A 19-year-old adolescent with short stature and scrotal tumor

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Background

Presentation of an adolescent for diagnostics of Primordial Short Stature PSS (= SGA with birth length and/or -weight -2 standard deviations (SD) or more and without catch up growth into the normal range until 2nd birthday). Differential diagnoses: 1) maternal causes (e.g. placental insufficiency), 2) fetal factors (prenatal infections), 3) genetic syndromes (Turner syndrome or mixed gonadal dysgenesis (MGD) with 45X/46XY mosaic, 4) skeletal disorders (achondroplasia) or 5) idiopathic PSS. Follow up is necessary a) to exclude underlying a systemic disease or growth hormone deficiency and b) to provide for diagnostics of increased risk of diabetes and cardiovascular diseases or of malignancy, e.g. in MGD.

Case presentation

19 year old adolescent boy with short stature

History:
Birth: spontaneous delivery after uneventful pregnancy; GA 40 weeks
- BW 2500g (SD -2.55), BL 47 cm (SD -2.38)
- recurrent urinary tract infections with double kidneys on the left side in earlier childhood
- overweight as a child
- mildly impaired intelligence, special school; training as a gardener
- delayed puberty with an onset at the age of 16 years

Clinical investigation:
- male phenotype
- well-proportioned short stature, except for a broad chest
- dysmorphic signs: low-hairline, low-set ears, multiple pigmented naevi
- no cardiovascular or renal anomalies
- normally formed penis
- two intrascrotal hypotrophic testes with a volume of 5 ml each, in the presence of nearly adult pubertal stages P5, G4-5
- caudal to the right testis in the right scrotal space an indolent scrotal tumour well separated from the testes was palpable

Endocrinological investigations:

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
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</thead>
<tbody>
<tr>
<td>Testosterone</td>
<td>14.85 nmol/l</td>
</tr>
<tr>
<td>FSH</td>
<td>15.41 U/l</td>
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<tr>
<td>LH</td>
<td>3.37 U/l</td>
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<tr>
<td>DHEA-S</td>
<td>8.0 µmol/l</td>
</tr>
</tbody>
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Karyogram: mixed gonadal dysgenesis with 46,XY(75%)/45,X(25%)

Ultrasound:
- tubular structure, suspected to be a hypoplastic uterus

Histological investigation:
- rudimentary uterus 3.7 cm in length without a cervix
- lined with inactive endometrium with minimal signs of proliferation
- typical expression of CD10 in the endometrial stroma and nuclear expression of oestrogen, progesterone and androgen receptor
- no evidence of malignancy in testicular biopsy
- but preserved active spermiogenesis

Conclusions

- In children with PSS and discrete dysmorphic signs of UTS, a karyogram should be performed in time, to rule out MGD
- A wide phenotypical range may be found in MGD, comprising the entire spectrum from normal testes or ovaries, unilateral streak gonads with contralateral testis, ovary or uterus to bilateral streak gonads
- The proceeding in male patients with MGD is controversially discussed because of the increased risk of testicular malignancy.
- Orchietomy was not performed in this patient since there were no indicators of malignancy, a well-accessible intra-scrotal location of the testes and an otherwise normal gonadal function
- Regular clinical and ultrasound examinations should detect malignant degeneration in time

Notes & References
Conversion from S-units: Testosterone: 14.85 nmol/l x 288 = 4276.8 ng/l