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

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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 ...

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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.

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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, ²Taichi Kitaoka, Shinji Takeyari, Yasuhisa Ohata, Takuo Kubota, Keiichi Ozono

Abstract

BACKGROUND

³McCune-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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