



What is CTX?
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What is CTX? – case presentation

- A 44 year old male from a Moroccan descent is brought to the internal medicine department for investigation of fever.
- The patient is non communicable and totally disabled for a few years D/T CTX disease.
- What is CTX?
- Is it related to the fever?

CTX - CerebroTendinous xanthomatosis

- AR Genetic Disease
- 1.9: 100,000
- More prevalent in north African Jews
- Lipid storage disease. CTX is one of the leukodystrophies

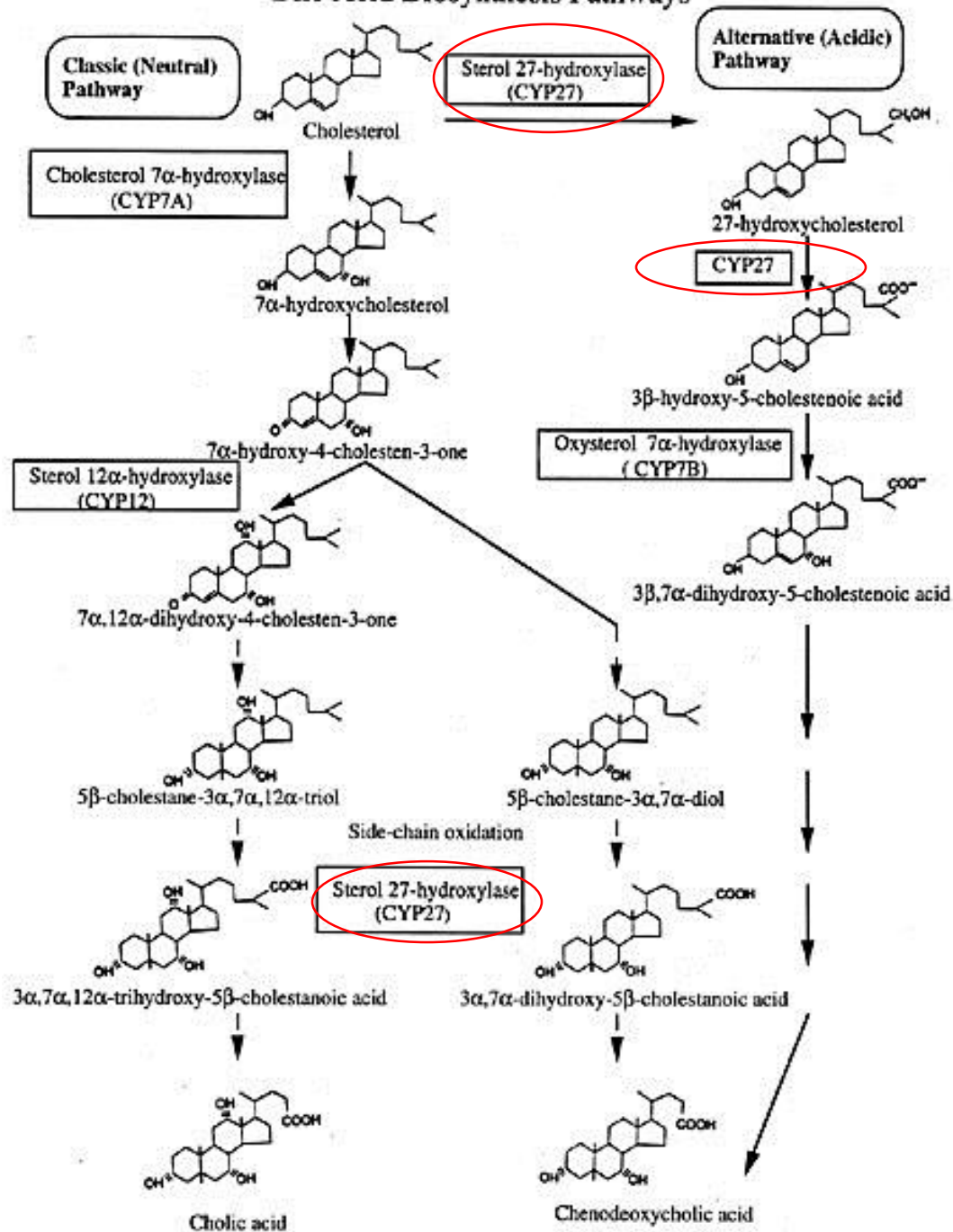
CTX - CerebroTendinous xanthomatosis



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- A deficiency of the enzyme sterol 27-hydroxylase causes the accumulation of cholesterol and cholestanol in virtually all tissues.
- Xanthomas, nodules, or plaques form in the central nervous system, tendons, skin, lungs, and bones.

Bile Acid Biosynthesis Pathways



CerebroTendinous xanthomatosis - Pathogenesis

- pathogenesis unknown
 - Cholesterol deficiency in membrane?
 - Cholestanol toxicity ?
 - Mitochondrial dysfunction?



CerebroTendinous xanthomatosis – Clinical presentation

- Neurologic dysfunction involving cerebral, cerebellar, neuropsychiatric, myelopathic, or peripheral manifestations is usually apparent by late childhood or early adulthood, and progresses during adulthood.



CerebroTendinous xanthomatosis – Clinical presentation

- Xanthomas develop in 70 percent of patients. They can form on the Achilles, extensor elbow, extensor hand, patellar, and neck tendons.
- Xanthomas can also develop in the brain, lungs, and bones.

CerebroTendinous xanthomatosis – Clinical presentation



Fig 1. Achilles tendon xanthomas (arrows): case 1 (A) and case 2 (B).

CerebroTendinous xanthomatosis – Clinical presentation

- Myopathic faces , generalized muscle weakness.
- Chronic infantile diarrhea
- Impaired calcium absorption
- Premature aging
- Hypothyroidism
- Atherosclerosis, mitral valve insufficiency, and lipomatous hypertrophy of the atrial septum

CerebroTendinous xanthomatosis – accessory tests

- Serum and tissue levels of cholestanol whereas serum cholesterol levels are normal or decreased.
- Increased serum lactate is also seen.
- Neuroimaging

CerebroTendinous xanthomatosis - summery

When to suspect?

- Infantile diarrhea
- Cataracts in childhood
- Tendon xanthomas presenting during adolescence.
- Progressive neurologic dysfunction beginning in late childhood.

CerebroTendinous xanthomatosis - treatment

- The mainstay of treatment is chenodeoxycholic acid (CDCA).
- Treatment with statins may be beneficial, but only when combined with CDCA.
- Symptomatic therapies are also important

THANKS



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