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.

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 (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.

Related Tests

- ELN-related Disorders: Sequencing Panel