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OPIS PRZYPADKU
CASE REPORT

Complete androgen insensitivity syndrome as a condition requiring interdisciplinary care: A case report

Zespół całkowitej niewrażliwości na androgeny jako schorzenie wymagające opieki interdyscyplinarnej – opis przypadku

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ABSTRACT

Complete androgen insensitivity syndrome (CAIS; also known as Morris syndrome) is a rare disorder associated with mutations in the androgen receptor gene located on the X chromosome. Individuals with CAIS, despite having a male karyotype, usually identify as female and exhibit female external genitalia. Management of CAIS patients requires a complex, interdisciplinary approach involving specialists in clinical genetics, gynecology, endocrinology, psychiatry and psychology. The aim of this study is to present the case of a 40-year-old patient diagnosed with an X chromosome mutation, including diagnostic procedures as well as pharmacological and surgical management.

KEYWORDS

disorders of sex development, androgen insensitivity syndrome, primary amenorrhea, CAIS, Morris syndrome

STRESZCZENIE

Całkowita niewrażliwość na androgeny (*complete androgen insensitivity syndrome* – CAIS; znana również jako zespół Morrisa) to rzadka jednostka chorobowa, związana z mutacją genu kodującego receptor androgenowy, znajdujący się na chromosomie płciowym X. Osoby z CAIS mimo męskiego kariotypu zazwyczaj identyfikują się jako kobiety, posiadając żeńskie zewnętrzne narządy płciowe. Opieka nad pacjentką z CAIS jest procesem złożonym, wymagającym

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zaangażowania interdyscyplinarnego zespołu, składającego się ze specjalistów z zakresu genetyki klinicznej, ginekologii, endokrynologii, psychiatrii i psychologii. Celem pracy jest zaprezentowanie przypadku 40-letniej pacjentki z pierwotnym brakiem miesiączki – u której rozpoznano mutację chromosomu X – w tym procedur diagnostycznych, a także leczenia farmakologicznego i chirurgicznego.

SŁOWA KLUCZOWE

zaburzenia rozwoju płci, zespół niewrażliwości na androgeny, pierwotny brak miesiączki, CAIS, zespół Morrisa

INTRODUCTION

Disorders/Differences of sex development (DSD) are rare conditions, with an incidence of approximately 1 in 20,000–99,000 live births. Based on the 2006 consensus of the Lawson Wilkins Pediatric Endocrine Society and the European Society for Pediatric Endocrinology, DSDs can be categorized into DSD associated with sex chromosome anomalies, DSD with a 46,XX karyotype, and DSD with a 46,XY karyotype. In this last group, disorders are further classified into abnormalities of gonadal development and disorders of androgen synthesis or action, including androgen insensitivity syndrome (AIS; Figure 1) [1].

