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Case Study

CEREBROTENDINOUS XANTHOMATOSIS: HEARD OF IT?

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Abstract

Cerebrotendinous Xanthomatosis (CTX) is a rare, autosomal recessive, lipid storage disorder, characterized by accumulation of cholesterol and cholestanol, predominantly in the brain, spinal cord, peripheral nerves, tendons, lungs, liver and kidneys. The usual conundrum is an amalgam of the primary symptom, as mentioned above, along with features of cerebellar ataxia, juvenile cataract and subcutaneous swellings over tendons. Mental retardation is a very striking and prominent feature in this syndrome.

Prompt diagnosis is vital, as patients benefit from therapy with chenodeoxycholic acid and progress of the disease can be halted prior to irreversible neurological dysfunction.

This is our experience.

Key words: mental retardation, soft tissue swellings, autosomal recessive disorder

Case summary:

A 20-year old male was brought to the hospital with complaints of painless, indolent swellings over his body. It started with one over each of his tendoachilles, 6 years ago, which later progressed to multiple firm non-tender swellings of varying sizes over his legs and hands. He was not able to progress beyond 6th grade because of his learning disability and was advised further evaluation of his condition. He had cataract surgery performed on his right eye 6 years ago for juvenile cataract.

On clinical examination, the swellings were firm, overlying tendons over the extremities (tendoachilles, patellar tendon, extensor aspect of digits and dorsum of hands and feet) and appeared to be arising from the tendons with free mobility over the underlying skin. Each swelling measured from a range between 3 x 4cms to 15 x 14cms in size.

He underwent a basic pre-operative hematological, biochemical and radiological workup, which were essentially normal. The swellings appeared as soft tissue shadows on X-Rays (Figure 2). The swellings were

excised and specimen was sent for histo-pathological examination (HPE).

Retrospective enquiry into the history revealed that, as an infant, he had recurrent bouts of diarrhea that subsided spontaneously. Neurological examination revealed a positive Romberg's sign and ankle clonus, which was otherwise unremarkable.

The pathology was reported as 'Juvenile Xanthogranuloma'. He turned out to be one of the few people afflicted with the syndrome Cerebrotendinous Xanthomatosis. He was followed up on outpatient basis and did not show any recurrence of swellings or progression of symptoms.

Cerebrotendinous Xanthomatosis or Van Bogaert-Scherer-Epstein Disease (Thiébaut's syndrome) was described by van Bogaert and colleagues Hans Joachim Scherer and Emil Epstein. Von Bogaert, a Viennese biochemist documented the neurodegerative disorder in two afflicted cousins and published a 30 year follow up of one of his original patients in 1969.

Cerebrotendinous Xanthomatosis is a lipid storage disease characterized by infantile-onset diarrhea, childhood-onset cataract, adolescent onset tendon xanthomas, and adult-onset progressive neurologic dysfunction[1] (dementia, psychiatric disturbances, pyramidal and/or cerebellar signs, dystonia, atypical parkinsonism, peripheral neuropathy[2], and seizures). Xanthomas appear in the second or third decade; they occur on the Achilles tendon, the extensor tendons of the elbow and hand, the patellar tendon, and the neck tendons. It is a potentially treatable disease if diagnosed prior to neurological insult and treated with chenodeoxycholic acid supplementation[3].

References:

1. Kuriyama M, Fujiyama J, Yoshidome H, et al. Cerebrotendinous xanthomatosis: Clinical and biochemical evaluation of 8 patients and review of literature. *J Neurol Sci*. 1991;102 :225-32.
2. Lee Y, Lin PY, Chin NM, Chang WN, Wen JK. Cerebrotendinous Xanthomatosis with psychiatric disorders: Report of three siblings and literature review. *Chang Gung Med J*.2002;25 :334-40.
3. Berginer VM, Salen G, Shefer S. Long-term treatment of cerebrotendinous xanthomatosis with chenodeoxycholic acid. *N Engl J Med*. 1984;311 :1669-1652.

