Androgen receptor CAG and GGN polymorphisms in boys with isolated hypospadias

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Background: The etiology of hypospadias is multifactorial. Abnormal androgenic secretion and/or action during the development of external genitalia may be involved in the etiology of this congenital malformation. This study explored CAG and GGN polymorphisms in the androgen receptor (AR) gene, which may affect its transcriptional activity, in patients with isolated hypospadias. Methods: The length of the CAG/GGN polymorphisms was determined in 44 boys with non-severe (glandular) or severe (penile or penoscrotal) isolated hypospadias and with a normal hormonal evaluation. In addition, 79 healthy men, as controls, were studied. Results: Mean CAG repeats were signifi cantly higher in total and severe cases compared to controls (24.4 ± 2.8 and 24.7 ± 3.1 vs. 22.7 ± 3.3, respectively; p > 0.05, Student ' s t and Bonferroni test). In addition, a frequency of CAG alleles < 23 was signifi cantly different in total and severe cases compared to controls (70.5 % and 74.1 % vs. 39.2 % , respective