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A Novel Mutation Causing Pseudohypoparathyroidism 1A ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3005748

Various **inactivating mutations** in **guanine nucleotide–binding protein**, alpha–stimulating activity polypeptide1 (**GNAS1**) **gene** have been described with poor phenotype correlation. Pseudohypoparathyroidism type 1a (PHP1a) results from an **inactivating mutation** in the **GNAS1 gene**.

Boy With Pseudohypoparathyroidism Type 1a Caused by ...

https://www.researchgate.net/publication/26295497_Boy_With_Pseudohypoparathyroidism...

We report on a 6-month-old boy with **craniosynostosis**, **pseudohypoparathyroidism type 1a** (PHP1A), and a **GNAS gene mutation**. He had synostoses of the coronal, frontal, and sagittal sutures ...

Pseudohypoparathyroidism Type 1a and the GNAS p.R231H ...

https://www.researchgate.net/publication/47556969...

Pseudohypoparathyroidism Ia (**PHP-Ia**), is an inherited disease with **clinical hypoparathyroidism** caused by **parathyroid hormone resistance** (PTH), and shows the phenotype of Albright hereditary ...

Ectopic Calcification as Discernible Manifestation in ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2778176

Pseudohypoparathyroidism type 1a (PHP1a) is a genetic disorder caused by maternally inherited **mutations** of **GNAS gene**, which encodes the α-subunit of the stimulatory G protein Gs [1, 2]. Common feature in PHP1a includes Albright hereditary osteodystrophy (AHO) such as brachydactyly [3] and resistance to multiple **Gs protein-coupled hormones** (e.g., parathyroid hormone (PTH) and ...

Cited by: 6 **Author:** Masanori Adachi, Koji Muroya, Yumi Asa...

Publish Year: 2009

(PDF) Diagnosis and management of ...

https://www.researchgate.net/publication/326074744_Diagnosis_and_management_of...

The two **main subtypes of pseudohypoparathyroidism** (**PHP**), **PHP-Ia and -Ib**, are caused by mutations in **GNAS exons 1–13** and methylation defects in the imprinted GNAS cluster, respectively.

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 51342

Manuscript Type: CASE REPORT

Cutaneous nodules and a novel *GNAS* mutation in a Chinese boy with pseudohypoparathyroidism type Ⅰa: A case report and review of literature

Yun-Ling Li, Ting Han, Fang Hong

Abstract

BACKGROUND

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Cutaneous nodules and a novel GNAS mutation in a Chinese b

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A Novel Mutation Causing Pseudohypoparathyroidism 1A ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3005748

We herein **report a case** of PHP1a presenting with congenital hypothyroidism, PHP, osteoma cutis, obesity and psychomotor retardation. These manifestations of PHP1a resulted from a **novel mutation** in the guanine nucleotide–binding protein, alpha–stimulating activity polypeptide 1 (GNAS1) gene.

Cited by: 5 **Author:** Lubell T, Garzon M, Anyane Yeboa K, Sh...
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Publish Year: 2009

Mutations in the Gsα gene causing hormone resistance

https://www.researchgate.net/publication/6639036_Mutations_in_the_Gsa_gene_causing...

Objective: We report an atypical association of primary **adrenal insufficiency** and pseudohypoparathyroidism (**PHP**) **and a novel GNAS1 gene mutation in a Caucasian female** who initially presented with ...

Diagnosis and management of pseudohypoparathyroidism ...

https://www.nature.com/articles/s41574-018-0042-0

Jun 29, 2018 · Several clinical and genetic features can be suggestive of the **diagnosis**, such as **a mutation** involving exons 1–13 of the **paternal GNAS allele**, radiographic evidence of **a reticular pattern** ...

Cited by: 16 **Author:** Giovanna Mantovani, Murat Bastepe, Davi...

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Cutaneous nodules and a novel GNAS mutation in a Chinese b

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https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3005748

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Novel nonsense GNAS mutation in a 14-month-old boy with ...

https://www.researchgate.net/publication/260148284_Novel_nonsense_GNAS_mutation_in_a...

Novel nonsense **GNAS mutation in** a 14-month-old **boy with** plate-like osteoma cutis and medulloblastoma Article in The Journal of Dermatology 41(4) · ...

Ectopic Calcification as Discernible Manifestation in ...

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