Special Feature:

Breast tumour in a male with complete androgen insensitivity

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A 17-year-old phenotypically female patient presented with chief complaints of not attaining menarche and also of a swelling in the left breast. Physical examination revealed normal breast development, normal female external genitalia, sparse pubic hair, absent axillary hair and bilateral inguinal masses. Ultrasonography of abdomen showed absence of uterus and ovaries in the pelvis. Her hormonal evaluation revealed elevated testosterone of 6.9 ng/dL with raised leutinizing hormone (LH) 33.0 IU/L and normal follicular stimulating hormone (FSH) 5.6 IU/levels. Subsequent cytogenetic analysis confirmed 46 XY karyotype. She underwent excision of the breast swelling at a local hospital. Histopathological examination of the excised breast tissue was suggestive of multiple papilloma with hyperplasia of unusual type and immunohistochemistry markers positive for CK-5, p 63 and actin (Figures 1, 2 and 3).

Androgen insensitivity syndrome, previously called as testicular feminization syndrome, is a rare X-linked recessive condition. Morris et al, described this entity in 1963, and it has also been known as 'Morris syndrome'.[1] It is a form of male pseudohermaphroditism, i.e., male karyotype (XY) with a female phenotype. The defect is in androgen receptor that is normally present in the androgen responsive tissue which results in failure of normal masculinization of external genitalia in chromosomally male individuals.[2] This failure of masculinization can be either complete or partial depending on the amount of residual receptor function.[3] In

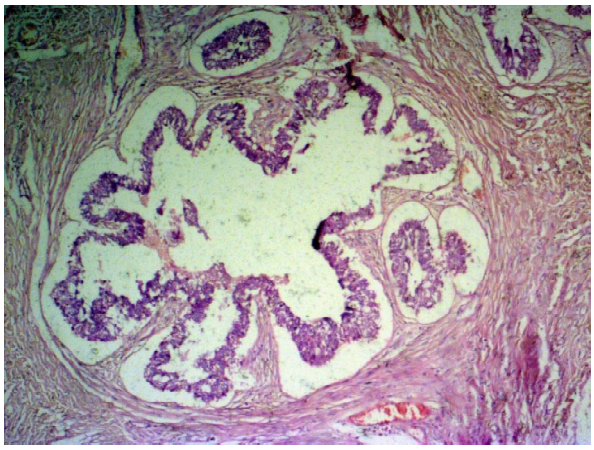

Figure 1: Photomicrograph showing ducts lined by stratified cuboidal epithelial cells, arranged in the form of ill-defined papillary projections with surrounding selerosis (Haematoxylin and eosin, × 200)

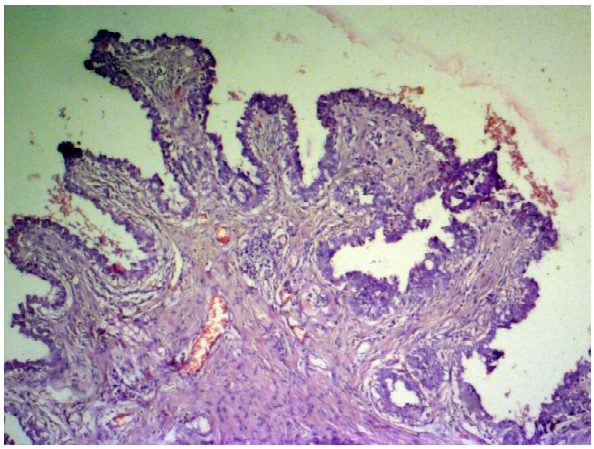

Figure 2: Photomicrograph showing mammary ducts with hyperplastic papillae lined by stratified cuboidal epitelial cells (Haematoxylin and eosin, × 400)

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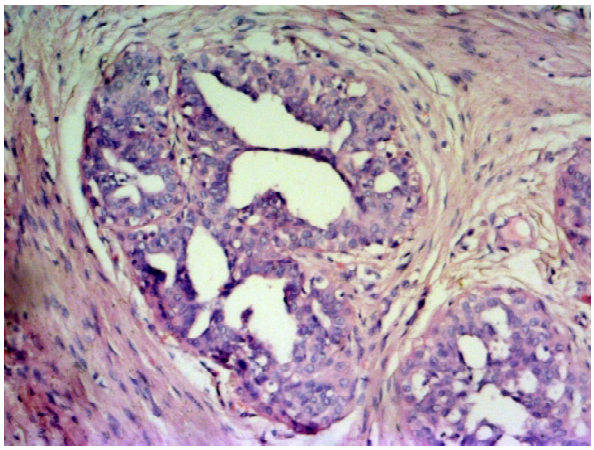

Figure 3: Photomicrograph showing the mammary ducts with cribriform pattern, lined by hyperplastic cuboidal epithetical cells (Haematoxylin and eosin, × 400)

complete androgen insensitivity syndrome (CAIS), the individual has adequate breast development, normal female external genitalia, a vagina of variable depth, absent uterus, and sparse or absent pubic hair and axillary hair.[4] The gonad (undescended testes) may be intra-abdominal, inguinal, or labial. They most often present in late adolescence with primary amenorrhoea. Breast tumours have not been previously reported in CAIS.

Breast tumours are rare in males but the risk is increased in those with androgen insensitivity caused by longer glutamine repeats in the androgen receptor seen with partial or minimal androgen insensitivity syndrome. Breast cancer has not been reported in CAIS.[5] The above case report illustrates a rare presentation of breast papilloma with hyperplasia of unusual type in a male with complete androgen insensitivity syndrome. All papillary lesions with atypical hyperplasia require surgical excision owing to the cancer risk in future.[6]

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