

SEE THE SIGNS

What to know about early-onset bilateral cataracts

If your child or loved one is experiencing or has experienced early cataracts in both eyes, it may be a rare, but treatable, genetic disorder called cerebrotendinous xanthomatosis (CTX). With available testing, you may be able to find out the underlying issue.

**BILATERAL
CATARACTS**



Many people don't realize they have CTX

Because CTX is a rare disorder with many symptoms, it usually is not discovered in patients until 20 to 25 years after symptoms start.

Symptoms of CTX

In people with CTX, cholesterol doesn't break down properly, causing cholestanols—toxins and bile alcohols—to build up in blood and tissues throughout the body. When cholestanols build up in the tissue near the lens of the eye, they may lead to cataracts. The cataracts may cause blurry vision.

While cataracts can and should be surgically removed, that will only resolve one symptom of CTX. This disorder causes different symptoms in different people at different ages and can lead to serious health problems over time.

These are some other possible symptoms of CTX:

- Cholestasis/prolonged jaundice (yellowing of the skin or eyes) in infants
- Frequent diarrhea
- Neurologic deterioration (developmental delay or behavioral problems)
- Tendon xanthomas (bumps on elbows, knees, or heels)

Approximately

85%

of patients with CTX experience early-onset cataracts in both eyes, which can appear as early as childhood or infancy

Early treatment of CTX may lead to better outcomes

Testing early on is so important because it can help find the underlying cause, which means doctors could start treating sooner. Early treatment may help prevent serious health problems and potentially lead to better outcomes for people with CTX.

Testing for CTX

Mirum Pharmaceuticals and PreventionGenetics are offering a genetic test to help find the cause of early cataracts in both eyes. It is offered at no cost to patients.



No actual patients shown.

Your ophthalmologist (eye doctor) may not have heard of CTX or the genetic testing for the condition. Therefore, you may need to encourage your doctor to learn more by ordering the genetic test for you. With a genetic test kit sent directly to your home, you could have answers in a matter of weeks.

If the test comes back positive for CTX, you will likely be referred to a neurologist or metabolic geneticist for CTX management.

Scan the QR code to
learn more about CTX.





Explore a No-Cost Testing Option*

Scout by Mirum: Early-Onset Bilateral Cataracts Genetic Panel



If you or your child is experiencing early cataracts in both eyes, Mirum Pharmaceuticals and PreventionGenetics are offering a genetic test for CTX at no cost*¹ to patients to help find the cause of early cataracts. **Talk with your ophthalmologist or geneticist about ordering a no-cost test.**

Criteria for No-Cost CTX Testing



AND



AND



***Program may be canceled or changed at any time. Eligibility criteria may apply.**

¹Note 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.

⁴Not known to be due to infectious causes, trauma, etc.

To learn more about the no-cost Scout by Mirum: Early-Onset Bilateral Cataracts Genetic Panel, visit [AboutCTX.com](https://www.AboutCTX.com)

