Date : January 28, 2023

GENOMICS

Change in 22q11.2 testing

Date effective: June 27, 2023

Background Information:

The chromosome 22q11.2 region contains a cluster of repetitive sequences called low-copy repeats (LCRs) that result in recurrent microdeletions and microduplications of various sizes. The most common microdeletion is approximately 3 Mb in size and results in chromosome 22q11.2 deletion syndrome (previously known as DiGeorge syndrome OMIM #188400 or velocardiofacial syndrome OMIM #192430). The reciprocal microduplication results in chromosome 22q11.2 duplication syndrome (OMIM #608363). Traditionally, fluorescent in situ hybridization (FISH) has been used to detect these copy number variants.

Change in Test Procedure:

- Due to the recent repatriation of chromosomal microarray (CMA), the 22q11.2 FISH test for blood specimens has been replaced with this targeted microarray analysis.
- This targeted microarray test examines a larger portion of the 22q11.2 region, allowing for a higher detection rate.

References/Resources:

- Burnside R.D. 2015;146(2):89-99. PMID: 26278718

Patient Impact:

- Higher detection rate

System Improvements:

- Patients with smaller atypical or nested copy number variants will be identified.

More Information:

- GeneReviews 22q11.2 Deletion Syndrome at https://www.ncbi.nlm.nih.gov/books/NBK1523/
- Lab Information Manual (LIM) at https://apps.sbgh.mb.ca/labmanual/test/view?seedId=34840

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