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Congenital nephrogenic diabetes insipidus due to the mutation in AVPR2 (c.541C>T)in a neonate:A case report

Congenital nephrogenic diabetes insipidus in a neonate

Fatao Lin, Jing Li, Bangli Xu, Xiuxiu Yang, Fang Wang

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Text-Only Report

Congenital nephrogenic diabetes insipidus due to the mutation in A

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A Novel Mutation in the AVPR2 Gene Causing Congenital ...

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Objective: **Congenital nephrogenic diabetes insipidus** (CNDI) is a rare inherited disorder characterized by a renal insensitivity to arginine vasopressin (AVP). In the majority of the cases, CNDI is caused by **mutations** in the arginine vasopressin receptor 2 (**AVPR2**) gene.Our objective is to **report** a novel **mutation** in the **AVPR2** gene causing CNDI in a 6-year-old boy, presenting with growth failure ...

Cited by: 1 **Author:** Aslı Çelebi Tayfur, Tuğçe Karaduman, Me...
Publish Year: 2018

Novel AQP2 Mutation Causing Congenital Nephrogenic ...

https://pubmed.ncbi.nlm.nih.gov/23950570

Congenital nephrogenic diabetes insipidus (NDI) is a rare inherited disorder, mostly caused by **AVPR2 mutations**. Less than 10% of cases are **due** to **mutations** in the aquaporin-2 (AQP2) gene. Diagnosis and management of this condition remain challenging especially during infancy. Here, we **report** two unr ...

Cited by: 4 **Author:** Rottanat Rugpolmuang, Asma Deeb, You...
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A Novel Mutation in the AVPR2 Gene Causing Congenital ...

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About 90 percent of all cases of hereditary **nephrogenic diabetes insipidus** result from **mutations** in the **AVPR2** gene. To date, more than 250 **mutations** have been identified comprising missense, nonsense, small insertions and deletions, large deletions and complex rearrangements **in AVPR2** gene.

Cited by: 1 **Author:** Aslı Çelebi Tayfur, Tuğçe Karaduman, Me...
Publish Year: 2018

Severe congenital nephrogenic diabetes insipidus in a ...

https://onlinelibrary.wiley.com/doi/10.1002/mgg3.568

1 INTRODUCTION. **Congenital nephrogenic diabetes insipidus** (NDI) is a rare disease that is characterized by the excretion of abnormally large volumes of urine, **due** to the inability of the kidneys to concentrate urine in response to arginine vasopressin (AVP) (Sasaki, 2004; Wesche, Deen, & Knoers, 2012).Patients with **congenital** NDI typically present in infancy with severe polyuria, polydipsia ...

Cited by: 1 **Author:** Ramón Peces, Rocío Mena, Carlos Pece...
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A Novel Mutation in the AVPR2 Gene Causing Congenital ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6280332

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Novel AQP2 Mutation Causing Congenital Nephrogenic ...

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