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MRI Features of Androgen Insensitivity Syndrome: Case Report

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Abstract | **Case Report**

Androgen insensitivity syndrome or testicular feminization is a genetic condition caused by the defect of androgen receptors and resulting in a lack of response to androgen. In the complete form patients have a female phenotype with a male karyotype (XY). In most cases it is revealed by primary amenorrhea. Imaging modalities are very contributive in the initial diagnosis but also to localize gonads. We report the case of complete androgen insensitivity in a 12 years old girl, discovered accidentally during an ultrasound for acute abdominal pain.

Keywords: Androgen Insensitivity Syndrome, Testicular feminization, Magnetic Resonance Imaging.

INTRODUCTION

Androgen insensitivity syndrome is an X linked recessive disorder, that was first described by Morris, characterized by the inability of organs to respond to androgen due to the absence or defect of the androgen receptors. Patients are phenotypically females and genetically males, they present normal female external genitalia however there is an agenesis of the Müllerian duct derivatives, resulting in the absence of the Fallopian tubes, uterus, cervix, and the proximal part of the vagina and the presence of testes along their path of descent [1]. Early surgical removal of the gonads helps preventing malignant transformation. When the diagnosis is suspected, initially an ultrasound is performed to localize the gonads. MRI on the other hand is more efficient in detecting intra-abdominal gonads, characterizing them and depicting complications. MRI is very contributive prior to surgical treatment.

CASE REPORT

We report the case of a 12 years old girl that consulted primarily for acute abdominal pain due to gastroenteritis. An ultrasound was performed that demonstrated the absence of uterus and ovaries as well as the presence of testes bilaterally in the inguinal canal (Figure 1).

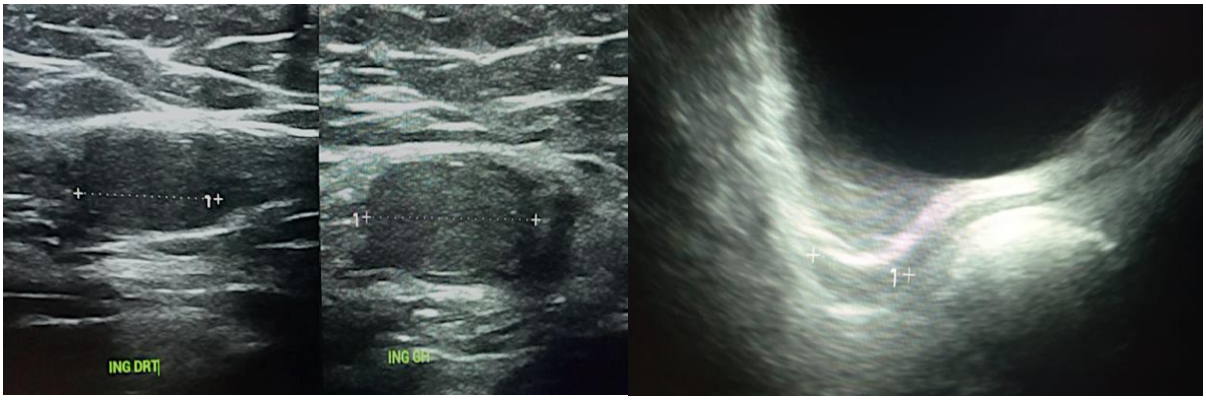

Figure 1: Androgen insensitivity syndrome in a 12-year-old girl with female phenotype. Abdominopelvic ultrasound shows the presence of both testicles in the inguinal canal (a) and the lower of the vagina (b)

MRI of pelvis showed well defined ovoid structures, with intermediate signal intensity in T2 weighted images bilateral in the inguinal canal suggestive of undescended testes. It also confirmed absence of uterus and ovaries (Figure 2).

Patient underwent karyotyping that proved the male XY karyotype. Bilateral gonadectomy was performed, and the patient was planned for hormonal substitution with estrogens. Histopathology showed seminiferous tubules lined by Sertoli cells with Leydig cell hyperplasia.

