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Cholesteryl ester storage disease - ScienceDirect.com

Cholesteryl ester storage disease (CESD) is caused by deficient lysosomal acid lipase ... in the **literature**, mostly in single **case reports** or small series of biochemically, ... Therefore, the **clinical**, pathologic, biochemical, and molecular **genetic** ... to develop ERT awaited the generation and/or **characterization** of murine CESD ...

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Cholesteryl ester storage disease: review of the ... - NCBI

Feb 26, 2013 - **Cholesteryl ester storage disease** (CESD) is caused by deficient lysosomal ... in 135 CESD patients described in the **literature** are **reviewed**. Diagnoses were based on liver biopsies, LAL deficiency and/or LAL **gene** (LIPA) mutations. ... in animal models, and recently, a phase I/II **clinical** trial demonstrated its ...

Missing: characterisation: case

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Targeting Wolman Disease and Cholesteryl Ester Storage ...

Jan 30, 2017 - Keywords: Wolman disease, **Cholesteryl ester storage disease**, Lysosomal ... In contrast to CESD, WD is an infantile onset disease and **clinical** presentation ... The LIPA **gene** contains 10 exons, 9 introns and is 36kb long. ... In a **review** of 135 **cases** of CESD reported in the **literature**, Bernstein et al. noted ...

books.google.co.kr › books

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 54228

Manuscript Type: CASE REPORT

Cholesteryl ester storage disease of clinical and genetic characterisation: A case report and review of literature

Rashu EB *et al.* Clinical and genetic characterisation of CESD

Elias Badal Rashu, Anders Ellekær Junker, Karen Vagner Danielsen, Emilie Dahl, Ole Hamberg, Line Borgwardt, Vibeke Brix Christensen, Nicolai J Wewer Albrechtsen, Lise L Gluud

Abstract

BACKGROUND

Cholesteryl ester storage disease (CESD) is a rare genetic disease. Its symptoms and severity are highly variable. CESD is a systemic disease that can lead to the accumulation of fat and inflammation in the liver, as well as gastrointestinal and cardiovascular disease. The majority of patients require liver transplantation due to decompensated cirrhosis. Enzyme replacement therapy has been approved based on a randomized trial. Our study aims to

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Cholesteryl ester storage disease: a rare and possibly ...

<https://jcp.bmj.com/content/66/11/918> ▼

Nov 01, 2013 · **Cholesteryl ester storage disease (CESD)** is an **autosomal recessive lysosomal storage disorder** caused by a variety of mutations of the LIPA gene. These cause reduced activity of lysosomal acid lipase, which results in accumulation of **cholesteryl esters** in lysosomes.

Cited by: 30

Author: Tim Reynolds

Publish Year: 2013

Cholesteryl Ester Storage Disease - an overview ...

<https://www.sciencedirect.com/.../medicine-and-dentistry/cholesteryl-ester-storage-disease>

Wolman disease and **cholesteryl ester storage disease** (also called '**cholesterol**' **ester storage disease** in older literature) are two disorders caused, respectively, by absent or by reduced (3–8%) activity of the enzyme lysosomal acid lipase. 1486,1487 The enzyme is essential for the intralysosomal metabolism of **cholesterol esters** and triglycerides, namely, their uptake by ...

Cholesteryl Ester Storage Disease: An underdiagnosed cause ...

<https://www.sciencedirect.com/science/article/pii/S1092913417300382>

Cholesteryl Ester Storage Disease (CESD), is a rare **multisystem autosomal recessive disorder** and belongs to the broad family of **lysosomal storage disorders**. It can present anytime from infancy and childhood to even adulthood. The **clinical manifestations** are generally severe in infants and with milder forms in adults.

Cited by: 5

Author: Mamta Pant, Kiyoko Oshima

Publish Year: 2017



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Cholesteryl ester storage disease (CESD) is a type of lysosomal acid lipase (LAL) deficiency; a rare genetic disorder characterized by a deficiency of the lysosomal acid lipase (LIPA or LAL) enzyme. This enzyme is essential for hydrolysis of triglycerides and cholesteryl esters in lysosomes.

Cholesteryl Ester Storage Disease - NORD (National ...

rarediseases.org/rare-diseases/cholesteryl-ester-storage-disease/

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Cholesteryl ester storage disease (CESD) is a type of **lysosomal acid lipase (LAL) deficiency**; a rare genetic disorder characterized by a **deficiency** of the **lysosomal acid lipase (LIPA or LAL)** enzyme. This enzyme is essential for hydrolysis of triglycerides and cholesteryl esters in lysosomes.

Cholesteryl ester storage disease | Genetic and Rare ...

<https://rarediseases.info.nih.gov/diseases/12099/cholesteryl-ester-storage-disease> ▼

Apr 14, 2017 · Cholesteryl ester storage disease is a type of **lysosomal acid lipase deficiency**. It is an inherited **disease** that causes a buildup of fats (lipids) in the tissues and organs of the body and calcium deposits in the adrenal glands. The **liver** is most severely affected in most cases.

Cholesteryl Ester Storage Disease (CESD) due to novel ...

<https://www.ncbi.nlm.nih.gov/pubmed/19307143>

Cholesteryl Ester Storage Disease (CESD) is a rare recessive disorder due to mutations in **LIPA gene** encoding the **lysosomal acidic lipase (LAL)**. CESD patients have **liver disease** associated with mixed hyperlipidemia and low plasma levels of high-density lipoproteins (HDL).

Cited by: 79

Author: Livia Pisciotta, Raffaele Fresca, Antonel...