Hyper IgE Syndromes: Deletion/Duplication Panel

Test Code: MD370
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Hyper IgE syndromes (HIES) are characterized by highly elevated levels of IgE, eczema, and recurrent skin and respiratory tract infections. An autosomal dominant form of HIES is caused by pathogenic variants in \textit{STAT3} and also involves connective tissue, vascular, and skeletal abnormalities. The autosomal recessive form is caused by pathogenic variants in \textit{DOCK8} and causes an increased incidence of neurological abnormalities and viral infections of the skin. Other disorders with elevated IgE and similar symptoms include tyrosine kinase 2 deficiency caused by pathogenic variants in \textit{TYK2} and Netherton syndrome caused by pathogenic variants in \textit{SPINK5}.

References:
- GeneReviews.
- OMIM.

Genes

\textit{DOCK8, SPINK5, STAT3, TYK2}

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of Hyper IgE Syndromes.

Methodology

\textbf{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

\textbf{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\textsuperscript{TM} 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

- Hyper IgE Syndromes: Sequencing Panel