Cerebrotendinous Xanthomatosis

Other names that are used for this condition are:
- 27 Hydroxylase Deficiency
- CTX

In this disorder there is a deficiency of the enzyme sterol 27-hydroxylase. This enzyme converts certain forms of cholesterol and cholestanol into bile acids. This leads to the accumulation of cholesterol and cholestanol (a substance that is similar to cholesterol) throughout the body. Though these deposits are particularly prone to accumulating in brain and spinal cord, as fatty deposits in tendons (tendon xanthomata), in the lens of the eyes, and as plaques in the arteries (atherosclerotic plaques).

A medication called chenodeoxycholic acid has been found to lower cholesterol and cholestanol levels in the body. This seems to prevent any further progression of the disorder and may also lead to improvements. Sometimes HMG-CoA reductase inhibitors are also given but there can be adverse side effects with this medication. The life span of individuals with this disorder can be shortened due to heart attacks (myocardial infarctions) or progressive neurological disease.

This information is fully sourced and referenced, for more detailed information and references please contact CLIMB by email, letter or telephone.

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