

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.

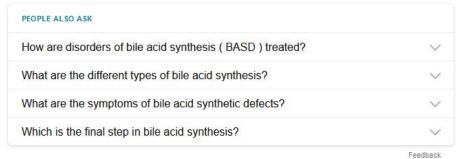
Cited by: 12 Author: James E. Heubi, Kenneth D.R. Setch...
Publish Year: 2018

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.

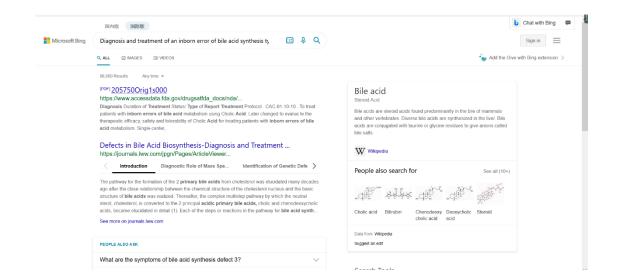
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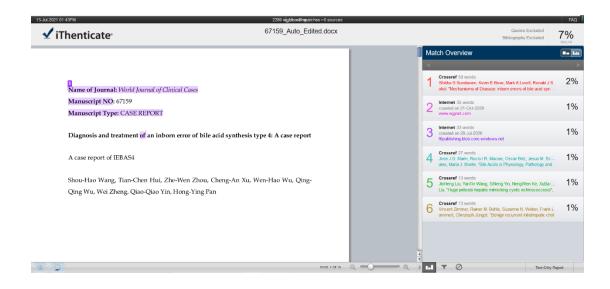


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Defects in Bile Acid Biosynthesis-Diagnosis and Treatment ... https://journals.lww.com/jpgn/Pages/ArticleViewer...

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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



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