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CASE REPORT

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A Case of Androgen Insensitivity Syndrome

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BACKGROUND

Androgen insensitivity syndrome is a rare X-linked recessive disorder affecting sexual development. The prevalence is estimated at between 1 in 20,000 and 1 in 60,000 live male births. This condition involves genotypic males that present with phenotypic characteristics of female sexual development; however they fail to menstruate at puberty. This is the third most common cause of primary amenorrhea after gonadal dysgenesis and müllerian agenesis (1).

Laboratory evaluation will reveal a 46, XY genotype, a testosterone level in the low to normal male range, and a relatively elevated LH level. The karyotype and elevated testosterone level are used to differentiate androgen insensitivity syndrome from müllerian agenesis, which can present with a similar phenotype (2).

CASE PRESENTATION

A 19 year-old phenotypic female presented to the gynecology clinic with a chief complaint that she was told that she does not have a uterus or ovaries and that she “may have boy parts inside.” She stated that she was told by another physician that she needed surgery. Further history revealed that the patient had previously been evaluated for primary amenorrhea, and imaging had revealed the absence of a uterus or ovaries. The patient had no significant past medical or surgical history. She had never menstruated, never been sexually active, and never had an abnormal pap smear. She was taking no medications and had no allergies. She smoked about 4 cigarettes a day and drank 2-4 beers a weeks.

Her physical exam was significant for Tanner stage III breasts and Tanner stage II pubic hair. Speculum and bimanual exam revealed a blind-ending vaginal pouch that was about 5 cm long. There was no evidence of a uterus on rectal exam.

Magnetic resonance imaging, CT scan, and ultrasonography indicated the absence of uterus and ovaries. There were bilateral structures at the inguinal canals that were suspected to be intra-abdominal testicles. Laboratory testing revealed a 46 XY karyotype and a serum testosterone in the male range, which confirmed the diagnosis of androgen insensitivity syndrome.

The patient was counseled about the risk of malignant transformation of the intra-abdominal testicles. The patient consented to, and underwent a laparoscopic removal of the intra-abdominal testicles.

Three days prior to surgery the patient attempted suicide by lying down in the middle of the street. She was escorted to the hospital where she was admitted to the inpatient psychiatric floor. On the scheduled day of the procedure, the patient was counseled once again about the procedure. The patient voiced understanding and desired laparoscopic removal of the intra-abdominal testes. The testes were found bilaterally at the internal ring of the inguinal canal and were removed laparoscopically without complication. After a short recovery, the patient was transferred back to the inpatient psychiatric floor. The patient was discharged after 10 days with a diagnosis of Mood Disorder NOS.

Final pathology: Cryptorchid testes with no malignant changes identified

The patient failed to attend the schedule post-operative office appointment.

DISCUSSION

Androgen insensitivity syndrome involves genotypic males that present with phenotypic characteristics of female sexual development. Due to dysfunctional or absent androgen receptors on target organs, there is a lack of male differentiation of the external and internal genitalia. The external genitalia remain feminine, as occurs in the absence of sex steroids. The classic finding is a female that presents with primary amenorrhea. This patient is found to have a blind vaginal pouch and the diagnosis is made by karyotype (1,2,3,4,5).

The testes in these individuals may be located intra-abdominally, along the inguinal canal, or in the labial pouch. The incidence of malignancy, like any cryptorchid testes, is elevated. Incidence of gonadal tumors ranges from 5-20% (2,4). With this in mind, optimal treatment is removal of the testes, which can usually be achieved laparoscopically. Because the androgens produced by the testes are converted to estrogens, which stimulate breast development, it is preferable to leave the testes in situ until puberty is complete (1,2,5). Estrogen replacement should be started thereafter (3).

As exemplified by this patient, this disorder can result in severe psychological trauma and confusion about gender identity. Although these individuals should be counseled that they are infertile, it should also be reinforced that they are completely female in their gender identity. Truthful education and appropriate psychological counseling should be advocated. ² With proper

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therapy and reassurance, these patients can lead a functional and normal life. There is a national androgen insensitivity support group that educates physicians as well as affected patients about special concerns and problems of women with androgen insensitivity syndrome (1).

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