

## WHO SHOULD CONSIDER EXOME GENETIC TESTING?

The Invitae Boosted Exome is often most appropriate for patients who have:

- symptoms suggestive of an underlying genetic condition
- complex medical history
- symptoms that affect more than one body system
- symptoms that do not seem to be easily explained by a single known genetic condition
- a suspected genetic diagnosis with negative results on previous genetic tests

In some cases, such as when symptoms seem to be explained by a single known genetic condition, genetic testing for a single gene or a multi-gene panel may be more appropriate than an exome. Discuss these options with your physician, who can guide you to the most appropriate type of genetic testing.

Invitae's mission is to bring comprehensive genetic information into mainstream medical practice to improve the quality of healthcare for everyone. Our goal is to aggregate most of the world's genetic tests into a single service with higher quality, faster turnaround time, and lower price than many single-gene tests today. For more information about Invitae's genetic tests, please visit [www.invitae.com/patients](http://www.invitae.com/patients).

For additional details on testing with Invitae, please contact Client Services at [clientservices@invitae.com](mailto:clientservices@invitae.com) or 800-436-3037.



INVITAE

Finding answers  
through exome testing

## THE INVITAE BOOSTED EXOME

Knowledge is a powerful tool when it comes to healthcare. People with certain types of genetic conditions may benefit from diagnostic genetic testing to better understand risks, confirm a diagnosis, or inform an appropriate medical management plan—or a combination of all three.

DNA in your cells contains a “genetic code” which tells the body how to make all of the many proteins required for proper functioning of a human being. The Invitae Boosted Exome is a genetic test that analyzes segments of your DNA that code for proteins, called exons, in approximately 20,000 human genes simultaneously.

The vast majority of disease-causing DNA changes are currently known to exist within exons,<sup>1</sup> which is why the test focuses on these regions.

## WHAT ARE THE BENEFITS OF GENETIC TESTING?

Genetic testing produces information that may help you or your healthcare provider:

- establish or confirm your specific diagnosis
- provide an explanation of the underlying cause of your symptoms
- uncover potential risks of developing a condition that affects your health
- make informed medical decisions and develop a comprehensive care plan
- identify other at-risk relatives for whom genetic testing is recommended
- make informed family planning decisions

<sup>1</sup>Pussegoda KA. Exome sequencing: locating causative genes in rare disorders. *Clin Genet*. 2010 Jul;78(1):32-3.

## HOW DOES IT WORK?

The Invitae Boosted Exome must be ordered by a healthcare professional. Once your healthcare provider orders the test, you will provide either a blood or a saliva sample. Ideally, both of your biological parents will also provide a blood or saliva sample; this is called a “trio” and is the most informative way to interpret the genetic sequence results. If one biological parent is affected with symptoms similar to yours, then a sample can be helpful from that parent, even if your other biological parent cannot provide a sample. This is called a “duo.” If neither of your biological parents can provide a sample, Invitae can still test your DNA alone; this is called a “proband-only.”

Invitae will conduct the testing at its San Francisco laboratory, and return the results to your healthcare provider within 6–8 weeks on average.

Your healthcare provider will then discuss the results with you, along with any new medical recommendations for you or your family members.

Insurance billing or patient pre-pay options are available for the Invitae Boosted Exome. We also offer payment plans and financial assistance based on poverty guidelines. Please consult your provider for the latest information.

## NOTES:

## WHAT ARE THE POTENTIAL RESULTS?

### POSITIVE

If testing identifies a variant associated with your symptoms, work with your healthcare provider to create a management plan and to identify relatives who may need to be tested.

### NEGATIVE

If testing does not detect disease-causing variants, the underlying cause of your condition remains unknown. However, as our knowledge of genetics improves, additional genetic testing may become available that could give you more information about the underlying cause of your condition.

### VARIANT OF UNCERTAIN SIGNIFICANCE

In some cases, testing can identify a genetic variant, but based on our current knowledge of genetics, it is not clear whether this variant is associated with your symptoms. If new information about the variant becomes available, this could change what the test results mean for you and your relatives. Always let your healthcare provider know if there are any updates regarding your family medical history and update them if your contact information changes so that they can relay any new findings to you.

### ACMG SECONDARY FINDINGS

Certain genetic changes identified during exome testing may not be directly related to your primary symptoms. When these genetic changes are associated with a serious medical condition for which there are treatments, such as inherited susceptibility to cancer or cardiac conditions, the American College of Medical Genetics (ACMG) recommends this information be made available to you. It is your choice whether or not you would like to know about these “secondary findings.” Please speak with your clinician about what you might learn and whether these results make sense for you.