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[PDF] Fetal akinesia deformation sequence: a case report and review of ...

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作者: R Abraham - 2014 - 相关文章

Case Report. Fetal ... a **case report** and **review** of **literature** ... 12,000 reflecting causal heterogeneity.² Various labels ... ultrasound scan.⁴ The **syndrome** is rare, about 100 **cases** ... **Fetal Akinesia Deformation Sequence** (FADS) is a condition characterised by ... studies identifying novel disease genes and (or) **mutations**.

Fetal akinesia deformation sequence | Genetic and Rare Diseases ...

<https://rarediseases.info.nih.gov/diseases/.../fetal-akinesia-deformation-sequ...> ▼ 翻译此页

Fetal akinesia deformation sequence (FADS) is a condition characterized by ... manner in some **cases** and may sometimes be **caused** by **mutations** in the RAPSN or DOK7 genes ... This table lists **symptoms** that people with this disease may have. of prenatal diagnosis--**report** of 21 **cases**, antenatal findings and **review**.

(PDF) Pena-Shokeir syndrome: current management strategies and ...

https://www.researchgate.net/.../328511635_Pena-Shokeir_syndrome_curren... - 翻译此页

2018年12月7日 - Pena-Shokeir **syndrome** (PSS) type 1, also known as **fetal akinesia deformation sequence**, is a rare genetic Keywords: **fetal akinesia deformation sequence**, ultrasound, comfort care et al **reported** the **case** of a **brother** and a **sister** in a non- ... They **both** had **compound heterozygous mutations** of.

(PDF) Identification of a Dutch founder mutation in MUSK causing fetal ...

https://www.researchgate.net/.../270004639_Identification_of_a_Dutch_fou... - 翻译此页

2018年7月31日 - PDF | **Fetal akinesia deformation sequence** (FADS) refers to a ... Homozygosity mapping in **two fetuses** revealed **MUSK** as a ... Other **mutations** previously **reported** to **cause** CMS are also indicated. **Mutations** found in **compound heterozygotes** are color-matched^{23, 24, 25,} multiple pterygium **syndromes**.

4 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

Li N *et al.* A mutation of *MUSK* causing FADS

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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MuSK: A new target for lethal fetal akinesia deformation ...

https://www.researchgate.net/publication/271537496_MuSK_A_new_target_for_lethal_fetal...

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

Prenatal diagnosis and genetic analysis of fetal akinesia ...

<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

Identification of a Dutch founder mutation in MUSK causing ...

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

Fetal akinesia: review of the genetics of the ...