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[AN INTERESTING CASE OF ATAXIA]

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INTRODUCTION

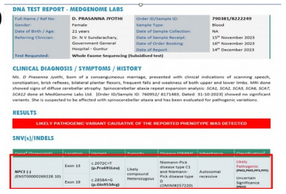
Niemann-Pick disease (NPD), also called sphingomyelin-cholesterol lipidosis, is a group of autosomal recessive disorders associated with splenomegaly, variable neurologic deficits, and the storage of lipids including sphingomyelin and cholesterol. Niemann-Pick disease type C is caused by pathogenic variants of the *NPC1* and *NPC2* genes and it is a lysosomal storage disorder that result in impaired cellular processing and transport of low-density lipoprotein (LDL) cholesterol and other macromolecules, including glycosphingolipids.

AIMS / OBJECTIVES

This is to highlight the importance of Metabolic ataxia apart from Hereditary ataxia in a patient presenting with Chronic progressive ataxia

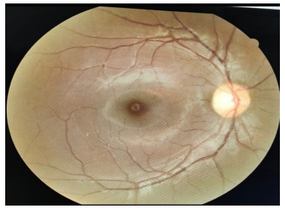
MATERIALS / METHODS

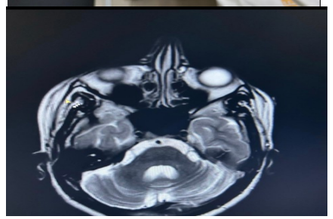
A 23 year old female born out of 2nd degree consanguineous parentage presented to our Department with Unsteadiness while walking since 3 years and tremulousness of both upper limbs on holding the objects ,slurred speech with undue stressing of syllables and weakness of both lower limbs associated with stiffness since 2 years and without any sensory, cranial nerve, autonomic and HMF involvement. She was examined and found to have dystonic posturing of both index fingers, cherry red spots on macula ,absent vertical saccades with intact VOR,grade 3 spasticity in both lower limbs and lower limb power 4/5 on both sides with brisk reflexes and scanned speech,and both limb ataxia and gait ataxia were p


DNA TEST REPORT - MEDGENOME LABS

RESULTS

LIKELY PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED.





RESULTS & DISCUSSION

In view of Early onset ataxia ,consanguineous parentage ,Autosomal recessive ataxia ,cherry red spot on macula ,loss of vertical saccades ,dystonic posturing of fingers .A Clinical diagnosis of Niemann-Pick disease was made and sent for MRI Brain which shown Diffuse Cerebellar atrophy ,USG abdomen : no organomegaly and remaining lab investigations were normal and Whole Exome Sequencing suggestive of Niemann-Pick disease type C1

NPD type C — can present from the perinatal period until late adulthood

Most patients with NPD-C have disease onset in middle to late childhood after normal early development. The clinical manifestations of types Niemann–Pick types C1 and C2 are similar because the respective genes are both involved in egress of lipids, particularly cholesterol, from late endosomes or lysosomes.

The *NPC1* gene is located on CHROMOSOME 18 (18q11-q12)

These patients typically have **cerebellar involvement** characterized by clumsiness and gait problems progressing to frank ataxia, and slow cognitive deterioration .**Vertical supranuclear ophthalmoplegia** is another early manifestation . Progressive **dystonia, dysarthria, and dysphagia** occur, eventually impairing oral feeding, and approximately one- third of patients develop seizures. Death typically occurs from aspiration pneumonia in the second or third decade of life.Adult onset is usually similar to juvenile / childhood cases ,but with slower progression

CONCLUSION

The diagnosis of Niemann-Pick disease can be challenging as clinical features overlap with other more common hereditary ataxia's.The rarity of adult NPC and lack of defined phenotypic spectrum make the clinical diagnosis challenging.So suspicion of NPC should be raised for adult patients presenting with ataxia and vertical supranuclear gaze palsy,to prompt further genetic and biochemic investigation.