**3β-HSD2 deficiency due to compound heterozygosity of a missense mutation (p.Thr259Met) and a frameshift deletion (p.Lys273ArgFs*7) in an under-virilized infant male with salt wasting**

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**Objectives**

Describe clinical, hormonal and genetic findings of an undervirilized infant male, 13 month old, Afghan origin, presented at the emergency department with salt-wasting adrenal crisis.

**Presenting case**

A 13 month old male infant presented at the emergency department with complaints of vomiting and fever.

- **Physical examination / Personal history**
  - Physical examination was significant for severe dehydration along with perineal hypospadias, bifid scrotum, penile chordee and undescended left testicle (fig. 1)
  - He was he was under cortisone treatment that was stopped 15 days before referral to our hospital.

- **Laboratory evaluation**
  - Revealed hyponatremia, Na: 130 mEq/L (135-145 mEq/L) with concurrent hyperkalemia, K: 5.5 mEq/L (3.5-4.5 mEq/L).
  - Steroid hormone testing obtained before treatment revealed a complex pattern suggestive of congenital adrenal hyperplasia (CAH) due to 3β-HSD deficiency (table 1).

- **Scrotal u/s** revealed the right testicle in the scrotal area (1.82x1.34, 0.92) cm (Vol: 1.12 cm3) and the left testicle in the upper inguinal area (1.38x0.89x0.93) cm (Vol: 0.58 cm3), both harboring microcalcifications.

<table>
<thead>
<tr>
<th>DHEA-S</th>
<th>13,35 (&lt;0,600)μgr/ml</th>
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<tbody>
<tr>
<td>17-OH-Progesterone</td>
<td>35,07 (0.200-0.800)ng/ml</td>
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<tr>
<td>Testosterone</td>
<td>0,946 (&lt;0,025)ng/ml</td>
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<tr>
<td>Cortisol</td>
<td>1,15 (6,2-23,00)μgr/dl</td>
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<tr>
<td>Chromosomal karyotype</td>
<td>46,XY</td>
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**Table 1**

**Conclusions**

Deficiency of 3-beta hydroxysteroid dehydrogenase type II (3β-HSD2) is a rare autosomal recessive form of congenital adrenal hyperplasia (CAH). More than 40 mutations have been found in the HSD3B2 gene causing 3BHSD2D2. Our patient is the first undervirilized male with severe salt wasting presenting compound heterozygosity of missense c.776C>T, p.Thr259Met and frameshift deletion: c.818-819delAA, p.Lys273ArgFs*7 maybe due to a founder effect for people leaving in that area.

The authors declare that they have no conflict of interest.

**References**