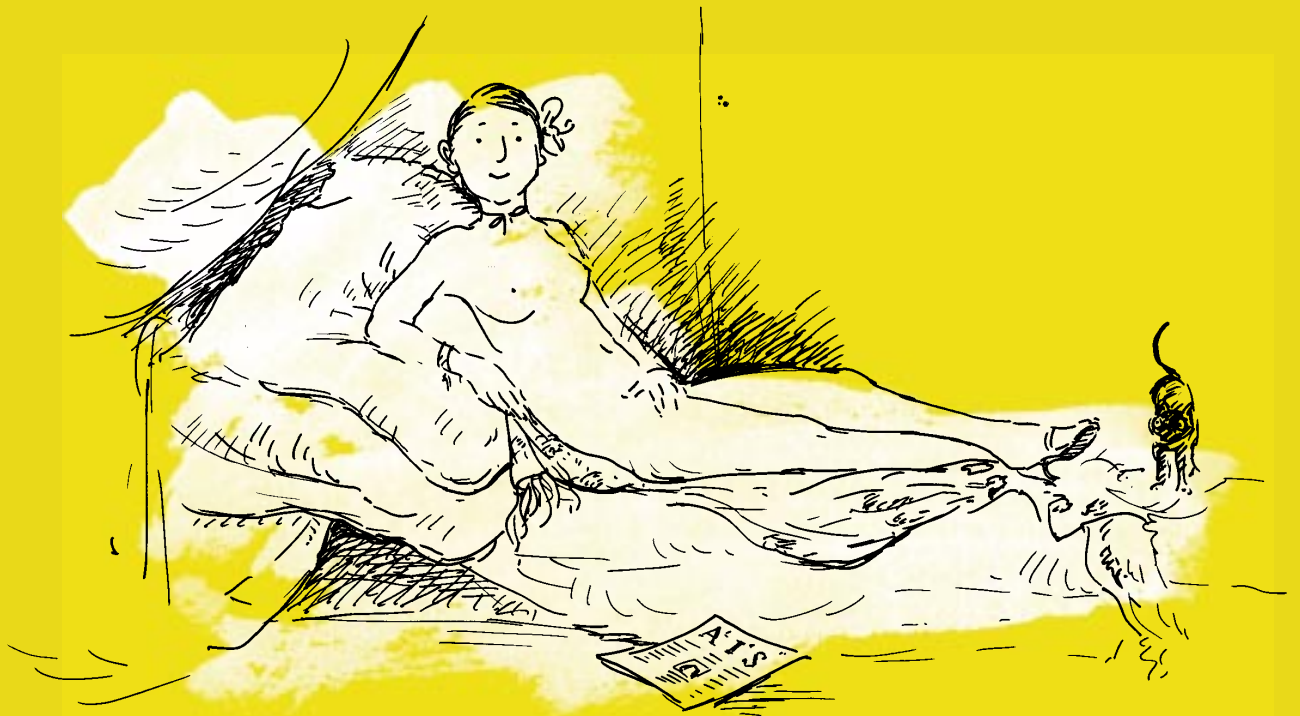


COMPLETE
ANDROGEN
INSENSITIVITY
SYNDROME



Garry L. Warne

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COMPLETE ANDROGEN
INSENSITIVITY
SYNDROME

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Parkville, Victoria, Australia



A U T H O R ' S N O T E

This book is the fruit of discussion with a number of people: the Editor of the AIS Support Group (UK) newsletter (who wishes to remain anonymous), and the members of the AIS Study Group: Dr Jennifer Batch, Department of Paediatrics, University of Melbourne; Dr Sonia Grover, Gynaecologist, Centre for Adolescent Health, Royal Children's Hospital; Professor John Hutson, Director of General Surgery, Royal Children's Hospital; Elizabeth Loughlin, Endocrine Social Worker, Royal Children's Hospital; Dr Sheila Park, Liaison Psychiatrist, Mental Health Service, Royal Children's Hospital; David Pereira, Child Psychotherapist, Mental Health Service, Royal Children's Hospital; Dr Glynis Price, Department of Endocrinology and Diabetes, Royal Children's Hospital; Dr Susan Weigall, Department of Psychiatry, Royal Melbourne Hospital; Associate Professor Jeffrey Zajac, Department of Medicine, Royal Melbourne Hospital.

