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作者：X Li - 2018

A case of a novel CACNA1G mutation from a Chinese family with SCA42: A case report and literature review. Li X(1), Zhou C(1), Cui L(1), Zhu L(2), Du H(1), Liu ...

缺少字词： ~~ATL1~~ ~~HSP~~

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Auton Neurosci. 2014 Oct;185:141-3. doi: 10.1016/j.autneu.2014.06.001. Epub 2014 Jun 9. Case Reports; Research Support, Non-U.S. Gov't.

(PDF) Hereditary spastic paraplegia due to a novel mutation of the ...

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Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 46157

Manuscript Type: CASE REPORT

Novel *ATL1* mutation in a Chinese family with hereditary spastic paraplegia: A case report and review of literature

Xue-Wen Xiao, Juan Du, Bin Jiao, Xin-Xin Liao, Lu Zhou, Xi-Xi Liu, Zhen-Hua Yuan, Li-Na Guo, Xin Wang, Lu Shen, Zhang-Yuan Lin

Abstract

BACKGROUND

Hereditary spastic paraplegias (HSPs) refer to a group of heterogeneous

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PDF | RATIONALE: **Hereditary spastic paraplegia** (**HSP**) is a heterogeneous group of **diseases little** known in clinical practice due to its low prevalence, slow progression, and difficult diagnosis.

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Rationale: **Hereditary spastic paraplegia** (HSP) is a heterogeneous group of diseases little known in clinical practice due to its low prevalence, slow progression, and difficult diagnosis. This results in an underestimation of HSP leading to belated diagnosis and management. In depth diagnosis is ...

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Exon 8–17 deletions of SPAST in a Chinese family with ...

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We presented a **Chinese HSP family** with **exon** 8–17 deletions of the **SPAST** gene. The patients with deletions of exons in the **SPAST** gene showed pure HSP. Age at onset of the patients with **exon** deletion of the **SPAST** gene showed variations. **Exon** deletion should be examined routinely in **HSP** patients.