# Rare Disease EXOME ANALYSIS

## TEST CODE: PR51017, PR51025, PR51030

### Overview

MyOme uses a whole-genome sequencing (WGS) backbone to evaluate protein coding regions of the genome, allowing enhanced coverage of exonic regions and higher resolution copy number variant (CNV) analysis compared to traditional exome sequencing. The whole-genome backbone enables the ability to re-query a patient's genome as new information becomes available and to reflex to genome analysis.

### Clinical Use

This test is for individuals with clinical features suggestive of a genetic cause, including neurodevelopmental disorders, multiple congenital anomalies, and epilepsy.

### 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
- Identification of single-nucleotide variants (SNVs) and small insertions and deletions (indels) in coding regions (±20 bp) of the genome plus genome-wide CNVs
- Interpretation and reporting based on ACMG guidelines, patient clinical indication, and familial samples (when provided)

### Sample Types

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

### Turn Around Time

- 5 to 6 weeks*

### Included

- 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
- Option to receive Secondary Findings
- One complimentary reanalysis (starting one year after the initial order)

### Test Performance**

- >99.5% exonic regions covered by ≥10X
- >99% sensitivity for SNVs and indels
- 98% sensitivity for benchmark CNVs >1 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.

 