Hypohidrotic Ectodermal Dysplasia: Deletion/Duplication Panel

Test Code: MD420
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Most cases of hypohidrotic ectodermal dysplasia are thought to be caused by mutations in the \textit{EDA}, \textit{EDAR}, and \textit{EDARADD}.

These genes produce proteins that form part of a signaling pathway critical for the interaction between the ectoderm and mesoderm cell layers. Ectoderm-mesoderm interactions are essential for the formation of several structures that arise from the ectoderm, including the skin, hair, nails, teeth, and sweat glands.

Hypohidrotic ectodermal dysplasia has several different inheritance patterns. Most cases are caused by mutations in the \textit{EDA} gene, which are inherited in an X-linked recessive pattern. \textit{EDAR} mutations can have an autosomal dominant or autosomal recessive inheritance pattern, while \textit{EDARADD} mutations have an autosomal recessive inheritance pattern.

Reference:
- GeneReviews.

Genes

\textit{EDA}, \textit{EDAR}, \textit{EDARADD}

Indications

The test is indicated for:
- Individuals with a clinical or suspected diagnosis of hypohidrotic ectodermal dysplasia.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that 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

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

\textbf{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: Ship sample at room temperature with overnight delivery.

\textbf{Type: Isolated DNA}

Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.
Related Tests

- Hypohidrotic Ectodermal Dysplasia: Sequencing Panel