Proactive Health
SINGLE-GENE RISK™
84 GENES

TEST CODE: PR22020

Overview

MyOme Proactive Health Plus Single-Gene Risk™ report, 84 Genes uses a Blended Genome-Exome backbone built from whole-exome sequencing and low coverage whole-genome sequencing (WGS) to identify a range of variant types. This allows MyOme to re-query a patient's data as healthcare needs change and new information about the genome is discovered.

Clinical Use

This 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[1], and reports pathogenic or likely pathogenic variants. Genetic testing may provide information to assess 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 whole-genome library is constructed and a sub-aliquot is taken through PCR amplification and exome selection. The blended genome-exome libraries are sequenced to generated 2x150 bp paired-end reads, resulting in low-coverage whole-genome and higher coverage exome data. In-house pipeline analysis allows identification of single-nucleotide variants (SNVs) and small insertions and deletions (indels). Variant interpretation is conducted by qualified scientists based on guidelines by the ACMG.

Sample Types

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

Turnaround Times

- From sample received, most results are delivered in 5-6 weeks.*
- Follow-up testing or re-requisitions are typically completed in under 2 weeks, often within just a few days.

Included

- Analysis of SNVs and indels
- Confirmation of Pathogenic/Likely Pathogenic variants by orthogonal method (e.g., Sanger sequencing)
- Cohesive report with actionable recommendations
- 84 genes included: *ACTA2, ACTC1, ACVRL1, APC, APOB, ATM, ATP7B, BAG3, BMPR1A, BRCA1, BRCA2, BTD, CACNA1S, CALM1, CALM2, CALM3, CASQ2, CHEK2, COL3A1, DES, DSC2, DSG2, DSP, ENG, FBN1, FLNC, GAA, GLA, HFE, HNF1A, KCNH2, KCNQ1, LDLR, LDLRAP1, 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[2]

- ≥60x average exome-wide coverage
- ≥1x average genome-wide coverage
- ≥90% of exonic regions at ≥20x depth
- >99.5% sensitivity for SNVs
- >97.5% sensitivity for indels

*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.

1. American College of Medical Genetics and Genomics. SF v3.2 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the ACMG. Genet Med. June22, 2023. doi: 10.1016/j.gim.2023.100866. **2.** MyOme Inc, Data on File

CONDITION-GENE RELATIONSHIP

The genes listed below are analyzed in this report. MyOme selected them based on our Gene Inclusion Framework Guidelines. Genes are prioritized based on clinical validity, actionability, penetrance/prevalence, and feasibility.

Cardiovascular	
Condition	**Gene(s)**
Arrhythmogenic right ventricular cardiomyopathy	*DES, DSC2, DSG2, DSP, PKP2, TMEM43*
Brugada syndrome	*SCN5A*
Catecholaminergic polymorphic ventricular tachycardia	*CALM1, CALM2, CALM3, CASQ2, RYR2, TRDN*
Dilated cardiomyopathy	*ACTC1, BAG3, DES, FLNC, LMNA, MYH7, SCN5A, TNNC1, TNNI3, TNNT2, TPM1, TTN, RBM20*
Ehlers-Danlos syndrome, vascular type	*COL3A1*
Emery-Dreifuss muscular dystrophy	*LMNA*
Fabry disease	*GLA*
Familial hypercholesterolemia	*APOB, LDLR, LDLRAP1, PCSK9*
Familial thoracic aortic aneurysm and dissection	*ACTA2, MYH11, SMAD3*
Hereditary transthyretin-related amyloidosis	*TTR*
Hypertrophic cardiomyopathy	*ACTC1, MYBPC3, MYH7, MYL2, MYL3, PRKAG2, TNNC1, TNNI3, TNNT2, TPM1*
Intrinsic Cardiomyopathy	*ACTN2, PLN*
Loeys-Dietz syndrome	*TGFBR1, TGFBR2, SMAD3*
Long QT syndrome	*CALM1, CALM2, CALM3, KCNH2, KCNQ1, SCN5A, TRDN*
Marfan syndrome	*FBN1*
Myofibrillar myopathy	*BAG3, DES, FLNC*
Short QT syndrome	*KCNH2, KCNQ1*

Other	
Condition	**Gene(s)**
Biotinidase deficiency	*BTD*
Hereditary hemochromatosis	*HFE*
Hereditary hemorrhagic telangiectasia	*ACVRL1, ENG, SMAD4*
Malignant hyperthemia	*CACNA1S, RYR1*
Monogenic diabetes	*HNF1A*
Ornithine transcarbamylase deficiency	*OTC*
Pompe disease	*GAA*
RPE65-related retinopathy	*RPE65*
Wilson disease	*ATP7B*

Cancer	
Condition	**Gene(s)**
Familial adenomatous polyposis	*APC*
Familial ovarian cancer	*PALB2*
Gastrointestinal stromal tumor	*KIT*
Hereditary breast cancer	*ATM, CHEK2, PALB2*
Hereditary breast and ovarian cancer	*BRCA1, BRCA2*
Hereditary nonpolyposis colon cancer	*ATM*
Hereditary paraganglioma-pheochromocytoma syndrome	*MAX, SDHAF2, SDHB, SDHC, SDHD, TMEM127*
Juvenile polyposis syndrome	*BMPR1A*
Juvenile polyposis with hereditary hemorrhagic telangiectasia	*SMAD4*
Li-Fraumeni syndrome	*TP53*
Lynch syndrome	*MLH1, MSH2, MSH6, PMS2*
Multiple endocrine neoplasia	*MEN1, RET*
MUTYH-associated polyposis	*MUTYH*
Neurofibromatosis type 2	*NF2*
Peutz-Jeghers syndrome	*STK11*
PTEN hamartoma tumor syndrome	*PTEN*
Retinoblastoma	*RB1*
Tuberous sclerosis complex	*TSC1, TSC2*
Von Hippel-Lindau syndrome	*VHL*
WT1-related Wilms tumor	*WT1*

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.

