Beta-Mannosidosis: MANBA Gene Deletion/Duplication

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

Condition Description

Beta-mannosidosis is a rare autosomal recessive disorder that is due to deficiency in the lysosomal enzyme beta-mannosidase. The enzyme is responsible for catalyzing the removal of mannose sugar residues from proteins that contain sugar groups (called glycoproteins), such as oligosaccharides. Deficiency of the beta-mannosidase activity results in accumulation of mannose-rich oligosaccharides chains, leading to swelling of the lysosome and impairment of normal cellular functions.

Patients with beta-mannosidosis have coarse facial features, mild bone disease, delayed speech development, hyperactivity, and mental retardation (1). There is significant variability in clinical presentation.

Mutations in the MANBA are responsible for beta-mannosidosis (2). Only 13 cases in 10 families have been identified with beta-mannosidosis. Two mutations in the MANBA gene have been described thus far (3).

For questions about testing for beta-mannosidosis, call EGL Genetics at (470) 378-2200 or 855-831-7447. For further clinical information about lysosomal storage diseases, including management and treatment, call the Emory Lysosomal Storage Disease Center at (404) 778-8565 or (800) 200-1524.

References:

Genes

MANBA

Indications

- Confirmation of a clinical diagnosis of beta-mannosidosis
- Prenatal testing for known familial mutation(s).
- Assessment of carrier status in high risk family members - known mutation analysis.

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.

Detection

Detection is limited to duplications and deletions. Array CGH will not detect point mutations 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

* 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.

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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

Submit copies of diagnostic biochemical test results with the sample. Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside EGL Genetics, please submit a copy of the sequencing report with the test requisition. Contact the laboratory if further information is needed.

Related Tests

- Known mutation analysis (Custom Diagnostics) is available to test family members.
- Prenatal testing is available for known familial mutations only. Please call the Laboratory Genetic Counselor for specific requirements for prenatal testing before collecting a fetal sample.