally and paternally imprinted genes. Inheritance of both copies of chromosome 14 from the mother (matUPD14) results in pre- and post-natal growth failure, developmental delay, hypotonia, obesity, short stature, small hands and feet, and early onset of puberty [1,2]. Inheritance of both copies of chromosome 14 from the father (patUPD14) results in a more severe phenotype with facial abnormalities, severe neurological involvement, a bell-shaped thorax, abdominal wall defects, growth retardation, and polyhydramnios [2].

This analysis uses methylation-specific PCR for a differentially methylated region (DMR), upstream of the MEG3 gene, on chromosome 14q32.2. In cases where there is biparental inheritance of chromosome 14, epimutations and deletions within the imprinted region can result in a UPD14-like phenotype, which will not be detected by conventional UPD14 analysis.

References:

Indications

This test is indicated for:

- Confirmation of a clinical/biochemical diagnosis of matUPD14 or patUPD14.

Methodology

DNA methylation specific PCR assay targeting the differentially methylated region (DMR) upstream of the MEG3 gene on chromosome 14q32.2 is used to test for maternal or paternal uniparental disomy of chromosome 14 (matUPD14, patUPD14). Parental samples are NOT required for patUPD14 analysis, but may be requested to confirm a diagnosis.

Detection

Specimen Requirements

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

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Special Instructions

Submit copies of diagnostic biochemical test results with the sample, if appropriate. Contact the laboratory if further information is needed.

Related Tests