The Nigerian Journal of GENERAL PRACTICE

Original Article

An Intriguingly Rare Presentation of Androgen Insensitivity Syndrome in a Female Adult in South- South Nigeria Presenting as a Diagnostic Dilemma- A Case Report and Literature Review

Dr ET Omu[1], Dr A Dinyain[2], Dr YE Oku[3], Dr RJ Biowe[4], Prof SF Brisibe[5].

[1]Department of Surgery, Familycare Hospital Yenagoa, [2]Department of Pathology, NDUTH
[3]Department of Surgery, Federal Medical Centre, Yenagoa, [4]Department of surgery, NDUTH
[5]Department of Family Medicine NDUTH.

Abstract

Androgen insensitivity syndrome (AIS) previously known as testicular feminization syndrome is a rare inherited form of disorders of sexual development which can be complete or partial. The complete type occurs in phenotypically normal women with adequate breast development, normal external genitalia, a vagina of variable depth, absent uterus, and sparse or absent pubic and axillary hair. These patients have male karyotype (XY) and negative sex chromatin. The gonad (undescended testis) may be intra-abdominal, inguinal, or labial and may be bilateral. We present patient SGJ, a 30-year-old unmarried female National Youth Corp Member who presented with a one-day history of a severely painful left groin mass. She had normal breast development, scanty growth of pubic and axillary hair, normal vulva and vagina but no cervix from a speculum examination. She had been amenorrhoeic since birth but sexually active. Groin examination revealed a tender soft mass measuring 3 by 2cm at the left inguinal region with an initial diagnosis of obstructed hernia. The uterus and ovaries were absent on ultrasonography. Due to the severity of the mass in the groin, she was counselled for emergency surgery under spinal anaesthesia and written consent obtained following which an emergency groin surgical exploration was done. A normal testicle was resected to avoid gonadal tumours in later life and confirmed histopathologically two weeks later as a cryptorchid testes. Hormonal assay result which also came two weeks later revealed elevated testosterone, normal FSH and LH levels. Chromosomal study (genetic karyotype) could not be done due to patient's decision despite counselling. The aim of this report is to review the current understanding of the diagnosis and treatment of complete androgen insensitivity syndrome.

Keywords: Spleen, Anthropometry, Nnewi, Ultrasound Scan, Weight, Height.

Address for correspondence author: Dr E.T.Omu, Department of Surgery, Familycare Hospital Yenagoa

How to cite: Omu ET, Dinyain A, Oku YE, Biowe RJ, Brisibe SF. An intriguingly rare presentation of Androgen Insensitivity Syndrome in a female adult in South- South Nigeria presenting as a diagnostic dilemma- A case report and literature review. NJGP. 2025:23(1):11-15.
DOI: https://doi.org/10.60787/njgp.v23i1.249 (https://doi.org/10.60787/njgp.v23i1.249)

Quick Response Code:

Introduction

Androgen insensitivity syndrome (AIS) can be defined as a disorder resulting from complete or partial resistance to the biological actions of androgens in an XY man or boy with normal testis determination and production of age-appropriate androgen concentrations[1-6]. AIS is caused by genetic defects on the X chromosome. These defects make the body unable to respond to the hormones that produce a male appearance [7-9]

The pathophysiology of this syndrome is based on the mechanism of action of androgens which gives rise to two types: the complete and partial androgen insensitivity syndrome.

An individual with complete AIS has the physical traits of a woman, but the genetic makeup of a man. The typical presentation for complete androgen insensitivity syndrome is either primary amenorrhoea in adolescence, or inguinal swellings in an infant. A female adolescent with the disorder has breast development and a pubertal growth spurt at the appropriate age, but no menses. No precise figures are available for the prevalence of complete androgen insensitivity syndrome, but estimates range from one in 20 to one in 99. Development of oestrogen-dependent secondary sexual characteristics occurs as the result of excess aromatisation of androgens. Pubic and axillary hair is usually absent or can be present in sparse amounts. In infancy, complete androgen insensitivity syndrome presents as an inguinal hernia or labial swelling containing a testis in an apparently female infant. The uterus, cervix, and proximal vagina are absent in complete androgen insensitivity syndrome because of the action of antimüllerian hormone produced by Sertoli cells of the testis. The vagina varies from a dimple in the perineum to normal length, but is always blind-ending

The clinical presentation of partial androgen insensitivity syndrome depends on the degree of responsiveness of the external genitalia to androgens. The typical phenotype is micropenis, severe hypospadias (perineoscrotal), and a bifid scrotum that might contain gonads. Extensive genetic and biochemical investigations are needed to confirm diagnosis.

