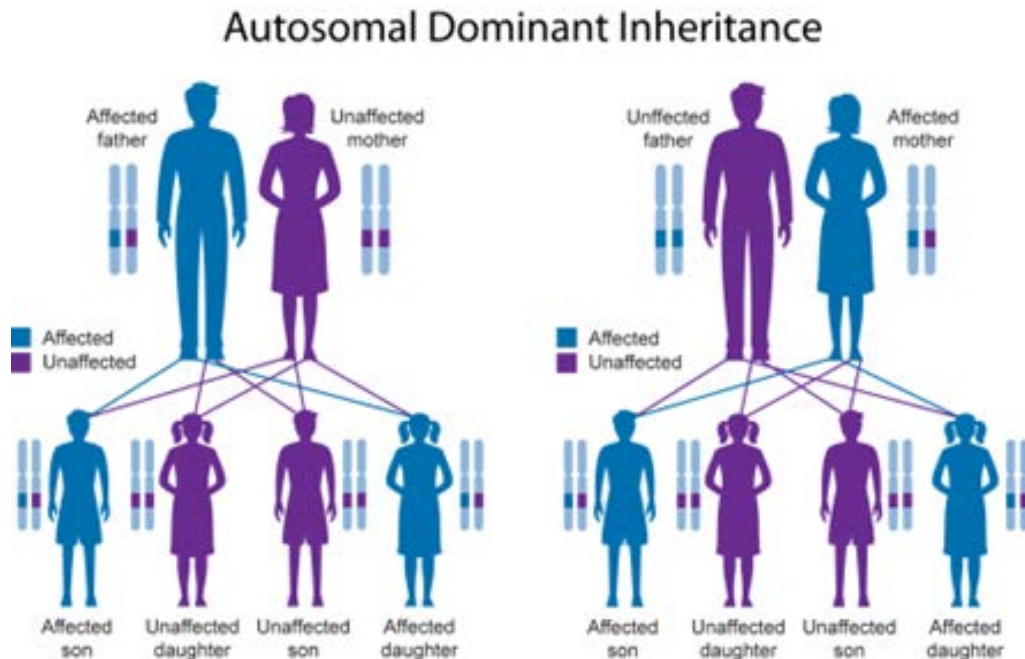


A Family Guide to Genetic Testing of the VCP Gene

VCP disease is caused by a bad copy of the VCP gene. This bad copy is called a “pathogenic variant.” The VCP gene is passed down through generations in an autosomal dominant pattern. This means that if you have one parent with a bad copy of VCP, you have a 50% chance of inheriting it, regardless of sex. A genetic counselor can help you trace family members at risk of having the VCP pathogenic variant.



What are my chances of getting the symptoms of VCP disease if my genetic test is positive?

If your VCP genetic test is positive, you will most likely develop symptoms at some point. Many types of conditions can develop in a person with VCP disease, even within the same family. VCP disease can affect a person’s muscles, brain, bones, and nerves. The most common conditions of VCP disease are inclusion body myopathy, Paget’s disease of bone, and frontotemporal dementia. According to scientific studies on VCP disease, the age people first develop symptoms varies widely, with some developing symptoms in their 20s and others not developing any symptoms until their 60s. A team of doctors and therapists can help you develop a care plan and help you manage symptoms over your lifetime.

If a family member is “at-risk” of VCP disease, should they get genetically tested?

When and if a person gets genetically tested is a personal choice. Discussing the considerations and risks with a genetic counselor is strongly encouraged by the National Society of Genetic Counselors. Below are some considerations.



CURE VCP DISEASE

- For symptomatic individuals: In a family with an established diagnosis of VCP disease, it is appropriate to consider testing if an individual is experiencing symptoms, regardless of age.
- For asymptomatic at-risk relatives: Predictive testing for at-risk relatives is possible once the pathogenic variant of VCP has been identified in an affected family member. A proper diagnosis before developing symptoms can allow for disease monitoring, lifestyle changes, and family planning, but all the risks should be considered before testing.
- For minors: In most cases, it is encouraged to defer VCP genetic testing until the child is an adult and can choose for themselves. The National Society of Genetic Counselors encourages deferring predictive genetic testing of minors for adult-onset conditions when results will not impact childhood medical management or significantly benefit the child.

Why should I consider genetic testing?

A genetic test is the only conclusive way to diagnose VCP disease. If you have symptoms, a proper diagnosis can help you receive care, support, and treatments as you navigate living with VCP disease.

A diagnosis confirmed by a genetic test also provides opportunities to:

- Screen for conditions that may develop over the course of VCP disease. For example, one-half of patients with VCP disease can develop Paget's disease of bone, and there is a treatment if detected early enough.
- Consider lifestyle changes like diet, exercise, and mental well-being.
- Plan modifications to your home to optimize your living situation.
- Understand family planning options. For example, in vitro fertilization is available for families wishing to have pregnancies that do not carry a familial VCP mutation.
- Seek genetic counseling and diagnosis for at-risk family members.
- Participate in research and learn more about VCP disease.

Early diagnosis enables timely treatment and intervention.

What are the risks of genetic testing?

You should take into consideration the social, financial, life insurance, and disability insurance implications prior to genetic testing. A genetic counselor can help guide you through whether genetic testing suits you now.

- Financial considerations: You should consider how a DNA test result might impact your employment status and insurance qualifications. It is essential to allow sufficient time before testing to make necessary financial or insurance arrangements. The Genetic Information Nondiscrimination Act (GINA) of 2008 protects health insurance and employment discrimination based on genetic information.

However, this law does not cover life, disability, or long-term care insurance and does not apply to military members.

- Psychological preparation: Before getting a genetic test, evaluating the reasons for testing can help you process how positive and negative results would affect your life. Discussing the implications of testing with a genetic counselor will allow you to make a fully informed decision about your options.

What if I can't find a genetic counselor?

As patients and families with VCP disease, we understand. There is a lot to navigate, and sometimes it is hard to find the right provider. We can help. Contact us at info@curevcp.org; we would love to help you and share our experience.

Did you know:

De Novo: A small percentage of the time, a family history cannot be determined in a person with VCP disease, and a doctor may determine that you have a de novo variant. It's known as "de novo," which means "new," because your parent did not have the mutation and was created in you.

VUS: If the words "*variant of unknown significance*" are on your genetic test results, there is not enough known to determine if the VCP variant you have would cause disease. This may change as new cases are discovered.

