

How to talk to family members about OTC

This resource will help you start a conversation with family members about what it means to be a carrier of ornithine transcarbamylase deficiency (OTC) and why they may want to consider getting a genetic test.

If your child was recently diagnosed with OTC or you just learned that you are an OTC carrier, it's important to let your family members know. UCDs are genetic, which means other people in your family could also be affected. They should be offered genetic testing and information on what being an OTC carrier may mean for their health. Talking with a doctor or genetic counselor can also help them understand if they have been experiencing UCD symptoms. If this is the case, a doctor can work with them to develop a plan to help manage their symptoms.

It's important to give family members the information they need about OTC. If possible, it may be best to do this in person. Being able to talk openly with your family members and answer their questions might give them a better understanding of UCDs and help them feel supported.

If you don't feel comfortable sharing this information in person or over the phone, a letter or an email may be a good place to start. This can help you gather your thoughts and share the necessary information. If you use a letter to start the conversation, it might be helpful to follow up with an in-person or phone conversation.

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If you're not sure how to start the conversation or what to say, use the ideas below to help you. Anything in red should be filled in so it applies to you.

- I recently found out that [I have/my child has/my child and I both have] a defective gene that causes a health condition called ornithine transcarbamylase deficiency, or OTC. OTC is a type of urea cycle disorder (UCD).
 - People with UCDS have an enzyme or transporter in their liver that is missing or isn't working. This means that when their bodies break down protein and turn it into ammonia, they're not able to get rid of the ammonia correctly. This can lead to health issues.
- OTC is genetic, which means it runs in the family. It can be passed down from parent to child.
- Because [I have/my child has/my child and I both have] OTC, it's important for other members of our family to be tested.
- If you have, or carry, the gene that causes OTC, you may or may not experience symptoms. If you do experience symptoms, they may need to be managed by a doctor.

Some common symptoms of OTC are:

- Feeling very tired or sluggish
 - Staring or zoning out
 - Nausea or vomiting
 - Losing touch with reality/hallucinating
 - Feeling unusually irritable or uncooperative
 - Aggression
 - Lack of appetite
 - Slurring words
 - Headaches
 - Trouble with balance/walking
 - Seizures
- These symptoms can come and go but might occur more frequently after eating a meal high in protein, doing strenuous activity, or being very stressed. Symptoms are different for everyone, and not everyone will experience all of these symptoms.
 - If you experience any of these symptoms, it's important to talk to a doctor about what you can do to manage them and stay healthy.
 - A genetic test can tell you if you have OTC. Even if you don't have symptoms, it's important to find out if you carry the defective gene because it can be passed on if you have children. A doctor can order a genetic test, which is usually done by drawing your blood.
 - It can be helpful to talk to a metabolic geneticist and/or a genetic counselor about OTC. You can ask your primary care doctor for a referral to one of these specialists.
 - You can also find a genetic counselor near you by searching on the National Society of Genetic Counselors website: [nsgc.org](https://www.nsgc.org).

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Below is a sample letter that you can use to help you start the conversation. Starting with a letter may help you feel more comfortable following up in person. You can copy this into an email, or use it to guide your own written letter. Anything in red should be filled in so it applies to you.

[Date]

Hello [Name],

I wanted to let you know that I recently found out that [I have/my child has/my child and I both have] the defective gene that can cause a health condition called ornithine transcarbamylase deficiency, or OTC. OTC is a type of urea cycle disorder (UCD). People with UCDs have an enzyme or transporter in their liver that is missing or isn't working. This means their bodies can't break down and get rid of protein correctly, which can lead to health issues.

OTC is a genetic condition. Having or "carrying" the defective gene that causes OTC means a couple of important things:

- 1) People who carry the gene may have inherited it from their parents. This means other people in their family (like siblings, grandparents, aunts, cousins, and nieces) may also have the defective gene.
- 2) Carriers may or may not experience symptoms of OTC. Some common symptoms are feeling very tired, staring or zoning out, nausea or vomiting, slurring words, and headaches. These symptoms might occur after eating a meal high in protein, doing strenuous activity, or being very stressed. People who do experience symptoms can work with their doctor to come up with a management plan that helps them stay healthy.

Because [I have/my child has/my child and I both have] OTC, it's important for other members of our family to be tested. A genetic test can tell you if you have or carry OTC. Even if you don't have symptoms, it's important to find out if you carry the gene because it can be passed on if you have children. A doctor can give you a genetic test, which is usually done by drawing your blood. It can be helpful to talk to a metabolic geneticist and/or a genetic counselor about OTC. You can ask your primary care doctor to help you get in touch with one of these specialists. I can also give you the contact information for [my/my child's/our] doctor if you'd like.

If you have any questions about OTC or [my/our] experience, please feel free to reach out. You can [call or email me] at [phone number or email address]. You can also find a lot of helpful information about OTC and other UCDs at UCDinCommon.com. Thank you for reading this, and I hope this information was helpful.

Sincerely,

[Name]