Garry L. Warne, 1997

F O R E W O R D

In the past, parents found it difficult to explain AIS (androgen insensitivity syndrome) to their daughters because they did not understand it properly themselves and even the doctors appeared to be confused about some aspects of the condition. It was therefore difficult for them to know where to start an explanation, and they also found choosing the right words very hard. So girls and women with AIS had to do without either accurate medical information or proper counselling. We know that many of them experienced intense feelings of fear and embarrassment. Somehow, they felt different from their friends and other women, and for many, it seemed best to keep their emotional turmoil hidden. And when you think that no one is giving you any support, you have low self-esteem regarding your sexuality. For some of us, this has coloured our lives.

For us, it has been a great relief to have gained an understanding of AIS, and many feelings of being different have disappeared. It helps just to know that our unusual condition

has a name, that it is well known, and that there are other people all around the world who know what it is to suffer from the effects of this condition. The truth seems easier to accept than all the half-truths and unanswered questions. And after all, it is our body!

This book, written by Associate Professor Garry Warne, contains the sort of clear, factual information that we wish had been available earlier in our lives. Our suffering has also been greatly alleviated by the explanations and counselling we have received at the Royal Children's Hospital.

An AIS Support Group has been established and we welcome all who wish to share their concerns or simply meet others who are getting on with their lives. To find out how to contact your nearest AIS Support Group, turn to page 24 of this book.

Kylie and Jocelyn

I N T R O D U C T I O N

Some medical conditions are easy to talk about and some are more challenging. A condition like Androgen Insensitivity Syndrome (AIS), which affects the development of the genital and reproductive system, raises some very uncomfortable issues about gender identity that are difficult to put into words. How is it that a woman can be born without a womb and with testes? For generations, women with AIS, and their parents, have struggled to understand this apparent contradiction. Many would have had no idea that there were others with the same condition, unless the condition was 'in the family'. Even then, discussion about a subject relating to genital development and sexual feelings may have been taboo.

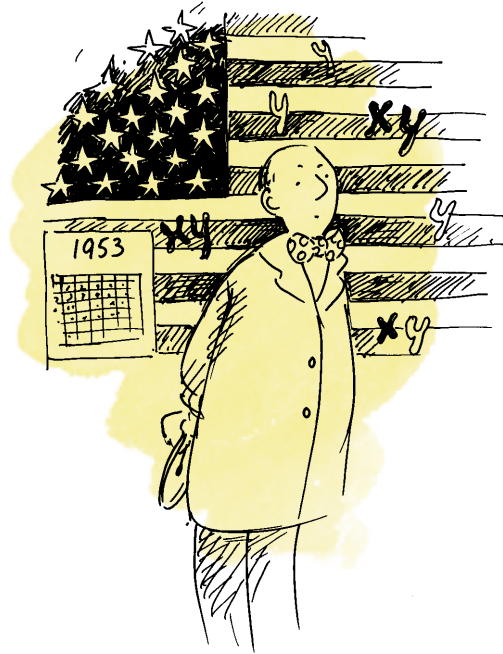
We are now in an age when sexual matters are openly discussed in the media. This freedom is presenting opportunities for the community at large to be given some information about the existence and nature of AIS. The community will become more

understanding and accepting of unusual medical conditions when they are better informed about them and the problems they cause. The same applies to women with AIS. It is a difficult condition to accept, but women will be helped if they have access to both good information about it and adequate opportunities to discuss the complex feelings that are bound to arise as this information is being absorbed.

This book has been prepared to provide women with AIS, and their families, with accurate medical information about this condition and guidance on how to find help.

HOW AIS WAS FIRST RECOGNISED

The first medical report on AIS was published in 1953 by J. M. Morris, an American gynaecologist. His patient was a woman who had never menstruated. She had well-developed breasts and the external genitals of a normal woman, but she had only very sparse pubic hair and a swelling in each groin. An operation was performed, and in each of the swellings was a testis. The woman had no uterus and no ovaries. It is now known that women with AIS have the XY chromosome pattern usually associated with the male gender, and the Y chromosome causes testes to develop. In addition, they have a relatively short vagina. Because there is no uterus, there is also no cervix, and so the vagina ends as a pouch with no internal connections.



T H E N A T U R E O F A I S

AIS is just one of a number of medical conditions that affect the development of the reproductive and genital organs. It is caused by an alteration in a gene (a physical particle containing genetic information that is found in every cell in the body). This alteration in the gene blocks the body's response to masculinising hormones (androgen) during foetal development and after birth. In other words, it makes the body *insensitive to androgen*, and masculine development that would occur in the presence of a normal gene is impossible. The whole *syndrome* (the combination of physical changes that are characteristic of AIS) results from this alteration in a single gene. The body can respond to feminising hormone (oestrogen) but not androgen.

