

FACT SHEET: CEREBROTENDINOUS XANTHOMASTOSIS (CTX)

What causes CTX?

Cerebrotendinous Xanthomatosis is caused by a mutation in a gene called CYP27A1, which produces an enzyme called sterol 27-hydroxylase. Sterol 27-hydroxylase is required to turn cholesterol into bile acids, which are important in the absorption of fats in the intestine. In addition, when sterol 27-hydroxylase is not working properly, cholesterol accumulates in the tissues.

How is CTX diagnosed?

Diagnosis can be performed with a biochemical analysis of certain molecules in the blood and urine. Because the gene that is involved in CTX is known, molecular tests are also available to confirm the diagnosis.

What are the symptoms of CTX?

In infancy:

- Diarrhea
- Cataracts
- Psychomotor retardation
- Pyramidal/cerebellar signs: pyramidal and cerebellar are parts of the brain/central nervous system

In adulthood:

- Neurologic dysfunction with variable time of onset
- Mental retardation leading to dementia
- Psychiatric symptoms, including behavioral changes, hallucinations, agitation, aggression, depression, suicide attempts
- Cataracts
- Optic disk paleness: paleness in a particular part of the eye
- Premature retinal aging

- Xanthomas: Skin lesions characterized by accumulations of lipid-laden cells. Xanthomas can be a reflection of lipid metabolism alteration or a result of local cell dysfunction.
- Premature arteriosclerosis
- Coronary artery diseases
- Cough
- Chronic intractable diarrhea
- Gallstones
- Osteoporosis
- Increased risk of bone fracture
- Epileptic seizures
- EEG abnormalities

What is the treatment for CTX?

If CTX is diagnosed before there has been too much deterioration of the brain, proper treatment can potentially prevent the brain damage that can lead to severe mental dysfunction and eventually death. Treatment with a molecule called chenodeoxycholic acid inhibits abnormal bile acid synthesis and can halt, slow, and in some cases reverse the course of the disease.

How is scientific research on CTX progressing towards improvement treatment or diagnosis?

The identification of the gene responsible for Cerebrotendinous Xanthomatosis has allowed scientists to develop better ways to diagnose the disease. In addition, they can use this information to try to improve treatments for CTX.

Other Clinical Names for Cerebrotendinous Xanthomatosis

- CTX
- Cerebral Cholesterinosis