Multiple Sulfatase Deficiency: SUMF1 Gene Deletion/Duplication

**Condition Description**

Multiple sulfatase deficiency (MSD) is an autosomal recessive inborn error of metabolism. Enzyme activity of all sulfatases is reduced or absent in MSD. The deficiency of the entire enzyme family is caused by a defect affecting a posttranslational modification of sulfatases that is required for catalytic activity. Mutations in the SUMF1 gene (3p26.1) cause MSD.

MSD has similar clinical characteristics to other sulfatase deficiencies, such as metachromatic leukodystrophy, the mucopolysaccharidoses, chondrodysplasia punctata type I, and X-linked ichthyosis. Clinical features include neurological deterioration, developmental delay, dysmorphism, organomegaly, skeletal abnormalities, and skin findings. The age of onset of features of MSD distinguish the subtypes: neonatal, late infantile, and juvenile disease. While neonatal MSD has the most severe phenotype, the majority of MSD cases are late infantile.

For patients with suspected MSD, sequence analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

**References:**
- OMIM #607939: SUMF1 gene
- OMIM #272200: MSD

**Genes**

SUMF1

**Indications**

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

**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 72 hours of collection. Do not 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.

**Special Instructions**

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition.
Related Tests

- Sequence analysis of the *SUMF1* gene is available and is required before deletion/duplication analysis.
- 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.