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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...](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 1a (PHP-1a), 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  $\alpha$ -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...](https://www.researchgate.net/publication/326074744_Diagnosis_and_management_of...)

The two main subtypes of pseudohypoparathyroidism (PHP), PHP-1a and -1b, 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 I a: A case report and review of literature**

Li YL *et al.* Novel *GNAS* mutation of PHP I a with subcutaneous nodules

Yun-Ling Li, Ting Han, Fang Hong

**Abstract**

BACKGROUND

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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...

Publish Year: 2009

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Publish Year: 2009

## Mutations in the Gs $\alpha$ gene causing hormone resistance

[https://www.researchgate.net/publication/6639036\\_Mutations\\_in\\_the\\_Gsa\\_gene\\_causing...](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 ...

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

[https://www.researchgate.net/publication/260148284\\_Novel\\_nonsense\\_GNAS\\_mutation\\_in\\_a...](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) · ...

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