

Chylomicron retention disease

Subjects: Genetics & Heredity

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Chylomicron retention disease is an inherited disorder that impairs the normal absorption of fats, cholesterol, and certain vitamins from food.

Keywords: genetic conditions

1. Introduction

The features of chylomicron retention disease primarily affect the gastrointestinal system and nervous system.

Chylomicron retention disease begins in infancy or early childhood. Affected children have slow growth and weight gain, frequent (chronic) diarrhea, and foul-smelling stools (steatorrhea). They also have reduced blood cholesterol levels (hypocholesterolemia). Some individuals with chylomicron retention disease develop an abnormal buildup of fats in the liver called hepatic stenosis and can have an enlarged liver.

Other features of chylomicron retention disease develop later in childhood and often impair the function of the nervous system. Affected people may develop decreased reflexes (hyporeflexia) and a decreased ability to sense vibrations. Rarely, affected individuals have heart abnormalities or muscle wasting (amyotrophy).

2. Frequency

Chylomicron retention disease is a rare condition with approximately 50 cases described worldwide.

3. Causes

Mutations in a gene called *SAR1B* cause chylomicron retention disease. The *SAR1B* gene provides instructions for making a protein that is needed for the transport of molecules called chylomicrons. During digestion, chylomicrons are formed within cells called enterocytes that line the small intestine and absorb nutrients. Chylomicrons are needed to absorb fat-soluble vitamins and carry fats and cholesterol from the small intestine into the bloodstream.

SAR1B gene mutations cause the retention of chylomicrons within enterocytes and prevent their release into the bloodstream. Impaired chylomicron transport causes severely decreased absorption (malabsorption) of dietary fats and fat-soluble vitamins, leading to nutritional and developmental problems in people with chylomicron retention disease. Affected individuals are unable to absorb sufficient fats, cholesterol, and vitamins that are necessary for normal growth and development.

3.1. The Gene Associated with Chylomicron Retention Disease

- *SAR1B*

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- Anderson disease
- Anderson syndrome

- CMRD
 - hypobetalipoproteinemia with accumulation of apolipoprotein B-like protein in intestinal cells
 - lipid transport defect of intestine
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