### Condition Description

Mutations in the \( NKX2-5 \) gene have been associated with atrioventricular (AV) conduction block, septal defects, conotruncal abnormalities (particularly Tetrology of Fallot), and AV valve formation defects. Mutations in \( NKX2-5 \) have been observed in autosomal dominant pedigrees and isolated cases of congenital heart disease. Studies suggest that \( NKX2-5 \) mutation may be a frequent cause (up to 4%) of sporadic and familial congenital heart defects.

\( NKX2-5 \) mutation analysis is appropriate for patients with an atrioventricular conduction block or structural heart defects with or without a family history of congenital heart defects. Analysis includes sequencing the entire \( NKX2-5 \) coding region (2 exons) and immediate exon/intron boundaries. Mutations in other regulatory regions and large deletions will not be detected by this assay. Variants of unknown clinical significance may be detected. Custom mutation detection is available for known familial mutations.

### Genes

**\( NKX2-5 \)**

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of \( NKX2-5 \)-related congenital heart disease in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of \( NKX2-5 \)-related congenital heart disease in whom sequence analysis was negative.

### 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.

### Detection

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: 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.

#### 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 72 hours of collection. Do not freeze.

### Special Instructions

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