找到约 4,510 条结果（用时 0.56 秒）

## Google 学术：A novel heterozygous missense mutation of SLC12A3 gene in Gitelman syndrome: A case report

... cotransporter gene in patients with Gitelman syndrome ... - Lemmink - 被引用次数：169

Gitelman's syndrome: a pathophysiological and clinical ... - Nakhoul - 被引用次数：54

... in SLC12A3 associated with Gitelman syndrome and ... - Godefroid - 被引用次数：19

## A novel compound heterozygous variant of the SLC12A3 gene ... - NCBI

https://www.ncbi.nlm.nih.gov/pubmed/29378538 - 翻译此页

作者：Y Chen - 2018 - 相关文章

2018年1月29日 - A novel compound heterozygous variant of the SLC12A3 gene in Gitelman syndrome pedigree. ... BACKGROUND: Gitelman syndrome (GS) is an autosomal recessive disorder caused by genic mutations of SLC12A3 (Solute carrier ... In this study, we report a case of a GS pedigree, including analysis of ...

## A novel SLC12A3 homozygous c2039delG mutation in Gitelman ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6296056/ - 翻译此页

作者：W Yang - 2018

2018年12月17日 - Keywords: Gitelman syndrome, SLC12A3 gene mutation, ... Here, we report a case of a new mutation of SLC12A3 in an Asian pedigree. .... A heterozygous mutation in the same site was observed in the ... To date, more than 400 mutations of SLC12A3 have been identified, including missense/ nonsense ...

## A novel SLC12A3 gene homozygous mutation of Gitelman syndrome ...

https://www.researchgate.net/.../280967709_A_novel_SLC12A3_gene_hom... - 翻译此页

2018年9月2日 - In this study, we reported a case of GS pedigree and... ... gene mutations have been discovered including missense, shear, ... A novel compound heterozygous variant of the SLC12A3 gene in Gitelman syndrome pedigree.

## A case report of Gitelman syndrome resulting from two novel mutations ...

https://www.researchgate.net/.../307615438_A_case_report_of_Gitelman_syndrome_res...

2018年8月1日 - A case report of Gitelman syndrome resulting from two novel mutations in ... Genetic testing was performed and two novel heterozygous ..... gene. mutation. SLC12A3. gene. encodes. the. thiazide-sensitive ..... Novel nonsense mutation in the SLC12A3 gene in a Spanish case of Gitelman syndrome. Article.

**Name of Journal:** *World Journal of Clinical Cases*

**Manuscript NO:** 45831

**Manuscript Type:** CASE REPORT

# Novel heterozygous missense mutation of SLC12A3 gene in Gitelman syndrome: A case report

Cheng-Lin Wang

## Abstract

### *BACKGROUND*

To screen for possible pathogenic loci in a patient with Gitelman syndrome (GS) by high-throughput exome sequencing and to explore the relationship

国内版 国际版

Novel heterozygous missense mutation of SLC12A3 gene in Gitelman syndrome: A

Sign in

All Images Videos 翻译成中文 关闭取词 Make Bing your se

90,600 Results Any time

## Novel mutation in the SLC12A3 gene in a Sri Lankan family ...

https://bmcnephrol.biomedcentral.com/articles/10.1186/s12882-017...

Apr 26, 2017 · **Gitelman syndrome** (GS) is a rare autosomal recessively inherited salt-wasting tubulopathy associated with **mutations** in the **SLC12A3 gene**, which encodes for NaCl cotransporter (NCC) in the kidney. In this **report**, we describe two siblings from a Sri Lankan non-consanguineous family presenting with hypokalaemia associated with renal potassium wasting, hypomagnesemia, ...

**Cited by:** 2 **Author:** Chandrika Jayakanthi Subasinghe, Nirmal...
**Publish Year:** 2017

## [PDF] Novel mutation in the SLC12A3 gene in a Sri Lankan ...

https://link.springer.com/content/pdf/10.1186/s12882-017-0563-0.pdf

CASE REPORT Open Access **Novel mutation** in the **SLC12A3 gene** in a Sri Lankan family with **Gitelman syndrome &** coexistent diabetes: **a case report** Chandrika Jayakanthi Subasinghe1, Nirmala Dushyanthi Sirisena2, Chula Herath3, Knut Erik Berge4, Trond Paul Leren4, Uditha Bulugahapitiya1 and Vajira Harshadeva Weerabaddana Dissanayake2* Abstract

**Cited by:** 2 **Author:** Chandrika Jayakanthi Subasinghe, Nirmal...
**Publish Year:** 2017

## [PDF] A novel compound heterozygous variant of the SLC12A3 ...

https://bmcmedgenet.biomedcentral.com/track/pdf/10.1186/s12881-018...

