Cerebrotendinous Xanthomatosis (CTX)

WHAT YOU SHOULD KNOW

Cerebrotendinous xanthomatosis (CTX) is a rare inherited disorder that causes the body to not be able to break down cholesterol. This leads to a buildup of cholesterol and a related product, cholestanol, in the nervous system and other organ systems. CTX has many clinical symptoms.

Symptoms of CTX

Infants

» Chronic diarrhea
» Neonatal cholestasis (poor flow of bile at any point from the liver cells into the intestine of a newborn)

Children

» Premature cataracts
» Thickening of tendons, especially the Achilles tendon, called xanthomas
» Developmental delays, cognitive impairment, intellectual disability, and learning difficulties

Adults

» The build-up of fat in the brain leads to progressive problems with the nervous system, resulting in seizures, abnormal muscle movements, trouble maintaining balance, difficulty speaking clearly, loss of feeling in the arms and legs, dementia, hallucinations, and depression.
» Other symptoms include an increased risk of bone fractures, due to loss of calcium, and an increased risk of developing heart or lung failure at an early age.

What causes CTX?

People with CTX are unable to make bile acids correctly. Bile acids are produced in the body to help break down fats, allowing for normal daily activities and good health. Because the body lacks proper amounts of bile acid in those with CTX, there is a buildup of fat in many organs, including the brain, which causes problems with normal function and may lead to organ failure. In addition, there is often a buildup of other chemicals that can damage the body's organs.

How Common is CTX?

It is thought that between 1 in 72,000 to 1 in 150,000 Americans have CTX.

How is CTX Diagnosed?

CTX is usually diagnosed based on clinical symptoms, biochemical tests showing elevated levels of cholestanol, and confirmed through genetic testing for mutations in the CYP27A1 gene.

Lifestyle Recommendations

Patients with CTX can benefit from:

» Low Cholesterol Diet: Avoiding foods high in cholesterol.
» Regular Exercise: Promotes overall health and may help manage symptoms.
» Regular Monitoring: Regular check-ups to measure cholestanol levels, screen for complications, and monitor overall health.

Treatment Options:

Chenodeoxycholic Acid (CDCA): Long-term treatment with the oral bile acid replacement therapy, chenodeoxycholic acid (CDCA,) has been effective in helping to reduce cholestanol levels and manage symptoms. Early treatment in young people without symptoms appears to prevent development of clinical signs and symptoms. Early treatment in symptomatic patients has also been shown to limit progression and to reverse neurologic deficits in some cases. The recommended dose of CDCA for adults is 250 mg three times a day and 15 mg/kg per day divided into three doses for children.

Symptomatic Treatment: This includes treating symptoms such as doing cataract surgery for vision issues or providing physical therapy for problems with movement. Neurologic complications of epilepsy, spasticity, and parkinsonism are treated symptomatically by a neurologist. Xanthomas may be surgically removed for cosmetic reasons.

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