A Case of True Hermaphroditism with 45X/46XY Mosaicism

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Summary. The authors describe a case of true hermaphroditism of mainly female phenotype, ambiguous external genitalia, and ovotestis. The cytogenetic studies revealed 45X/46XY mosaicism and an absence of Barr bodies.

Subjects with 45X/46XY mixoploidy manifest an extremely variable phenotype that ranges from Turner's syndrome to male pseudohermaphroditism and includes all clinical pictures found between these two extremes; however, only 2% of these can be considered true hermaphrodites (Gorlin, 1974).

As this form of intersexuality is rare, we think the case we observed merits both a clinical and genetical report.

Case Report

S. R., aged 16 years, was born naturally at full-term. She was the second-born of non-consanguineous parents, her ascendants and collaterals were in apparently good health. At 8 years of age the development of her clitoris was abnormal. She was first seen at the clinic of obstetrics and gynaecology of the University of Perugia for primary amenorrhea and because of the ambiguity of her external genitalia. Her height (149 cm) was below normal and she weighed 50 kg. Clinical examination (Fig. 1 a and b) revealed bilateral cubitus valgus, hypotrophy of the breast, and only moderate hair development, except in the pubic area where it was of the mixed-type distribution.

In the vulva area (Fig. 1 c) only the labia majora were perceptible; they bordered a furrow which originated from the junction of the labia minora. The labia minora, in their turn, joined together to form a fraenulum which at the upper limits terminated in a large clitoris, which in formation and size was not unlike the penis of a 12-year-old male, with a large gland and an abundant foreskin partially covering it.

A number of laboratory examinations were performed on the patient during her hospitalization.

1. Morphologic and hematologic examinations were within the normal range.
2. Radiologic examinations revealed L5 left hemisacralization and widespread scoliosis with right convexity. No abnormalities were noted in the cranium, thorax, and abdomen.
3. Urographic examination showed the right side to be normal. On the left there was an anomalous rotation of the kidney. There were no filling defects and no intrinsic alterations in the bladder walls. The excretory urography revealed a short opaque tract in the urethra but no canalization defects.
4. Endocrine examinations: a) urinary 17 ketosteroids = 10.08 mg/24 h; b) urinary 17 ketogenics = 7.14 mg/24 h; c) total urinary estrogens = 9.388 mg/24 h; d) pregnandiol = 0.05 mg/24 h; radioimmunologic sampling of the hypophysial gonadotropins = LH 19.3 UI/24 h, FSH 52.2 UI/24 h.
Fig. 1. (a and b) Phenotype of patient. (c) External genitalia. (d–g) Surgical findings showing internal genitalia.

Fig. 2. Histologic section of ovotestis showing seminiferous tubules and ovarian stroma with primordial follicles.