



Rare Disease COPY NUMBER ANALYSIS

TEST CODE: PR5004

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 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)

Turn Around Time

- 5 to 6 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 post-test genetic counseling

Test Performance

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



Genetic Counseling & Billing Support

MyOme provides post-test genetic counseling via a third-party telehealth provider

To learn more, contact us at support@myome.com

*Turn around time starts after MyOme, Inc. receives all samples and required information; **MyOme, Inc. (Data on File)

This test was developed, and its performance characteristics were determined, by MyOme, Inc., a clinical laboratory certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) and College of American Pathologist (CAP) accredited to perform high complexity clinical laboratory testing. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). Test results should always be interpreted by a clinician in the context of clinical and familial data with the availability of genetic counseling when appropriate. MyOme is not responsible for the content or accuracy of third-party websites.