

Severe Neonatal Cholestasis as an Early Presentation of ...<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6398189>

Feb 20, 2019 · **McCune-Albright syndrome (MAS)** is a rare genetic disorder characterized by café-au-lait macules, polyostotic fibrous dysplasia and multiple endocrinopathies. Liver involvement, although described, is a rare complication. We review the case of a child with MAS whose initial presentation was characterized by severe neonatal cholestasis.

Author: Nicole Coles, Ian Comeau, Tatiana Mu... Publish Year: 2018

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Severe Neonatal Cholestasis as an Early Presentation of **McCune-Albright Syndrome** - PubMed. **McCune-Albright syndrome (MAS)** is a rare genetic disorder characterized by café-au-lait macules, polyostotic fibrous dysplasia and multiple endocrinopathies. Liver involvement, although described, is a rare complication.

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Sep 04, 2015 · Case presentation. We report a case of **McCune–Albright syndrome** with multi-organ manifestations in the neonatal period. A newborn preterm black girl was referred to our Neonatal Intensive Care Unit at the age of 17 days for suspected extrahepatic cholestasis.

Cited by: 12 Author: Rita Lourenço, Patrícia Dias, Raquel Gou...

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Neonatal McCune-Albright Syndrome: A Unique Syndromic ...

https://asbmr.onlinelibrary.wiley.com/doi/full/10.1002/jbm4.10134

Dec 14, 2018 - Somatic gain-of-function mutations of GNAS cause a spectrum of clinical phenotypes, ranging from McCune-Albright syndrome (MAS) to isolated disease of bone, endocrine glands, and more rarely, other organs. In MAS, a syndrome classically characterized by polyostotic fibrous dysplasia (FD), café-au-lait (CAL) skin spots, and precocious puberty, the heterogeneity of organ involvement ...

Author: Alessandro Corsi, Natasha Cherman, D... Publish Year: 2019

McCune-Albright syndrome: A case report and review of the ...

https://www.researchgate.net/publication/11252691...

We propose that McCune-Albright syndrome be included in the list for differential diagnosis of neonatal cholestasis and chronic cholestasis of infancy, as a rare cause. View Show abstract

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McCune-Albright syndrome

Complex Genetic Disorder

McCune-Albright syndrome is a complex genetic disorder affecting the bone, skin and endocrine systems. It is a mosaic disease arising from somatic activating mutations in GNAS, which encodes the alpha-subunit of the G, Heterotrimeric G protein. These mutations lead to constitutive receptor activation.

Wikipedia

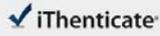
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Name of Journal: *World Journal of Clinical Pediatrics*

Manuscript NO: 61740

Manuscript Type: CASE REPORT

Neonatal cholestasis can be the first symptom of McCune-Albright syndrome: A case report

Satomura Y *et al.* McCune-Albright syndrome with neonatal cholestasis

Yoshinori Satomura, Kazuhiko Bessho, Taichi Kitaoka, Shinji Takeyari, Yasuhisa Ohata, Takuo Kubota, Keiichi Ozono

Abstract

BACKGROUND

McCune-Albright syndrome (MAS) is caused by postzygotic somatic mutations of the *GNAS* gene. It is characterized by the clinical triad of fibrous dysplasia, café-au-lait skin spots, and endocrinological dysfunction. Myriad complications in MAS, including

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Sep 04, 2015 · In the neonatal period the diagnosis of McCune–Albright syndrome depends on having a high index of suspicion and café-au-lait spots may be the clue for the diagnosis. McCune–Albright syndrome is a rare sporadic disease characterized by fibrous bone dysplasia, café-au-lait skin spots