Name of Journal: *World Journal of Hepatology*

ESPS Manuscript NO: 30559

Manuscript Type: Case Report

Case of hepatocellular carcinoma in a patient with hereditary tyrosinemia in the post-newborn screening era

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Abstract

Hereditary tyrosinemia type 1 (HT-1) is a metabolic disorder caused by a defect in tyrosine degradation. Without treatment, symptoms of hepatomegaly, renal tubular dysfunction, growth failure, neurologic crises resembling porphyrias, rickets and

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作者：LS King - 2014 - 被引用次数：11 - 相关文章

Synonyms: FAH Deficiency, Hepatorenal Tyrosinemia, Hereditary Tyrosinemia Type I, ... Initial Posting: July 24, 2006; Last Update: July 17, 2014. ... before age ten years, typically from liver failure, neurologic crisis, or hepatocellular carcinoma. .... (2) Elevated tyrosine concentration on newborn screening can be the result of ...

Case Studies - Laboratory Medicine

https://labmed.oxfordjournals.org/content/labmed/37/11/665.full.pdf - 翻译此页

blood sample obtained at 36 hours post-delivery. Family History. A sibling died at age 3 ... often found in cases of hereditary tyrosinemia type I (HTI). Image 1_At .... dence of hepatocellular carcinoma observed in affected patients. In addition, FAA is .... newborn screening program includes evaluation of SA by GC-. MS using ...

Newborn Screening for Hepatorenal Tyrosinemia: Tandem Mass ...

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作者：J Sander - 2006 - 被引用次数：50 - 相关文章

Here we report our experience using this test for newborn screening. .... Because the tyrosine concentration was unusually low for a case of HT, amino acid .... disease and possibly may lead to a lower rate of hepatocellular carcinoma in patients with HT. ... Laberge C. Hereditary tyrosinemia in a French Canadian isolate.

Two Novel FAH Gene Mutations in a Patient with Hereditary ...

www.annclinlabsci.org/content/44/3/317.full - 翻译此页

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Newborn Screening for Tyrosinemia Type I: Further Evidence that ...

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作者：G la Marca - 2011 - 被引用次数：5 - 相关文章

2011年6月22日 - In many countries, newborns are screened for tyrosinemia type I ... The outcome of patients detected in the MS/MS screening is ... rickets, failure to thrive, and hepatocellular carcinoma (Mitchell et al. ... An elevated concentration of tyrosine, however, is not sensitive enough to detect all cases (Wilcken et al.

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blood sample obtained at 36 hours post-delivery. Family History. A sibling died at age 3 ... often found in cases of hereditary tyrosinemia type I (HTI). Image 1_At .... dence of hepatocellular carcinoma observed in affected patients. In addition, FAA is .... newborn screening program includes evaluation of SA by GC-MS using ...

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