



Genetic Carrier Screening Frequently Asked Questions

1) Why should I be screened?

Every individual carries at least 8-10 different genetically inherited conditions. Therefore, the reality is that anyone can be a carrier of a common genetic condition, such as Tay-Sachs or Cystic Fibrosis. So, regardless of an absence of family history of a genetic disease, there remains a chance for any individual to be a carrier of a common genetic condition, or further yet, for any couple to have a child with a genetic condition. A majority of individuals report this knowledge is helpful in just knowing the risk, providing an opportunity to learn more about a genetic condition, a family's specific risk, and the ability to prepare or make decisions as that couple sees fit.

2) Do I need to be screened?

No, some individuals do not want to know this information. Opting to pursue or not pursue screening is a personal choice with positive and negative implications both ways. No one choice is correct, the only correct choice is the one you are comfortable with.

3) What will I be screened for?

Individuals pursuing carrier screening can screen for one genetic condition, such as cystic fibrosis, a panel of conditions more common based on a person's ancestry, or an even broader panel of over 100 genetic conditions. Individuals can make a personalized choice on what they screen for based upon how much information they wish to know.

4) When should I be screened?

The ideal time to undergo carrier screening is prior to pregnancy, if utilizing the information for reproductive decisions. However, carrier screening can be performed at any time to provide information for an individual and their family members.

5) Can we screen my children too?

No. Carrying a genetic condition does not impact or cause childhood diseases or illnesses. Just as you have a chance to make a decision to pursue or decline genetic carrier screening, we believe that choice should also be provided to a child when they become an adult. They are then able to consider and answer the question whether they believe testing is right for them or not. Various national committees, such as the American College of Medical Genomics and Genetics also agree and recommend genetic testing of children be pursued only if the results will impact childhood care. Finally, as noted in Question 11, there are insurance ramifications to consider.

6) What is the cost of screening?

At NxGen MDx, we believe all patients should have access to genetic technology without the limitations of costs to hold them back. For that reason, we have created the Peace of Mind program, which limits expenses for qualified patients.

7) How long will it take to get the results from screening?

Results are typically available 7-10 business days from sample collection. Additionally, once released, genetic counseling is available for individuals who wish to discuss the results, positive or negative, in greater detail.

8) What happens if I am a carrier?

Genetic counseling is available to all individuals who undergo testing to help answer questions regarding negative or positive test results. Typically, the next step after an individual is noted to be a carrier of a genetic condition is to screen their partner for the same genetic condition to determine their risk, as a couple, to have a child with a genetic condition. Additionally, individuals who are noted to be positive are encouraged to share this information with family members, as they too would have an increased risk to carry the same condition. In rare cases, as some genetic conditions can be very mild, an individual can test positive not to be a carrier, but rather, to have a genetic condition. This can be helpful in guiding whether additional medical management or guidance is needed for that individual. Further yet, in the case of some genetic conditions, an individual who carries a genetic condition may have a slight increased risk for medical concerns. While this may increase anxiety or concern, it can also help appropriately inform an individual and their health care providers to any additional screening or medical management necessary.

9) How successful is screening in finding genetic disorders?

It is estimated that individuals have 20,000 genes in each cell of their body. Carrier screening typically looks at anywhere from one to hundreds of genes, therefore, it cannot tell an individual everything about their genetic make-up. On the other hand, 1 in 4 to 1 in 5 individuals of Ashkenazi Jewish descent will test positive to carry at least one of the genetic conditions screened for on a panel looking at genetic conditions more frequent in the Ashkenazi Jewish population. NxGen MDx's cutting-edge Next Generation Sequencing (NGS) technology allows us to look at an entire gene. This means our test is superior in detecting less frequently observed mutations, regardless of their prevalence in a patient's genetic heritage. This technology provides one of the best sequencing sensitivities available compared to other genetic testing laboratories.

10) Who has access to the results?

Laboratories report results to the ordering provider and patient. However, if in one's medical record, any entity that may pull information from a medical chart, such as insurance companies, would also be privy to the information. There are laws, such as the Genetic Information Non-Discrimination Act (GINA), that protect individuals from some, but not all, types of genetic discrimination. Many states also have specific, more stringent laws in place, protecting individuals.

11) Will this impact my ability to obtain insurance now or in the future?

Again, there are laws, such as the Genetic Information Non-Discrimination Act (GINA), that protect individuals from some, but not all, types of genetic discrimination. Many states also have specific, more stringent laws in place, protecting individuals. However, neither GINA nor all state laws apply to insurance an individual can obtain on the private market, such as life insurance, disability insurance or long-term care insurance. And while genetic carrier status of an autosomal recessive condition most often won't affect one's ability to obtain these types of insurances, there are some situations where it may. Some carrier conditions may slightly increase the risk for medical concerns later in life, or some individuals may test and find they actually have a genetic condition, in a very mild form. And genetic discrimination may or may occur for private market insurance in these situations.



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Does my insurance cover Carrier Screening?

NxGen Genetic Carrier Screening is most likely NOT a participating provider with your health insurance company. However we have taken innovative measures to create flexible and affordable payment options that will likely benefit you. Please call Diane Spruill at 855-776-9436 ext. 221 with any questions about your out of pocket expense.

If you have any questions regarding the following, please contact NxGen MDx directly:

- Your bill from NxGen MDx LLC
- The Explanation of Benefits (EOB) from your health insurance company
- You would like to find out if NxGen MDx is a participating provider for your health plan

What if I receive a check directly from my health plan?

If you choose to receive a check, please forward to NxGen MDx as payment for the laboratory services rendered. To do this, simply endorse the check by writing "Pay to the order of NxGen MDx" on the back of the check and signing your name on the line below. Please include a copy of any insurance explanation of benefits so we can properly adjust your account. Mail to:

NxGen MDx
PO Box 72512
Cleveland, OH 44192

You may also pay on-line at www.nxgenmdx.com.

If you have already cashed or deposited the check, please call our billing support line at 855-77-NxGen and we will send you a statement that you can then pay via check, cash or credit card.

Need Additional answers?

Our NxGen customer service department can be reached at 855-77-NxGen between 9am and 5 pm EST to address any of your concerns.

Please do not call your physician's office, as we are your best resource for information.