ELN-related Disorders: Deletion/Duplication Panel

Test Code: MD440
Turnaround time: 2 weeks
CPT Codes: 81228 x1

### Condition Description

Numerous genetic disorders, including connective tissue, cardiac, pulmonary, and seizure, are associated with pathogenic variants found in the *ELN* gene. This gene provides instructions for making proteins called tropoelastin. These proteins attach to one another and are processed to form a mature protein called elastin. Elastin is the major component of elastic fibers; slender bundles of proteins that provide strength and flexibility to connective tissue. Elastic fibers are found in the intricate lattice that forms in the spaces between cells, where they give structural support to organs and tissues such as the heart, skin, lungs, ligaments, and blood vessels.

Reference:
- GeneReviews.

### Genes

**ELN**

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of an *ELN*-related disorder.

### 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

**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.

**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

- *ELN*-related Disorders: Sequencing Panel