Testicular feminization syndrome as a rare cause of tumor-level serum testosterone in women

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OBJECTIVE
The aim of our work was to evaluate the number and the final diagnosis of women with an elevation of total testosterone levels within tumor range, i.e. ≥ 1.6 ng/ml by a direct RIA (normal range: 0.2-0.8 ng/ml).

RESULTS
In the 17 patients with a total testosterone level > 3 ng/ml, diagnosis were: pregnancy with normal index of free testosterone (n = 2), congenital adrenal hyperplasia (CAH) (n = 3), androgen therapies in post-menopausal women (n = 3), men misclassified as women by data processing (n = 2), unknown for 3 hyperplasia (CAH) (n = 3), androgen therapies in post-menopausal women (n = 305-310) and FSH levels.

Karyotype analysis revealed a male chromosomal sex (46,XY) in the presence of a female phenotype confirming the diagnosis of complete androgen insensitivity syndrome. Finally, FISH analysis using a SRY gene probe confirmed the presence of SRY gene on Y chromosome.

DISCUSSION
Complete androgen insensitivity syndrome is an X-linked recessive disorder with an estimated incidence between 1/20000 and 1/99000 live male births. It is caused by mutation in the androgen receptor (AR) gene which is located on the long arm of the X chromosome (Xq11-12).

The syndrome was first described in 1953 by John Morris as “testicular feminization syndrome”, who reviewed the clinical features of 82 patients. The syndrome was later given the name of androgen insensitivity syndrome. Finally, a systematic analysis of a lab database could allow to spot unusual cases that were possibly undiagnosed.

CONCLUSION
A systematic analysis of a lab database could allow to spot unusual cases that were possibly undiagnosed. In this case of a woman with CAIS, management is dependent on a multidisciplinary team (endocrinologist, gynaecologist, clinical psychologist) in order to address functional, sexual and psychological issues such as disclosure to the patient, gonadectomy and subsequent hormone replacement, creation of a functional vagina and genetic counseling.

REFERENCES
2. Hughes IA, Deeb A. Androgen resistance. Best Pract Res Clin Endocrinol Metab 2006; 20: 570-585

PATIENTS AND METHOD
Analysis of the lab database (between 2004 and 2013) identified 132 women.

Only 17 patients had serum testosterone concentrations above 3 ng/ml.

In the 17 patients with a serum level < 2 ng/ml and 85% ≤ 3 ng/ml.

Biochemistry showed elevated total testosterone (8.4 ng/ml) with normal SHBG conferring elevated index of free testosterone, low estradiol and normal LH and FSH levels.

Karyotype analysis revealed a male chromosomal sex (46,XY) in the presence of a female phenotype confirming the diagnosis of complete androgen insensitivity syndrome. Finally, FISH analysis using a SRY gene probe confirmed the presence of SRY gene on Y chromosome.