Testicular adrenal rest tumors in two young patients with congenital adrenal hyperplasia
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Introduction

Testicular adrenal rest tumours (TART) may develop in males with congenital adrenal hyperplasia (CAH), due to overstimulation of aberrant adrenal cells (1) or originate from the adrenal cortex tissue: ectopic tissue in the testes-stimulated by ACTH excess. All pathways causing an elevation of ACTH levels may be associated with adrenal inclusions (2). Widely variable prevalence, congenital adrenal hyperplasia by 21-hydroxylase deficiency being by far the most common (3).

Case reports

First case

- 15 years old, diagnosed with CAH at the age of 3 weeks due to 21-OH deficiency
- Successfully treated with glucocorticoids and salt supplementation until the age of six, then interrupted without consulting a specialist
- Gradual increase of testicular size, consults for ‘testicular infertility’ (Figure 1)
- Physical examination:
  - Short stature (147.5 cm, -2DS), puberty P VI, testicles ~ 50 ml, hard
- Imaging:
  - Testicular ultrasound: RT 79/46 mm, inhomogeneous hypoechoic; LT 68/33 mm, contains 3 hypoechoic formations, isomorphic, of 21/13, 30/15 and 20/12 mm (Figure 2)
- Adrenal ultrasound (Figure 3): increased dimensions, macronodular aspect
- Replacement therapy with glucocorticoids was given:
  - Prednisone 10 mg/day in 2 divided doses
  - The follow-up revealed an improvement of hormonal and imagistic parameters:
    - Clinical - reduction in testicular size of 25% (Figure 4)
    - Biological - improved gonadal axis
  - Morphologic: ultrasound RT 60/50 mm, 58/34 mm LT, normal adrenal ultrasound (Figures 5 and 6).

Second case

- 10 years old, diagnosed with CAH at the age of 3 weeks due to 21-hydroxylase deficiency
- Received intermittent steroid supplementation over the course of his life, because of poor compliance
- Long-term evolution of adrenal hyperplasia, inconsistent and incomplete therapy resulted in early onset of puberty
- Puberty stage IV, G IV
  - Advanced stature (+1.26 SD)
  - Advanced bone age (~ 14 years)
- At the age of 8, the scrotal ultrasound revealed increased size of testes, rich blood supply, with bilateral TRATs (Figure 7)
- RT: 46/31/26 mm, LT: 51/31/27 mm
- Given the elevated gonadotropins, advanced bone age and stature, the therapy with GnRH analogues was introduced, with improved evolution.

Conclusions and Discussions

- TART is by far the most likely diagnosis in the presence of testicular nodules in the following cases:
  - Patient with congenital adrenal hyperplasia
  - Bilateral involvement
  - Ultrasound aspect (hilarious location, preserved vascular architecture)
  - Anomalies diminishes under well-balanced substitution treatment (5)
- TART is the most important cause of infertility in adult male patients with CAH, representing a diagnostic challenge (1)
- The reported prevalence by sonography varies between 24% and 94% (3)

Parameters

<table>
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<tr>
<th>Parameters</th>
<th>Normal</th>
<th>First case</th>
<th>Second case</th>
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<tbody>
<tr>
<td>FSH (mIU/ml)</td>
<td>1.5–11</td>
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<td>1.3</td>
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<td>LH (mIU/ml)</td>
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<td>TESTOSTERON (ng/ml)</td>
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<td>ESTRADIOL (pg/ml)</td>
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<td>ACTH (pg/ml)</td>
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<td>77.8</td>
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<td>CORTISOL (mg/ml)</td>
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<td>446.7 ± (7)</td>
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<td>rOH PROGESTERON (ng/ml)</td>
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References: