PATIENT GUIDE

Clinical Genomics

GENETIC TESTING FOR UNDIAGNOSED DISEASES

Taking the extra step to find answers

Ambry Genetics®
A Konica Minolta Company

Know the Basics

You are made up of about
20,000 genes
This is called your genome

Genomic testing
can find answers for up to half of all people with an undiagnosed disease

TYPES OF GENOMIC TESTING

Chromosomal Microarray

Chromosomal microarray (CMA) is a test designed to look for genetic changes in your genome. CMA is especially good at finding extra or missing genetic information. For many people, these imbalances can be the cause of an existing medical condition.

Exome Sequencing

Exome sequencing is a comprehensive test designed to look for genetic changes (mutations) in your exome that may be the cause of an existing medical condition. Some genetic tests just look for common mutations, while others may just look for changes in common genes. Exome sequencing looks at all of the genes and all of the mutations, making it a comprehensive genetic test.

Genome
Apple

Exome
Slice

1-2%

85% of genetic conditions are caused by changes in the exome

WHY IS GENOMIC TESTING IMPORTANT?

By knowing the underlying genetic cause of a condition, your doctors may be able to treat and manage your health better. They may also be able to better understand if your medical condition may also affect other family members. As researchers and doctors continue to learn more about genomics, we are able to find better treatments and cures for certain conditions.

Is Genomic Testing Right For You?

YOUR DOCTOR MAY CONSIDER EXOME SEQUENCING OR CMA

to identify the underlying genetic cause of your medical concerns when:

1. Previous genetic and medical tests have not yet found the cause of your medical concerns, and your doctor thinks it may have an underlying genetic cause.

2. There is no specific genetic testing available for the genetic condition you may have.

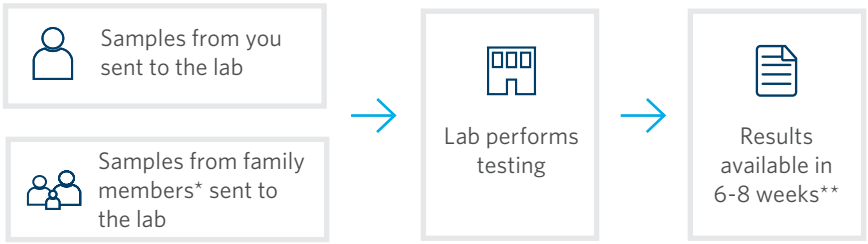
3. Your medical concerns may be caused by changes in more than one gene and genomic testing is a way to test for all of them at once.

Your healthcare provider may identify other reasons why you could consider genomic testing.

Understanding Disease Better Through Quality Testing

YOUR GENES CARRY A STORY THAT IS UNIQUE TO YOU AND MAKES YOU WHO YOU ARE. TAKING A CLOSE LOOK AT YOUR GENES CAN HELP YOU BETTER UNDERSTAND THE CAUSE OF YOUR MEDICAL CONCERNS.

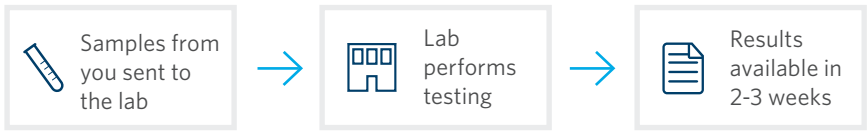
How is Exome Sequencing Performed?



** "Family members" refers to blood relatives, such as brothers/sisters/parents/grandparents/aunts/uncles/cousins*

*** Availability of results varies based on test ordered.*

How is CMA Performed?



Your healthcare provider has determined that the best test for you is:

☐ CMA (chromosomal microarray) ☐ ExomeNext

VISIT OUR WEBSITE

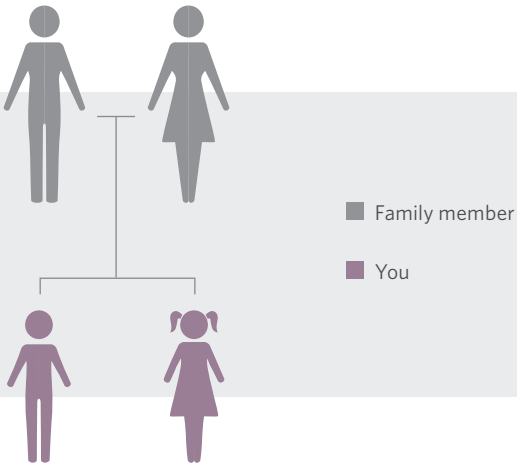
See updated information about the test your healthcare provider selected above: ambrygen.com/patient/genomicstest

Family-Based Genomic Testing

WHY MIGHT THIS TESTING INVOLVE MY FAMILY MEMBERS?

Exome sequencing and CMA may provide more accurate answers when your DNA is compared to your family members'. This helps sort through all the genetic changes and identify those that are common in your family vs. those that may be causing your medical condition. Or, if multiple people in your family have the same medical condition, it helps to see which of the same genetic changes each family member has.

This testing is still possible without your family members, but it just lowers the chance of finding the answer. To test your family members, all we need is blood or saliva sample from them.



What are the Benefits of Genomic Testing?

