

It involves minimal risk and expense, and results in a vagina that closely resembles a normal one. In some cases, the Vecchietti procedure, basically a semi-surgical way of accelerating dilation, has advantages. There are a number of plastic surgical methods of lengthening the vagina using skin grafts, sections of intestine, etc. These all have many disadvantages, and should be used only when less invasive treatments have been ruled out. Vaginoplasty in early childhood usually has poor results and should not be done.

### Facing the Diagnosis

Some clinicians and parents, in a misguided attempt to spare the patient an inner conflict, withhold the genetic and gonadal information from her, but most professional caregivers now recommend truth disclosure with psychological support/counselling. Otherwise, many patients will seek diagnostic information via medical libraries or the Internet, bearing the burden alone and in silence. Many will wrestle with perplexing half-truths, or reach false conclusions (e.g. that gonadectomy means they have cancer).

If the parents' emotional needs/anxieties are addressed first (via psychological support/counselling from professionally trained staff) it will be easier for them to provide effective support to their child. Everyone will feel better if there are no taboos about the subject. Talking, like grieving, is therapeutic, enabling feelings to be confronted and resolved. Pushing the matter under the carpet is just storing up psychological trouble for later. It is important that parents encourage discussion with their child, and actively seek out information on their behalf. Unfortunately, keeping the condition a secret can become more important to some parents than acknowledging their child's need for emotional support and appropriate clinical intervention. It wastes mental/emotional energy that is better spent in helping the child come to terms with the truth.

The rights of the patient to assign meaning and validity to her condition via a diagnosis, and to seek out a support group, must be considered. Meeting others who are affected is vital and is probably the single most useful therapeutic measure. Doctors may have over-emphasized the extent to which the knowledge of their genetic/gonadal status causes lasting distress for AIS women. CAIS adults tell us that, in the long-term, their XY chromosomes and testes would have been of no material relevance to them were it not for the isolation, sense of freakishness and stigma which results from an apparent unacceptability of their biological status in the eyes of society - since they have a normal feminine gender identity.

In CAIS, the person will look like a girl and problems of psychosexual identity as a biologically-determined feature of the condition are unlikely. In CAIS, leanings towards heterosexuality, lesbianism or bisexuality seem no different from females in general.

Over-emphasis on a CAIS patient's femaleness with an unwillingness to allow exploration of her very real female deficiencies (lack of internal female organs, pubic hair, menstruation, with possibly a diminished vaginal length) will suggest to her a very considerable anxiety and discomfort on the part of doctors/parents. Preparing the youngster for intimate personal relationships as an adult should be a priority, tempting as it may be to divert their attention toward substitute goals.

### Aims of the Group

- To reduce the secrecy, stigma, and taboo that has surrounded AIS and other intersex states, by encouraging doctors, parents and society to be more open.
- To encourage the provision of psychological support within the medical system, for young people with AIS and their parents.
- To put parents and people with AIS in touch with each other and to encourage them to seek support and information.
- To increase the availability of information on AIS both verbal (from the health professionals) and written (from the support group and other sources).
- To encourage improvements in treatment for vaginal hypoplasia, and research into why the extent of vaginal development can vary in AIS.
- To encourage retrospective studies on genital surgery, so as to evaluate whether it is an effective treatment for the patient.

### Membership/Meetings/Publications

In contact with around 300 affected families/individuals in USA/Canada. Group meetings in US from 1996 and in Canada from 1998. Publications developed in the UK from 1993. Newsletter called ALIAS, 3 per year. For details, please send large stamped S.A.E. to your national representative. Please refer to web site for contact details of all the overseas AIS groups.

## AIS Support Group

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A self-help group providing information and support to parents of AIS youngsters and to AIS adults. Started in the UK in 1988 by the mother of an AIS infant, and formalized in 1993.

### Introduction

Up to 8 weeks every fetus, whether of male or female genetic sex, has the capacity to develop either a male or female reproductive system, and in a genetically male (XY) fetus the active intervention of male hormones (androgens) is needed to produce a fully male reproductive system. A female body type with female external genitalia is the basic underlying human form.

### What is AIS?

Androgen insensitivity (syndrome) causes an interruption of fetal development of the reproductive system. In AIS the child is conceived with male (XY) sex chromosomes. Embryonic testes develop inside the body and start to produce androgens.

But these male hormones cannot complete the male genital development due to a rare insensitivity of the fetal body tissues to androgens. So the external genital development continues along female lines (the "backup" route) but the development of female internal organs has already been suppressed by a hormone (MIF or mullerian inhibiting factor) from the fetal testes.

Tissue androgen sensitivity is controlled by a gene on the X chromosome, and AIS is an "X-linked recessive" condition, inherited along the maternal line (or, in an estimated third of cases, results from a spontaneous mutation). A mother who carries the defective gene has a 1:2 chance of any XY child having AIS and a 1:2 chance of any XX child being a carrier like herself. XX individuals can be tested to see if they are carriers. AIS is an example of a biological intersex condition, in which the reproductive organs/genitalia are partly at variance with the person's genetic sex. This is not the same as transsexuality (gender dysphoria).

