

# Androgen Insensitivity Syndrome

## Courtesy the Intersex Society of North America

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Reviewed July 1<sup>st</sup> 2008

Androgen Insensitivity Syndrome, or A.I.S., is a genetic condition, inherited (except for occasional spontaneous mutations), occurring in approximately 1 in 20,000 individuals. In an individual with complete A.I.S., 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 A.I.S. and karyotype 46 XY, testes develop during gestation. The foetal testes produce mullerian inhibiting hormone (M.I.H.) and testosterone. As in typical male fetuses, the M.I.H. causes the foetal mullerian ducts to regress, so the foetus lacks a uterus, fallopian tubes, and cervix plus upper part of the 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 A.I.S. 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.

A.I.S. individuals are clearly women. At puberty, the oestrogen produced by the testes produces breast growth, though it may be late. She does not menstruate, and is not fertile. Most A.I.S. women have no pubic or underarm hair, but some have sparse hair.

When an A.I.S. 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, I.S.N.A. 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 A.I.S. 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.

I.S.N.A. 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 A.I.S. women about their sexual experience and about surgery in order to make a fully informed decision. Not all A.I.S. 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.

Physicians and parents have been most reluctant to be honest with A.I.S. girls and women about their condition, and this secrecy and stigma has unnecessarily increased the emotional burden of being different.

Because A.I.S. is a genetic defect located on the X chromosome, it runs in families. Except for spontaneous mutations, the mother of an A.I.S. individual is a carrier, and her XY children have a one in two chance of having A.I.S. Her XX children have a one in two chance of carrying the A.I.S. gene.

Most A.I.S. women should be able to locate other A.I.S. women among siblings or maternal relatives.

### **Is there a test for androgen insensitivity syndrome?**

The answer depends upon exactly what you are looking for – diagnostic information, or carrier status. If you were born with female genitals and testes, and have very sparse or absent pubic hair, you most likely have complete A.I.S.. If you were born with ambiguous genitals and testes, there are a number of possible etiologies, including partial A.I.S.

Testing for partial A.I.S. is more problematic than the complete form. Hormonal tests in a newborn with 46 XY karyotype and ambiguous genitals will show normal to elevated testosterone and L.H., and a normal ratio of testosterone to D.H.T. A family history of ambiguous genitals in maternal relatives suggests partial androgen insensitivity.

If you are wondering if you are a carrier, or if you know that you are a carrier and are wondering about the status of your foetus, genetic testing is possible.

A.I.S. has been diagnosed as early as 9–12 weeks gestation by chorionic villus sampling (sampling tissue from the foetal side of the placenta). By the 16th week it can be detected by ultrasound and amniocentesis. However, prenatal diagnosis is not indicated unless there is a family history of A.I.S.

### **What is partial androgen insensitivity syndrome?**

The extent of androgen insensitivity in 46XY individuals is quite variable, even in a single family. Partial androgen insensitivity typically results in "ambiguous genitalia." The clitoris is large or, alternatively, the penis is small and hypospadiac (these are two ways of labeling the same anatomical structure).

Partial androgen insensitivity may be quite common, and has been suggested as the cause of infertility in many men whose genitals are of typically male appearance.

Individuals with ambiguous genitals have typically been subjected to "corrective" surgery during infancy. Based on our own painful experiences, I.S.N.A. believes that such cosmetic surgery of the genitals is harmful and unethical.

Surgery is justified only when it is necessary for the health and well-being of the child.

Surgery which is intended to make the genitals appear more male or more female should be offered, but not imposed, only when the child is old enough to make an informed decision for her/himself.

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