Rare Disease GENOME ANALYSIS

TEST CODE: PR51224, PR51235, PR51243

Overview

MyOme's whole-genome sequencing (WGS) evaluates protein coding and non-coding regions of the genome and includes copy number variant (CNV) analysis, mitochondrial genome analysis, and select tandem repeat expansion (TRE) analysis. The whole-genome backbone enables 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 nuclear genome
- Identification of SNVs and small insertions and deletions across the mitochondrial genome
- TRE analysis in a select set of genes (see table on back for list of genes and associated reportable ranges)
- 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)

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

- Confirmation of all reported variants and TREs 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**

Nuclear Genome

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

Mitochondrial Genome

- Mean coverage depth of 3000X or greater (minimum acceptable is 1000X)

*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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TRE Reportable Ranges for Genes Analyzed

Key
= Potentially reportable based on patient phenotype
= No associated phenotype, not reportable

Gene	Pathogenic*	Premutation	Intermediate	Uncertain	Normal
AFF2	>200	61–200			≤60
AR	≥35				≤34
ATN1	≥48		36–47		≤35
ATXN1	≥39		36–38		≤35
ATXN2	≥33			31–32	≤30
ATXN3	≥60		45–59		≤44
ATXN7	≥34	28–33		20–27	≤19
ATXN8OS	≥80			51–79	≤50
C9orf72	≥60			25–60	≤24
CACNA1A	≥20	19			≤18
DIP2B	≥351			24–350	≤23
DMPK	≥50	35–49			≤34
FMR1	≥201	55–200			≤54
FXN	≥66	34–43		44–65	≤33
HTT	≥36				≤35
JPH3	≥40			29–39	≤28
LRP12	≥60			46–59	≤45
PABPN1	≥11				≤10
PPP2R2B	≥43			33–42	≤32
TBP	≥41				≤40

*Includes ranges associated with reduced penetrance

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.