

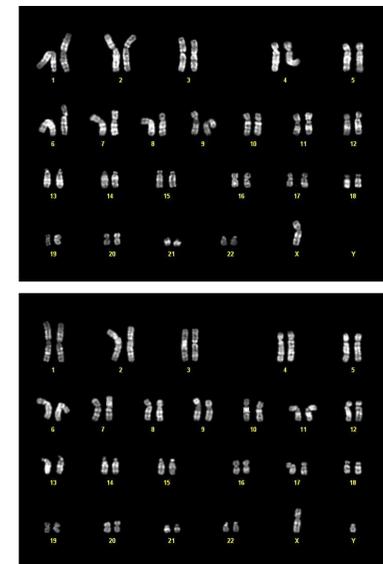
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BACKGROUND

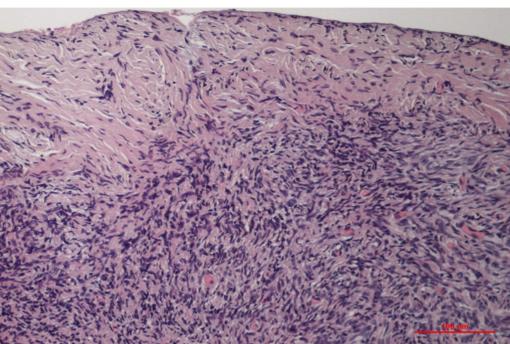
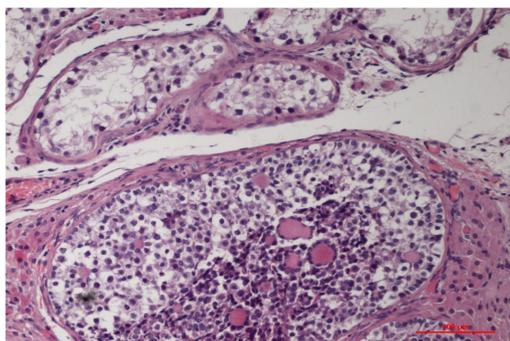
Male pseudohermaphroditism is a group of disorders in which, despite the existence of a Y chromosome, the fetus develops a female or an ambiguous phenotype. A not common etiology may be a gonadal dysgenesis (GD), which may be pure (46,XY) or mixed (Y chromosome mosaicisms).

Patients with GD typically present with one or two dysgenetic testis associated with a streak or an absent contralateral gonad. In the fetus, this gonadal picture is associated with no müllerian regression hence the presence of a uterus and one or both ovaric tubes. Streak gonads may be associated with the development of gonadoblastoma.

Among these patients, cases of peripheral PP have been reported; on the contrary, a central PP has been very rarely described.



CASE: 8 yr and 5 mo old girl remitted to our attention for suspected precocious puberty



- **Clinical evaluation:** mild clitoris hypertrophy, no dysmorphic features.
Tanner stage B3 AH2 PH3-4,
Height 129.3 cm (50th centile)
Weight 39.4 Kg (90th centile)
BMI 23.27 (>97th centile)
- **Bone age:** advanced (+ 18 mo).
- **Pelvic US:** pre-pubertal gonads and uterus.
- **Blood exams:** Tumoral markers were negative. Hormonal assessment was normal and the 17 β -Estradiol levels fitted the patient's Tanner stage. Surprisingly *Testosterone was very high (233 ng/dL)*.
- **GnRH test:** high levels of LH and FSH.
- **MRI:** apparently normal gonads and hypoplastic uterus.
- **Karyotype:** 45,X0/46,XY(50%/50%).
- The **videolaparoscopy** disclosed: hypoplastic uterus, a fallopian tube on the right and an epididymis on the left.
- The **histological exam** showed the presence of bilateral epididymis and fallopian tubes; bilateral gonadoblastoma with germinal elements and Leydig cells on the left gonad and ovarian cortex looking like tissue with no oocytes on the right one.
- **Bilateral salpingo-oophorectomy was performed.**

A) Gonadoblastoma, Leydig cells and germinomatous elements

B) Ovarian cortex with no oocytes

DISCUSSION

GnRH test revealed the activation of the HPG axis and a gonadotropine pattern hinting gonads failure. The presence of Leydig cell and lack of normal ovarian tissue suggested that estradiol production might depend on a peripheral conversion of testosterone by tissue aromatase. By the way, the gonadoblastoma itself could have produced the hormone. Another wonder is whether, at the very beginning, our patient experienced a gonadotropine-dependant or independent form of precocious puberty. The first one would result from an aleatory activation of the HPG axis, stimulating dysgenetic gonads to produce sexual steroids and possibly the tumor replication; the second one would result from an autonomous sexual steroids production leading, by chronic exposure, to HPG axis activation.

CONCLUSIONS

Our case underline the importance of taking into consideration, when dealing with suspect pubertal disorders, the possibility of a sexual development disorder – a male pseudohermaphroditism in our case – because of its difficult early diagnosis when not associated to ambiguous genitalia at birth and its liability to neoplastic evolution.