A **novel** compound **heterozygous** variant of the **SLC12A3 gene in Gitelman syndrome** pedigree Yixin Chen, Ziyi Zhang, Xihua Lin, Qianqian Pan, Fenping Zheng and Hong Li* Abstract Background: **Gitelman syndrome** (GS) is an autosomal recessive disorder caused by genic **mutations of SLC12A3**

**Author:** Yixin Chen, Ziyi Zhang, Xihua Lin, Qia... **Publish Year:** 2018

## Novel mutation in the SLC12A3 gene in a Sri Lankan family ...

## Gitelman syndrome

Autosomal Recessive Kidney Disorder

Gitelman syndrome is an autosomal recessive kidney 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. This symporter is a channel responsible for the transport of multiple electrolytes such as sodium, chloride, calcium, magnesium, and potassium.

Wikipedia

### People also search for

Bartter syndrome

Liddle's syndrome

Renal tubular acidosis

Fanconi syndrome

Pseudohypoaldosteronism

See more

Data from: Wikipedia

Suggest an edit

国内版 国际版

Novel heterozygous missense mutation of SLC12A3 gene in Gitelman syndrome: A

All Images Videos 翻译成中文 开启取词

85,100 Results Any time

## Novel mutation in the SLC12A3 gene in a Sri Lankan family ...

https://bmcnephrol.biomedcentral.com/articles/10.1186/s12882-017...

Apr 26, 2017 · **Case** presentation. **Genetic testing** showed that both were **homozygotes** for a **novel missense mutation** in **exon** 10 of the **SLC12A3 gene** [NM_000339.2, c.1276A > T; p.N426Y], which has not previously been **reported** in the literature in association with GS. Their **mother** was a **heterozygous** carrier for the same **mutation**.

**Cited by:** 2 **Author:** Chandrika Jayakanthi Subasinghe, Nirma...
**Publish Year:** 2017

## [PDF] A novel compound heterozygous variant of the SLC12A3 ...

https://bmcmedgenet.biomedcentral.com/track/pdf/10.1186/s12881-018...

RESEARCH ARTICLE Open Access. A **novel** compound **heterozygous** variant of. the **SLC12A3 gene** in **Gitelman syndrome**. pedigree. Yixin Chen, Ziyi Zhang, Xihua Lin, Qianqian Pan, Fenping Zheng and Hong Li*. Abstract. Background: **Gitelman syndrome** (GS) is an autosomal recessive **disorder** caused by genic **mutations of SLC12A3**.

**Author:** Yixin Chen, Ziyi Zhang, Xihua Lin, Qia... **Publish Year:** 2018

## Spectrum of Mutations in Gitelman Syndrome

**www.ncbi.nlm.nih.gov** › ... › J Am Soc Nephrol › v.22(4); 2011 Apr

**Gitelman's syndrome** (GS, MIM 263800) is a rare salt-losing tubulopathy characterized by hypokalemic metabolic **alkalosis**, hypomagnesaemia, and hypocalciuria. Loss of function **mutations** in the **SLC12A3 gene** encoding for the thiazide-sensitive NaCl cotransporter (NCC) are responsible for most of the **cases**.

**Cited by:** 128 **Author:** Rosa Vargas-Poussou, Karin Dahan, Dia...
**Publish Year:** 2011

## Novel mutation in the SLC12A3 gene in a Sri Lankan family ...

https://link.springer.com/article/10.1186/s12882-017-0563-0

Apr 26, 2017 · Genetic testing showed that both were homozygotes for a **novel missense mutation** in exon 10 of the **SLC12A3 gene** [NM_000339.2, c.1276A > T; p.N426Y], which has not previously been reported in the literature in association with GS. Their mother was a **heterozygous** carrier for the same **mutation**.

**Cited by:** 2 **Author:** Chandrika Jayakanthi Subasinghe, Nirma...
**Publish Year:** 2017

## A Pedigree with c.179 Cytosine to Threonine Missense ...

**www.ncbi.nlm.nih.gov** › ... › v.14(1); 2016 Jun

**Case Report**. Variations in the sequences between GS and matched normal sample were performed using the ABI 3730 DNA sequencer (Bionics Inc, Korea). The result revealed that the patient had **homozygous missense mutation** of nucleotide 179, cytosine to threonine (ACG to ATG) on **exon** 1 of the **SLC12A3 gene**.

**Author:** Yaerim Kim, Seong Sik Kang, Woo Ye... **Publish Year:** 2016