CAN ABETALIPOPROTEINEMIA PREVENT THE DEVELOPMENT OF CLINICAL FINDINGS OF NIEMANN PICK TYPE C2; A CASE OF CHOLESTEROL TRAFFIC

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•İNTRODUCTION

•Niemann pick C2 is a neurovisceral storage disease in which intracellular cholesterol transport is impaired and Ldl cholesterol accumulates in lysosomes. The main findings are splenomegaly, upward gaze paralysis, ataxia, dystonia and seizures.

•In Abetalipoproteinemia, another hereditary metabolic disease, there is a problem in the intercellular transport of cholesterol, unlike Niemann pick type c. Fat malabsorption, growth retardation, hepatosteatosis, as well as neurological problems such as cerebellar ataxia and spasticity can be seen.

•We would like to report a patient with abetalipoproteinemia and Nieman pick type C2 (NPC2) having homozygous variants for these diseases, and discuss the effect of the coexistence of two diseases on prognosis.
A 16-month-old female patient was admitted to our clinic with hypocalcemic convulsions. The patient's vitamin D was found to be low, and replacement therapy was given. She had steatorrhea and failure to thrive.

In the medical history of the patient, it was learned that she held his head when he was 5 months old, and sat without support at 10 months. There was first-degree consanguinity between the parents. The patient's height was 63 cm (3p) and his weight was below 6300 g (<3P). She did not have gait or speech.

In his family history, it was determined that his brother received enteral product support due to developmental delay.

In the patient's examinations, total cholesterol 20 mg/dl, triglyceride 23 mg/dl, Hdl cholesterol 24 mg/dl, Ldl cholesterol 1 mg/dl, vitamin D: 7.8 mg/dl, vitamin E: 0.15 mg/dl were found to be very low. Acontocytosis was present in peripheral blood smear.
## Treatment

Diet: A diet containing the followings was adjusted.

- 130 kcal/kg energy
- 61% carbohydrates
- 13% protein
- 26% fat (13% MCT)

Vitamins A, E, D and K supplements were provided by Coagulation parameters.

- 100 lu/kg of vitamin A
- 100 IU kg/day of vitamin E
- 100 IU/kg of vitamin D was started.

After diet and vitamins supplements, she gain weight and she hadn’t steatthore. Neurological development improved.
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Genetic Results

- A whole exome analysis was performed with a preliminary diagnosis of abetalipoproteinemia.
- We found homozygous c.537+5G>A variant in APOB gene and homozygous c.388C>T(p.Leu130Phe) variant in NPC2 gene.
- The same variants in the APOB and NPC2 genes were found to be heterozygous state in both parents.
- The same variant in the ABOP gene was found to be homozygous state in his 4-year-old brother, who was followed up due to developmental delay and no variant was detected in the NPC2 gene.

In the lysosomingolipid panel of the patient, lyso SM 509:4.7 nmol/l (RA 1.9-27.5), specific for niemann pick type C, was found to be normal.

CONCLUSION

We think that the presence of low Ldl cholesterol in the patient due to abetalipoproteinemia may have prevented the accumulation of intracellular Ldl cholesterol in Nieman pick type C2.

No case with these two diseases was reported in the literature review. In the follow-up of the patient, the clinical findings and the intermittent repetition of the lyso SM 509 measurement will be more enlightening. More cases and research on the subject are needed.