

Name of journal: *World Journal of Gastroenterology*

ESPS Manuscript NO: 17316

Manuscript Type: CASE REPORT

Novel variant syndrome associated with congenital hepatic fibrosis

Bayraktar B *et al.* New syndrome with congenital hepatic fibrosis

Yusuf Bayraktar, Ozlem Yonem, Kubilay Varlı, Hande Taylan, Ali Shorbagi, Cenk Sokmensuer

Yusuf Bayraktar, Ali Shorbag, Hacettepe University, Department of Gastroenterology, 312 Ankara, Turkey

Ozlem Yonem, Department of Gastroenterology, Cumhuriyet University, Sivas 346, Turkey

Match Overview

1	Internet 122 words crawled on 17-Apr-2010 www.wjgnet.com	4%
2	CrossCheck 108 words Gabriele I. Kirchner. "COACH syndrome associated with multifocal liver tumors", <i>The American Journal of Gastroenterology</i>	3%
3	Internet 73 words crawled on 25-Jun-2009 pevsnerlab.kennedykrieger.org	2%
4	CrossCheck 50 words Minami, K.. "Septo-optic dysplasia with congenital hepatic fibrosis", <i>Pediatric Neurology</i> , 200308	2%
5	Internet 50 words crawled on 29-Jul-2010 www.ncbi.nlm.nih.gov	2%
6	CrossCheck 26 words "The 21st Conference of the Asian Pacific Association for the Study of the Liver : Oral Presentations 17 February, 2003"	1%
7	Internet 24 words crawled on 16-Dec-2002 link2.springer.de	1%
8	CrossCheck 22 words Daniel Satran. "Cerebello-oculo-renal syndromes including Arima, Senior-Löken and COACH syndromes: More ..."	1%
9	CrossCheck 22 words Bisti, S.. "Degeneration/re-organization coupling in retinitis pigmentosa", <i>Clinical Neurophysiology</i> , 201003	1%

学术搜索

找到约 19,000 条结果 (用时0.11秒)

文章

我的图书馆

时间不限

2015以来

2014以来

2011以来

自定义范围...

按相关性排序

按日期排序

搜索所有网页

中文网页

英语网页

☒ 包括专利☒ 包含引用☐ 创建快讯**Mutations in a novel gene, NPHP3, cause adolescent nephronophthisis, tapeto-retinal degeneration and hepatic fibrosis**

H Olbrich, M Fliegauf, J Hoefele, A Kispert, E Otto... - Nature ..., 2003 - nature.com

... with NPHP and associated Leber congenital amaurosis called Senior-Løken syndrome (SLSN3) also ... the critical regions of NPHP3 (novel centromeric flank D3S1541) and SLSN3 (novel telomeric flank ... Alternative splice variants and transcripts involving exon 3b, 13 and 15 and ...

被引用次数: 260 相关文章 所有 12 个版本 引用 保存

nature.com 中的 [HTML]

Mutations in 3 genes (MKS3, CC2D2A and RPGRIP1L) cause COACH syndrome (Joubert syndrome with congenital hepatic fibrosis)

D Doherty, MA Parisi, LS Finn... - Journal of medical ..., 2010 - jmg.bmj.com

... These criteria were also applied to interpretation of historical reports of COACH syndrome. Mutation identification. The ... Biosystems). Sequence variants were identified using the software package, Variant Reporter (Applied Biosystems). Confirmation ...

被引用次数: 54 相关文章 所有 15 个版本 引用 保存

nih.gov 中的 [HTML]

Mutations in the cystic fibrosis gene in patients with congenital absence of the vas deferens

M Chillón, T Casals, B Mercier, L Bassas... - ... England Journal of ..., 1995 - Mass Medical Soc

... Frequencies of the 5T Allele DNA Variant of Intron 8 of CFTR. ... Cystic fibrosis transmembrane conductance regulator splice variants are not conserved and fail to produce chloride channels. ... A novel mutation in the cystic fibrosis gene in patients with pulmonary disease but normal ...

被引用次数: 791 相关文章 所有 7 个版本 引用 保存

ub.edu 中的 [PDF]

MKS3/TMEM67 mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement

F Brancati, M Iannicelli, L Travaglini... - Human ..., 2009 - Wiley Online Library

... between 1.5 and 2.5 (values >1.0 are considered predictive of a variant being damaging ... C) Conservation across species (shaded in yellow) of residues affected by novel missense variants. ... two distinct bands, corresponding to the wild type fragment and to a novel fragment of ...

被引用次数: 50 相关文章 所有 10 个版本 引用 保存

nih.gov 中的 [HTML]

[PDF] Review Joubert Syndrome and related disorders

F Brancati, B Dallapiccola, EM Valente - Orphanet J Rare Dis, 2010 - biomedcentral.com

... An infantile variant of NPH manifests within the first years of life, with a more rapid ... classification system of JSRD is still evolving due to the discovery of novel genes and ... renal syndromes including Arima Senior-Løken and COACH syndromes: more than just variants of Joubert ...

被引用次数: 152 相关文章 所有 15 个版本 引用 保存 更多

biomedcentral.com 中的 [PDF]



Novel variant syndrome associated with congenital hepatic fibrosis



网页 图片 新闻 视频 购物 更多 ▾ 搜索工具

找到约 281,000 条结果 (用时 0.73 秒)

Google 学术 : Novel variant syndrome associated with congenital hepatic fibrosis

... , tapeto-retinal degeneration and hepatic fibrosis - Olbrich - 被引用次数 : 260

... syndrome (Joubert syndrome with congenital hepatic ... - Doherty - 被引用次数 : 54

... in the cystic fibrosis gene in patients with congenital ... - Chillón - 被引用次数 : 791

Congenital hepatic fibrosis - Wikipedia, the free encyclopedia

https://en.wikipedia.org/wiki/Congenital_hepatic_fibrosis ▾ 翻译此页

Congenital hepatic fibrosis is an inherited fibrocystic liver disease associated with proliferation of interlobular bile ducts within the portal areas and fibrosis that ...

Caroli disease - UpToDate

www.uptodate.com/contents/caroli-disease ▾ 翻译此页

2014年3月26日 - Caroli disease is a congenital disorder characterized by multifocal, segmental dilatation of ... Caroli initially described two variants, wh. ... in which bile duct dilatation is associated with congenital hepatic fibrosis [3]. ... PKHD1, the polycystic kidney and hepatic disease 1 gene, encodes a novel large protein ...

Congenital Hepatic Fibrosis: Background, Pathophysiology ...

emedicine.medscape.com/article/927984-overview ▾ 翻译此页

2014年3月31日 - Congenital hepatic fibrosis (CHF) is an autosomal recessive disease ... It is characterized by hepatic fibrosis, portal hypertension, and renal cystic disease. ... Congenital hepatic fibrosis is associated with an impairment of renal functions, Analysis of missense variants in the PKHD1-gene in patients with ...