Stocco Dos Santos XLMR Syndrome: SHROOM4 Gene Deletion/Duplication

Test Code: DKIAA
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

A mutation in the SHROOM4 gene (Xp11.2) has been reported in a Brazilian family in which males had severe intellectual disability, bilateral congenital hip luxation and short stature. The SHROOM4 gene was also found to be disrupted in two unrelated females with mild to moderate intellectual disabilities. Other features included delayed or no speech, seizures, kyphosis and hyperactivity. Carrier females displayed seizures and depression. No mutations in SHROOM4 were identified in more than 1000 control X chromosomes.

For patients with suspected Stocco Dos Santos X-linked mental retardation syndrome, 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:


Genes

KIAA1202, SHROOM4

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Stocco Dos Santos X-linked mental retardation syndrome in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of Stocco Dos Santos X-linked mental retardation 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.

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

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 24 hours of collection. Do not refrigerate or 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

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

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 SHROOM4 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.