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Liver and the defects of cholesterol and bile acids biosynthesis: Rare disorders many diagnostic pitfalls

Corso G *et al.* Cholesterol and bile acids biosynthesis defects

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Abstract

In recent decades, biotechnology produced a growth of knowledge on the causes and mechanisms of metabolic diseases that have formed the basis for their study, diagnosis and treatment. Unfortunately, it is well known that the clinical features of metabolic diseases can manifest themselves with very different characteristics and escape early detection. Also, it is well known

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Bile Acid Synthesis Disorders - NORD (National Organization for Rare ...

<https://rarediseases.org> > For Patients and Families > Rare Disease Information ▼ [翻译此页](#)

Bile acids are formed from cholesterol and, therefore, bile acid synthesis ... In many cases, symptoms or signs are present at birth or during the newborn period. ... Vitamin E deficiency from undiagnosed liver disease may contribute to neurological disease. Defects in bile acid biosynthesis – diagnosis and treatment.

Mechanisms of Disease: inborn errors of bile acid synthesis - NCBI

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作者: SS Sundaram - 2008 - 被引用次数: 51 - 相关文章

2008年6月24日 - Inborn errors of bile acid synthesis are rare genetic disorders that can present as ... Failure to diagnose any of these conditions can result in liver failure or ... Importantly, many bile acid synthesis defects are readily treatable and ... the different pathways involved in the modification of cholesterol to bile acids.

Inherited Disorders of Bile Acid Transport or Synthesis - NCBI

<https://www.ncbi.nlm.nih.gov> > NCBI > Literature > PubMed Central (PMC) - [翻译此页](#)

作者: WF Balistreri - 2007 - 被引用次数: 3 - 相关文章

Molecular Defects in Bile Acid Transport or Synthesis that Manifest as ... G&H Could you describe how these defects affect liver function? ... of bile acids (the main motive force driving bile formation) from cholesterol or ... WB There is a notion that these are extraordinarily rare disorders that are only detected in newborns.

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Bile Acid Synthesis Disorders - NORD (National Organization for Rare ...

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Bile acids are formed from cholesterol and, therefore, bile acid synthesis ... In many cases, symptoms or signs are present at birth or during the newborn period. ... Vitamin E deficiency from undiagnosed liver disease may contribute to neurological disease. Defects in bile acid biosynthesis – diagnosis and treatment.

Mechanisms of Disease: inborn errors of bile acid synthesis - NCBI - NIH

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作者：SS Sundaram - 2008 - 被引用次数：54 - 相关文章

2008年6月24日 - Inborn errors of bile acid synthesis are rare genetic disorders that can present as ... Failure to diagnose any of these conditions can result in liver failure or ... Importantly, many bile acid synthesis defects are readily treatable and ... the different pathways involved in the modification of cholesterol to bile acids.

Bile Acid Synthesis Disorders - WebMD

www.webmd.com › Children's Health › Reference ▼ [翻译此页](#)

2014年2月25日 - Bile acid synthesis disorders (BASDs) are a group of rare metabolic disorders characterized by defects in the creation (synthesis) of bile acids. Bile acids are chemical compounds found in the liver that have several roles ... In many cases, symptoms or signs are present at birth or during the newborn period.

Bile Acid Synthesis Disorders and Zellweger Spectrum Disorders ...

www.retrophin.com/content/patients/bile-acid-synthesis-disorder.php ▼ [翻译此页](#)

Defects in bile acid synthesis are caused by inherited disorders affecting the enzymes ... It is important to diagnose and treat BASD as early as possible, since ... in the well-being of patients with rare liver diseases and has partnered with Emory ... bile acid synthesis that have overlapping signs and symptoms affecting many ...

[PDF] bile acid synthesis and metabolism defects - Childhood Liver Diseases...

<https://childrennetwork.org/download.aspx?f=bile.pdf> ▼

Liver and the defects of cholesterol and bile acids biosynthesis: Rare disorders n

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Bile acids are formed from **cholesterol** and, therefore, **bile acid synthesis** ... In **many** cases, symptoms or signs are present at birth or during the newborn period ... Vitamin E deficiency from undiagnosed **liver disease** may contribute to neurological **disease**. **Defects in bile acid biosynthesis** – **diagnosis** and treatment.

[Mechanisms of Disease: inborn errors of bile acid synthesis](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3888787/)<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3888787/>

24 Jun 2008 ... Inborn errors of **bile acid synthesis** are **rare** genetic **disorders** that can present as ... Failure to **diagnose** any of these conditions can result in **liver** failure or ... Importantly, **many bile acid synthesis defects** are readily treatable and ... the different pathways involved in the modification of **cholesterol** to **bile acids**.

[Disorders of bile acid synthesis. - NCBI](https://www.ncbi.nlm.nih.gov/pubmed/21229319)<https://www.ncbi.nlm.nih.gov/pubmed/21229319>

13 Jan 2011 ... Both types of **disease** can often be treated very effectively with **bile acid** ... and it is therefore important to **diagnose** these **disorders** as early as possible. ... for **many** of these **disorders** is analysis of urinary cholanoids (**bile acids** and ... and the amidation **defects**, **bile acid-CoA: aminoacid N-acyltransferase** ...

[Bile Acid Synthetic Defects and Liver Disease: A Comprehensive ...](https://link.springer.com/article/10.1007/PL00021514)link.springer.com/article/10.1007/PL00021514

Specific **diagnosis** is made by analysis of body fluids (bile, blood, and urine) using fast ... factor in **defects** of side-chain **synthesis**, peroxisomal abiogenesis and S-L-O syndrome. ... with BASD than with the **many** other **liver diseases** in infants where it occurs. ... **Bile acids** enzyme **defects liver disease** giant cells cholestasis.

[Bile Acid Synthesis Disorders - WebMD](#)