Could intramuscular testosterone differentiate between partial androgen insensitivity and 5 alpha reductase deficiency?

Shaymaa Raafat¹, Saber Mohammed Waheeb²

¹Pediatric Endocrinology and Diabetology Unit, Department of Pediatrics, Faculty of Medicine, Alexandria University, Egypt.
²Pediatric Surgery Department, Faculty of Medicine, Alexandria University, Egypt.

INTRODUCTION

• Disorders of sex development are challenging for patients, parents, and the interdisciplinary health-care team. Sex assignment of these patients needs optimal integrated hormonal profile, imaging and molecular diagnosis.
• Testosterone replacement is one of the cornerstone of management of DSD cases. When molecular diagnosis is unavailable, the DSD team searches for other diagnostic clues helping diagnosis as response to testosterone treatment.

AIM

This study aimed at finding a new tool for differentiation between partial androgen insensitivity and 5 alpha reductase deficiency especially when the molecular diagnosis is highly expensive or even unavailable.

METHOD

Forty-four boys were included in this study through a year form January 2020 to January 2021. They were divided into two groups: partial androgen insensitivity (22 patients) and 5 alpha reductase deficiency (22 patients)

All patients were assessed with emphasis on
• Detailed history
• Thorough clinical examination including stretched penile length and glans diameter.
• Karyotype
• Hormonal profile was done for all of them. Imaging studies including ultra-sound for pelvis and inguino-crural region were done.

All included boys received three intramuscular injections of testosterone enanthate 25 mg every four weeks as a preparation for masculinizing genitoplasty.

RESULTS

Among boys of the study cohort, there were 22 patients with partial androgen insensitivity and 22 patients of 5 alpha reductase deficiency. Their ages ranged from one month to 56 months. Six patients were initially assigned as females then finally assigned as males after their complete assessment. (Table 1)

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Partial androgen insensitivity</th>
<th>5 alpha reductase deficiency</th>
<th>U</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Min. – Max.</td>
<td>12.0 – 76.2</td>
<td>11.11 – 145.0</td>
<td>95.0</td>
<td>0.001</td>
</tr>
<tr>
<td>Mean ± SD</td>
<td>31.27 ± 19.33</td>
<td>67.26 ± 34.20</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Median (IQR)</td>
<td>29.33 (20.40-40.0)</td>
<td>60.0 (31.00-100.10)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

% of increase in length

- 150
- 100
- 50
- 0

Patients with partial androgen insensitivity had variable while patients with 5 alpha reductase deficiency had significant increase in SPL after injections in comparison with partial androgen insensitivity boys. (Table 3, Figure 1)

CONCLUSIONS

• Preoperative testosterone injections can help the treating team to reach an accurate diagnosis and proper sex assignment as they can differentiate between partial androgen insensitivity and 5 alpha reductase deficiency

• Patients with partial androgen insensitivity may need more than one course of testosterone injections or increasing the dose of injection to 50 mg.

REFERENCES


CONTACT INFORMATION

drshimoraafat@yahoo.com
S_elsaid00@Alexmed.edu.eg