There are two forms of AIS: *complete* AIS (CAIS) and *partial* AIS (PAIS). CAIS is a condition in which the external genitals are completely female in appearance, but the internal female reproductive organs are missing. PAIS is a variant of AIS in which affected children are born with masculinised genitalia. The extent of this is variable: some babies have a slightly enlarged clitoris and a

smaller than usual cleft between the labia, while others may have an almost fully formed penis and scrotum. Some children with PAIS are raised as girls, others as boys. The presence of testes and the absence of a uterus is characteristic of both CAIS and PAIS.

The problems associated with CAIS are in many ways different from those of PAIS. In CAIS, the sex at birth is clearly female, whereas the sex of a newborn infant with PAIS may be unclear and a decision must be made about whether to raise the child as a girl or a boy. Therefore, to avoid confusion, this book will only consider CAIS, and PAIS will be discussed in a separate publication.

There are many other disorders in which the development of the external and internal genital and reproductive organs is incomplete or in which both male and female organs are found together. One in every 4,500 newborn babies has external genitals that differ significantly from the standard male and female appearance. Most people would assume that such conditions are terribly rare because they do not read about them in the newspapers, but they are not.

In common parlance, much is made of the

The nature of AIS

difference between the sexes. How different are they really? According to Eastern philosophy, the female and male attributes *Yin* and *Yang* are considered to coexist in perfect balance and harmony. Most people have some qualities that may be regarded as feminine and some that are masculine. In bodily structure and function, there are more similarities than differences between females and males. For example, all women produce masculinising hormone (androgen) and all men produce feminising hormone (oestrogen).

In the development of the human foetus, there is a stage when the internal and external genitalia of females and males appear identical. This means that all differences between the sexes develop from a common starting point, a completely neutral stage. From there, development may proceed either along female or male lines. The genetic and hormonal control of female and male sexual development in the foetus is well understood, and much more is known about it than about how most other aspects of development are controlled. This detailed knowledge provides a solid framework on which to build a clear understanding of AIS.

Hormones

Hormones are the chemical messengers in the body. They are produced in one place by a gland, and act somewhere else in the body. The hormone insulin, for example, is made in the pancreas and acts in the liver and the muscles to regulate the level of glucose in the bloodstream. Growth hormone is made in the pituitary gland near the brain and acts to promote bone growth in children.

Androgen

Androgen is a masculinising hormone, that in males is responsible for the growth of the penis, development of facial and body hair, growth of the muscles and skeleton, and deepening of the voice. Both males and females have androgen. In males, and in females with AIS, it comes from two sources: the testes and the adrenal glands (a pair of organs that lie above the kidneys, on the back wall of the abdomen). Nearly all of the androgen in females with ovaries comes from the adrenal glands with only a small contribution from the ovaries.

The nature of AIS

Androgen and masculine sexual development

When babies begin their development in the womb, it is impossible to distinguish male genitalia from female. In both sexes there is a midline cleft, with a button-like swelling at the top representing the future penis or clitoris. These 'neutral' genitalia develop into either the male or the female genitalia, as shown in the following figures.

In Figure 1, the 'neutral' appearance is shown on the left. The right-hand side of the diagram shows what happens in boys at 12–15 weeks after conception, as their testes start producing androgen. The button grows into a penis, the cleft closes over, starting from the bottom. This causes the urinary opening to move progressively towards the tip of the penis, and the scrotum to form.

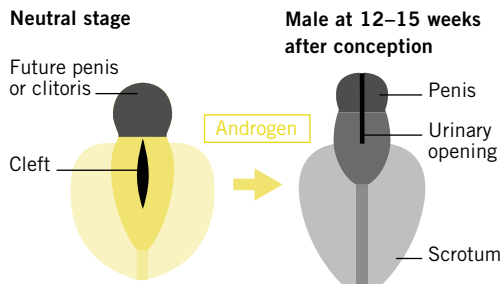


Figure 1 Androgen causes the 'neutral' genitalia to become male.

Figure 2 shows how the 'neutral' genitalia develop into normal female genitalia. Because androgen is normally not produced in the female foetus, the clitoris remains small and the cleft remains open.

