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Manuscript Type: CASE REPORT

Clinical characteristics on manifestation and gene mutation of a transient neonatal cyanosis: A case report

Jing Yuan, Xue-Ping Zhu

Abstract

BACKGROUND

We analyzed the main features of an infant diagnosed with temporary neonatal

国内版 国际版

Clinical characteristics on manifestation and gene mutation of

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Pseudoaldosteronism due to mutation of SCNN1A gene: a ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4429115

Pseudohypoaldosteronism type 1 (PHA1) is a rare inherited disease characterized by resistance to the actions of aldosterone. It was first described in 1958 by Cheek and Perry, and common clinical manifestations include **salt wasting**, **hyperkalaemia**, **metabolic acidosis** and **elevated plasma aldosterone levels** in the neonatal period.

Author: Can Thi Bich Ngoc, Vu Chi Dung, Bui ... **Publish Year:** 2015

Identification of the first large deletion in the CLDN16 ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4487846

Jul 02, 2015 · **Case** presentation. We here **report** the phenotype and molecular analysis of a female Brazilian patient with a novel large homozygous deletion in the CLDN16 **gene**. The proband, born from consanguineous parents, presented the first symptoms at age 20.

Cited by: 2 **Author:** Paulo Marcio Yamaguti, Pollyanna Almei...
Publish Year: 2015

AB130. Pseudoaldosteronism due to mutation of SCNN1A ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4563464

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Author: Ngoc Thi Bich Can, Dung Chi Vu, Tha... **Publish Year:** 2015

Biallelic SCN2A Gene Mutation Causing Early Infantile ...

https://journals.sagepub.com/doi/full/10.1177/1179573519849938

May 15, 2019 · Herein, we **report** the first patient with a biallelic missense **mutation** in the SCN2A **gene** with an autosomal recessive inheritance. **Case Report** The patient is a 30-month-old Saudi girl born full term via elective cesarean section to first cousin parents with an uneventful pregnancy.

Author: Shahad AlSaif, Muhammad Umair, Ma... **Publish Year:** 2019

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Jul 02, 2015 · The **hypothesis** of the influence of modulators and epigenetic factors in the clinical spectrum of FHHNC is reinforced by studies that report unusual clinical findings in FHHNC patients with CLDN16 **mutations** [27–29] and different clinical courses in siblings with the same CLDN16 **mutation** [17, 30]. One case report presented a boy with a **truncating mutation in claudin-16** (p.W237X) with **early-onset renal insufficiency**...

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Iranian neonatal diabetes mellitus due to mutation in PDX1 ...

https://jmedicalcasereports.biomedcentral.com/articles/10.1186/s13256-019-2149-x