Androgen Insensitivity Syndrome (AIS)

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Androgen Insensitivity Syndrome, or AIS, is a genetic condition, inherited (except for occasional spontaneous mutations), occurring in approximately 1 in 20,000 individuals. In an individual with complete AIS, the body’s cells are unable to respond to androgen, or “male” hormones. (“Male” hormones is an unfortunate term, since these hormones are ordinarily present and active in both males and females.) Some individuals have partial androgen insensitivity.

In an individual with complete AIS and karyotype 46 XY, testes develop during gestation. The fetal testes produce mullerian inhibiting hormone (MIH) and testosterone. As in typical male fetuses, the MIH causes the fetal mullerian ducts to regress, so the fetus lacks uterus, fallopian tubes, and cervix plus upper part of vagina. However, because cells fail to respond to testosterone, the genitals differentiate in the female, rather than the male pattern, and Wolffian structures (epididymis, vas deferens, and seminal vesicles) are absent.

The newborn AIS infant has genitals of normal female appearance, undescended or partially descended testes, and usually a short vagina with no cervix. Occasionally the vagina is nearly absent. AIS individuals are clearly women. At puberty, the testes are stimulated by the pituitary gland, and produce testosterone. Because testosterone is chemically very similar to estrogen, some of the testosterone converts back to estrogen (“aromatizes”) in the bloodstream. This estrogen produces breast growth, though it may be late. Women with AIS do not menstruate, and are not fertile. Because the development of pubic and underarm hair, in women as well as in men, depends upon testosterone, most AIS women have no pubic or underarm hair, but some have sparse hair.

When an AIS girl is diagnosed during infancy, physicians often perform surgery to remove her undescended testes. Although removal of testes is advisable, because of the risk of cancer, ISNA advocates that surgery be offered later, when the girl can choose for herself. Testicular cancer is rare before puberty.

Vaginoplasty surgery is frequently performed on AIS infants or girls to increase the size of the vagina, so that she can engage in penetrative intercourse with a partner with an average size penis. Vaginoplasty surgery is problematic, with many failures. ISNA advocates against vaginal surgery on infants. Such surgery should be offered to, not imposed on, the pubertal girl, and she should have an opportunity to speak with adult AIS women about their sexual experience and about surgery in order to make a fully informed decision. Not all AIS women will choose surgery.

Some women have successfully increased the depth of their vagina with a program of regular pressure dilation, using aids designed for that purpose. Contact the AIS Support Group.