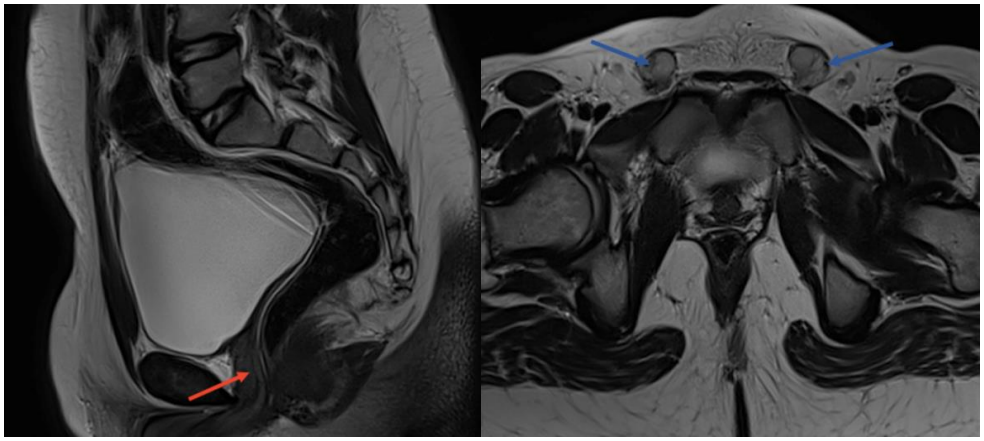

Figure 2: MRI with sagittal T2 (a), axial T2 (b) image: Absence of uterus and ovaries (red arrow) with testes in the inguinal canals (blue arrow)

DISCUSSION

The prevalence for androgen insensitivity syndrome (AIS) is estimated to be between 1 /20,400 and 1/ 99,100 [2]. This disorder is due to a mutation in the androgen receptor gene. Two entities can be distinguished: complete androgen insensitivity syndrome and partial androgen insensitivity syndrome depending on the degree of virilization. In the complete form, the patient develops external female genitalia as well as secondary sexual characteristics. In both entities the psycho-sexual gender of the patient is female [2, 3]. The clinical presentation is variable depending on the age. In the adult population primary amenorrhea is the primary cause of consultation. Infants on the other hand may present inguinal hernia or labial swelling [2, 4]. In our case AIS was discovered fortuitously during an ultrasound for acute abdominal pain. The testes can be located in the abdomen, inguinal canal or in the sub-labial soft tissue due to an incomplete descent. The main risk is the malignant transformation, which is dominant in the adult population. Differential diagnoses include cryptorchidism, lymphadenopathy, Mayer Rokitansky kuster Hauser. Imaging modalities are very contributive. Ultrasound is the first prescribed examination; it is performed to localize testicular tissue; however its efficiency decreases when the testes are located above the inguinal ring. The absence of uterus and ovaries are also noted. MRI has better soft tissue contrast, it can detect the exact localization and characteristic of the testes, it's also important in ruling out differential diagnoses. Transverse and sagittal T2 and T1 weighted images are used to assess the absence of the uterus, ovaries, vagina as well as the presence and location of the testes and signs of testicular malignancy [5, 6].

CONCLUSION

Imaging techniques are crucial in both the initial diagnosis as well as preoperative planning in androgen insensitivity disorder.

REFERENCES

1. Morris, J. M. (1953). The syndrome of testicular feminization in male pseudohermaphrodites. *American Journal of Obstetrics & Gynecology*, *65*(6), 1192-1211. doi:10.1016/0002-9378(53)90359-7.
2. Tank, J., Knoll, A., Gilet, A., & Kim, S. (2015). Imaging characteristics of androgen insensitivity syndrome. *Clinical Imaging*, *39*(4), 707-710. http://dx.doi.org/10.1016/ j.clinimag.2015.02.002
3. Koren, A. T., Lautin, E. M., Kutcher, R., Rozenblit, A., & Banerjee, T. D. (1996). Testicular feminization: radiologic considerations in a unique formof cryptorchidism. *Abdominal imaging*, *21*(3), 272-274.
4. Hughes, I. A., Davies, J. D., Bunch, T. I., Pasterski, V., Mastroyannopoulou, K., & MacDougall, J. (2012). Androgen insensitivity syndrome. *Lancet*, 380(9851), 1419-1428.
5. Nezzo, M., De Visschere, P., T'sjoen, G., Weyers, S., & Villeirs, G. (2013). Role of imaging in the diagnosis and management of complete androgen insensitivity syndrome in adults. *Case Reports in Radiology*, *2013*. Doi:10.1155/2013/158484
6. Elagwany, A. S., Eltawab, S., & Abouzaid, Z. S. (2016). 46, XY complete gonadal dysgenesis (Swyer syndrome): Report of two different cases. *Apollo medicine*, *13*(1), 63-66. http://dx.doi.org/10.1016/j.apme.2013.10.005.