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Original Research Paper

Pathology

ROLE OF CYTOLOGY IN THE EARLY DIAGNOSIS OF CEREBROTENDINOUS XANTHOMATOSIS -A CASE REPORT

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ABSTRACT Cerebrotendinous xanthomatosis (CTX) is a rare autosomal recessive lipid storage disease associated with abnormally cholesterol (cholestanol) accumulates in the nervous system and other organ systems. Early diagnosis helps in the therapeutic and prognostic outcomes. Here We would like to report one such case of rare occurrence of Cerebrotendinous xanthomatosis . Here cytopathological examination played important role.

KEYWORDS : Cerebrotendinous xanthomatosis, cytopathological examination.

INTRODUCTION -

Cerebrotendinous xanthomatosis (CTX) is a rare autosomal recessive lipid storage disease associated with abnormal cholesterol (cholestanol) accumulates(1) in the nervous system and other organ systems. Early diagnosis helps in the therapeutic and prognostic outcomes. Most cases are diagnosed based on clinico-radiological findings. Here we reported a case of CTX where cytomorphology has played an important role in the early diagnosis of the syndrome.

Case Report -

A 26 year old male came for FNAC from multiple, firm, smooth swellings of both legs, insidious in onset for past five years. (Figure 1) He had bilateral presenile cataracts, subnormal intelligence, progressive cerebellar ataxia and behavioural change from an early age. There was no history of consanguinity or similar illness in the family. X-ray showed soft tissue shadows without any bony involvement. (Figure 2) Blood cholesterol level was within normal range. Stained smears showed foamy histiocytes admixed with touton giant cells having multiple nuclei arranged in a garland like fashion surrounded by foamy cytoplasm. (Figure 3, 4) Some foreign body type of giant cells, few polymorphs and a large number of extracellular cholesterol crystals were present in the background.

Figure 1

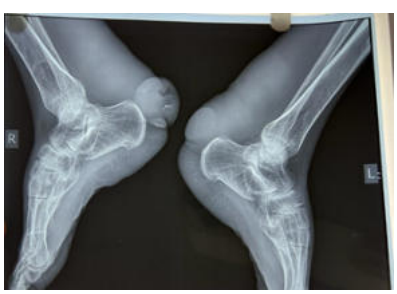

Figure 2

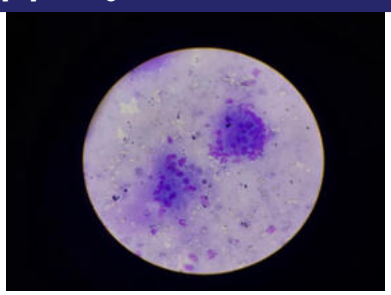

Figure 3

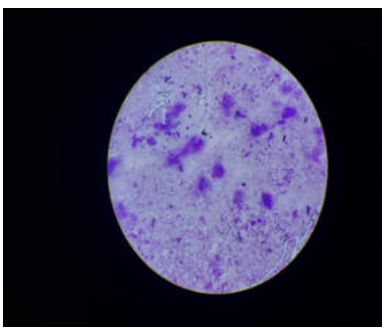

Figure 4

DISCUSSION-

CTX was first described in the medical literature in 1937. It occurs due to a mutation that causes deficiency in an important enzyme in the bile acid synthesis pathway (2) ; sterol 27 hydroxylase. This disease is exceptionally rare in the Indian population. Only about 300 cases have been reported worldwide. We have made the diagnosis on the basis of typical clinical history, normal cholesterol level and characteristic radiological appearance. Clinically CTX resemble Familial Hypercholesteromia & Marinesco-Sjogren syndrome. Normal cholesterol level excludes the former and presence of tendonxanthoma excludes the second one.

CONCLUSION-

In our Indian scenario, where poor patients cannot afford confirmatory genetic analysis, here lies the utility of aspiration cytology for its early diagnosis. Since CTX is a treatable disease and long term therapy with chenodeoxycholic acid (750mg/d) can improve the symptoms, FNAC seems to play a very important role in its better diagnosis

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