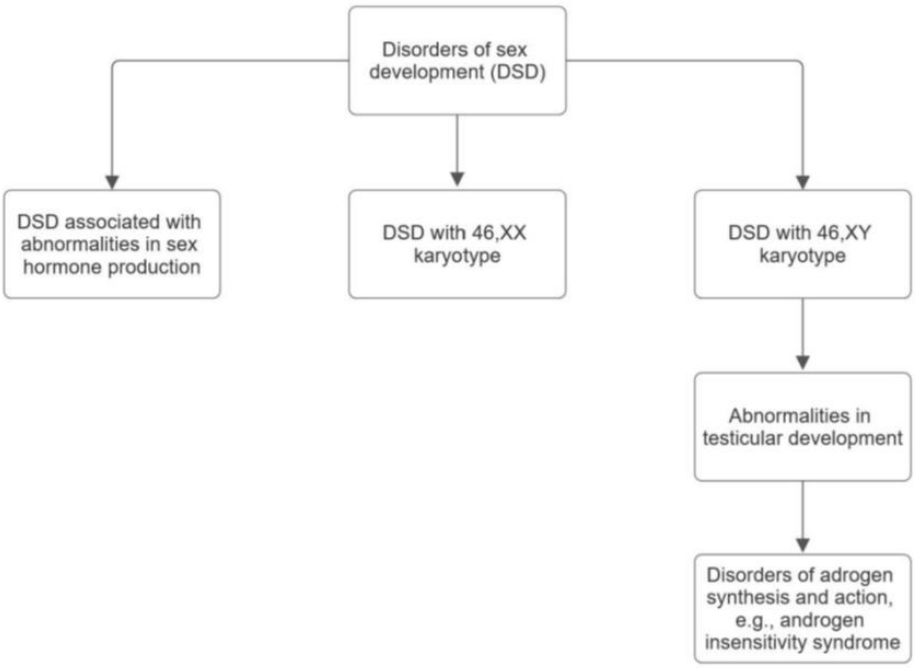

Fig. 1. Classification of disorders of sex development

AIS (also known as Morris syndrome or testicular feminization syndrome) is a congenital disorder caused by mutations in the androgen receptor (AR) gene. AIS is inherited in an X-linked recessive manner, with the *AR* gene located on the proximal long arm of the X chromosome (locus Xq11-12). Its incidence is estimated at 2–5 per 100,000 male births [2]. In complete androgen insensitivity syndrome (CAIS), the body is either completely or partially unresponsive to androgens, impairing the biological action of testosterone and dihydrotestosterone during fetal development. Consequently, this disrupts the formation of Wolffian duct-derived male internal genital structures (the epididymis, vas deferens, seminal vesicles, and prostate). Meanwhile, Sertoli cells of the testes produce anti-Müllerian hormone (AMH), inhibiting the development of Müllerian structures (fallopian tubes, uterus, and upper vagina) [1,2]. Through the aromatization of testosterone into estradiol, female external genitalia, breasts, and a typically female body shape develop [2]. Morris syndrome presents in three forms: complete (CAIS), partial (PAIS), and mild (MAIS; Table I) [2,3].

Table I. Forms of Morris syndrome and associated phenotypes

Property	Type of AIS		
	CAIS	PAIS	MAIS
Androgen sensitivity	Absent	Partial	Reduced
Karyotype	46,XY	46,XY	46,XY
Phenotype	Female: primary amenorrhea, intra-abdominal or inguinal testes, female external genitalia, normal breast development, female body shape, sparse or absent pubic/axillary hair, short blind-ending vagina	Ambiguous: hypospadias, micropenis, undescended testes, gynecomastia, inguinal hernia	Male: micropenis, gynecomastia, high-pitched voice, alopecia

AIS – androgen insensitivity syndrome; CAIS – complete androgen insensitivity syndrome; PAIS – partial androgen insensitivity syndrome; MAIS – mild androgen insensitivity syndrome

CASE REPORT

A 40-year-old patient was admitted to the Department of Endocrinology and Neuroendocrine Tumors at the University Clinical Center Prof. K. Gibiński in Katowice for the evaluation of primary amenorrhea. In 2022, cytogenetic analysis of peripheral blood T lymphocytes revealed a 46,XY karyotype. Fluorescence in situ hybridization (FISH) testing confirmed the presence of the *SRY* gene, indicating a male genotype despite a female phenotype. The analysis was performed due to a significant family history: the patient's sister had previously been diagnosed with CAIS and had undergone testicular resection. On physical examination, the patient weighed 62 kg and measured 168 cm in height. Findings included normal breast development, an absence of axillary and lower leg hair, and sparse pubic hair. Transvaginal ultrasound failed to visualize the uterus and ovaries. Laboratory tests revealed elevated levels of free testosterone, total testosterone, androstenedione, and AMH relative to female reference ranges. Selected laboratory results are shown in Table II. Further imaging with abdominal and pelvic magnetic resonance imaging (MRI) revealed an absence of the uterus and adnexa and a left adrenal nodule containing a small amount of adipose tissue, but no residual gonadal tissue was detected. Consequently, the patient was referred for surgical exploration. During laparoscopy, residual gonadal tissue was located and excised. Histopathological examination confirmed testicular tissue (Figure 2). Hormone replacement therapy was initiated postoperatively. Although the patient accepted the diagnosis, she decided not to change her gender. She reported low mood, prompting a psychiatric consultation.

