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Gitelman syndrome caused by a rare homozygous mutation in the SLC12A3 gene: a new case report and review of literature

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Abstract

BACKGROUND

Gitelman syndrome (GS) is an unusual, autosomal recessive salt-losing tubulopathy characterized by hypokalemic metabolic alkalosis, hypomagnesemia and hypocalciuria. It is caused by mutations in the Solute Carrier Family 12 Member 3 (SLC12A3) gene resulting in improper function on the thiazide-sensitive NaCl co-transporter. To date, many kinds of mutations in the SLC12A3 gene have been discovered that trigger different

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GS is **caused** by an **inactivating mutation in the SLC12A3 gene**, which is located on the long arm of chromosome 16 (16q13) and encodes a thiazide-sensitive sodium chloride cotransporter (NCCT). **Graves' disease** (GD) is a common **cause** of **hyperthyroidism**. In our department, we diagnosed a patient with GD and GS.

Cited by: 2 **Author:** Haiyang Zhou, Xinhuan Liang, Yingfen Qi...
Publish Year: 2018

Novel heterozygous missense mutation of SLC12A3 gene in ...

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Jun 26, 2019 · Sequencing showed a **novel heterozygous missense mutation** (a G to A transition at nucleotide 2582) in exon 22 of the **SLC12A3 gene**, which resulted in a substitution of histidine for arginine at position 816 of the **LRP1B protein and caused** the **occurrence of disease**.

Author: Cheng-Lin Wang **Publish Year:** 2019

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https://link.springer.com › article › 10.1186 › s12902-018-0298-3

Nov 08, 2018 · Gitelman syndrome (GS) is an inherited **autosomal recessive renal tubular disorder** characterized by low levels of **potassium and magnesium** in **the blood**, decreased **excretion of calcium** in **the urine**, and **elevated blood pH**.

Cited by: 2 **Author:** Haiyang Zhou, Xinhuan Liang, Yingfen Qi...
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Nov 08, 2018 · **GS** is caused by an inactivating mutation in the SLC12A3 gene, which is located on the long arm of chromosome 16 (16q13) and encodes a **thiazide-sensitive sodium chloride cotransporter** (NCCT). **Graves' disease** (**GD**) is a common **cause** of **hyperthyroidism**. In our department, we diagnosed a patient with GD and GS.

Cited by: 3 **Author:** Haiyang Zhou, Xinhuan Liang, Yingfen Qi...
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Gitelman syndrome is a **rare salt-losing renal tubular disorder** associated with **mutation** of **SLC12A3 gene**, which encodes the Na-Cl co-transporter (NCCT). **Gitelman syndrome** is characterized by **hypokalemia**, **metabolic alkalosis**, hypomagnesemia, hypocalciuria, and **renin-angiotensin-aldosterone system** (RAAS) activation.

Gitelman syndrome | Genetic and Rare Diseases Information ...

https://rarediseases.info.nih.gov/diseases/8547/gitelman-syndrome

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A case of **hypokalemia and proteinuria** with a new mutation in the SLC12A3 Gene This case is the first to report a homozygous mutation in the 841th nucleotide of exon 6 on the SLC12A3 gene (p.Trp281Arg), which may cause Gitelman syndrome.

Author: Qin Chen, Yaqin Wu, Jingya Zhao, Ying Jia, Wei Wang

Cited by: 2

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Feedback

New SLC12A3 disease causative mutation of Gitelman's syndrome
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5099602

Nov 06, 2016 · Gitelman's syndrome (GS) is an autosomal **recessive salt-wasting disease prevalently caused** by mutations occurring in the SLC12A3gene coding for the **thiazide-sensitive NaCl cotrasporter** (NCC)[1]. This **disorder** is a tubulopathy characterized by **hypokalemic metabolic alkalosis**, hypocalciuria and hypomagnesemia.

Gitelman syndrome

Gitelman syndrome is an autosomal recessive kidney tubule disorder characterized by low blood levels of potassium and magnesium, decreased excretion of calcium in the urine, and elevated blood pH. The disorder is caused by genetic mutations resulting in improper function of the thiazide-sensitive sodium-chloride symporter located in the distal convoluted tubule of the kidney. The distal convoluted tubule of the kidney serves a minimal role in salt absorption and a greater role in managing the excretion of electrolytes like magnesium and calcium to produce more concentrated urine.

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