Case Summary

We present a case of a 30-year-old unmarried female National Youth Corp Member who presented at the gynae clinic with a one-day history of a severely painful left groin mass and was referred to the surgery clinic. The mass had been noticed for more than 18years, painless but progressively enlarged prior to presentation. There was no associated vomiting or constipation. She had been amenorrhoeic and sexually active, having a satisfactory sexual life without medical intervention. The patient was 165cm tall and weighed 64kg. There was normal breast development but scanty pubic and axillary hair growth. Groin examination revealed a tender soft mass measuring 3 by 2cm at the left inguinal region which was reducible with a positive cough impulse and a right supra inguinal scar said to be for the resection of a similar mass at the age of 12years but she could not tell if it was sent for histology. She had normal vulva and vagina but no cervix from a speculum examination. The uterus and ovaries were absent on ultrasonography. The groin was explored surgically as an emergency procedure and a normal testicle was resected and confirmed histopathologically as a cryptorchid testes. A hormonal assay result which was obtained later revealed elevated testosterone as well as normal FSH and LH levels. Chromosomal study (genetic karyotype) was not done due to patient's decision despite counselling.

Discussion

An individual with Androgen insensitivity syndrome (AIS) is genetically male with one X and one Y chromosome but due to defects on the X chromosome, the body is resistant to

male hormones called androgens.[14-15] As a result, the person has some of the physical traits of a woman.

The complete form of the syndrome occurs in as many as 1 in 20,000 live births.[3, 5,7, 15]A person with complete AIS as the case presented appears to be female with a vagina but no cervix or uterus. An inguinal hernia with testes can be felt during a physical exam as in this case which was also reported by other authors.[5,16-18]The gonad (undescended testis) may be intra-abdominal, inguinal, or labial. It was inguinal in the index patient. They have normal female breasts and very little axillary and pubic hair as seen in this case. Complete AIS is rarely discovered during childhood. Most people with this condition are not diagnosed until they do not get a menstrual period or they have trouble getting pregnant as found in other reports.[5, 6, 19-20]

In partial AIS, people have different patterns of male traits.[8-11] Partial AIS can include other disorders, such as: Failure of one or both testes to descend into the scrotum after birth, hypospadias, a condition in which the opening of the urethra is on the underside of the penis, instead of at the tip, Reifenstein syndrome (also known as Gilbert-Dreyfus syndrome or Lubs syndrome). People with partial AIS may have both male and female physical characteristics. Many have partial closing of the outer vagina, an enlarged clitoris, and a short vagina. Partial AIS is often discovered during childhood because the person may have both male and female physical traits.

Treatment and gender assignment can be a very complex issue,[16, 21-23] and must be targeted to each individual person.[3,5, 6, 15, 23]Treatment includes surgical resection of the testes as was done in this case, to prevent development of cancer. Another very important treatment is oestrogen replacement after puberty to prevent the regression of secondary sexual characteristics and consequences of estrogen deficiency; also, to preserve normal sexual activity.[21] This advice was given to the case presented and she was referred to see the endocrinologist. Possible Complications of AIS include: Infertility, psychological and social issues and testicular cancer.[16, 22-23]In terms of prognosis, the outlook for complete AIS is good if the testicular tissue is resected at the right time to prevent cancer.

Conclusion

A 30year with the rare complete androgen insensitivity syndrome is presented who required multispecialty expert care from the gynaecologist, surgeon, endocrinologist and psychologist. This case highlights the diagnostic dilemma that could be faced with the myriads of presentations which calls for a high index of suspicion.

References

1. Fulare S, Deshmukh S, Gupta J. Androgeninsensitivitysyndrome: A rare genetic disorder. Inter J Case Rep. 2020;6:371-373.
2. Batista RL, Costa EMF, Rodrigues AS, et al. Androgen insensitivity syndrome: a review. Arch Endocrinol Metab.2018;62(2):227-235. https://doi.org/10.20945/2359-3997000000031
3. Guo M, Huang JC, Li CF, Liu YY. Complete androgen insensitivity syndrome: a case report and literature review. J Int Med Res. 2023 Feb;51(2):3000605231154413. doi: 10.1177/03000605231154413. PMID: 36851849; PMCID: PMC9983103.
4. *Oakes MB, Eyvazzadeh AD, Quint E, Smith YR. Complete androgen insensitivity syndrome—a review. J. Pediatr. Adolesc. Gynecol. 200821December;6:305–310.*

