Assessment of the gonadotrophin–gonadal axis and Sertoli cell function in partial androgen insensitivity syndrome

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Objectives:

Androgen insensitivity syndrome (AIS) is the largest single entity that leads to male under-masculinization. Although adequate serum concentrations of testosterone exclude a defect in testosterone biosynthesis, a low testosterone value at baseline does not always exclude PAIS. OBJECTIVE To study the value of measuring basal and human chorionic gonadotropin (HCG) stimulated testosterone level, Dihydrotestosterone, anti-mullerian hormone (AMH) and Inhibin levels in 9 prepubertal children with the final diagnosis of partial androgen insensitivity syndrome (PAIS)

Methods:

Retrospective study of patients in Alexandria University Ped Endocrine clinic, Alexandria, Egypt. Patients included 9 cases of PAIS (mean age = 8.2 months ± 2.3) A single dose HCG stimulation protocol was used (1500U/m2). Measurements included **pre-HCG and post-HCG serum testosterone values, serum DHT values, and serum AMH and inhibin were measured and analyzed.**

Results:

The mean testosterone rise following fixed dosage of HCG was 94.5 times the basal value. 5/9 patients had low basal testosterone. The mean stimulated testosterone: DHT ratios were 11.3. AMH was High to normal in 8/9 patients and Inhibin was high to normal in 7/9 patients and low in 2/9 patients.

Conclusions:

Basal testosterone may not be raised during early infancy in patients with PAIS; however testosterone rise after HCG stimulation is adequate. The elevation of serum AMH and inhibin level appears to be an interesting marker of androgen resistance in sexually ambiguous male infants.

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