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www.ncbi.nlm.nih.gov › articles › PMC6339326 이 페이지 번역하기

Novel compound heterozygote mutations of TJP2 in a ... - NCBI

T Ge 저술 - 2019 - 5회 인용 - 관련 학술자료

2019. 1. 18. - Genetic mutations in **tight junction protein 2** (TJP2) are linked to PFIC **4** with low or normal levels gamma-glutamyltransferase (GGT), and most of reported cases were homozygous mutations [6].

Abstract · Background · Discussion and conclusions · Abbreviations

www.ncbi.nlm.nih.gov › articles › PMC4061468 이 페이지 번역하기

Mutations in TJP2 cause progressive cholestatic liver disease

M Sambrotta 저술 - 2014 - 144회 인용 - 관련 학술자료

2014. 3. 9. - Common to all patients is early-onset **progressive** liver disease. PFIC may be subdivided based on levels of serum γ-glutamyl transferase activity (GGT)[2-4]. ... FIC1 (**familial intrahepatic cholestasis protein** 1) deficiency is **caused** by mutations in ATP8B1, and BSEP (bile salt export pump) deficiency by mutations in ABCB11.

누락된 검색어: ~~adults~~ | 다음 정보가 포함되어야 합니다. adults

Abstract · Online Methods · Acknowledgments

omim.org › entry 이 페이지 번역하기

OMIM Entry - * 607709 - TIGHT JUNCTION PROTEIN 2; TJP2

The TJP2 gene encodes **tight junction protein-2**, which belongs to a family of ... and breast cancer cell lines and in pancreatic adenocarcinoma of the ductal **type**. ... In a patient with **progressive**

Name of Journal: *World Journal of Gastroenterology*

Manuscript NO: 51702

Manuscript Type: CASE REPORT

New tight junction protein 2 variant causing progressive familial intrahepatic cholestasis type 4 in adults

Chun-Shan Wei, Naja Becher, Jenny Blechingberg Friis, Peter Ott, Ida Vogel, Henning Grønbæk

Abstract

BACKGROUND

Progressive familial intrahepatic cholestasis (PFIC) encompasses a group of autosomal

国内版 国际版

New tight junction protein 2 variant causing progressive f

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12,100 Results Any time

Progressive familial intrahepatic cholestasis

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2647530

Jan 08, 2009 · **Cholestasis** is **due** to impaired transport of unconjugated bile acids into bile and to bile leakage into plasma through abnormal **canalicular tight junctions** increasing paracellular permeability. Another category of **progressive cholestastic liver disease** of childhood could be **due** ...

Cited by: 371 **Author:** Anne Davit-Spraul, Emmanuel Gonzales, ...
Publish Year: 2009

Progressive familial intrahepatic cholestasis: diagnosis ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6136920

Sep 10, 2018 · In the patients, pruritus is probably induced by the stimulation of nonmyelinated subepidermal free nerve ends because of increased serum bile acids.2. **Progressive familial intrahepatic cholestasis** (PFIC) is a heterogeneous group of liver disorders of autosomal recessive **inheritance**, characterized by an early onset of **cholestasis** (usually during infancy) with pruritus and malabsorption, which rapidly progresses and ends up as liver failure.3,4 ...

Cited by: 1 **Author:** Mithat Gunaydin, Asudan Tugce Bozkurt...
Publish Year: 2018

Progressive Familial Intrahepatic Cholestasis | Children's ...

https://childliverdisease.org/.../progressive-familial-intrahepatic-cholestasis

Recently mutations in the **tight junction protein 2** (**TJP 2**) gene have been described in children who behaved like **PFIC 1** or **2** with a normal GGT. This has also been called **PFIC 4**. A mild form of **liver disease** associated with mutations in the **TPJ 2 gene** was previously called **familial hypercholanaemia**.

Familial intrahepatic cholestasis: New and wide ...

https://www.sciencedirect.com/science/article/pii/S1590865819305535

Progressive familial intrahepatic cholestasis (PFIC) represents a heterogeneous group of autosomal recessive disorders of childhood, characterized by **intrahepatic cholestasis due** to defects of both bile synthesis and transport [1,2]. PFIC can evolve rapidly leading to portal hypertension, liver failure, liver cancer and/or extrahepatic manifestations within the first ten years of life.

Cited by: 2 **Author:** Giovanni Vitale, Stefano Gitto, Ranka Vu...
Publish Year: 2019

(PDF) Familial intrahepatic cholestasis: New and wide ...

https://www.researchgate.net/publication/333158018_Familial_intrahepatic_cholestasis

Progressive familial intrahepatic cholestasis

Progressive familial intrahepatic cholestasis is a group of familial cholestatic conditions caused by defects in biliary epithelial transporters. The clinical presentation usually occurs first in childhood with progressive cholestasis. This usually leads to failure to thrive, cirrhosis, and the need for liver transplantation.

Wikipedia

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Alpha 1-antitrypsin deficiency

Stargardt disease

Dubin–Johnson syndrome

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Data from: Wikipedia

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