Original Research Paper

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General Surgery

A CASE REPORT ON ANDROGEN INSENSITIVITY SYNDROME

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ABSTRACT Androgen Insensitivity Syndrome is an X linked disorder resulting in normal masculinization of external genitalia due to loss of function mutation in AR gene. Case Report : A 18 year old female presented with C/O not attaining menarche. Patient gives a history of lack of development of pubic and axillary hair. No history of abnormal breast development,cyclical abdominal pain or excessive weight gain. No relevant family history. O/E No palpable swellings in abdomen or bilateral inguinal region. External genitalia appear like a normal female external genitalia. The patient's karyotyping was done and it is of male genotype. The patient is diagnosed as Complete AIS as the phenotype is female and genetically male. The patient is managed by laparoscopic B/L orchidectomy with B/L deep ring closure.

KEYWORDS : Complete Androgen Insensitivity Syndrome ; male genotype ; X linked disease ; P/W - Presented with ; O/E - On Examination ; B/L - Bilateral ; AIS - Androgen Insensitivity Syndrome ; AR - Androgen receptor gene

INTRODUCTION

Androgen Insensitivity Syndrome is typically characterised by evidence of feminization of external genitalia at birth,abnormal secondary sexual development in puberty, and infertility in individuals with a 46,XY karyotype due to the loss of function mutation in the AR gene. Incidence is 1 in 20,000 to 64,000 male births. AIS represents a spectrum of defects in androgen action and can be subdivided into three broad phenotypes [1] 1) Complete AIS with typically female external genitalia 2) Partial AIS with predominantly female or male or ambiguous external genitalia 3) Mild AIS with typical male external genitalia. Complete AIS are phenotypically normal women and have a tendency to develop gonadal malignancies. In either case, affected individuals have normal testes with normal production of testosterone and normal conversion to dihydrotestosterone. Because the testes produce normal amounts of müllerian-inhibiting factor, affected individuals do not have fallopian tubes, a uterus, or a proximal (upper) vagina.

PATHOPHYSIOLOGY

The basic pathology of androgen insensitivity syndrome is a loss-of-function mutation in the androgen receptor (AR) gene. This AR gene has been localized to the long arm of the X chromosome.These mutations can cause a variety of functional defects, ranging from a complete loss of receptors on the cell surface because of incomplete protein synthesis to alterations in substrate binding affinity.Loss of AR function means that, despite normal levels of androgen synthesis, the typical postreceptor events that mediate the effects of hormones on tissues do not occur. This results in the phenotype of prenatal undervirilization of external genitalia, absence of pubic and axillary hair, lack of acne, and absence of voice changes at puberty.

CASE PRESENTATION

18 year old female presented with c/o not attaining menarche. Patient gives a history of lack of pubic and axillary hair. No history of abnormal breast development,cyclical abdominal pain or excessive weight gain. No relevant family history. O/E No palpable swellings in abdomen or bilateral inguinal region. External genitalia appear like a normal female external genitalia.

CLINICAL COURSE

DISCUSSION

Patients with Complete AIS are chromosomally and gonadally male but phenotypically female. In an adolescent patient, notable findings include primary amenorrhea with B/L Inguinal masses, having no pubic or axillary hair, although breasts are normal due to peripheral conversion of testosterone to estradiol[2]. The diagnosis was confirmed after karyotyping and the patient managed by laparoscopic B/L Orchidectomy with B/L deep ring closure. Intra operative findings include 3×2 cm mass at deep rings with omental herniation bilaterally. Uterus and Ovaries were absent.

BLOOD TESTS	Hb - 13 gm/dl WBC - 10,700/mm3 PLC - 3.4 lakh/mm3 T.Bilrubin - 0.9mg/dl ALT - 9.9 U/L ALP - 134 U/L
USG ABDOMEN & PELVIS	Uterus is not visualised. Ovaries couldn't be evaluated. 2.8 × 1.9 cm Iso to hypoechoic area noted in B/L Inguinal region likely Testes.
KARYOTYPING	46,XY
HPE	Section studied shows preserved architecture of seminiferous tubules with various stages of spermatogonia, moderate to abundant interstitial fibrosis with leydig cell hyperplasia. Epididymis also shows fibrosis . S/o Cryptorchid testes.

Figure 1 :- Intra op picture of testes on Laparoscopy.

CONCLUSION

The prognosis in these patients is excellent with appropriate medical and psychological support.

Patients with AIS Benefit with a multi disciplinary approach including :-

- Gonadectomy at puberty as the risk of testicular malignancies like Germ Cell tumour[3] is significantly high.
- Psychological counseling as these patients may have psychosocial problems ranging from identity issues to problems dealing with gender perceptions.
- Hormone replacement therapy.

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