Talk to your doctor about getting tested for CTX

Cerebrotendinous xanthomatosis (CTX) is a rare, genetic, lipid storage and metabolic disease 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, which may get worse without treatment.

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

-  Cholestasis in infants ☐
-  Frequent diarrhea ☐
-  Early cataracts in both eyes ☐
-  Neurological problems ☐
-  Tendon xanthomas ☐

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:

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

There are several tests available to help confirm CTX.
Your doctor can order one of these tests through testCTX.com.[a]

[a]Your doctor can visit testCTX.com for more information. Program may be canceled or changed at any time.

Prepare for a discussion about CTX

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

What to keep in mind

- CTX is a rare disease and can be mistaken for other diseases
- 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

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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 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 disease

Or

- Mirum-sponsored test: Mirum Pharmaceuticals, Inc., has partnered with Oregon Health and Science University's Sterol Analysis Laboratory to offer a free test program to patients to help diagnose CTX. This program uses a biochemical test to screen and a genetic test to confirm a diagnosis using a single blood sample

To order this Mirum-sponsored test, ask your doctor to visit testCTX.com to learn more.

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