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COMPLETE ANDROGEN INSENSITIVITY SYNDROME: REPORT OF A CASE WITH FOCAL SEGMENTAL GLOMERULOSCLEROSIS

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ABSTRACT Complete Androgen Insensitivity Syndrome, commonly known as the Testicular Feminization Syndrome, is characterized by a 46, XY karyotype, bilateral testes, absent or hypoplastic Wolffian ducts, and female-appearing external genitalia with diminished axillary and pubic hair development. Although initial diagnosis in the child is difficult, the syndrome must be suspected after puberty if primary amenorrhea is present. Coexistence of genital defects with urologic abnormalities is expected in these cases because of close embryologic origin. However, renal involvement in form of Nephrotic Syndrome does not seem so common. We report a case of testicular feminization syndrome with Focal Segmental Glomerulo Sclerosis.

KEYWORDS :

Testicular feminization (the syndrome of complete androgen insensitivity) is characterized clinically by a 46, XY karyotype; bilateral testes which may be found in the labia, inguinal canal or abdomen, female-appearing external genitalia, and absence of Mullerian duct derivatives. The syndrome is caused by androgen resistance as a result of several types of receptor abnormalities starting from in utero. The disease has an incidence of 1 in 20,000 to 1 in 60,000 males and is transmitted as an X-linked trait.

In the prepubertal child, the diagnosis is difficult, but the diagnosis may readily be made in the postpubertal patient on the basis of clinical and hormonal findings of amenorrhea and/or inguinal hernias containing testes (1).

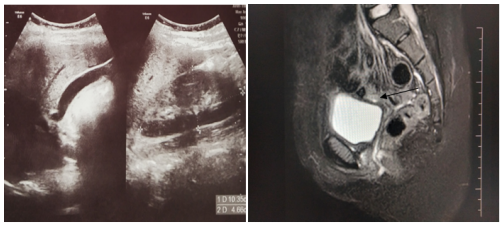
The syndrome of partial androgen resistance, which is another variant of testicular feminization, is characterized by ambiguity of the external genitalia. The classic phenotype is a male with hypospadias, cryptorchidism, rudimentary Wolffian structures, gynecomastia, and infertility. But endocrine profile is similar to that of the complete androgen insensitivity syndrome (1). The psychosexual orientation of the patient is usually female, which is the rule for complete forms.

Since the Mullerian ducts develop in association with the Wolffian ducts and their derivatives, it is not surprising that renal abnormalities occur with the failure of Mullerian development. Nephrotic Syndrome in form of FSGS was recognized in our patient.

Case Report

A 21-year-old female was initially admitted to the hospital with primary amenorrhea, facial puffiness and pedal edema. She was evaluated in Nephrology Department and was referred to Endocrinology in view of Primary Amenorrhea. On general examination , she had tall stature 178 cms, Breast B3 with minimal pubic hair and absent axillary hair. External genitalia appeared normal superficially, but during the gynecological examination, atrophic, short (5-6 cm in length) and blind-ending vagina was seen. During examination of the inguinal area, no inguinal hernia or palpable testes was observed. There was no family history of congenital abnormality or female infertility.

Blood biochemical analysis was normal. Serum hormonal parameters including prolactin (PRL), follicle stimulating hormone (FSH), luteinizing hormone (LH), progesterone and testos-terone, were as follows (PRL: 8.17 ng/ml; FSH: 97.99 mIU/ml; LH: 29.87 mIU/ ml; progesterone: 9.3 ng/ml; testosterone: 0.59 ng/ ml).USG Abdomen and Pelvis revealed Hypoplastic Uterus.



USG Abdomen and Pelvis – Hypoplastic Uterus MRI Abdomen & Pelvis – Hypoplastic Uterus

However, leukocyte chromosome analysis revealed a 46, XY male karyotype. Renal Biopsy suggested Focal Segmental Glomerulo sclerosis with no immune deposits. In addition, bilateral hyperintense structures anterolateral to epigastric vessels were noted , which was thought to be the immature testicular tissue on MRI..

DISCUSSION

During embryogenesis, androgen resistance prevents masculinization of the external genitalia and differentiation of the Wolffian ducts. Secretion of anti-Mullerian hormone by the fetal Sertoli cells leads to regression of Mullerian ducts. Therefore, affected patients are born with female appearance.

One of the syndromes of androgen resistance, known as testicular feminization, is characterized by normal levels of serum testosterone but external features that resemble totally the female phenotype with hypoplastic clitoris and labia minora, a blind vaginal pouch, and absent Mullerian derivatives. Most of the patients were diagnosed after puberty during the evaluation of primary amenorrhea. The syndrome is mostly contrasted with the Mayer-Rokitansky-Kuster-Hauser syndrome in which chromosome analysis of the patient reveals a 46, XX karyotype.

Use of MRI in the diagnosis of these disorders has some advantages: a lack of ionizing radiation, perfect contrast resolution, and soft tissue visualization – features that allow exact localization of undescended testes or immature gonadal structures in these patients. In 1997, Reinhold and colleagues accurately localized gonadal structures in 86% of patients with testicular feminization or true hermaphroditism (4).

Although it is well known that undescended testes are more prone to neoplastic changes than descended testes, the incidence of tumor development in complete androgen insensitivity syndrome is reported as 3.6% at the age of 25 years (2), which is only slightly higher than for a cryptorchid testis (3). Unfortunately, this risk reaches 33% at the age of 50.

CAIS follow up requires the team work of Endocrinologist, Urologist and Pychiatrist.

In the present case, the patient wants to be assigned female gender only

and blind vagina is present. So there is no requirement of neovagina creation. Gonadectomy is planned and if required, low dose Estrogen for Breast development and Bone health

CONCLUSION

Our case reveals the association of Renal involvement in form of FSGS with complete androgen insensitivity syndrome. Involvement of genital with urinary disorders must alert the clinician and radiologist to the need for a detailed and cooperative evaluation of both the genital and urinary system, which can be accomplished easily by MRI.

REFERENCES

1. Diamond DA. Sexual differentiation: normal and abnormal. In: Walsch PC, et al., editors. Campbell urology. Philadelphia: Saunders; 2002. P. 2417–18.
2. Manuel M, Katayama K, Jones H. The age of occurrence of gonadal tumors in intersex patients with a Y chromosome. Am J Obstet Gynecol 1976;124:293–300.
3. Muller J, Skakkebaek NE. Testicular carcinoma in situ in children with the androgen insensitivity (testicular feminization) syndrome. Br Med J 1984;288:1419.
4. Reinhold C, Hricak H, Forstner R, Ascher SM, Bret PM, Meyer WR, et al. Primary amenorrhea: evaluation with MR imaging. Radiology 1997;203: 383–90.