Climb™

Supporting those affected by Inherited Metabolic Disorders

Chylomicron Retention Disease

Chylomicron Retention Disease is a rare disorder caused by a defect in the SAR1B gene which provides instructions for the Sar1b protein. This is needed to help molecules called chylomicrons to carry fats and cholesterol (lipids) from the intestine to the bloodstream and to absorb certain vitamins. This disorder causes malabsorption of dietary fats and fat-soluble vitamin which are vital for normal growth and development. Chylomicron Retention Disease usually presents in infancy or early childhood with chronic diarrhoea, vomiting, abdominal swelling and failure to grow or gain weight (failure to thrive). Neurological symptoms may appear during late childhood. Fatty liver disease is common and an enlarged liver is seen in approximately 20% of cases.

Synonyms

Alternative names for this condition are:

- Anderson Disease
- CRD

Further information about this condition is available from Climb.

Disclaimer

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