

A Family Case Report of Niemann Pick C with New Mutation and Different Presentations

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NPC is a metabolic disorder with wide ranges of clinical presentation. There is not definite cause to this variety. In this case presentation, there are three patients in one family with similar mutation in exon 16 of the NPC1 gene, p.Ser826_Leu828del, but different clinical presentations.

We detected a previously unreported mutation in exon 16 of the NPC1 gene, c.2476_2484del (p.Ser826_Leu828del). This is an in-frame mutation that causes the loss of 3 amino acid residues. To date, this variant is not described in the Exome Aggregation Consortium, Exome Sequencing Project or the 1000 Genomes Browser and it is the first time we detect this variant based on Centogene's mutation/variation database (CentoMD®).

Based on this family genetic counseling and other relevant family members, to explain the results and address any concerns by testing a second independent sample from the patient in order to confirm the results (CMGS best practice guidelines) and testing the parents to confirm homozygosity by excluding the presence of a large deletion, also the concentration of the biomarker NPC - 509 is pathologically increased, therefore we considered this mutation as disease-causing.

The first case is a 27 year old female that complaints progressive unsteadiness since 10 years ago, the additional symptoms are dysphagia, dysphonia and impaired eye movement in vertical gaze that causes frequent fallings. In recent neurologic exam she had impaired cerebellar sign in both hands and feet with truncal ataxia normal proprioceptive sense and normal visual acuity. Neurological exam showed normal motor exam and normal DTR. In cranial nerve exam, slow vertical saccade and pursuit in eyes that confirmed by "eye see camera" test and bulbar signs in palatal movements is positive findings also she had near normal mental activity. Paraclinic findings include mild cerebellar atrophy in brain MRI and normal electromyography and nerve conductive study normal electroencephalography normal laboratory findings in liver and thyroid functions and electrolytes and peripheral blood cells and copper metabolism.

The second case is a 25 year old male that is the brother of the first case. His disease presented in psychiatric feature after he was 18 and was graduated in high school, with a psychotic attack then he is on the antipsychotic drugs up to now, he is psychologically control except mild obsession in thought and mental activity is near to normal. In neurologic findings ; no positive findings except impaired vertical eye movement in downward gaze. Only abnormal paraclinical finding is slow downward saccade in "eye see camera" test.

The third case is a 45 year male old that is the son of aunt of the both previous

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cases with the different presentation. His motor milestone was delayed in toddler age but the serious problem that starts in his 4th decade after normal developing goal was frequent and progressive seizure type fallings then follow with speech problem in word finding. Neurologic exam, 10 years after disease starting showed, impaired vertical eye movements in both eyes and mild cognitive impairment and mild dysphonia and dysphagia but there was severe cortical and brain stem atrophy in his brain MRI, no any other paraclinical findings.

Nobody of these cases had history of liver dysfunction in their lifetimes.

Keywords: NPC; New Mutation; Different presentation