5 α -Reductase Type 2 Deficiency — Response to Dihydrotestosterone Gel: Correspondence

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To The Editor: In the article by Madhavarao et al., recently published in Indian Journal of Pediatrics, the authors have described response to Dihydrotestosterone (DHT) gel in a case with 5 α -reductase 2 deficiency (5 α RD). The child was put on three monthly intramuscular testosterone injections (25 mg), which increased the stretched penile length (SPL) to 2.1 cm from initial SPL of 1.3 cm and both the testes descended to labioscrotal folds with increase in rugosity. The authors also reported that the application of DHT gel increased SPL to 3 cm within 1 mo and later to 3.4 cm after 5 mo [1].

We also observed increase in SPL after application of DHT cream in one of our patients with 5 α RD. Our patient, born of a consanguineous marriage, was reared as female until 13 y when diagnosis of 46, XY DSD was made. He had small phallus (SPL 2.5 cm) with chordee, bifid scrotum and penoscrotal hypospadias. USG revealed left testis was in labioscrotal fold and right testis in inguinal canal. His basal hormonal levels were LH 5.96 mIU/mL, FSH 15.83 mIU/mL, Testosterone (T) 4.35 ng/mL, DHT 210 pg/mL, T/DHT 20.7, Androstenedione (A) 0.92 ng/mL, T/A 4.7. DHT and A were

estimated by radio immuno assay (RIA) after Celite-chromatography. He was advised to apply DHT cream over the phallus. On next follow up at 7 mo, the SPL had increased to 5 cm. Psychological evaluation revealed male gender identity. Most of our peri-pubertal children with 5 α RD had opted for male genitoplasty [2]. Molecular genetic studies revealed homozygous missense mutation of R246Q on exon 5 of *SRD5A2* gene. This mutation has been reported by some centers from India [3, 4].

To correct cryptorchidism, alternative treatment with hCG is recommended [5].

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