

TWO CASES OF PARTIAL ANDROGEN INSENSITIVITY- SOCIAL ISSUES

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Androgen insensitivity syndrome is an X linked disorder of male sexual differentiation with incidence of 1 in 20,00-60,000 male births. Here we describe two siblings who presented to us at the age of 5 and 15 years, with partial androgen insensitivity syndrome (PAIS).

The eldest sibling who is now 31 years of age, presented initially at the age of 5-6 years with ambiguous genitalia, for which corrective surgery was performed at 7 years of age. He developed gynecomastia at the age of 12 years for which surgery was advised. His basal hormonal profile showed a LH of 7.2U/L and S testosterone of 800 ng/dl and on LHRH stimulation he had a peak LH of 12.2U/L and a peak S.testosterone of 800 ng/dl.

The second sibling who was last in birth order presented initially at the age of 15 years with gynecomastia, on examination he had absent testis on the left side and hypospadias. His basal hormonal profile showed a LH of 6U/L and a S testosterone of 445 ng/dl and on LHRH stimulation test he had a peak LH of 30IU/L and S.testosterone was 690 ng/dl. Karyotype was 46XY in both siblings.

In India home deliveries are common, ambiguous genitalia often go unnoticed. There is lack of awareness among the public regarding the medical centers or specialists caring for these patients. The lack of privacy due to overcrowding in out patient departments and fear of lack of confidentiality, deter patients from availing the medical facilities. There is also a tendency among the patients and their parents to conceal the phenotype, so as to avoid social isolation and segregation in the community. All of these factors results in a delay in seeking medical attention.

These cases highlight the fact that there is a pressing need to improve the awareness among the public on inter sex states in India, so that affected children present for appropriate treatment at an early age.

Presentation Type: Oral Presentation

Authorize Publishing: Yes

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