STAT FISH: 22q11 Microdeletion

Condition Description

Analysis by Fluorescence In Situ Hybridization (FISH) allows for the most rapid detection of the most common chromosome deletion syndromes. Results can typically be reported in 24-48 hours from the time of receipt.

Individuals with 22q11.2 deletion syndrome have a range of features including congenital heart defects, palate abnormalities, feeding difficulties, characteristic facial features, hypocalcemia and learning disabilities. 75% of individuals with 22q11.2 deletion syndrome also have immune deficiency and recurrent infections. Characteristic facial features can include auricular abnormalities, hooded eyelids, hypertelorism, cleft lip and palate. However, in individuals with 22q11.2 deletion syndrome, features can vary widely, even among affected family members.

Children with 22q11.2 deletion syndrome can have developmental delays. They often achieve motor and speech milestones later than children in the general population. The mean age for walking is 18 months and many 2-3 year olds are nonverbal. They also can have learning disabilities and are more likely to have attention deficit hyperactivity disorder (ADHD). Recent studies have shown an increase in the rates of mental illness such as bipolar disorder and schizophrenia in these individuals as they age.

22q11.2 deletion syndrome is inherited in an autosomal dominant manner. Approximately 93% of cases are a de novo occurrence and approximately 7% of cases are inherited from a parent. In inherited cases, other family members may also have features of the syndrome as well, but may not be diagnosed. This deletion is present in approximately 1 in 4000 live births; however, because of the variability seen with this condition, it may be under diagnosed.

References:
- 22q11.2 deletion syndrome

Indications

This test is appropriate for the following indications:

- Cleft palate
- Characteristic facial features such as hypertelorism
- Heart defect
- Learning disabilities
- Immune deficiencies
- Autism spectrum disorder

Methodology

Detection

FISH is very sensitive in the detection of 22q microdeletions. This probe set is specific to chromosome 22 and other types of chromosomal imbalances will not be detected. Validation for specificity and sensitivity are performed on each probe. Control probes are present in all probe sets.

Specimen Requirements

Type: Whole Blood

Specimen Requirements:

In sodium heparin (green top) tube:
- Infants (Children less than 6 months): 3-5 ml
- Older Children & Adults: Metaphase FISH analysis is required

Specimen Collection and Shipping: Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Related Tests

- Chromosomal Microarray, EmArray Cyto (VA)
- Chromosome Analysis (CA/CB)