Analysis of clinical manifestations and gene mutations of 5α-reductase type 2 deficiency in 16 cases

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Objectives:

5α-reductase type 2 deficiency is a rare autosomal recessive hereditary disease. SRD5A2 gene defects lead to dysfunction of 5α-reductase type 2, that impair the conversion of testosterone to dihydrotestosterone and cause clinical features. In this report, we we try to analyze the clinical manifestations and gene mutations of 5α-reductase type 2 deficiency in childhood.

Methods:

The clinical features, laboratory tests and gene mutational analysis of 16 cases of 5α-reductase type 2 deficiency in our hospital were analyzed retrospectively.

Results:

In 15 of 16 cases with gender as male, 1 case of gender as female. All had genital dysplasia in clinical history and ambiguous genitalia in physical examination, including microphallus, penile curvature, hypospadias, bifid scrotum and clitoridauxe, etc. The test of hCG stimulation suggested the existence of testicular tissue and the function of secretion of testosterone. 10 mutations in the SRD5A2 gene were detected in 16 patients, including c.282-1G>C, c.586G>A,p.(Gly196Ser), c.680G>A,p.(Arg227Gln), c.159G>A,p.(Trp53X), c.607G>A,p.(Gly203Ser), c.650C>A,p.(Ala217Glu), c.665G>A,p.(Cys222Tyr), c.196G>A,p.(Gly66Arg), c.656delT,p.(Phe219fs) and c.560C>T,p.(Thr187Met). The most common detected mutatons was c.680G>A,p.(Arg227Gln), that 13 out of 16 patients carried the mutation, including 5 homozygous and 8 heterozygous. The second most common one was c.607G>A,p.(Gly203Ser), that 4 out of 16 cases were detected, including 2 homozygous and 2 heterozygous.

Conclusions:

Patients of 5α-reductase type 2 deficiency in childhood had clinical features of genital dysplasia. The test of sexual hormone and hCG stimulation helped to estimate the function of testicular tissue, and SRD5A2 gene mutational analysis was necessary for precise diagnosis. This study found that the mutatons of c.680G>A,p.(Arg227Gln) and c.607G>A,p.(Gly203Ser) may be the hotspot mutations in Chinese patients of 5α-reductase type 2 deficiency.

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