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One case of apert syndrome diagnosed by prenatal ultrasound combined with magnetic resonance image and whole exome sequencing: A case report

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Apert syndrome : A case report - Perinatal Journal / ... 翻译此页

Objective To present a **case of Apert syndrome diagnosed by prenatal** ultrasonography. **Case(s)** In **prenatal ultrasound** examination, a 29-year-old G2P1 patient has been found to have a fetus with craniosynostosis, hypertelorism, frontal bossing, nasal bridge depression, syndactyly in the hands and feet, moderate ventriculomegaly.

www.perinataljournal.com/Archive/Article/20120201003 ▼

Comparison of ultrasound and magnetic resonance ... 翻译此页

Comparison of **ultrasound** and **magnetic resonance** imaging in the **prenatal** diagnosis of **Apert syndrome**: Report of a **case** Article in Child s Nervous System 30(8) · February 2014 with 50 Reads

https://www.researchgate.net/publication/262046636_Comparison_of_ultrasound_and...

Apert Syndrome - NORD (National Organization for ... 翻译此页

In some instances, features of **Apert syndrome** may be detected before birth. This would be done through **prenatal** 2D or 3D **ultrasound** or **magnetic resonance** imaging (MRI). An **ultrasound** is a noninvasive procedure that can see an **image** of the fetus. This can detect differences in ...

<https://rarediseases.org/rare-diseases/apert-syndrome> ▼

Diagnosis of Apert syndrome in the second-trimester ... 翻译此页

Apert syndrome can be **diagnosed** in ... of the corpus callosum were **diagnosed by prenatal ultrasound** and **magnetic resonance** imaging in two of 30 patients with **Apert syndrome**. ... a **case of Apert's** ...

https://www.researchgate.net/publication/6333829_Diagnosis_of_Apert_syndrome_in_the...

(PDF) Apert syndrome with FGFR2 758 C > G ... 翻译此页

Background: **Apert syndrome** is considered as one of the most common craniosynostosis syndromes with

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Apert syndrome. Ultrasound images demonstrating frontal bossing and midface hypoplasia on two-dimensional (2D) (A) and 3D (B) images, as well as by **magnetic resonance** imaging (C).

Ultrasound image also demonstrated syndactyly of the foot (D), which is characteristic of **Apert syndrome**.

[\(PDF\) Apert syndrome with FGFR2 758 C > G mutation: A ...](https://www.researchgate.net/publication/325196179)

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imaging in the **prenatal** diagnosis of **Apert syndrome**: report of a case. ... for **Apert syndrome**.

Magnetic resonance excluded agenesis of corpus callosum and confirmed bilateral mild ventriculomegaly ...

[Apert Syndrome: a Case Report | Request PDF](https://www.researchgate.net/publication/26588261)

[https://www.researchgate.net/publication/26588261...](https://www.researchgate.net/publication/26588261)

Apert syndrome is a rare autosomal dominant genetic disorder characterized by irregular craniosynostosis, symmetric syndactylia of hands and feet, and mid-line hypoplasia.

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Our **case** underlines the complementary role of **ultrasound** and **magnetic resonance** imaging in the early **prenatal diagnosis** of **Apert syndrome**. ... and skin fragility. Using **whole exome sequencing** ...

[\(PDF\) Whole exome sequencing of fetal structural anomalies ...](#)

<https://www.researchgate.net/publication/345352624...>

Nov 06, 2020 · The objective of this study was to evaluate the efficacy of **whole exome sequencing** (WES) for the genetic **diagnosis** of cases presenting with **fetal** ...

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Apert syndrome is a rare autosomal dominant genetic disorder characterized by irregular craniosynostosis, symmetric syndactylia of hands and feet, and mid-line hypoplasia.

[Prenatal exome sequencing in fetuses with congenital heart ...](#)

<https://onlinelibrary.wiley.com/doi/full/10.1111/cge.13774>

May 14, 2020 · The aim of our study was to evaluate the clinical value of **whole exome sequencing** (WES) in the **prenatal diagnosis** of CHDs in a large cohort. Trio-based WES was performed in 260 fetuses with CHDs negative for karyotype and chromosome microarray analysis results. ... **Apert syndrome** (101200), Crouzon **syndrome** (123500), Saethre-Chotzen **syndrome** ...

Author: Ru Li, Fang Fu, Qiuxia Yu, Dan Wang, Xia... **Publish Year:** 2020

[Three-dimensional helical computed tomography in prenatal ...](#)

<https://obgyn.onlinelibrary.wiley.com/doi/full/10.1002/uog.12298>

Ultrasound examination alone is generally insufficient to make an etiological **diagnosis** in the **case** of abnormalities such as bowed long bones, narrow thorax or complex abnormalities. In such cases, it is often necessary to examine the **fetal** skeleton by helical computed tomography (CT) or three-dimensional