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Frequency of CNKSR2 mutation in the X - linked epilepsy - aphasia ...

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作者: JA Damiano - 2017 - 被引用次数: 1 - 相关文章

2017年1月18日 - Frequency of **CNKSR2** mutation in the **X-linked epilepsy - aphasia** spectrum **syndrome** (LKS) to the mild condition of childhood **epilepsy** with centrottemporal spikes. ... **CNKSR2** (connector enhancer of KSR2),6-10 was shown to **cause** **cases**.6-10 This is the first **report** of BECTS and febrile **seizures** in ...

CNKSR2 deletions: A novel cause of X-linked intellectual disability ...

onlinelibrary.wiley.com/doi/10.1002/ajmg.a.36902/abstract - 翻译此页

2015年3月5日 - Previous article in issue: A germline MTOR **mutation** in Aboriginal ... sign of 17q21.31 microdeletion **syndrome**: Preaxial polydactyly of hands with broad thumbs ... **CNKSR2** deletions: A novel **cause** of **X-linked** intellectual disability and **seizures** ... Disclosure: Our institution does not consider a **case report** as ...

Frequency of CNKSR2 mutation in the X-linked epilepsy-aphasia ...

<https://www.ncbi.nlm.nih.gov/pubmed/28098945> - 翻译此页

作者: JA Damiano - 2017 - 被引用次数: 1 - 相关文章

2017年1月18日 - Frequency of **CNKSR2** mutation in the **X-linked epilepsy-aphasia** ... In this **study**, we sequenced 112 patients with phenotypes within the ...

缺少字词: **causes syndrome ease**

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Frequency of **CNKSR2** mutation in the **X-linked epilepsy-aphasia** spectrum. ... Thauvin-Robinet C. The DYRK1A gene is a **cause** of syndromic intellectual ... Chawala S, Trump D. **Report** of two brothers with

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 40627

Manuscript Type: CASE REPORT

CNKS2 mutation causes the X-linked epilepsy-aphasia syndrome: A case report and review of literature

Ying Sun, Yi-Dan Liu, Zhi-Feng Xu, Qing-Xia Kong, Yan-Ling Wang

Abstract

The mutation in CNKS2 leads to a broad spectrum of phenotypic variability and manifests as an X-linked intellectual disability. However, we reported that the male patient in this study not only had intellectual disability but also

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Frequency of CNKSR2 mutation in the X-linked epilepsy-aphasia ...

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作者: JA Damiano - 2017 - 被引用次数: 3 - 相关文章

2017年1月18日 - Frequency of **CNKSR2 mutation** in the **X-linked epilepsy-aphasia** spectrum. ... (1)
Department of Medicine, Epilepsy Research Centre, Austin Health, University of ... In this **study**, we sequenced 112 patients with phenotypes within the ... Aphasia/physiopathology; Cohort Studies; DNA Mutational **Analysis** ...

缺少字词: **causes syndrome case literature**

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2017年1月18日 - Frequency of **CNKSR2 mutation** in the **X-linked epilepsy-aphasia** spectrum
syndrome (LKS) to the mild condition of childhood **epilepsy** with ... by **CNKSR2** (connector enhancer of KSR2),6-10 was shown to **cause** ... Medical Centre Institutional **Review** Board approved this **study**.
Citing **Literature** ...

Electrical status epilepticus in sleep, a constitutive feature of ...

<https://www.sciencedirect.com/science/article/pii/S1090379817319803> - 翻译此页

2018年7月21日 - Christianson **syndrome** (CS) is a **X-linked** neurodevelopmental **disorder**, ... The two published **case reports** and our observation suggests that ESES could ... as GRIN2A and **CNKSR2**-related developmental epileptic encephalopathies. 'GRIN2A **Mutations Cause Epilepsy-Aphasia Spectrum Disorders**'.

[PDF] Genetic etiologies of the electrical status epilepticus ... - ResearchGate

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Frequency of CNKSR2 mutation in the X-linked epilepsy-aphasia ...

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缺少字词: causes syndrome case literature

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 related developmental epileptic encephalopathies. 'GRIN2A **Mutations Cause Epilepsy-Aphasia**
 Spectrum Disorders'.

Genetic etiologies of the electrical status epilepticus during slow wave ...

<https://bmcbgenet.biomedcentral.com/articles/10.1186/s12863-018-0628-5> ▾ 翻译此页

作者: M Kessi - 2018

2018年7月6日 - We aimed to perform a systematic **review** of all genetic etiologies which ... Furthermore,
 few genetic **causes** have been reported including monogenic **mutations** and ... We included **case**
reports, **case** series, and cohort studies. Frequency of **CNKSR2 mutation** in the **X-linked epilepsy-**
aphasia spectrum.

Sara Kivity's research works | Schneider Children's Medical Center of ...

https://www.researchgate.net/scientific-contributions/38821608_Sara_Kivity

Genetic **analysis** of known GEFS+ genes was carried out where possible. Frequency of **CNKSR2**
mutation in the **X-linked epilepsy-aphasia** spectrum **RARS2 mutations cause** early onset epileptic
 encephalopathy without ponto-cerebellar This is the first **report** of familial Ohtahara **syndrome** due to