

## **46XY TRUE HERMAPHRODITISM SPANNING FIVE GENERATIONS**

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This is a study of a family of true hermaphrodites. All affected members had a karyotype of 46XY. The phenotype was variable. Only 4 affected members could be studied in detail as these came to the hospital. A visit to their village revealed several more members of the family having similar features on clinical examination. Only one of the members consented to have blood collected for karyotyping, which was 46XY. One of the members had died of gonadal malignancy while 3 others were treated for gonadal malignancy. Tracing the history back into 4 previous generations, it is surmised that in all, 12 members were affected over 5 generations. The transmission was always found to be by normal males. All normal females gave birth to normal children. The third generation was found to have skipped the defect. Of the 4 patients who were hospitalised, 2 were children, reared as boys, and underwent masculinizing genitoplasty with removal of uterus and adnexa and the gonads as required. The other two were adults admitted for malignancy. To our knowledge, this is the first time, that familial true hermaphroditism having karyotype of 46XY is reported.

**Presentation Type:** Oral Presentation

**Authorize Publishing:** Yes

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