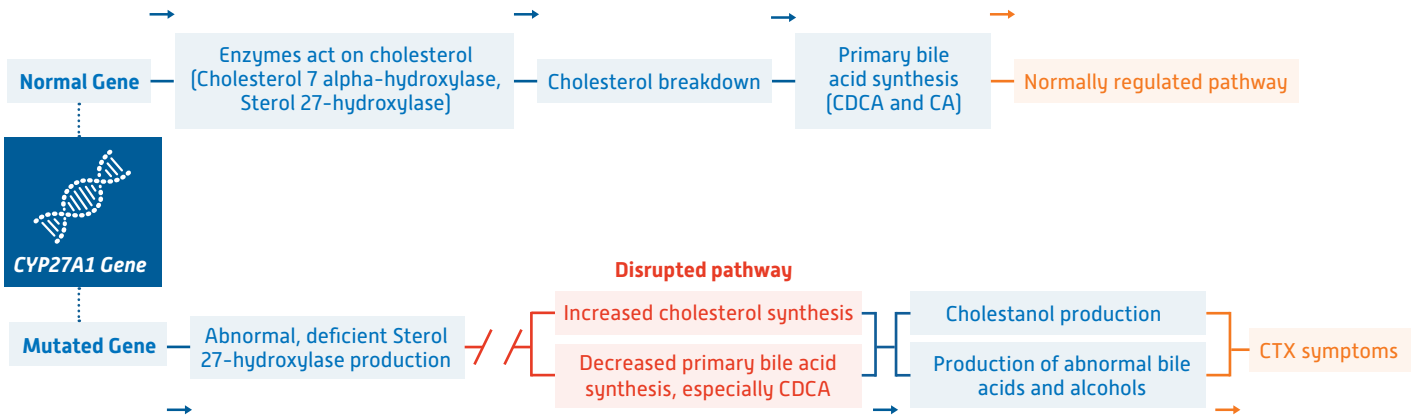


# CTX Fact Sheet

## Understanding Cerebrotendinous Xanthomatosis (CTX)

**Cerebrotendinous Xanthomatosis (CTX)** is a rare genetic disorder. The disrupted normal breakdown of cholesterol into primary bile acids leads to the abnormal accumulation of cholesterol and cholestanol throughout the body's tissues, especially eyes, tendons and brain. CTX affects both children and adults.

## CTX Results from the *CYP27A1* Gene Mutations Which Lead to a Deficiency in sterol 27-hydroxylase<sup>1</sup>



## Under-diagnosed: Exact Prevalence Unknown

200–425 cases worldwide reported<sup>2,3</sup>

## Clinical Features<sup>4</sup>

The signs and symptoms of CTX are diverse and may impact many systems in the body.

### Knowing the Symptoms

#### Infancy and early childhood

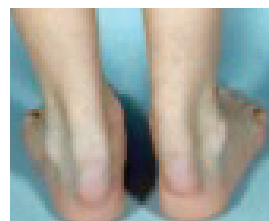
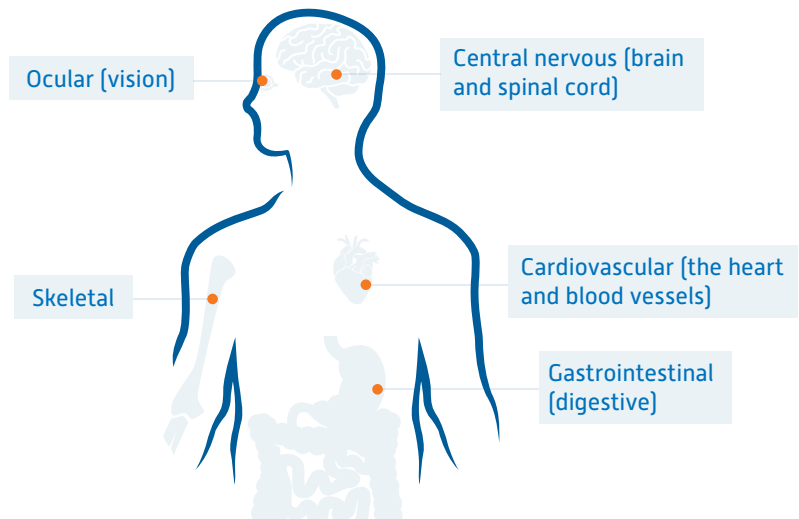
- Neonatal cholestatic jaundice (hepatitis of infancy)
- Chronic diarrhea in infancy and childhood
- Juvenile cataracts
- Developmental delay/mental retardation

