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Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 45079

Manuscript Type: CASE REPORT

Compound heterozygous mutation of *MUSK* in a fetus causing fetal akinesia deformation sequence syndrome: Case report and review of literature

Abstract

BACKGROUND

Fetal akinesia deformation sequence (FADS) is a broad spectrum disorder with absent fetal movements as the unifying feature. The etiology of FADS is heterogeneous, and mostly still unknown. A prenatal diagnosis of FADS has relies on clinical features obtained by ultrasound and fetal muscle pathology. However, the recent advance of next-generation sequencing (NGS) can effectively provide a definitive molecular diagnosis.

CASE SUMMARY

A fetus presented after 24 wk and 6 d of gestation with absent fetal movements and multiple abnormal ultrasonographic signs. The mother had had a previous abortion due to a similarly affected fetus a year before. A clinical diagnosis of FADS was made. The

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Fetal akinesia deformation sequence syndrome (FADS, OMIM 208150) is characterised by **decreased fetal movement** (**fetal akinesia**) as well as **intrauterine growth restriction**, arthrogryposis, and ...

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https://www.sciencedirect.com/science/article/pii/S1028455912000058

Fetal akinesia deformation sequence may phenotypically overlap with the lethal type of multiple pterygium **syndrome**. This article provides a comprehensive **review** of prenatal diagnosis and genetic analysis of **fetal akinesia deformation sequence** and multiple pterygium **syndrome** associated with neuromuscular junction disorders.

Cited by: 21 **Author:** Chih-Ping Chen
Publish Year: 2012

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europepmc.org/articles/PMC4538208

Fetal akinesia deformation sequence (FADS; **MIM 208150**) represents a group of disorders characterized by decreased or **absent fetal movements**. **Affected infants** may die in utero or shortly after **birth due to severe pulmonary hypoplasia**.

Cited by: 18 **Author:** M Brigita Tan-Sindhunata, Inge B Mathijs...
Publish Year: 2015

Massive parallel sequencing identifies RAPSN and PDHA1 ...

https://www.sciencedirect.com/science/article/pii/S1090379817300089

The second patient represents the first case of **AMC due** to a PDHA1 **mutation**, advocating that **pyruvate dehydrogenase deficiency** should be considered in the **differential diagnosis of fetal akinesia**. This study illustrates the relevance of a **disease-associated-gene** panel as a diagnostic tool in pregnancies complicated by this genetically heterogeneous condition.

Cited by: 6 **Author:** Lore Winters, Evelien Van Hoof, Luc De ...
Publish Year: 2017

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