

Proximal hypospadias in a male patient with 5 α -reductase deficiency: A case reports

Erol Basuguy, Selcuk Otcu

Abstract

Hypospadias is a congenital disorder of male external genital. The newborn showed penoscrotal hypospadias with chordee and microphallus. Endocrine data and a normal male karyotype were suggestive of 5 α -reductase deficiency. Penoscrotal hypospadias repair of the patient was made.

Key Words

Hipospadias; Penis; Fetal development; 5 α -reductase deficiency

From the Department of Pediatric Surgery, University of Dicle, School of Medicine, Diyarbakir, Turkey.

Corresponding Author: Erol Basuguy

Dicle University, School of Medicine,

Department of Pediatric Surgery, Diyarbakir 21280, Turkey.

E-mail: erbas.80@hotmail.com

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Introduction

Hypospadias is a congenital disorder of male external genital with an incidence of 0.03–0.4%. However, epidemiological evidence suggests that in developed Western countries the incidence is increasing [1]. The male human urethra develops in the course of fetal wk 8–16, and any hesitation in this process causes hypospadias. However, the etiology of the hypospadias is still not clearly understood [2]. It may be categorized as simple (glandular or penile) or severe (penoscrotal, scrotal or perineal) according to the anatomical location of the urethral meatus [3].

The development of the external male genitalia occurs by the induction, growth and differentiation of the genital tubercle by the effect of androgens [4]. Dihydrotestosterone (DHT) synthesized by 5 α -reductase is more potent than testosterone (T) and is essential for normal development of the human male external genitalia [5]. Here, we report a boy with 5 α -reductase deficiency and proximal hypospadias.

Case reports

The newborn showed penoscrotal hypospadias with chordee and microphallus (2.3 cm length). Palpable gonads were located in the scrotum. Endocrine data and a normal male karyotype were suggestive of 5 α -reductase deficiency. The testosterone was 540 pg/ml and dihydrotestosterone was 16 pg/ml and the basal T:DHT ratio was 33. After administration of hCG plasma testosterone was 4.58ng/ml and

dihydrotestosterone was 22.7 pg/ml. The length of the phallus increased by 5 mm (from 2.3 to 2.8 cm) after administration of testosterone enanthate once per month for 3 months. Penoscrotal hypospadias of the patient was repaired and in the patient did not develop any problems (Fig. 1).



Figure 1. Post-operative appearance of the penis.

Discussion

The enzyme 5α -reductase is a nicotinamide adenine dinucleotide phosphate dependent protein that catalyses the conversion of testosterone into DHT. It is 50-fold more potent than testosterone [6].



DHT is essential for formation of the male phenotype such as the male external genitalia, urethra and prostate during embryogenesis [7]. A rare disease, the enzyme steroid 5α -reductase deficiency represents 46,XY genetic male subjects who have incomplete virilization of their external genitalia and testes [8]. However, the clinical spectrum is heterogeneous, varying from a female to a fully male phenotype with hypospadias or only microphallus [9,10]. Spermatogenesis is generally normal if the testes are descended in the scrotum [11]. Hormonal values include normal or increased testosterone levels, with low DHT levels in relation to T and a high ratio of 5β - to 5α -reduced urinary steroid metabolites. Stimulation with human chorionic gonadotropin (hCG) reveals more this altered ratio [12]. Hypospadias can be induced experimentally by androgen receptor antagonists, inhibitors of 5α -reductase, and

inhibitors of enzymes involved in steroid hormone synthesis [13-15].

Different of types of the hypospadias can be observed according to the original site of the opening of the urethral meatus and to other associated anomalies. Proximal hypospadias are a less frequently seen and correspond to 20% of all cases of hypospadias [16]. Proximal hypospadias usually found together with chordee. Chordee is formed by residual fibrous tissue of the corpus spongiosum and is located on the ventral side of the urethra [17]. Additionally, proximal hypospadias with chordee is the most challenging type of hypospadias to reconstruct^[18]. Complications of the hypospadias repair are generally complete disruption, fistula formation, stenosis, post operative bleeding, and recurrence of chordee or excess skin at the glans. The most common complication is fistula formation [19]. Optimal age for hypospadias repair is between 8 and 12 months of age [17]. In the present case, endocrine data were followed. Our patient was operated on a year old and was used flap from scrotum for repair of the hypospadias, chordee was corrected.

As a result, in patients with proximal hypospadias and micropenis, endocrine data should be obtained and a multidisciplinary approach to the treatment of patients is important.

CONFLICT OF INTEREST

None declared.

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