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Frontiers | Next-Generation Sequencing Analysis Reveals ...

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Next-Generation Sequencing Analysis Reveals Novel **Pathogenic** Variants in Four Chinese Siblings With Late-Infantile Neuronal Ceroid Lipofuscinosis Xiao-Tun Ren 1 , Xiao-Hui Wang 1 , Chang-Hong Ding 1 , Xiang Shen 2 , Hao Zhang 2 , Wei-Hua Zhang 1 , Jiu-Wei Li 1 , Chang-Hong Ren 1 and Fang Fang 1 *

Cited by: 1 **Author:** Xiao-Tun Ren, Xiao-Hui Wang, Chang-Hong ...

Publish Year: 2019

The impact of next-generation sequencing on the diagnosis ...

<https://www.ncbi.nlm.nih.gov/pubmed/29691679>

1. Neurogenetics. 2018 May;19(2):111-121. doi: 10.1007/s10048-018-0545-9. Epub 2018 Apr 24. The impact of **next-generation sequencing** on the diagnosis of pediatric-onset hereditary spastic paraplegias: new genotype-phenotype correlations for rare HSP-related **genes**.

Cited by: 15 **Author:** Lorena Travaglini, Chiara Aiello, Fabrizia Str...

Publish Year: 2018

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 60876

Manuscript Type: ORIGINAL ARTICLE

Retrospective Study

Study on pathogenic genes of dwarfism disease by next-generation sequencing

Pathogenic Genes of Dwarfism

Abstract

BACKGROUND

There are many factors that lead to dwarfism, and the mechanism has not yet been elucidated. Next-generation sequencing may identify candidate-related gene mutations, which may clarify the molecular cause.

AIM

Candidate-related gene mutations were screened to analyze the genetic variation by

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Dec 12, 2018 · Here, we have constructed a targeted **next-generation sequencing** (NGS) panel of selected **genes** that are suspected to be associated with **dwarfism** for **genetic screening**. **Genetic screening** of 91 children with short stature of unknown etiology was performed with the help of ...

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Diagnostic utility of a targeted next-generation ...

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More than half of patients do not carry any **pathogenic** variant in formerly associated **disease genes**. Here, we report a cross-sectional **study** on targeted Next-Generation **Sequencing** (NGS) screening in patients with suspected sAIDs to determine the diagnostic utility of **genetic screening**.

Cited by: 8 **Author:** İlker Karacan, İlker Karacan, Ayşe Balam...

Publish Year: 2019

Frontiers | Next-Generation Sequencing Analysis Reveals ...

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