HYPOGONADOTROPIC HYPOGONADISM IN A PATIENT WITH VANISHING TESTIS SYNDROME – CASE REPORT

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OBJECTIVES

Vanishing testis syndrome leads to hypergonadotropic hypogonadism.

We report a case detected with hypogonadotropic hypogonadism after puberty time.

CASE HISTORY

The 28-years-old patient was diagnosed with bilateral cryptorchidism and phymosis at birth. At **1, than 3 and 7-years-of-age** surgical explorations have not found the testes.
Meanwhile one cure of hCG therapy was applied, without any success.
Endocrine investigations continued just at **24 years-of-age** (in **2011**):
- infantile external male genitalia, undeveloped male secondary sexual characters
Surprisingly, **very low LH (0.13 mIU/mL)** and **FSH (0.7 mIU/mL)** with **low free testosterone level (1.64 pg/mL**, normal: 5.5-42) were detected.
- normal hypothalamic-pituitary system on **MRI**
- abdominal and pelvic MRI: the gonads were not found, hypoplastic prostate and seminal vesicles
- Karyotype: 46,XY

Table 1. Hormonal assessment in 2016

Hormonal assessment	Value	Reference range for age and gender
TSH	1.23 mIU/L	0.35-4.94
free-T4	1.10 ng/dL	0.7-1.48
Baseline cortisolaemia at 8:00 AM	12.8 microg/dL	5-23
Prolactin	5.46 ng/mL	3.46-19.4
LH	**0.03** mIU/mL	1.5-15
FSH	**0.13** mIU/mL	0.95-11.95
Total testosterone	**37.4** ng/dL	142.4-923.1
SHBG	**20.8** nmol/L	13.5-71.4
Albumin	40.34 g/dL	35-52
Free testosterone	**0.85** ng/dL – 2.28%	5-19.8 1.1-2.5%
Bioavailable testosterone	**20.2** ng/dL – 54%	83-257 35%
AMH (anti-müllerian h.)	**2.02** ng/mL	1.43-11.6
DHEAS	370.5 microg/dL	167.9-591.9

Table 2. Human chorionic gonadotropin stimulation test

hCG stimulation test	Total testosterone	Androstene-dione	Dihydro-testosterone	hCG (normal<2.5)
Day one	**26 ng/dL** (241-827)	6.7 nmol/L (2.09-10.82)	67 ng/L (250-1,000)	–
2+1/2 amp. hCG 1,500 U im. (1,500 U/m2 SQ)				
Day three	**30 ng/dL**	4.92 nmol/L	61 ng/L	33.9 mIU/mL
2+1/2 amp. hCG 1,500 U im. (1,500 U/m2 SQ)				
Day six	**22 ng/dL**	4.81 nmol/L	80 ng/L	25.2 mIU/mL

Interpretation: no testosterone response to hCG → no functional testicular tissue

INVESTIGATIONS, RESULTS

Due to **non-compliance** he returned to endocrine evaluation just at **28-years of age** (in **2016**), complaining of intense hypogastric pain and micturition disturbance.
Urological diagnosis and therapy: severe phymosis with chronic incomplete urinary retention and frequent urinary infections. Chronic prostatitis.
After circumcision urinary complains ceased.

Physical exam: Weight 112 kg, Height 185 cm, W/H: 0,97, BSA 2.4 m2, Armspam 189 cm, no testes in the scrotum, micropenis, stage Tanner 1, pubic hair stage Tanner 2, absence of secondary sexual characters, gynaecomastia, normosmia

Endocrine investigations:
Hormonal assessment is presented on Tables 1. and 2.

Imagistic findings:
Hypothalamic-pituitary MRI: hypothalamus, pituitary stalk, adeno- and neurohypophysis, and other structures with normal appearance.
Abdominal and pelvic MRI: undetected testicles, atrophic seminal vesicles, hypoplastic prostate, inflammatory inguinal lymph node enlargement.
DXA osteodensitometry: L1-L4 vertebral T score: -1,42 SD
Other investigation: Total PSA <0.04 ng/mL (normal < 4)

DISCUSSION, CONCLUSIONS

The almost **undetectable basal testosteron** level, **without any elevation after hCG** stimulation test, and **undetectable testicles on repeated abdominal and pelvic MRI** prove the **absence of both testicles** (1). During puberty time an **isolated hypogonadotropinaemia** was detected with **normal anatomy of hypothalamus-pituitary system on MRI**. **Testosterone replacement therapy** was started.
Final diagnosis: 46,XY DSD: Vanishing testis syndrome, Hypogonadotropic hypogonadism, Secondary osteopenia. Gynoid obesity grade I.
We report a case with bilateral anorchia associated with hyposecretion of gonadotropins. The occasional coexistence or correlation of these two etiologies need to be studied, in the literature being reported only in few cases (2, 3).

References

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