Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Cerebrotendinous Xanthomatosis

Cerebrotendinous Xanthomatosis (CTX) is a rare disorder that affects bile acid synthesis. It is caused by a defect in a gene called CYP27A1 which is essential to produce an enzyme known as sterol 27-hydroxylase. A problem in the production of this enzyme means that cholesterol cannot be converted into a bile acid (chenodeoxycholic acid) which is required for the proper absorption of fats in the intestine. As a result, cholestanol (produced from the breakdown of cholesterol), accumulates in the tissues, tendons nerve cells and membranes which causes damage to various parts of the body and the organs. More than 300 cases of CTX are known worldwide.

Symptoms of CTX may begin in infancy with one of the first signs most commonly being chronic diarrhoea or Cholestasis a condition in which when bile is unable to flow from the gallbladder to the duodenum. Other symptoms may include liver problems, cataracts, and intellectual problems. Fatty growths (xanthomas) are characteristic of this disorder often occurring around the ages of 20-30. Progressive neurological and psychiatric symptoms are also commonly seen in adulthood.

Synonyms

Alternative names for this condition are:

- Cerebral Cholesterinosis
- CTX
- Sterol 27-hydroxylase Deficiency

Further information about this condition is available from Climb.

Disclaimer

Updated 01/08/17

Telephone: **0845 241 2173** Freephone: **0800 652 3181**
Email: contact@climb.org.uk Website: www.climb.org.uk