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A rare case report of Turner Syndrome with SRY gene and Non-classical Congenital Adrenal Hyperplasia and literature review

Turner Syndrome with SRY gene and NCAH

Meinan He, Shanchao Zhao, Jimin Li, Lulu Tong, Xinzhao Fan, Yaoming Xue, Xiaohong Lin, Ying Cao

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A rare case report of Turner Syndrome with SRY gene and Non-clas

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A Rare Combination: Congenital Adrenal Hyperplasia Due To ...

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A **combination of Turner syndrome (TS)** and **classical congenital adrenal hyperplasia (CAH)** is rare. A one-day-old newborn was referred to our hospital with **ambiguous genitalia.** The parents were third-degree relatives. The infant's weight was 3350g (50-75p), and the head circumference was 34.5cm (50p). The gonads were nonpalpable.

Cited by: 1 **Author:** Havva Nur Peltek Kendirci, Zehra Aycan, Se...

Publish Year: 2012

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PEOPLE ALSO ASK

Is genetic testing available for congenital adrenal hyperplasia?

Is genetic testing available for at risk relatives?

What is congenital adrenal hyperplasia?

Is adrenal hyperplasia hereditary?

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Congenital Adrenal Hyperplasia

Medical Condition

A group of rare genetic conditions that reduce certain hormone production in the adrenal glands.

- Rare (Fewer than 200,000 cases per year in US)
- Requires lab test or imaging
- Treatments can help manage condition, no known cure
- Can be lifelong

Caused by the deficiency of proteins required to make specific hormones. Enlarged penis, ambiguous genitalia and salt-wasting shock are the commonly noted symptoms. Treatment includes medication and reconstructive surgery.

Symptoms

Symptoms of classic CAH in infants are:

- Enlarged genitals or clitoris in females
- Enlarged penis in male infants

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Turner syndrome with SRY gene and non-classical congenital adren

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Congenital Adrenal Hyperplasia, Non-Classical Form ...

https://pediatricendocrinologynj.com/congenital...

Non-classic **congenital adrenal hyperplasia** due to 21-hydroxylase deficiency (NCAH) is a milder and later onset form of a genetic condition known as **congenital adrenal hyperplasia**. Some people affected by the condition have no associated signs and symptoms while others experience symptoms of androgen (male hormone) excess.

PEOPLE ALSO ASK

Is genetic testing available for congenital adrenal hyperplasia?

Can congenital adrenal hyperplasia be cured?

What are the symptoms of congenital adrenal hyperplasia?

Is genetic testing available for at risk relatives?

Feedback

Nonclassic congenital adrenal hyperplasia misdiagnosed as ...

www.jhrsonline.org/article.asp?issn=0974-1208;year=...

We present a patient with **nonclassic congenital adrenal hyperplasia (NCAH)** misdiagnosed as mosaic Turner syndrome. She presented with complaints of primary infertility. Short stature, the presence of facial hair and hoarse voice was also noted.

Clinical implications of the detection of Y-chromosome ...

https://www.sciencedirect.com/science/article/pii/S0015028207035583

Oct 01, 2008 - Clinical implications of the detection of Y-chromosome mosaicism in **Turner's syndrome**: **report** of 3 cases ... virilization and high levels of testosterone (T). **Congenital adrenal hyperplasia** previously had been investigated and ruled out. ... et al.Screening for Y-derived sex determining **gene SRY** in 40 patients with **Turner syndrome**. J Clin ...

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Symptoms

Symptoms of classic CAH in infants are:

- Enlarged genitals or clitoris in females
- Enlarged penis in male infants

In adults and children, symptoms of classic CAH are:

- Early pubic hair growth
- Rapid growth

Symptoms of Non- classic CAH in adult and teenage females are:

- Absence of menses
- Deep voice, and excessive facial hair, body hair
- Acne

In both males and females, symptoms of Non- classic CAH include:

Early pubic hair appearance

Treatments

Treatment depends on the type of CAH and severity of symptoms.

Medication

- Corticosteroids: Given to replace the hormone cortisol. Hydrocortisone · Cortisol