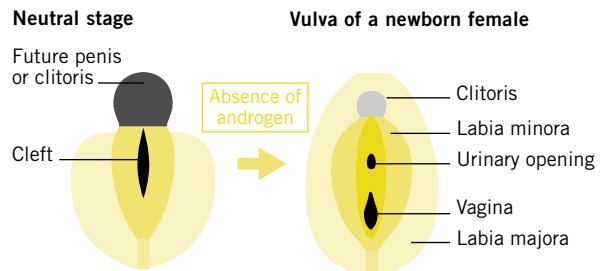


Figure 2 In the absence of androgen, the genitalia change from neutral into female.

In CAIS the testes produce androgen but neither the skin covering the genital structures nor the other genital tissues are able to respond in the normal way, which is to develop into a boy. It is as though the androgen was not produced at all. The external genitalia develop as in normal females because the natural direction of development in the foetus is to become female. Masculine changes only occur in response to androgen.

The nature of AIS

The internal reproductive organs

At the beginning of foetal development, males and females have identical internal reproductive organs. In both sexes there is a pair of tubes on the back wall of the abdomen that are capable of developing into the female uterus, fallopian tubes and the upper part of the vagina. These organs do not develop in males because the foetal testes produce a hormone (MIS) that blocks this process. The same thing

happens in females with CAIS: their testes produce MIS which prevents development of the uterus, fallopian tubes and the upper part of the vagina.

The length of the vagina is variable between women with CAIS, for reasons that are still unclear. Even sisters who appear to have the same external features of CAIS may have quite different degrees of vaginal development.

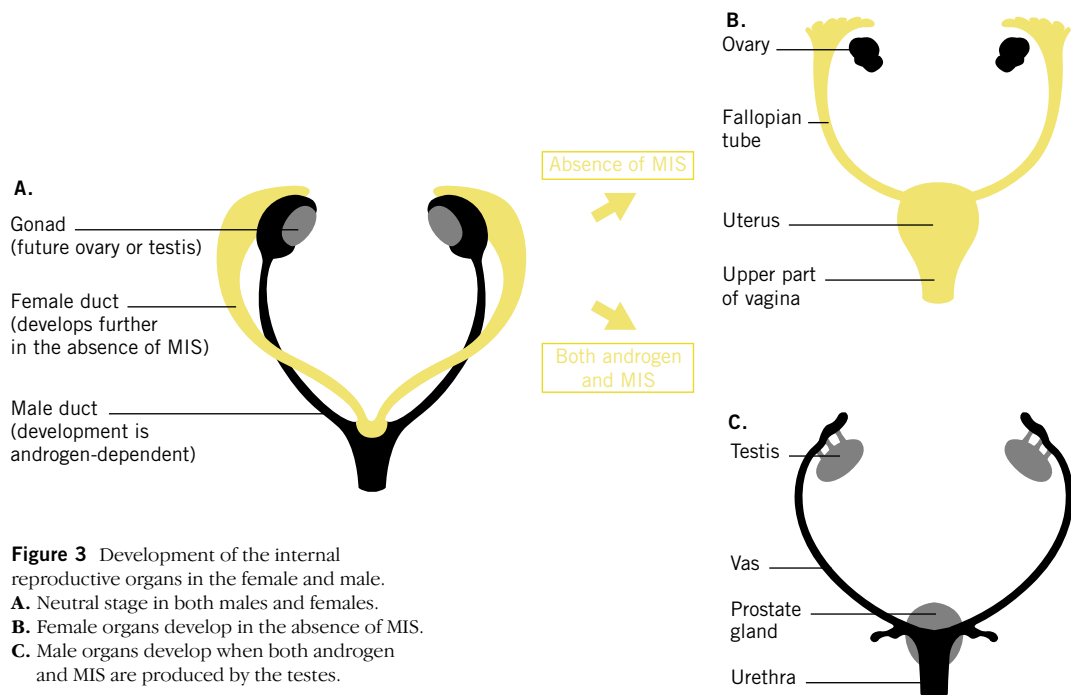


Figure 3 Development of the internal reproductive organs in the female and male.
A. Neutral stage in both males and females.
B. Female organs develop in the absence of MIS.
C. Male organs develop when both androgen and MIS are produced by the testes.

