

What causes a UCD?

A UCD is inherited, meaning it is something a person is born with. Inherited UCDs occur when one or both parents pass a copy of an abnormal UCD-causing gene on to their child. However, it is also possible for a UCD to occur randomly while a developing baby's cells are forming, even if the parents are not carriers. (A carrier is a person with one normal copy of a gene and one abnormal copy.)

Do you have a family member with a UCD?

If you have a family member with a UCD, talk to your doctor about what this means for you. The most common type of UCD, ornithine transcarbamylase (OTC) deficiency, is usually passed on from a mother to her child. In rare cases, OTC deficiency can also be passed down from a father to his daughter. In many cases, affected adults do not know they carry the OTC gene until their child is diagnosed. It is especially important for mothers, aunts, and sisters of an affected family member to consider UCD testing because they are more likely to be at risk.

Learn more

about urea cycle disorders at
www.UCDinCommon.com.

Learn more

about sponsored testing at
www.invitae.com/UCD.



About Invitae

Invitae is a genetics company whose mission is to bring comprehensive genetic information into mainstream medicine to improve healthcare for billions of people. Invitae testing provides answers to essential health questions—understanding disease risk, guiding a healthy pregnancy, or finding a diagnosis—at high quality, fast turnaround, and low prices.

We strive to make testing affordable and accessible.

About Horizon Therapeutics

Horizon Therapeutics is focused on researching, developing and commercializing innovative medicines that address unmet treatment needs for rare and rheumatic diseases. By fostering a growing pipeline of medicines in development and exploring all potential uses for currently marketed medicines, we strive to make a powerful difference for patients, their caregivers, and physicians. For us, it's personal: by living up to our own potential, we are helping others live up to theirs. For more information, please visit www.horizontherapeutics.com, follow us @HorizonNews on Twitter, like us on Facebook or explore career opportunities on LinkedIn.

While Horizon Therapeutics provides financial support for this program, tests and services are performed by independent third parties. Healthcare professionals must confirm that patients meet certain criteria to use the program. Horizon Therapeutics receives de-identified patient data from this program, but at no time does Horizon Therapeutics receive patient identifiable information. Horizon Therapeutics receives contact information for healthcare professionals who use this program. Genetic testing and counseling are only available in the US. Healthcare professionals and patients who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any Horizon Therapeutics product.

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UCD Genetic Testing Program

Genetic testing for urea cycle disorders (UCDs)

SPONSORED BY HORIZON THERAPEUTICS



UCD Genetic Testing Program

Sponsored, no-charge genetic testing for urea cycle disorders (UCDs)

The UCD Genetic Testing Program, sponsored by Horizon Therapeutics and performed by Invitae, provides access to genetic testing for patients in the US suspected of having a UCD and their family members, at no charge to patients.

What is UCD?

- A UCD is a rare genetic disease that affects about 1 in 35,000 births in the United States.
- UCDs occur when a person's urea cycle does not work correctly and the body is unable to change ammonia into urea so it can be removed from the body through urine. This leads to toxic levels of ammonia in the body that can cause symptoms like vomiting, confusion, and swelling in the brain.

To learn more about UCDs, visit www.UCDinCommon.com.

It is important for you to get tested for a UCD if you experience any of these common symptoms:

- Feeling very tired or sluggish
- "Zoning out"
- Headaches, fatigue, nausea, or vomiting

Note: Children who exhibit these symptoms or developmental delays, including autism, should also be tested.

How long will it take to get results?

Once Invitae receives your blood or saliva sample, your results will be available in 10 to 21 calendar days, on average.

What do I need to know when I get my results?

Whether your results are positive or negative, they provide important information that can help you make decisions about your healthcare and family planning. Knowing this information can also help you educate others in your family about UCDs and the importance of genetic testing.

What are the potential results?

POSITIVE means that you carry the abnormal gene that causes a UCD.

If you have a positive result, talk with your doctor about the UCD gene you tested positive for and what having the gene means for your health.

NEGATIVE means that the test did not find the abnormal gene that causes a UCD.

If you have a negative result, the genetic test did not find an abnormal gene that causes a UCD. However, if you are still experiencing health problems, your doctor may still suspect you have a similar disorder. He or she may want to discuss further testing and possible management options with you.

UNCERTAIN means that the test found something abnormal but it is not medically known whether the specific abnormality found causes a UCD.

In this case, your doctor may want to order additional testing to find out if you have a UCD.

Genetic testing with Invitae



- 1. Visit your healthcare provider**
Talk to your healthcare provider about testing through the UCD Genetic Testing Program.



- 2. Provide a sample**
Provide a sample of your DNA. This is usually done through a blood draw or saliva sample, which will be sent to Invitae's lab for analysis.



- 3. Get your results**
Receive your results. Invitae will send your results to your doctor, who will share them with you.



- 4. Make your plan**
Talk to your healthcare provider to understand your results and what they mean for you.