Multiple Sulfatase Deficiency: *SUMF1* Gene Deletion/Duplication

**Test Code:** DSUMF  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### 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. 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.

### Specimen Requirements

#### Additional Specimen Collection/Handling Instructions Required for this Test

Arylsulfatase A and B enzyme activity studies are recommended. To order Arylsulfatase A enzyme activity, please order Metachromatic Leukodystrophy, test code LA. To order Arylsulfatase B enzyme activity, please order Mucopolysaccharidosis Type VI, Arylsulfatase B enzyme activity, test code BMPS6.

Additionally, GAGs may be ordered, test code GA.

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

### 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.

### 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.

**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.