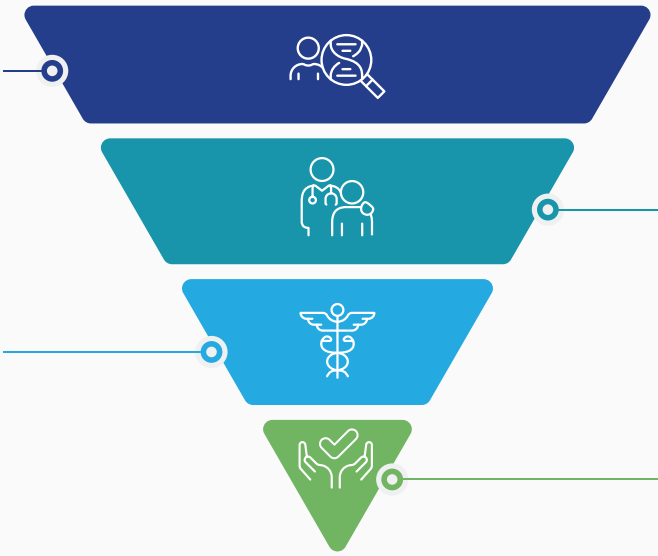
myome Personal Genome™

MyOme Personal Genome™ Gene Inclusion Framework

At MyOme, our mission is to empower healthcare providers and patients with clinically relevant, actionable insights that improve care. To achieve this, we developed a gene selection framework for the MyOme Personal Genome Proactive Health™ report, that prioritizes genes based on clinical validity, actionability, penetrance/prevalence and feasibility, along with other supporting data.

We utilized this framework to identify additional genes for version 2 of the report beyond the 81 genes deemed medically actionable by the American College of Medical Genetics and Genomics (ACMG)[1]. Below are the selection criteria we use for the genes included in our reports.

MyOme Personal Genome™ Gene Inclusion Framework



CLINICAL VALIDITY

Clinical validity measures the strength of the association between a gene and a disease. Using the ClinGen framework[2], only genes with moderate or higher evidence are included.

PENETRANCE / PREVALANCE

Penetrance is the proportion of individuals with a pathogenic variant who show clinical symptoms. Ideally, it should be at least 40%. However, diseases with lower penetrance may be included if they have high burden and actionability. We also consider the prevalence of disease, when available, to weigh the impact of testing at a population level.

ACTIONABILITY

Actionability is based on the potential for medical intervention if a pathogenic variant is found. It considers disease severity and the effectiveness of available treatments. Using the ClinGen framework[3], genes are evaluated to ensure findings lead to preventive measures or treatments that can improve clinical outcomes in adults.

FEASIBILITY

Gene-specific features, such as genetic complexity or variant interpretation challenges, that may affect the feasibility of accurate and meaningful genetic reporting.

As scientific knowledge advances and testing evolves, we will continually evaluate both previously assessed and new genes to ensure we deliver clear clinical utility in every MyOme Personal Genome™ report.

Contact us to learn more about the MyOme Personal Genome™ platform.

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1. Miller, David T., 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 25.8 (2023): 100866.
2. Strande NT, Riggs ER, Buchanan AH, et al. "Evaluating the clinical validity of gene-disease associations: an evidence-based framework developed by the Clinical Genome Resource." Am J Hum Genet. 2017;100:895–906.
3. Hunter, Jessica Ezzell, et al. "A standardized, evidence-based protocol to assess clinical actionability of genetic disorders associated with genomic variation." Genetics in Medicine 18.12 (2016): 1258-1268.

