Chylomicron retention disease

Description

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

Frequency

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

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.

Learn more about the gene associated with Chylomicron retention disease

- SAR1B

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.

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

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chylomicron retention disease (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0795956/)

Genetic and Rare Diseases Information Center

- Chylomicron retention disease (https://rarediseases.info.nih.gov/diseases/9683/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Chylomicron retention disease%22)

Catalog of Genes and Diseases from OMIM

- CHYLOMICRON RETENTION DISEASE; CMRD (https://omim.org/entry/246700)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28chylomicron+retention+disease%5BTIAB%5D%29+OR+%28cmrd%5BTIAB%5D%29+OR+%28anderson+disease%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

References

- Burnett JR, Hooper AJ, Hegele RA. Chylomicron Retention Disease. 2022 Mar 24. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews(R) [Internet]. Seattle (WA): Universityof Washington, Seattle; 1993-2024. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK578949/ Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/35344313)
- Charcosset M, Sassolas A, Peretti N, Roy CC, Deslandres C, Sinnett D, Levy E, Lachaux A. Anderson or chylomicron retention disease: molecular impact of fivemutations in the SAR1B gene on the structure and the functionality of Sar1bprotein. Mol Genet Metab. 2008 Jan;93(1):74-84. doi: 10.1016/j.ymgme.2007.08.120.Epub 2007 Oct 22. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17945526)
- Hesse D, Jaschke A, Chung B, Schurmann A. Trans-Golgi proteins participate inthe control of lipid droplet and chylomicron formation. Biosci Rep. 2013 Feb22;33(1):1-9. doi: 10.1042/BSR20120082. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23033902) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3522472/)
- Levy E. Insights from human congenital disorders of intestinal lipidmetabolism. J Lipid Res. 2015 May;56(5):945-62. doi: 10.1194/jlr.R052415. Epub2014 Nov 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25387865) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4409285/)
- Sane AT, Seidman E, Peretti N, Kleme ML, Delvin E, Deslandres C, Garofalo C, Spahis S, Levy E. Understanding Chylomicron Retention Disease Through Sar1bGtpase Gene Disruption: Insight From Cell Culture. Arterioscler Thromb Vasc Biol.2017 Dec;37(12):2243-2251. doi: 10.1161/ATVBAHA.117.310121. Epub 2017 Oct 5. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/28982670)
- Silvain M, Bligny D, Aparicio T, Laforet P, Grodet A, Peretti N, Menard D,Djouadi F, Jardel C, Begue JM, Walker F, Schmitz J, Lachaux A, Aggerbeck LP,Samson-Bouma ME. Anderson's disease (chylomicron retention disease): a newmutation in the SARA2 gene associated with muscular and cardiac abnormalities.Clin Genet. 2008 Dec;74(6):546-52. doi: 10.1111/j.1399-0004.2008.01069.x. Epub2008 Sep 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18786134)

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