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

Doaa Khater, Magdy Omar, Shaymaa Raafat
Department of Pediatrics, Faculty of Medicine, Alexandria University, Egypt

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.

References: