Rare Disease
Copy Number Analysis

TEST CODE: PR51001, PR51333

Overview

This chromosomal microarray (CMA) replacement test produces indication-based testing results in reports with variants of the highest relevance to the patient's symptoms and phenotypes. MyOme uses whole-genome sequencing (WGS) to detect genome-wide copy number variants (CNVs) with high coverage and resolution. The whole-genome backbone enables the ability to add on Fragile X syndrome analysis to the initial order and reflex to exome or genome analysis.

Clinical Use

This test can be used as a primary screening tool for individuals with neurodevelopment disorders, multiple congenital anomalies, or suspected chromosomal imbalances. This test can be used as a replacement for traditional CMA testing.

Method

- PCR-free library preparation with 2x150 base pair (bp) paired-end WGS of genomic DNA extracted from submitted samples to an average depth of 30X or greater
- Detection of CNVs for deletions and duplications ≥50 kb
- Interpretation and reporting based on ACMG guidelines and patient clinical indication

Sample Types

- Blood (2 EDTA tubes)
- Buccal (2 swabs)
- Saliva (2 tubes)

Turnaround Times

- From sample received, most results are delivered in 5-6 weeks.*
- Follow-up testing or re-requisitions are typically completed within 2-3 weeks.

Included

- CNV report comparable to that of a CMA report
- Confirmation of all reported variants by a secondary technology
- Comprehensive report with pathogenic variants, likely pathogenic variants, and variants of uncertain significance (VUS) correlated with the patient's phenotype
- Option for *FMR1* expansion analysis (Fragile X syndrome) add-on
- Option for post-test genetic counseling

Test Performance

- 30X average coverage genome-wide
- >99.5% sensitivity for CNVs ≥50 kb in size**

* Turnaround times are provided as estimates and begin once sample(s) are processed at MyOme. Turnaround times may be extended in cases outside of MyOme's control, including delays related to confirmation testing or other unforseen circumstances. **MyOme, Inc. (Data on File)

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