Correlation of androgen receptor and SRD5A2 gene mutations with pediatric hypospadias in 46, XY DSD children

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ABSTRACT. We performed an exploratory study by analyzing the correlation of 46, XY disorders of sex development (46, XY DSD) with androgen receptor (AR) and steroid 5α-reductase-2 (SRD5A2) gene mutations and a safety analysis of dihydrotestosterone (DHT) gel treatment for pediatric micropenis. We collected samples from 76 pediatric patients with 46, XY DSD and 50 healthy adult men with normal fertility as the control group. The pediatric patients were treated with DHT gel (0.1-0.3 mg/kg/day) for three to six months. The extended penis length, testicular volume, and multiple blood parameters were collected before treatment and one, three, and six months after treatment. Of the 76 cases with 46, XY DSD, 31.58% had hypospadias with micropenis and 6.58% had male pseudohermaphroditism. Through AR gene screening, it was found that 14 patients had AR point mutations and 22 patients had SRD5A2 mutations. After treatment with DHT, the penis length of the patients significantly improved after one, three, and six months of treatment, with longer treatment times resulting in greater improvement. Before treatment with DHT, the average serum DHT value of patients with 46, XY DSD was 24.29 pg/mL. After one, three, and six months of treatment, this value increased to 430.71, 328.9, and 323.6...
pg/mL, respectively. We conclude that for pediatric patients who have male
hermaphroditism or hypospadias with micropenis, AR and SRD5A2 gene
mutation detection should be performed. Local application of DHT gel can
promote penis growth effectively without systemic adverse reactions.

**Key words:** 46,XY; AR gene; Hypospadias; Micropenis; SRD5A2 gene