Rickets and dysmorphic findings in a child with abetalipoproteinemia

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ABSTRACT

Abetalipoproteinemia (ABL) is characterized by acanthocytosis, hypcholesterolemia, and steatorrhea. Here, we describe a case of ABL associated with rickets and dysmorphic findings and the subsequent therapeutic course in an 18-month-old male referred for evaluation for failure to thrive and chronic diarrhea. Examination revealed a pale child, dysmorphic face, and signs of rickets. Laboratory examination revealed low hemoglobin (3.7 gm/dl), low albumin (28 gm/L), low cholesterol and triglyceride levels. The blood smear showed acanthocytes while the small bowel histology showed the enterocytes were distended with lipid droplets. He was diagnosed with ABL and treated with fat-soluble vitamins (ADEK), and hydrolyzed protein formula containing medium chain triglycerides. Three months later, his fatty diarrhea becomes normal stool, his serum fat-soluble vitamins normalized, and his weight increased from 4.1 kg to 5.9 kg.

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A betalipoproteinemia (ABL) is a rare autosomal recessive disorder of lipid metabolism characterized by the absence of very low-density lipoproteins (VLDLs) and low-density lipoproteins (LDLs) from plasma, acanthocytosis, and steatorrhea. We present this case to report both rickets and dysmorphic findings together as an initial presentation of ABL, with the subsequent therapeutic course.

Case Report. An 18-month-old Saudi male was referred for evaluation for failure to thrive and chronic diarrhea. He was the product of a full-term, uncomplicated pregnancy, and birth weight of 2.9 Kg. His stools were described as being “oily” since birth, not bloody, and consisted 4 times/day. There was a poor appetite and poor weight gain associated with abdominal distension. There was no history of vomiting, pulmonary complaints, feeding difficulties, or recurrent infections. There was no travelling history or contact with animals. His diet consisted of breast feeding and baby food. Other systemic review was unremarkable. Examination revealed weight of 4.1 Kg (below 5th
Child with abetalipoproteinemia ...

Hasosah et al

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Monogen), which contains 90% of fat as medium chain triglycerides (MCT). He was also received oral high dose of vitamin E 800 I.U/day. Three months later, his fatty diarrhea became normal stool, his serum fat-soluble vitamins were normalized, his hemoglobin of 3.7 gm/dl was improved to 8.7 gm/dl, and his weight increased from 4.1 kg to 5.9 kg.

Discussion. Abetalipoproteinemia (Bassen-Kornzweig syndrome) was first described in 1950 as a condition characterized by acanthocytosis, hypcholesterolemia, progressive combined posterior column degeneration, peripheral neuritis, mental retardation, retinitis pigmentosa, and steatorrhea. This autosomal recessive disorder results from mutations of the microsomal triglyceride transfer protein (MTP), the gene of which maps to 4q22–q24. Patients manifest defective assembly and secretion of apoprotein B (apoB)-containing lipoproteins, leading to the absence of chyomicrons, VLDL, and LDL in the plasma.
Apo(B) is the main apolipoprotein of chylomicrons and LDLs. It occurs in the plasma in 2 main forms: apoB48 and apoB100. The ApoB48 is synthesized exclusively by the gut and apoB100 by the liver. The low plasma cholesterol concentrations are due to the low levels of apoB-containing lipoproteins (VLDL and particularly LDL) that transport most of the cholesterol. In turn, low levels of apoB are due to low production rates of both mutant and wild-type forms of apoB in heterozygotes. Affected children usually present within the first year of life with failure to thrive. Fat malabsorption results in foul-smelling, bulky stools. Other gastrointestinal symptoms include abdominal distention, diarrhea, and vomiting. These symptoms are similar to our patient in addition to signs of rickets, which is an unusual initial presentation of ABL as rickets is due to vitamin D deficiency. Narchi et al described an unexpected initial manifestation of rickets in 2 children with ABL and hypobetalipoproteinemia. The dysmorphic features in our patient, which includes hirsutism, hypertelorism, short nose, long and slightly smooth philtrum, thin upper lip, large mouth, decreased subcutaneous fat and distended abdomen are unusual initial presentations of ABL. Solomon et al described dysmorphic findings in 2 cases of ABL/hypobetalipoproteinemia. Our patient was diagnosed with ABL based on the presence of acanthocytes on peripheral blood smear, low lipid profile (low VLDLs, cholesterol and triglyceride) and characteristic epithelial vacuolization on duodenal biopsy in the context of normal parental cholesterol profiles. Genetic testing to rule out other causes for the dysmorphic features included chromosome analysis on peripheral blood, testing for fragile X syndrome, and fluorescence in-situ hybridization testing around the MTP locus to assess for a small microdeletion, and fluorescence in-situ hybridization testing around the MTP locus to assess for a small microdeletion, and fluorescence in-situ hybridization testing around the MTP locus to assess for a small microdeletion, and fluorescence in-situ hybridization testing around the MTP locus to assess for a small microdeletion, and fluorescence in-situ hybridization testing around the MTP locus to assess for a small microdeletion, and fluorescence in-situ hybridization testing around the MTP locus to assess for a small microdeletion, and fluorescence in-situ hybridization testing around the MTP locus to assess for a small microdeletion, and fluorescence in-situ hybridization testing around the MTP locus to assess for a small microdeletion, and fluorescence in-situ hybridization testing around the MTP locus to assess for a small microdeletion. The low plasma cholesterol levels caused by a short deletion in the apolipoprotein B gene.

In summary, we report a rare lipid disorders (ABL), characterized by acanthocytosis, low lipid profile, and characteristic fat droplets on duodenal biopsy. In addition, our case reports the presence of both rickets and dysmorphic finding in ABL. Early diagnosis and initiation of treatment of ABL offers the best chance for improved outcome.

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References