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Study on pathogenic genes of dwarfism disease by next-generation

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Pathogenic gene screening in 91 Chinese patients with ...

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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**. Methods: **Genetic screening** of 91 children with short stature of unknown etiology was performed with the help of the **NGS** panel. All the coding regions and exon-intron boundaries of 166 **genes** were included in the panel.

Cited by: 2Author: Lulu Yang, Chenhui Zhang, Wei Wang, Junq...

Publish Year: 2018

Molecular analysis of inherited cardiomyopathy using next ...

<https://pubmed.ncbi.nlm.nih.gov/30165862>

A custom designed panel, including 64 **genes**, was screened using **next generation sequencing** on the Ion Torrent PGM platform. The best candidate **disease-causing** variants were verified by **Sanger sequencing**. Results: A total of 78 variants in 73 patients were identified. After excluding the variants predicted to be benign and VUS, 26 **pathogenic** or ...

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## Frontiers | Next-Generation Sequencing Analysis Reveals ...

<https://www.frontiersin.org/articles/10.3389/fgene.2019.00370> ▾

**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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**Cited by:** 2

**Author:** Lulu Yang, Chenhui Zhang, Wei Wang, J...

**Publish Year:** 2018

### Pathogenic gene screening in 91 Chinese patients with ...

<https://bmcmmedgenet.biomedcentral.com/articles/10.1186/s12881-018-0730-6> ▼

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 ...

**Cited by:** 2

**Author:** Lulu Yang, Chenhui Zhang, Wei Wang, J...

**Publish Year:** 2018

### Diagnostic utility of a targeted next-generation ...

<https://www.ncbi.nlm.nih.gov/pubmed/30783801>

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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