Table II. Selected preoperative laboratory results

Hormone	Value in patient	Female reference range
Free testosterone (pg/ml)	14.79	0.2–4.10
Total testosterone (ng/ml)	>15.00	0.084–0.481
Androstenedione (ng/ml)	2.14	0.49–1.31
LH (IU/l)	22.10	follicular: 2.4–12.6; ovulatory: 14.0–95.6; luteal: 1.0–11.4; postmenopausal: 7.7–58.5
FSH (IU/l)	8.85	follicular: 3.5–12.5; ovulatory: 4.7–21.5; luteal: 1.7–7.7; postmenopausal: 25.8–134.8
PRL (ng/ml)	5.04	4.79–23.30
AMH (ng/ml)	159.00	0.06–4.44
SHBG (nmol/l)	119.00	32.4–128.00

LH – luteinizing hormone; FSH – follicle-stimulating hormone; PRL – prolactin; AMH – anti-Müllerian hormone; SHBG – sex hormone-binding globulin

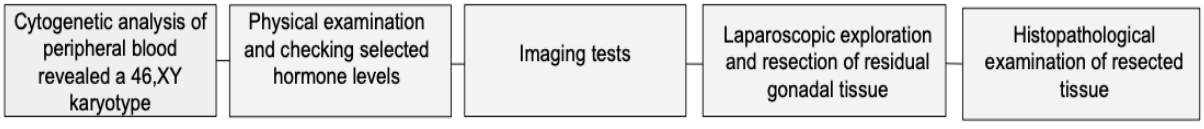

Fig. 2. Diagnostic process for the patient

DISCUSSION

The diagnosis of CAIS entails karyotype and phenotype assessment. Supporting assays include hormone levels, especially luteinizing hormone (LH) and total testosterone, both of which were elevated compared to normal female ranges. Ultrasound and MRI were employed to localize residual gonadal tissue; however, in this case, imaging failed to detect it, necessitating laparoscopic exploration [4]. CAIS is rarely diagnosed in infancy due to a normal female

genital appearance; it is most often discovered during adolescence when evaluating primary amenorrhea. In this case, diagnosis was delayed until age 40 following genetic testing. It is a rare instance when CAIS also affects a patient's sibling. Patients typically exhibit female external genitalia, but lack internal female reproductive organs due to AMH secretion by the testes, inhibiting the development of the uterus, fallopian tubes, and upper vagina. The lower vagina develops from the urogenital sinus, resulting in a short, blind-ending vaginal pouch, often measuring 2.5–8 cm. Inguinal hernias in female infants should raise suspicion for CAIS, since studies indicate that they occur in 57% of cases [1]. Additional features include sparse pubic and axillary hair, tall stature, and normal breast development due to peripheral aromatization of testosterone into estradiol during puberty [5]. After a diagnosis of CAIS, gonadectomy is recommended due to an increased risk of testicular germ cell tumors, with malignancy risk rising with age – from about 2% in adolescence to as high as 33% by age 50. It is advisable to perform gonadectomy after puberty to allow for natural development of secondary sexual characteristics mediated by endogenous hormone production. Post-gonadectomy, estrogen replacement therapy should be administered until approximately age 50 to maintain female secondary sex characteristics and to prevent osteoporosis [6,7,8]. In addition to surgical and hormonal management, comprehensive psychological support is crucial, including gender identity counseling. Most CAIS patients identify as female, attributed to brain insensitivity to androgens. Patients may also elect reconstructive procedures, such as labiaplasty or vaginal dilation therapy, to improve self-esteem and body image [9,10].

CONCLUSIONS

Although CAIS is a rare disorder, clinicians should maintain a high index of suspicion, especially in cases of primary amenorrhea. Early diagnosis enables timely intervention and the provision of holistic care, including psychological support. Interdisciplinary collaboration among endocrinologists, gynecologists, clinical geneticists, and mental health professionals is essential to optimize patient outcomes. Furthermore, educating patients and their families about CAIS can enhance understanding of the disease and improve quality of life.

Authors' contribution

Study design – D. Rost, J. Strzelczyk

Manuscript preparation – D. Rost, J. Gniewek, M. Jekiełek

Literature research – J. Gniewek, D. Rost

Final approval of the version to be published – J. Strzelczyk

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