Rare Disease GENOME ANALYSIS

TEST CODE: PR51049, PR51058, PR51063

Overview

MyOme's whole-genome sequencing (WGS) evaluates protein coding and non-coding regions of the genome and includes copy number variant (CNV) analysis and the ability to re-query a patient's genome as new information becomes available.

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), small insertions and deletions (indels), and CNVs in coding regions and non-coding regions across the genome
- 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.

