

GENDER IDENTITY IS ESTABLISHED EARLY IN CHILDHOOD IN 5 α REDUCTASE DEFICIENCY

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Male pseudohermaphroditism (MPH) due to 5 α reductase deficiency is not an uncommon endocrine entity. The disorder has an autosomal recessive pattern of inheritance. The disorder is diagnosed at puberty. Here we present 2 brothers of a family presenting with genital ambiguity

A.N., 15 years of age from eastern India was born of a full term normal vaginal delivery of a 1st degree consanguinous marriage. The genitalia at birth looked female. Therefore child was reared as a female. Childhood milestones were normal. However by the age of 4 years, the child preferred to wear male dresses and play with boys. The child did schooling till age 14 years in a girls' school. Recently, noticing the child's behaviour, parents have shifted him to boy's school and sought medical attention.

On physical examination, he is 178cms tall and weighs 60kg. He has muscular body built. There is no gynaecomastia; no facial hair. Genital examination revealed pubic hair stage 3 with perineoscrotal hypospadias. Stretched phallic length is 5 cms. Both the testes are palpable at the inguinal rings and measure 10ml in volume. Based on this a clinical diagnosis of MPH was made. Hormonal profile is as given in the table below.

LH (U/L)	4.3
FSH (U/L)	4.7
PRL (μ g/L)	16
Testosterone (ng/dl)	420
DHT (ng/dl)	14.4
T: DHT ratio	29.2
Post hCG tests	
Testosterone (ng/dl)	1050
DHT (ng/dl)	31
T: DHT ratio	34

With this we made a diagnosis of 5 α reductase deficiency. This child has 2 elder siblings – both girls with normal menstrual cycles; 1 younger sibling who was reared as female. We had the opportunity to see her clinically. She is 5 years old but dressed like a boy and the style of walking and general behaviour resembled a male child. Genital examination revealed small phallus with perineoscrotal hypospadias and both testes palpable in inguinal canals.

Discussion: This child with 5 α reductase deficiency was being reared as female. However, from the age of 3-4 years itself the behaviour and general outlook was like a male child. This means gender identity is established early in childhood and not at puberty as many believe.

Conclusion: This reemphasizes the importance of deference of genital surgery to a later age when gender identity can be assessed.

Presentation Type: Oral Presentation

Authorize Publishing: Yes

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