#### Late childhood

- Xanthomas of tendons and other organs
- Psychiatric disorders

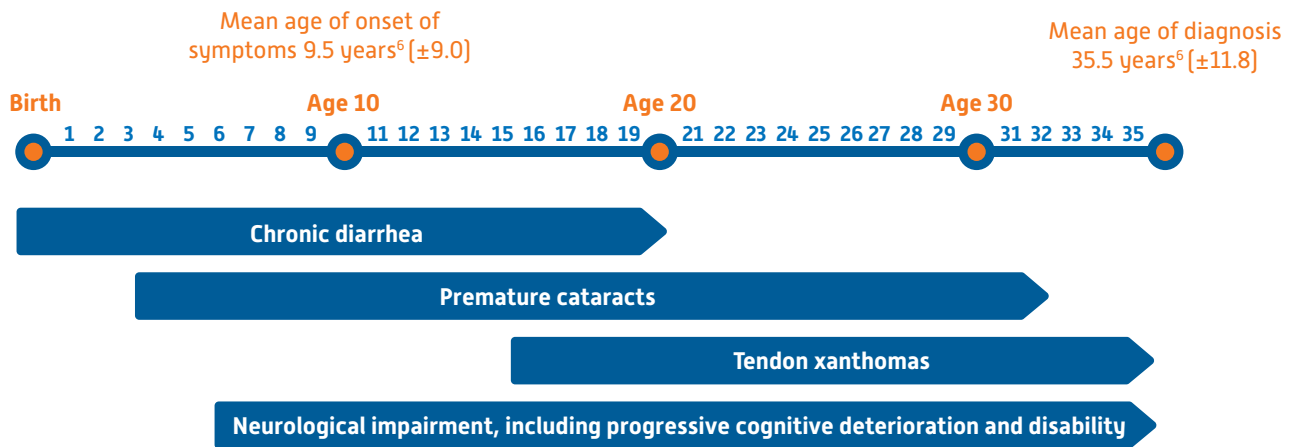
#### Adulthood

- Neurological manifestations
  - EEG: diffuse slow activity with intermittent discharges
  - Pyramidal signs: paresis, bulbar palsy
  - Epileptic seizures
  - Neuroimaging (MRI and CT): brain atrophy and bilateral focal lesions
  - Dementia or intellectual decline
  - Cerebellar syndrome
  - Peripheral polyneuropathy
- Myopathic-like facies
- Large paranasal sinuses
- Pes cavus
- Osteoporosis and bone fractures



One of the hallmark manifestations of CTX: tendon xanthomas (picture on the left).<sup>5</sup>

## Hallmark Clinical Manifestations of CTX



## Principal Diagnostic Tests for CTX<sup>7</sup>

Blood test: elevated plasma cholestanol levels

Urine test: elevated urine bile alcohol

Genetic testing



## CTX Management

CTX is typically treated with primary bile acid replacement therapy.

In addition, patients with CTX may be referred to other medical specialists to receive additional treatments or other supports to address the complications of the disease.

## Concerns About CTX?

If you are concerned about or are diagnosed with CTX, please seek your local healthcare practitioners for professional consultation, as practice and referral systems may vary from country to country.



For more information,  
visit [ctxawareness.com](http://ctxawareness.com).

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