J Neurol Neurophysiol 2017, 8:5 (Suppl)
DOI: 10.4172/2155-9562-C1-056

A novel gene in Neimann-Pick type C disease

Harsh Bhardwaj
Army Hospital, Research and Referral, India

Niemann-Pick disease type C (NP-C) is a rare, autosomal recessive and a progressive genetic lysosomal lipid storage disease which is caused by mutations in the NPC1 or NPC2 gene. NPC1 mutations accounts for the majority of the cases which are about 90%. This is a highly heterogeneous disease and has varied clinical manifestations in various combinations involving almost all the body systems. The age at onset and the course can vary greatly from one patient to another. The combination of multiple signs and symptoms shows more diagnostic specificity for NP-C, which may aid with disease detection. There are certain combinations of the signs and symptoms which could strongly favor the diagnosis. These include splenomegaly and Vertical Supranuclear Gaze Palsy (VSGP) among others. VSGP is a hallmark of NP-C and becomes highly specific of the disease when it occurs in combination with other manifestations. Our patient presented with a history of decline in scholastic performance since last 4 years along with multiple episodes of seizures since last one year. A clinical suspicion of Neimann-Pick disease type C was made based on the combination of the symptoms listed above and she was worked up for the same. Two germline variations were identified. One in genomic position chr18:21131633; C>C/G (HET), cDNA position c.1612G>G/C (ENST00000269228) with amino acid change of p.G538R at exon number 10. Second was in genomic position chr18:21118573- 21118575delCCT, cDNA position c.2972_2974delAGG (ENST00000269228) with amino acid change of p.Gln991_Gly992delinsArg at exon number 20. The first variation has been identified in the literature previously as a cause of Neimann-Pick disease type C. The second variation has not been identified so far in the reported cases of the disease and could be a novel variation causing this disease.

bhardwajharsh43@yahoo.com