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Oct 15, 2001 · **Genetic counseling**. **ATP8B1 deficiency** is **inherited** in an autosomal recessive manner. The parents of an affected individual are generally obligate carriers of a **pathogenic variant**. **Intrahepatic cholestasis** of **pregnancy** (**ICP**) has been **reported** occasionally in mothers of some individuals with **ATP8B1 deficiency**.

Characterization of mutations in ATP8B1 associated with ...

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Mutations in **PFIC** Patients. In 28 families, patients were **homozygous** for an **ATP8B1 mutation** (Table 2 A); in 9 families, patients were compound **heterozygotes** (Table 2 B); and in 2 families, we detected a **mutation** on only one allele (Table 2 C). None of these **mutations** was identified in control samples.

Cited by: 243 **Author:** Leo W. J. Klomp, Julie C. Vargas, Saski...
Publish Year: 2004

Benign recurrent intrahepatic cholestasis - Genetics Home ...

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Oct 29, 2019 · Mutations in the **ATP8B1 gene** cause **benign recurrent intrahepatic cholestasis** type 1 (BRIC1), and mutations in the ABCB11 **gene** cause **benign recurrent intrahepatic cholestasis** type 2 (BRIC2). These two **genes** are involved in the release (secretion) of bile, a fluid produced by the liver that helps digest fats.

Name of Journal: *World Journal of Hepatology*
Manuscript NO: 52712
Manuscript Type: CASE REPORT

Nonsense variant of *ATP8B1* gene in heterozygosis and benign recurrent intrahepatic cholestasis: A case report and review of literature

Mariano Piazzolla, Nicola Castellaneta, Antonio Novelli, Emanuele Agolini, Dario Cocciadiferro, Leonardo Resta, Loren Duda, Michele Barone, Enzo Ierardi, Alfredo Di Leo

Abstract

BACKGROUND

Benign recurrent intrahepatic cholestasis is a genetic disorder with recurrent cholestatic jaundice due to *ATP8B1* and *ABCB11* gene mutations encoding for hepato-canalicular transporters. Herein, we firstly provide the evidence that a nonsense variant of *ATP8B1* gene (c.1558A>T) in heterozygous form is involved in BRIC pathogenesis.

CASE SUMMARY

A 29-year-old male showed severe jaundice and laboratory tests consistent with intrahepatic cholestasis despite normal Gamma-glutamyltranspeptidase. Acute and chronic liver diseases with viral, metabolic and autoimmune etiology were excluded.

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2001. 10. 15. - Intrahepatic cholestasis of pregnancy (ICP) has been reported .... Inuit; see Molecular **Genetics**), **ATP8B1** sequence **analysis** can replace steps 2 and 3. .... have been termed "**benign recurrent intrahepatic cholestasis** type 1" ... A **case report** suggests that heterozygotes for an **ATP8B1** ..... **Literature** Cited.

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ATP8B1 mutations in British cases with intrahepatic ... - NCBI

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Homozygous mutations in the **ATP8B1 gene** cause cholestasis with a normal serum ... cholestasis type 1 (PFIC1) and **benign recurrent intrahepatic cholestasis** (BRIC). ... The **study** conformed to the guidelines outlined by the 1975 Declaration of ... Screening of 182 ICP **cases** for **ATP8B1 variants** previously reported in PFIC ...

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The Identification of Two New ABCB11 Gene Mutations and ...

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Publish Year: 2014

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[PDF] Benign Recurrent Intrahepatic Cholestasis with a Single ...

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