INVITAE

A trusted partner in genetic testing

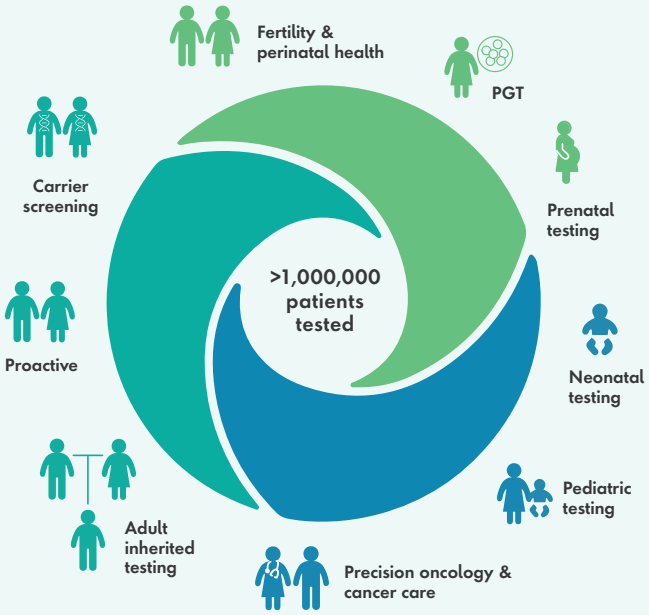
Invitae's team of medical genetics experts is dedicated to making affordable, high-quality genetic testing the standard of care in medicine.

Experience you can trust

We've tested **more than**

1,000,000 patients

across a broad range of indications, providing genetic information for all stages of life.



Dedicated to the highest quality

Invitae's testing is backed by peer-reviewed studies.[1]

1 in 7 **positive test results are challenging variants.[2]**

- These could be missed by standard next-generation sequencing (NGS) technology.
- Invitae's customized NGS-based methods detect complex variants with high accuracy.*

Committed to transparency

Invitae is the largest contributor to ClinVar with

>390,000 submissions.†

Comprehensive and accurate variant interpretation

SHERLOC

Our **variant interpretation process**:[3]

- Systematically strives to remove subjectivity
- Ensures reproducibility in classifications
- Is peer-reviewed and published

Functional Modeling Platform

Invitae's **Functional Modeling Platform** (FMP):

- Reduces uncertainty
- Provides a definitive result for an additional 1 in 40 patients
- Reduces variants of uncertain significance in real time

How does this compare to what some other labs use?

Other labs: Computational *(in silico)* evidence from publicly available models, such as PolyPhen2 and SIFT	**Invitae:** Computational *(in silico)* evidence from FMP incorporated into Sherloc
• Often outdated	• Dynamic and AI-enabled, continuously learning and improving with experience from Invitae's vast database of >1 million patients
• Single model for all genes: "one size fits all" approach	• Gene-specific: AI evaluates variants in each gene separately, taking gene-specific characteristics into account
• ~75–85% accuracy[4-6]	• >99% accuracy[7]

Invitae's demonstrated **quality**, combined with our commitment to making testing **affordable**, gives you the ability to make informed healthcare decisions.

Trust Invitae for all your genetic testing needs.

References

1. Data on file
2. Lincoln SE *et al.* One in seven pathogenic variants can be challenging to detect by NGS: An analysis of 450,000 patients with implications for clinical sensitivity and genetic test implementation. medRxiv doi: 10.1101/2019.12.11.12345678. July 2020.
3. Nykamp, K., Anderson, M., Powers, M. *et al.* Sherloc: a comprehensive refinement of the ACMG–AMP variant classification criteria. *Genet Med* 19, 1105–1117 (2017).
4. Sim NL *et al. Nucleic Acids Res.* 2012;40:W452-W457.
5. Adzhubei IA *et al. Nat Methods.* 2010;7(4):248-249
6. Loannidis NM *et al. Am J Hum Genet.* 2016;99(4):877-885
7. Data on file.

* Alu insertions and breakpoints within an exon. Invitae's algorithms are both read-depth and split-read based, following the approach taken by Nord *et al.* (BMC Genomics 2011)
† For a list of the largest contributors to ClinVar, visit http://www.ncbi.nlm.nih.gov/clinvar/docs/submitter_list.

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