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Familial Pure Gonadal Dysgenesis with 46, XY Karyotype in Three Siblings and Gonadoblastoma in the Youngest Sibling

M. Eunice, B. Kulshreshtha, A. Kriplani *, Rima Dada, S. Agarwal***,**

K. Kucheria, A. K. Karak***, S. Datta Gupta*** and A. C. Ammini**

Department of Endocrinology and Metabolism, *Department of Obstetrics and Gynaecology, **Department of Anatomy, ***Department of Pathology, All India Institute of Medical Sciences , New Delhi 110 029, India

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ABSTRACT The occurrence of gonadoblastoma in XY phenotypic females has been well documented. This condition may rarely present in a familial form. In this present study, we present a family of 5 siblings born of non-consanguineous parents. Three sisters (age 13, 15 and 17 years) with XY karyotype presented with complaints of primary amenorrhoea. They had a normal female phenotype. The younger two brothers were apparently normal. All the three affected siblings have undergone laparoscopic removal of the gonads. The histopathology of gonads in the elder sibling (II 1) revealed ovarian stroma along with rete testes like elements. Bilateral tubes and the gonads of the younger sibling (II 2) showed fallopian tubes (both sides) and a small portion of ovarian stroma. No focus of gonadoblastoma was identified in these two elder siblings. Histopathology of the youngest sibling (II 3) revealed presence of bilateral gonadoblastoma. The occurrence of gonadoblastoma in the youngest sibling of 3 affected XY siblings is unusual.