BENEFITS OF EXOME SEQUENCING AND CMA:

Comprehensive genomic tests may help find the underlying cause of your medical concerns. Finding a diagnosis through this test is beneficial because:

Quicker and more cost-effective diagnosis, since they study multiple genes at once

May guide or influence your treatment or medical health care decisions

Removes uncertainty related to having an undiagnosed disease

Provides awareness about health concerns in your family members and/or risk of passing on the genetic change to future children

Results

It will be important to discuss your results with your healthcare provider to determine how they may impact your medical care.

PRIMARY RESULTS FOR EXOME SEQUENCING AND CMA

+ POSITIVE/LIKELY POSITIVE

A mutation was found that is known to be associated with your medical concerns

Genetic testing for the specific mutation may be considered for family members

? UNCERTAIN

A DNA change(s) was found, but it is not clear if this is the cause of your medical concerns

Studying your genomic data in the future may provide useful information

- NEGATIVE

The underlying genetic cause of your medical concerns has not been found

Studying your genomic data again may be helpful in the future

SECONDARY FINDINGS RESULTS (EXOME SEQUENCING ONLY)

+ POSITIVE/LIKELY POSITIVE

A mutation was found in a gene that is unrelated to your medical concerns, but may affect your health in the future

Genetic testing for the specific gene mutation may be considered for family members

It will be important to discuss these findings with your doctor

- NEGATIVE

No mutations were identified among a group of common disease genes unrelated to your medical concerns

There may still be genetic changes that may affect your future health, however they were not identified through this test

Things to Consider

LIMITATIONS

In some cases results may be negative. Although these tests are designed to look at the entire exome/genome, the cause of your medical concerns may be in an area of the exome/genome that is not well understood or cannot be identified.

If your results are negative, your healthcare provider may choose to do additional genetic testing now or in the future. Negative results could also suggest that the medical issues you are concerned about are not inherited.

SECONDARY FINDINGS FOR EXOME SEQUENCING

Because this test looks at all of your genes at once, medically important mutations may be found that have nothing to do with your current medical concerns. If this happens, you could potentially learn new information about your health, for which there may be recommended medical follow-up. You can choose ahead of time if you wish to learn about this information.

TESTING FOR FAMILY MEMBERS

Genomic testing may find a genetic change in you that may also be found in your family. It is important to consider sharing your results; however some family members may not want to know if they are at risk for developing a genetic condition. Ideally, your family members should meet with their doctor or a genetic counselor to discuss their options for being tested. Genomic testing may also find unexpected information about family relationships. You can discuss these possibilities with your doctor/genetic counselor.

PRIVACY

You may wonder if your genomic test results could be used against you/or your family in the future. Your insurance company has access to all your test results in your medical record. However the Genetic Information Nondiscrimination Act of 2008 (GINA) is a U.S. law that states you cannot be denied a job or denied health insurance because of your genetic condition. Unfortunately, this law does not apply to all individuals and to some types of insurance, including life insurance. Your area or country may have other, more complete laws in this area.

Frequently Asked Questions

1 HOW IS GENOMIC TESTING PERFORMED AND HOW LONG DOES IT TAKE?

Genomic testing is done using a blood (blood or saliva samples can be used for your family members), which is collected using a special kit that is shipped overnight to Ambry (all coordinated by your healthcare provider). Samples are collected from you (the patient) and two other family members. Testing looks for mutations that that may be the cause of your medical condition. It takes about 1-8 weeks (depending on the test that is ordered) for the testing to be completed and results are sent to your healthcare provider.

2 WHAT WILL HAPPEN WHEN MY RESULTS ARE READY?

Your healthcare provider will receive your results; they will not be sent directly to you. Every healthcare provider may have a different method and time frame to contact you to discuss your results, so it is important to discuss this process with them. Based on your test results, your healthcare provider will discuss any next steps.

3 WILL MY GENOMIC TEST RESULTS AFFECT MY INSURANCE COVERAGE?

In the U.S., the Genetic Information Nondiscrimination Act (2008) prohibits discrimination by health insurance companies and employers, based on genetic information. Depending on where you live in the world, there may be additional or fewer laws about genetic testing. Visit ginahelp.org to learn more.

4 WILL GENOMIC TESTING BE COVERED BY MY INSURANCE?

Many insurance plans cover genetic testing and Ambry is contracted with the majority of U.S. health plans. Your out-of-pocket cost may vary based on your individual plan; therefore, we offer personalized verification of insurance coverage and financial options for your genetic testing. A team of dedicated specialists is available to help you get access to the genetic testing you need and answer any questions you have about our payment options. Call or email our Billing department at +1.949.900.5795 or billing@ambrygen.com with any questions.

5 WHAT IS AN EXPLANATION OF BENEFITS (EOB)?

Your insurance company sends you an EOB to explain any services paid on your behalf. You can contact us directly to speak with a Billing specialist with any questions or concerns about your EOB. Some genetic tests take weeks or months to process in order to receive the best results. In addition, insurance companies can take several weeks or even a couple of months to process claims.

STILL HAVE QUESTIONS?

Talk to your doctor or visit our website: ambrygen.com

Resources For You

Ambry's Patient Education Website

ambrygen.com/patient

Global Genes

rarediseasefoundation.org

National Organization For Rare Disorders

rarediseases.org

Genetic Information Nondiscrimination Act

ginahelp.org

FIND A GENETIC COUNSELOR

National Society of Genetic Counselors

nsgc.org

Canadian Association of Genetic Counsellors

cagc-accg.ca

Finding Answers.

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