

Inborn Errors of Bile Acid Metabolism - PubMed

<https://pubmed.ncbi.nlm.nih.gov/30266156>

Inborn errors of bile acid metabolism are rare causes of neonatal cholestasis and liver disease in older children and adults. The **diagnosis** should be considered in the context of hyperbilirubinemia with normal serum **bile** acids and made by urinary liquid secondary ionization mass spectrometry or DNA testing.

Cited by: 12

Author: James E. Heubi, Kenneth D.R. Setch...

Publish Year: 2018

Bile Acid Synthesis Disorders - NORD (National ...

<https://rarediseases.org/rare-diseases/bile-acid-synthesis-disorders> ▾

Bile acid synthesis disorders result from improper **synthesis of bile** acids, particularly the two primary **bile** acids, cholic **acid** and chenodeoxycholic **acid**. The principal **bile** acids are synthesized by the liver through a series of complex chemical reactions involving at least 17 enzymatic steps.

Estimated Reading Time: 10 mins

PEOPLE ALSO ASK

How are disorders of bile acid synthesis (BASD) treated? ▾

What are the different types of bile acid synthesis? ▾

What are the symptoms of bile acid synthetic defects? ▾

Which is the final step in bile acid synthesis? ▾

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Identification of Genetic D



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Inborn errors of bile acid metabolism

<https://pubmed.ncbi.nlm.nih.gov/1749214>

However, diagnosis of this disorder is problematical; a similar pattern of metabolite excretion can occur as a result of liver damage caused by viruses or inborn errors of pathways unrelated to bile acid synthesis. Defective side chain oxidation in patients with cerebrotendinous xanthomatosis (CTX) leads to synthesis of bile alcohols such as 5 ...

Cited by: 82

Author: P. T. Clayton

Publish Year: 1991

PEOPLE ALSO ASK

What causes bile to be yellow in color?



Why is differential diagnosis of cholestatic liver injury important?



What kind of bilirubin level is cholestasis?



How much bile is reabsorbed in the liver?



Feedback

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https://www.accessdata.fda.gov/drugsatfda_docs/nda/...

synthesis. The applicant obtained Orphan Designation for cholic acid treatment of patients with inborn errors of cholesterol and bile acid synthesis and metabolism from the Office of Orphan Products Development (OPPD) in 2007. This rare condition has an estimated prevalence of less than 100 total