ABETALIPOPROTEINEMIA, A RARE DISEASE
A CASE PRESENTATION

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- **BACKGROUND: Abetalipoproteinemia (ABL)** is a rare autosomal recessive disorder caused by biallelic pathogenic mutations in the MTTP gene (v<1/1000000).
  Appears in the first months of life, with delayed physical growth and steatorrhea. It is characterized by low cholesterol values, malabsorption of fat-soluble vitamins, retinal degeneration, neuropathy, coagulation disorders, hepatic steatosis.
- **OBJECTIVE:** To present a newborn with abetalipoproteinemia.
- **METHODS:** A male, full-term, neonate, was born by normal labor. The family's first child, a male newborn, was detected with abetalipoproteinemia (heterozygous form). Now, he is 7 years old and healthy. The second child, was a male newborn, with abetalipoproteinemia, too. During his second year of life, he was died, due to respiratory infection. The third child, was a healthy, male newborn, without abetalipoproteinemia. Our patient was the fourth child of the family. The newborn's first examination, was normal. Due to family history, the neonate was placed on in-hospital monitoring of bowel movements and weight gain, and laboratory examination of lipids and fat-soluble vitamins. Initially he was fed with Alfare milk, then with formula. The newborn showed insufficient weight gain, without vomiting or diarrheal stools, while he was in good general condition, hemodynamically stable. No infections were approved, and thyroid function test was normal. The other laboratory tests: Chol:55mg/dl, HDL:51mg/dl, LDL:2 mg/dl, TRIG:30mg/dl, ApoA :76mg/dl (normal ranges: 104-202mg/dl), ApoB:12mg/dl(normal ranges:66-133mg/dl) , LpA:1,1, 25(OH):11,6ng/dl, VitA:normal, VitE: normal. Genetic testing sent. He started feeding with special milk, high in MCT, and low in HCT. At the same time he received vitD: 1200IU/d, MVW: 0.5 ml/d. Brain ultrasound, ophthalmological, audiological examination were performed, and all were normal.
- **RESULTS:** Molecular testing detected the mutation in the MTP gene, NM_000253c.1813T>C, p.(Tyr605His) in homozygosity.
- **CONCLUSION:** The prognosis of abetalipoproteinemia varies. Early diagnosis and strict adherence to a diet contribute to the recovery of normal nerve function and halting the progression of the disease.