

Talk to your doctor about getting tested for CTX

Cerebrotendinous xanthomatosis (CTX) is a rare, genetic, lipid storage and metabolic disorder with many symptoms. In CTX, certain enzymes that help with the proper breakdown of fats and cholesterol are not working.

What happens when cholesterol doesn't break down?

TOXINS called cholestanols and bile alcohols build up in blood and tissues throughout the body over time, which can cause a lot of different problems.

BUILDUP in different parts of the body can take years and can cause different symptoms that vary from person to person, and may get worse without treatment.

SERIOUS LONG-TERM HEALTH PROBLEMS can happen over time, but early diagnosis and treatment are the first steps to prevention.

Symptoms to suspect CTX in you or your loved one

Check off any current and/or past symptom(s) to discuss with your doctor.

Which symptoms indicate you should be tested for CTX

If you have **1** of the following:

- Early cataracts in both eyes
- Tendon xanthomas

or

If you have **2 or more** of the following:

- Neurological problems
- Cholestasis in infants (prolonged jaundice)
- Frequent diarrhea



Early cataracts in both eyes



Tendon xanthomas



Neurological problems



Cholestasis in infants



Frequent diarrhea

There are several tests available to help confirm CTX. Your doctor can order one of these tests through AboutCTXHCP.com.*

Prepare for a discussion about CTX

If you think you or your loved one may have CTX, it's important to start a discussion with a doctor about this disorder and how to get the right diagnosis and care.

What to keep in mind

- CTX is a rare disorder and can be mistaken for other disorders
- There are many symptoms of CTX, but not all symptoms may happen in an affected person
- Because of these factors, it can take several doctor visits, and sometimes many years, to figure out what is wrong

1 Share current and past symptoms. Remember, every person experiences CTX differently throughout life, with certain symptoms starting earlier or later in life.

2 Ask the doctor—based on current and past symptoms, if CTX might be the cause.

- If no, ask what else could be causing all of these symptoms
- If yes, ask your doctor to run a test to help make a diagnosis

There are several tests to help identify CTX:

- Biochemical test: A cholestanol test can be used to look for elevated levels of cholestanol in blood
- Genetic test: The *CYP27A1* gene is responsible for the disorder
 - **Mirum Pharmaceuticals has partnered with PreventionGenetics to offer no-cost genetic tests*[†]** to help identify the causes of early-onset bilateral cataracts or movement disorders

To order this Mirum-sponsored test, ask your doctor to visit [AboutCTXHCP.com](https://www.AboutCTXHCP.com) to learn more.

*Program may be canceled or changed at any time.

[†]Note that Mirum Pharmaceuticals cites the above-named external testing resource for information purposes only and does not endorse or guarantee in any way the services or advice provided by them.

