Abetalipoproteinemia

Abetalipoproteinemia, also known as Bassen-Kornzweig syndrome, is an autosomal recessive medical condition that interferes with the normal absorption of fat and fat-soluble vitamins. This condition is caused by a deficiency of apolipoproteins B-48 and B-100 from a mutation in the microsomal triglyceride transfer protein gene, or MTTP. These patients develop a complete deficiency of chylomicrons, VLDL, and LDL. Early symptoms include malabsorption, steatorrhea, and failure to thrive, while late symptoms include ataxia, retinitis pigmentosa and spinocerebellar degeneration. Diagnostic signs include lipid-laden enterocytes on intestinal biopsy, acanthocytes, and decreased plasma cholesterol and LDL levels. Treatment includes large doses of oral vitamin E and restriction of long-chain fatty acids.



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Pathophysiology

Autosomal Recessive

Recessive-chocolate

This condition is inherited in an autosomal recessive manner. This means that for the child to manifest the disease, they must inherit two abnormal alleles.

Deficiency of Apolipoproteins B-48 and B-100

Broken Apple-lips-protein (B) Bee (40) oz and (B) Bee (100) Dollar-bill

This condition is caused by a mutation in microsomal triglyceride transfer protein resulting in deficiencies of apolipoprotein B-48 and apolipoprotein B-100. These proteins are used in the synthesis and transportation of chylomicrons and VLDL, respectively.

MTTP Gene Mutation

MounTain ToP Jeans Mutant

Mutation in microsomal triglyceride transfer protein (MTTP) is inherited in an autosomal recessive manner and results in the deficiency of apolipoprotein B-48 and apolipoprotein B-100.

Clinical Features

Malabsorption

Intestine-mallet

Malabsorption is defined as an abnormal absorption of nutrients in the gastrointestinal tract. Malabsorption of fat and fat-soluble vitamins (vitamins A, D, E, K) is particularly pronounced in patients with abetalipoproteinemia.

Steatorrhea

Steak-diarrhea

Steatorrhea is characterized by the excretion of abnormal quantities of fat with feces owing to reduced absorption of fat by the intestines. This is common in patients with abetalipoproteinemia due to the malabsorption of fat and fat-soluble vitamins (A, D, E, K).

Failure to Thrive

Very Skinny Baby

Infants with this condition often present with failure to thrive because of malabsorption of fat-soluble vitamins and calories.

Ataxia

A-taxi

Ataxia is one of the possible manifestations of abetalipoproteinemia.

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Retinitis Pigmentosa

Red-tin-eyes Piggy-bank

Retinitis pigmentosa is a group of genetic disorders that involves a breakdown and loss of cells in the retina. Common symptoms include difficulty seeing at night (nyctalopia) and a loss of peripheral vision. This condition is associated with abetalipoproteinemia.

Spinocerebellar Degeneration

Broken-silver-bell-tract

Spinocerebellar degeneration is one of the late manifestations of abetalipoproteinemia; this can lead to balance problems and ataxia.

Diagnosis

Lipid-Laden Enterocytes

Fat Lips Biopsy needle Stuck In Fatty Intestine

Intestinal biopsy can be used for the confirmation of the diagnosis and reveals lipid-laden enterocytes.

Acanthocytes

A-thorny-cell

Acanthocytes are abnormal red blood cells that have spiked cell membranes made up of thorny projections and are frequently found on peripheral blood smears of patients with abetalipoproteinemia. Acanthocytes can also be seen in patients with severe liver dysfunction and malnutrition.

Decreased Cholesterol and LDL

Down-arrow Ladybug with Cholesterol Burger

Due to the severe fat malabsorption, this condition is characterized by decreased levels of plasma cholesterol (< 30 mg/dl) and undetectable levels of LDL.

Treatment

Vitamin E (Alpha-Tocopherol)

Viking Electric with Afro-tacos-for-all

Large doses of oral vitamin E and restriction of long-chain fatty acids in the diet are used in the treatment of this condition.

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