Title (en)

NUCLEIC ACID CORRESPONDING TO MUTATION ASSOCIATED WITH CHOLESTASIS SYNDROMES

Title (de)

DER MIT CHOLESTASE SYNDROMEN ASSOZIIERTEN MUTATION ENTSPRECHENDE NUCLEISCHE SÄURE

Title (fr)

ACIDE NUCLEIQUE CORRESPONDANT A UNE MUTATION ASSOCIEE A DES CHOLESTASES

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Application

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Abstract (en)

[origin: WO9936533A2] The invention relates to the field of gastroenteral disease in humans, more specifically to the field of liver disease, bile formation and bile acid secretion. In particular, it relates to gastroenteral disorders characterised by cholestasis, as for example seen with benign recurrent intrahepatic cholestasis and progressive familial intrahepatic cholestasis, or other disorders related to impaired bile flow or bile acid secretion, particularly in mammals. The invention provides an isolated and/or recombinant nucleic acid, or a functional fragment or homologue thereof, derived from a gene, which gene comprises a mutation in patients with benign recurrent intrahepatic cholestasis (BRIC) or progressive familial intrahepatic cholestasis (PFIC), said nucleic acid in humans having a sequence substantially identical to a nucleic acid sequence as shown in Fig. 5.

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