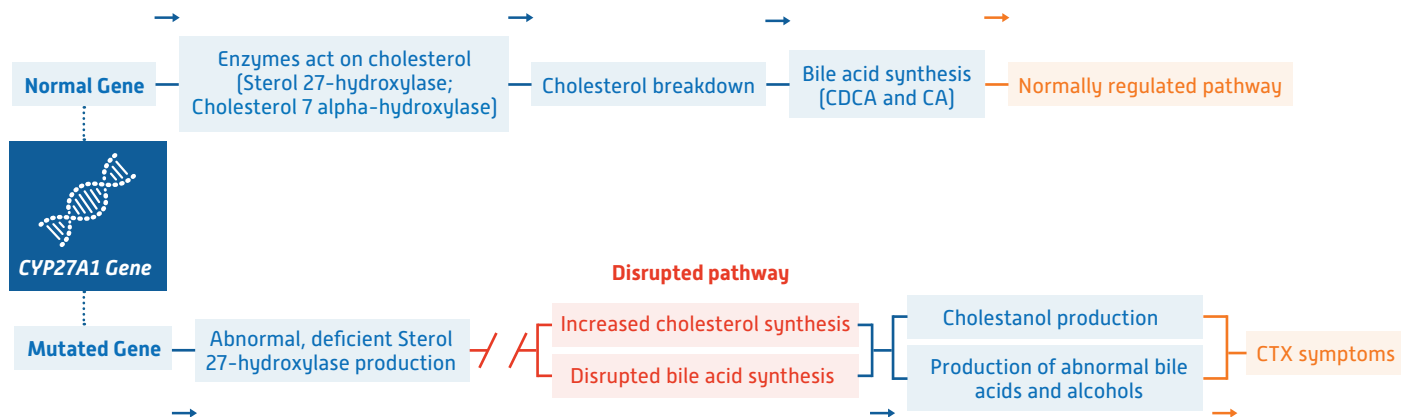


CTX Fact Sheet

Understanding Cerebrotendinous Xanthomatosis (CTX)

Cerebrotendinous Xanthomatosis (CTX) is a rare genetic disorder that disrupts the normal breakdown of cholesterol into bile acids causing the abnormal accumulation of lipids in certain areas of the body. CTX affects children and adults, and can lead to debilitating problems in the brain, eyes, tendons and other organs.

CTX Results from A Breakdown in Function of Sterol 27-Hydroxylase



Under-diagnosed: Exact Prevalence Unknown

200-425 cases worldwide reported^{1,2}

Clinical Features

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

Knowing the Symptoms

Infants and Young Children

Most common:

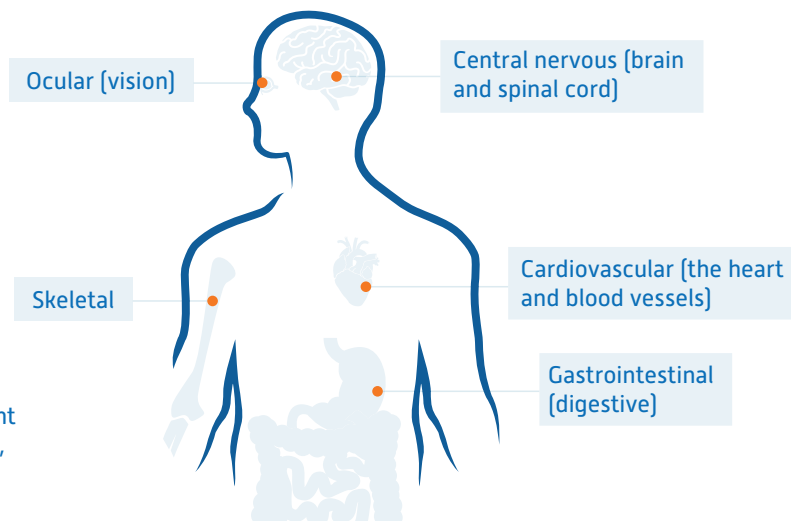
- Chronic diarrhea
- Cataracts

Less common:

- Prolonged jaundice in newborns
- Developmental delays and neurological issues
- Slowed physical movements, speech and thought
- Eye problems such as blurred vision, blind spots, or vision loss
- Autism Spectrum Disorder³
- Epilepsy

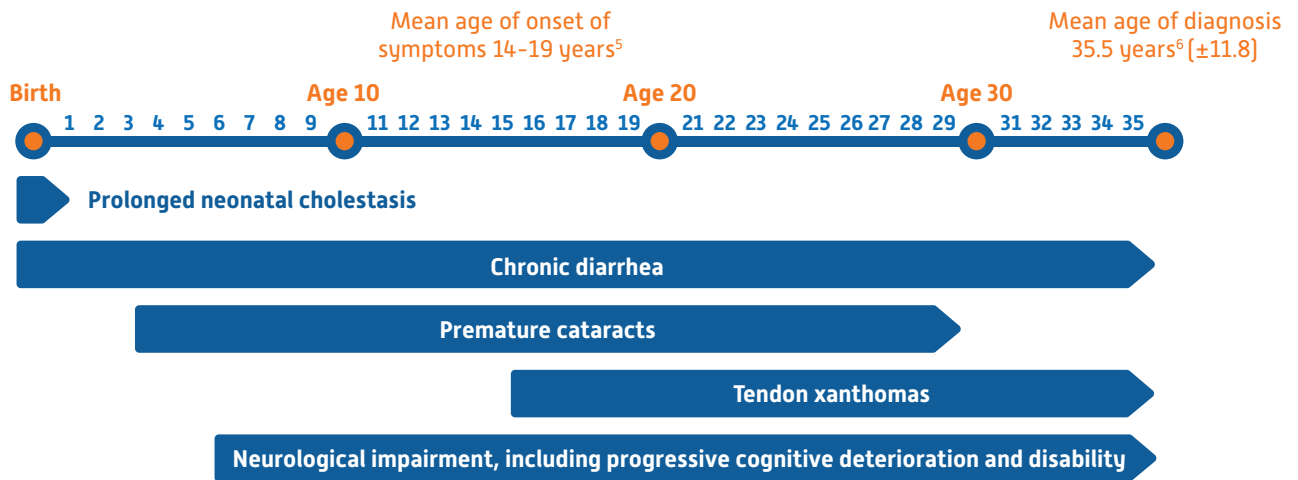
Adolescents and Adults

- Xanthomas
- Neurological problems including mental retardation, seizures and Parkinson-like tremors
- Psychiatric symptoms including behavioral changes, hallucinations, agitation, aggression, and depression
- Increased muscle tone and stiffness
- Atherosclerosis
- Cardiovascular disease
- Osteoporosis
- Bone fractures



The hallmark manifestations of CTX include tendon and tuberous xanthomas (pictured on the left), juvenile cataracts, and nervous system dysfunction.⁴

Clinical Presentation of CTX Symptoms



Principal Diagnostic Tests for CTX

Blood test: elevated plasma and serum cholestanol levels

Urine test: elevated urine bile alcohol

Genetic analysis

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

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