

Smith-Magenis Syndrome: *RAI1* Gene Deletion/Duplication

Test Code: DRAI1

Turnaround time: 2 weeks

CPT Codes: 81228 x1

Condition Description

Smith-Magenis syndrome (SMS) is characterized by behavioral abnormalities, including the “self-hug” and “lick and flip” behaviors, significant sleep disturbances, and self-injurious behaviors; distinctive facial features that progress with age, mild to moderate intellectual disability, and developmental delay. Additionally, individuals with SMS have mild to moderate infantile hypotonia with feeding difficulties and failure to thrive, minor skeletal anomalies, short stature, eye abnormalities, otolaryngologic abnormalities, early speech delays with or without hearing loss, and peripheral neuropathy. SMS is caused by deletions or mutations of the *RAI1* (17p11.2) gene.

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

References:

- GeneReviews
- [OMIM #607642](#): *RAI1* gene
- [OMIM #182290](#): SMS

Deletion/Duplication testing should be ordered as the first tier test.

Genes

[RAI1](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Smith-Magenis syndrome.
- Carrier testing in adults with a family history of Smith-Magenis syndrome.

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

~90% are detected by deletion/duplication analysis. 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 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/ μ L in TE buffer. Ship sample at room temperature with overnight delivery.

Related Tests

- Sequence analysis of the *RAI1* gene is available for those individuals in whom deletion/duplication analysis is negative.
- 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.