myome Personal Genome™

Proactive Health Report, 81 Genes

TEST CODE: PR-2008

Overview

MyOme Proactive Health Report, 81 Genes uses a PCR-free whole genome backbone that allows identification of a range of variant types. Whole genome sequencing (WGS) allows MyOme to re-query a patient's genome as healthcare needs change and new information about the genome is discovered.

Clinical Use

Test is intended for a wellness screening of germline heritable conditions in individuals from an asymptomatic population. MyOme annotates and interprets variants according to American College of Medical Genetics (ACMG) guidelines, and reports pathogenic or likely pathogenic variants. Genetic testing may provide information to provide individual risk, support a clinical diagnosis, and assist with the development of a personalized treatment and management strategy in conjunction with standard clinical assessment.

Method

PCR-free library prep followed by 2x150 bp paired-end whole genome sequencing is the backbone for this test. In-house pipeline allows identification of single-nucleotide variants (SNVs), small insertions and deletions (indels) and copy number variants (CNVs). Variant interpretation by qualified scientists based on guidelines by the ACMG.

Sample Types

- Blood (2 EDTA tubes)
- Saliva (2 tubes)
- Buccal (3 swabs)

Turn Around Time

- From initial sample, approximately 6 to 8 weeks

Included

- Analysis of SNVs, indels and CNVs (deletions and duplications)
- Confirmation of Pathogenic/Likely Pathogenic variants by orthogonal technology (e.g. Sanger sequencing)
- Cohesive report with actionable recommendations
- **81 Genes included**: *ACTA2, ACTC1, ACVRL1, APC, APOB, ATP7B, BAG3, BMPR1A, BRCA1, BRCA2, BTD, CACNA1S, CALM1, CALM2, CALM3, CASQ2, COL3A1, DES, DSC2, DSG2, DSP, ENG, FBN1, FLNC, GAA, GLA, HFE, HNF1A, KCNH2, KCNQ1, LDLR, LMNA, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, MYBPC3, MYH11, MYH7, MYL2, MYL3, NF2, OTC, PALB2, PCSK9, PKP2, PMS2, PRKAG2, PTEN, RB1, RBM20, RET, RPE65, RYR1, RYR2, SCN5A, SDHAF2, SDHB, SDHC, SDHD, SMAD3, SMAD4, STK11, TGFBR1, TGFBR2, TMEM127, TMEM43, TNNI3, TNNC1, TNNT2, TP53, TPM1, TRDN, TSC1, TSC2, TTN, TTR, VHL, WT1*

Test Performance[1]

- 30x average genome-wide coverage
- >99.5% of exonic regions at ≥10x depth
- >99.5% ClinVar P/LP variants covered by ≥10x depth
- >99% sensitivity for SNVs and indels
- 98% sensitivity for benchmark CNVs >1 kb in size

1. MyOme Inc, Data on File.

The MyOme Personal Genome Report was developed, and its performance characteristics were determined, by MyOme, Inc., a clinical laboratory certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) to perform high complexity clinical laboratory testing. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary for laboratory-developed tests.

DISORDER-GENE RELATIONSHIP

Findings of the following 81 genes are deemed medically actionable by the Amercian College of Medical Genetics and Genomics (ACMG).[2]

Cancer

Disorder	Gene
Familial adenomatous polyposis (FAP)	*APC*
Familial medullary thyroid cancer	*RET*
Hereditary breast and/or ovarian cancer	*BRCA1, BRCA2, PALB2*
Hereditary paraganglioma-pheochromocytoma syndrome	*MAX, SDHAF2, SDHC, SDHB, SDHD, TMEM127*
Juvenile polyposis syndrome (JPS)	*BMPR1A, SMAD4*
Li–Fraumeni syndrome	*TP53*
Lynch syndrome (HNPCC)	*MLH1, MSH2, MSH6, PMS2*
Multiple endocrine neoplasia type 1	*MEN1*
MUTYH-associated polyposis (MAP)	*MUTYH*
Neurofibromatosis type 2	*NF2*
Peutz-Jeghers syndrome (PJS)	*STK11*
PTEN hamartoma tumor syndrome	*PTEN*
Retinoblastoma	*RB1*
Tuberous sclerosis complex	*TSC1, TSC2*
von Hippel-Lindau syndrome	*VHL*
WT1-related Wilms tumor	*WT1*

Inborn Errors of Metabolism

Disorder	Gene
Biotinidase deficiency	*BTD*
Fabry disease	*GLA*
Ornithine transcarbamylase deficiency	*OTC*
Pompe disease	*GAA*

Cardiovascular

Disorder	Gene
Aortopathies	*ACTA2, FBN1, MYH11, SMAD3, TGFBR1, TGFBR2*
Arrhythmogenic right ventricular cardiomyopathy	*DSC2, DSG2, DSP, PKP2, TMEM43*
Cardiomyopathy	*BAG3, DES, RBM20, TNNC1*
Catecholaminergic polymorphic ventricular tachycardia	*CASQ2, RYR2, TRDN*
Dilated cardiomyopathy	*BAG3, DES, FLNC, LMNA, RBM20, TNNC1, TNNT2, TTN*
Ehlers-Danlos syndrome, vascular type	*COL3A1*
Familial hypercholesterolemia	*APOB, LDLR, PCSK9*
Hypertrophic cardiomyopathy	*ACTC1, MYBPC3, MYH7, MYL2, MYL3, PRKAG2, TNNI3, TPM1*
Long QT syndrome types 1 and 2	*CALM1, CALM2, CALM3, KCNH2, KCNQ1*
Long QT syndrome 3, Brugada syndrome	*SCN5A*

Miscellaneous

Disorder	Gene
Hereditary hemochromatosis	*HFE*
Hereditary hemorrhagic telangiectasia	*ACVRL1, ENG*
Hereditary TTR (transthyretin) amyloidosis	*TTR*
Malignant hyperthermia	*CACNA1S, RYR1*
Maturity-onset of diabetes of the young	*HNF1A*
RPE65-related retinopathy	*RPE65*
Wilson disease	*ATP7B*

2. Miller, DT, et. Al., ACMG SF v3.2 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG), Genetics in Medicine, V25, Issue 8, Aug 2023