5. Farah S, El Masri D, Hirbli K. Complete androgen insensitivity syndrome in a 13-year-old Lebanese child, reared as female, with bilateral inguinal hernia: a case report. J Med Case Reports. 2021;15:202-*5.* https://doi.org/10.1186/s13256-021-02738-0

6. *Malipatil V, Malipatil P, Chaitanya VM. Androgen insensitivity syndrome, a case report and literature review. Int. J. Res. Med. Sci. 2016;4:1830–1833.*
7. Hughes IA, Deeb A. Androgen resistance. Best Pract. Res ClinEndocrinol Metab.2006;20(4):577–598.
8. Ahmed SF, Cheng A, Dovey L, et al. Phenotypic features, androgen receptor binding, and mutational analysis in 278 clinical cases reported as androgen insensitivity syndrome. J ClinEndocrinol Metab.2000;85(2):658–665. https://doi.org/10.1210/jcem.85.2.6337
9. Yuan Z, Fan L, Wang Y, Li L, Ren X, Cao Set al. Analysis of genetic and clinical characteristics of androgen insensitivity syndrome: a cohort study including 12 families. European Journal of endocrinology. 2024;191(1):87-96. https://doi.org/10.1093/ejendo/lvae082
10. Gulía C,Baldassarra S, Zangari A, et al. Androgen insensitivity syndrome. Eur Rev Med Pharmacol Sci. 2018;22(12):3873–3887. https://doi.org/10.26355/eurrev_201806_15272
11. Hughes IA, Davies JD, Bunch TI, Pasterski V, Mastroyannopoulou K, MacDougall J. Androgen insensitivity syndrome. Lancet. 2012;380 (9851):1419-1428. doi:10.1016/S0140-6736(12)60071-3.

12. Mongan NP, Tadokoro-Cuccaro R, Bunch T, Hughes IA. Androgen insensitivity syndrome. Best Pract Res ClinEndocrinoMetab. 2015;29(4):569–580. https://doi.org/10.1016/j.beem.2015.04.005
13. Tadokoro-Cuccaro R, Hughes IA. Androgen insensitivity syndrome. CurrOpinEndocrinol Diabetes Obes. 2014;21(6):499–503. https://doi.org/10.1097/med.0000000000000107
14. Liu Q, Yin X, Li P. Clinical, hormonal and genetic characteristics of androgen insensitivity syndrome in 39 Chinese patients. ReprodBiol Endocrinol. 2020;18:34. https://doi.org/10.1186/s12958-020-00593-0
15. Souhail R, Amine S, Nadia A, Tarik K, Khalid E, Abdellatif K, Ahmed I. Complete androgen insensitivity syndrome or testicular feminization: review of literature based on a case report. Pan African Medical Journal. 2016;25:199 doi:10.11604/pamj.2016.25.199.10758
16. Dohnert U, Wünsch L, Hiort O. Gonadectomy in Complete Androgen Insensitivity Syndrome: Why and When? Sex Dev 2017;11:171-174, 2017/07/19. doi: 10.1159/000478082
17. Deeb A, Hughes IA. Inguinal hernia in female infants: a cue to check the sex chromosomes? BJU Int. 2005;96(3):401–3. https://doi.org/10.1111/j.1464-410x.2005.05639.x
18. Hurme T, Lahdes-Vasama T, Mäkela E, Iber T, Toppari J. Clinical findings in prepubertal girls with inguinal hernia with special reference to the diagnosis of androgen insensitivity syndrome. Scand J UrolNephrol. 2009;43:42–6.
19. Babu R, ShahU. Gender identity disorder (GID) in adolescents and adults with differences of sex development (DSD): a systematic review and meta-analysis. J Pediatr Urol. 2021;17(1):39–47. https://doi.org/10.1016/j.jpurol.2020.11.017.

20. Sharma S, Balwan WK, Kumar P, Gupta S. Androgen Insensitivity Syndrome, Testicular Feminization J ObstetGynaecol India 2012 Apr; 62(2):199–201. doi: 10.1007/s13224-011-0098-4

21. *Ko JK, King TF, Williams L, Creighton SM, Conway GS. Hormone replacement treatment choices in complete androgen insensitivity syndrome: an audit of an adult clinic. Endocr. Connect. 2017;6(6)):375–379.*

22. *Hines M, Ahmed SF, Hughes IA. Psychological outcomes and gender-related development in complete androgen insensitivity syndrome. Arch. Sex. Behav. 2003;32:93–101.*

23. *Niveditha V, Sudarshan CY. Psychological aspects of Androgen Insensitivity Syndrome – A case report. J. Psychol. Psychother. 2015;5(183) 2161-0487.*