

Uncover What's Behind the Blur

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 can find out the underlying issue.



BEHIND THE BLUR
Early-Onset Bilateral Cataracts

Not Actual Patients.



Many people don't realize they have CTX

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

Approximately

85%

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

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 these cataracts. The cataracts may cause blurry vision and look like snowflakes or flecks in the eyes.

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 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)

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 find the cause of early cataracts in both eyes. It is offered at no cost to patients.

Your ophthalmologist (eye doctor) will likely not have heard of this test—or even CTX. Therefore, you may need to encourage your doctor to learn what is Behind the Blur by ordering the genetic test. 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.



Point your camera at the QR code to scan to watch a video on how to provide a sample for testing.

Uncover the Cause of Early Cataracts in Both Eyes



18 months
to
35 years

Is available at no cost to US residents 18 months to 35 years of age with current cataracts or a history of cataracts without apparent cause



Uses saliva or cheek-swab samples you can collect in your home or a blood sample taken in your doctor's office



Provides results in about 2 to 4 weeks

To learn more about the no-cost Behind the Blur Early-Onset Bilateral Cataracts genetic test for patients, visit [BehindTheBlur.com](https://www.behindtheblur.com)



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