## Forms of AIS

There are two forms; a complete form (CAIS) where the tissues are completely insensitive to androgens and a partial or incomplete form (PAIS) where the tissues are in/sensitive to varying extents, forming a spectrum of genital appearances. At the CAIS end of the spectrum<sup>1</sup> the external genitalia are completely female (AIS Grades 6/7)<sup>2</sup> and the sex of rearing is invariably female. In PAIS the outward genital appearance can lie anywhere along a continuum from completely female (Grade 6),<sup>3</sup> through mixed male/female, to completely male (Grade 1) and can vary somewhat between affected siblings. Some PAIS babies may be raised as males. Slight androgen insensitivity may contribute to infertility in some otherwise normal men. CAIS has been sometimes referred to as "classical" testicular feminization, PAIS as incomplete testicular feminization. The complete and partial forms may be caused by different defects at the genetic/cellular level and do not occur in the same extended family.

## Synonyms

Androgen Insensitivity Syndrome, Androgen Resistance Syndrome, Testicular Feminization Syndrome, Male Pseudo-hermaphroditism, Goldberg-Maxwell Syndrome (CAIS), Morris,s Syndrome (CAIS), Lubs Syndrome (PAIS),

Reifenstein Syndrome (PAIS), Gilbert-Dreyfus Syndrome (PAIS), Rosewater Syndrome (PAIS).

Other XY conditions with some AIS-like features: 5-alpha-reductase deficiency, 17 keto-steroid reductase deficiency, XY gonadal dysgenesis (Swyer Syndrome), Anorchidism, Denys-Drash Syndrome, Smith-Lemli-Opitz Syndrome.

XX conditions with some AIS-like features: Mayer Rokitansky Kuster Hauser (MRKH) Syndrome, Mullerian dysgenesis.

## Incidence

The most accurate figure currently available for CAIS comes from an analysis (1992), of a nationwide Danish patient register, suggesting an incidence of 1 in 20,400 XY births (hospitalized cases only, so true incidence probably higher). PAIS may be only about 1/10 as common as CAIS.

## Effects (CAIS)

Even in the complete form (CAIS) there will be no ovaries, Fallopian tubes or uterus,<sup>4</sup> and the vagina will be blind-ending and possibly short or absent. The undescended testes can result in an inguinal (groin) hernia in infancy and this is when the condition may come to light in an apparently female child (~50% of cases). Otherwise CAIS may not be discovered until puberty as a result of failure to menstruate.

Female pubertal development occurs, because the testes produce some estrogen, but there will be no menstruation and no possibility of conceiving or bearing children. Some AIS girls may develop some dark, coarse pubic/underarm hair (AIS Grade 6) but this does not develop in true CAIS (Grade 7) because androgen action is needed for its growth. The nipples usually remain under-developed and pale in color. The vagina may need to be lengthened before sexual intercourse is possible.

The older literature sometimes states that girls with AIS are often tall, that the body form is "voluptuously female", i.e. with very adequate breast development, and that the skin maintains a good condition, not being prone to acne (which is linked to the action of male hormones). Many AIS women have been photographers' models.

There is a risk of cancerous changes occurring in the gonads (testes) after age 20, and removal before this time is recommended. Usually this is deferred until the late teens to allow a spontaneous feminizing puberty to occur, which may

have physical and psychological advantages over a puberty induced by exogenous hormones.

Although the risk of pre-adult cancer is too small to justify it before adulthood, gonadectomy is sometimes done in infancy or childhood, usually with the intention of avoiding a psychological crisis when the need for an operation later on must be explained. Arguably, this violates the patient's right to informed consent and to optimal treatment.

## HRT/Osteoporosis

When the testes are removed after puberty, immediate long-term female hormone replacement therapy (HRT) is needed to prevent menopausal symptoms and osteoporosis (bone thinning). In the case of gonadectomy in infancy/childhood, HRT is often started at 10 or 11, in order to initiate puberty.

Low bone density appears to be more common in AIS women than in XX women. The cause is not clear. It may be the direct result of lack of androgen effects. Lack of HRT is a risk factor, although some AIS adults have a low bone density in spite of regular HRT. Possibly, this is due to the fact that XY girls with testes have lower estrogen levels than XX girls with ovaries during the time when healthy bone should be laid down. XX girls start producing estrogen at around the age of 8 (i.e. a year of two before breast development starts) so supplementary low dose estrogen from this age, with or without gonads in place, may be advisable in AIS.<sup>5</sup>

Women with AIS should be aware of their increased risk of osteoporosis, especially if they have not used HRT continuously after gonadectomy.

## Vaginal Hypoplasia

Generally the top one-third of the vagina is missing in AIS but in some cases the vagina may be no more than a centimeter or two in length, or even just a dimple. Clinicians must not overlook vaginal hypoplasia in pubertal AIS patients, because some youngsters discover this by self-examination and can live in fear and isolation with this secret for many years.

Vaginal hypoplasia (in both CAIS and PAIS) can be treated by the non-surgical method of pressure dilation, performed by the girl herself at home. This is best deferred until she has gone through puberty and is sufficiently motivated.

<sup>1</sup>For grading details see issue No. 6 of ALIAS.

<sup>2</sup>Grade 7 = female without pubic hair (true CAIS).

<sup>3</sup>Grade 6 = female with pubic hair (severe PAIS).

<sup>4</sup>Although fragments of tubes/uterus have been reported in as many as 30% of cases.

<sup>5</sup>Advised by Dr. Richard Stanhope, Consultant Pediatric Endocrinologist, Gt. Ormond St. Children's Hospital, London.