Figure legend:

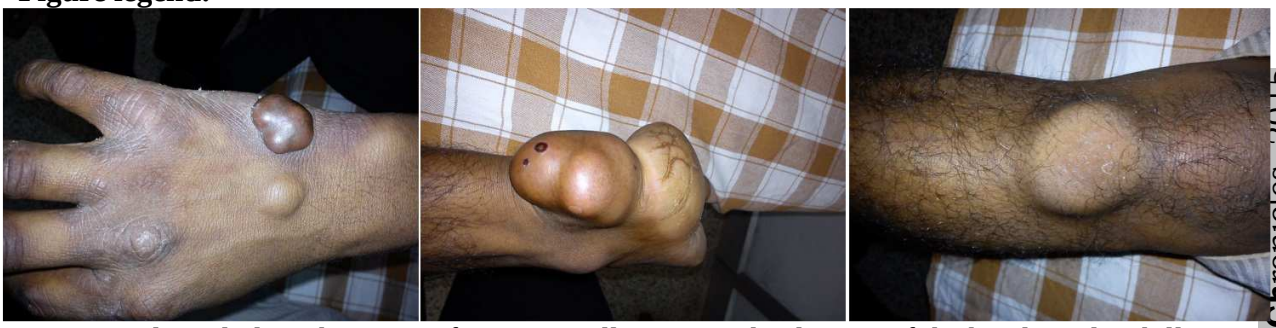

Figure 1: Clinical plate showing soft tissue swellings over the dorsum of the hand, tendoachilles and patellar tendon.

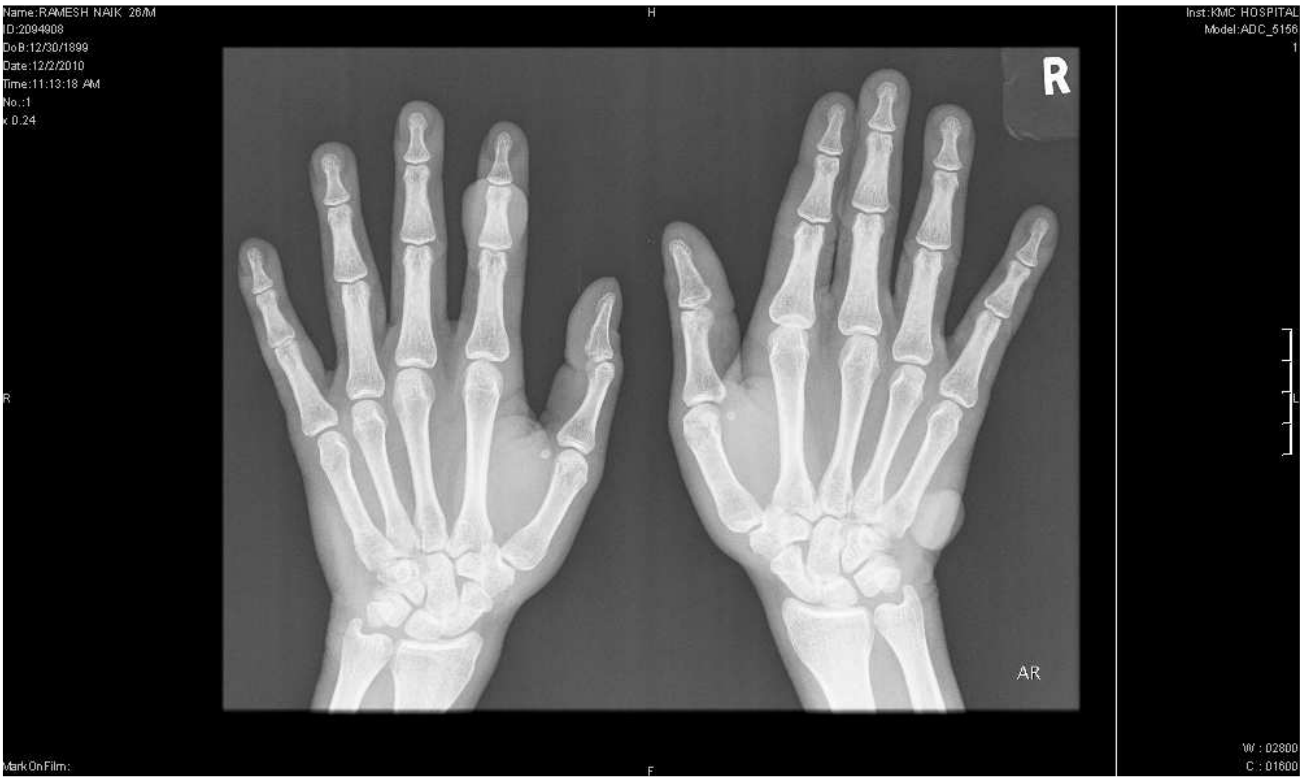

Figure 2: X-ray of both hands showing soft tissue shadow with no osseous involvement.