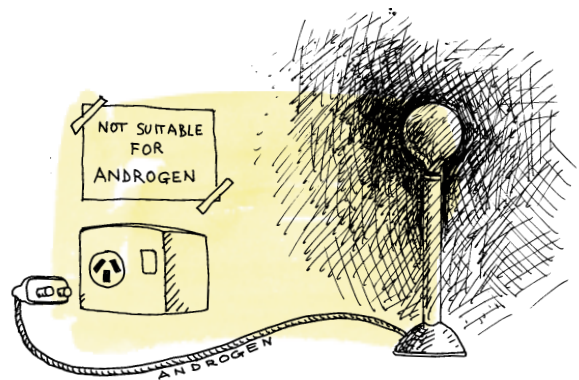
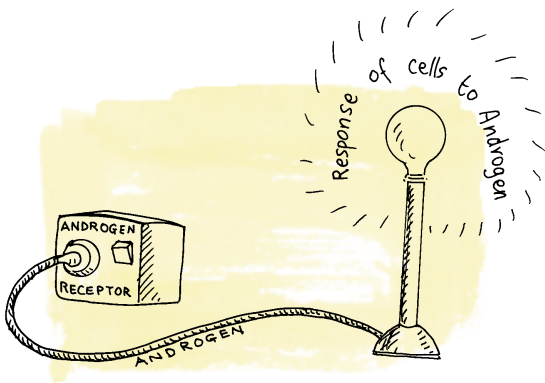
The nature of AIS

Androgen receptor abnormality

In a foetus with CAIS, androgen does not have the usual masculinising effect on genital development. The reason is that its body cells lack a special anchoring substance called *androgen receptor*, and without androgen receptor, the cells cannot respond to androgen, no matter how much is present. It is a bit like trying to plug an electrical appliance into the wrong sort of power point. Not all hormone receptors are abnormal in CAIS, only androgen receptor. The oestrogen receptor is

completely normal and therefore women with CAIS develop breasts and a feminine body shape.

Some females with CAIS have no androgen receptor at all, while in others it is abnormal and does not function as a receptor. A foetus with a small amount of *active* receptor (less than normal) will begin to develop as a boy but the process will not be completed, therefore the baby will have the features of PAIS.



THE GENETIC BASIS OF AIS

Chromosomes and genes

Every new human being results from the fertilisation of an egg by a sperm. The characteristics of the father are contained in each individual sperm, and those of the mother in each of her eggs. The sperm and egg cells contain about 100,000 tiny particles called genes. Each gene represents some characteristic of the parent that the child inherits.

When an egg has been fertilised by a sperm, the father's genes are added to the mother's, and the fertilised egg (the new human being) thus has two sets of genes.

There is, for example, a gene from the mother and a gene from the father for the colour of the eyes. In fact, there is a pair of genes for every separate characteristic. The genes from the mother and father are nearly always paired, and these pairs are reproduced in every cell of the new person.

Genes are linked together on structures called chromosomes. Every cell in the body contains exactly forty-six chromosomes. Two of these are sex chromosomes, which may be either X or Y. Females normally have two X chromosomes, while males normally have one X and one Y chromosome. The Y chromosome contains the gene that causes testes to develop.

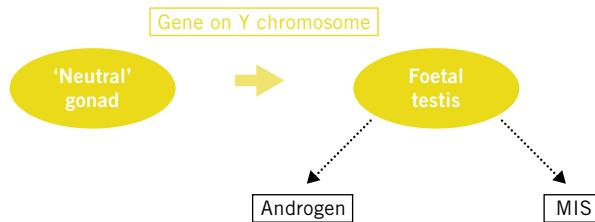


Figure 4 Triggered by a gene on the Y chromosome, the 'neutral' gonad becomes a testis. It later produces two hormones, androgen and MIS.

The genetic basis of AIS

The androgen receptor gene

Females with CAIS have an X and a Y chromosome. The Y chromosome is completely normal, therefore testes develop. The X chromosome is also normal, except that one of its thousands of genes has altered. This alteration is in the androgen receptor gene. A female will have two androgen receptor genes, one inherited from her mother and one from her father. If one of these is the altered androgen receptor gene that causes CAIS, the

normal gene on the other X chromosome will be sufficient to cause her to develop as a normal female. If, however, this woman has a child who is XY and who inherits the X chromosome bearing the altered gene, the abnormal gene will not be balanced by a normal gene and the child will be a female with CAIS. It should also be noted that in one-third of girls with CAIS, it is caused by a spontaneous genetic change in the newly formed foetus at the time of conception, rather than being inherited.

The genetic basis of AIS

The inheritance of AIS

The following diagram shows the different chromosome combinations that can result when a mother who carries the CAIS gene on one of her X chromosomes has children.

A couple who have a daughter with CAIS can use this genetic diagram to predict the odds of having other affected children. The

way the parents' genes were distributed at the time of a particular conception is completely independent of what will happen during the conception of a future child, just as each throw of dice is independent of every other throw. Therefore the risk of having another child with CAIS is 1 in 4 for *every* pregnancy, assuming the mother to be a carrier.

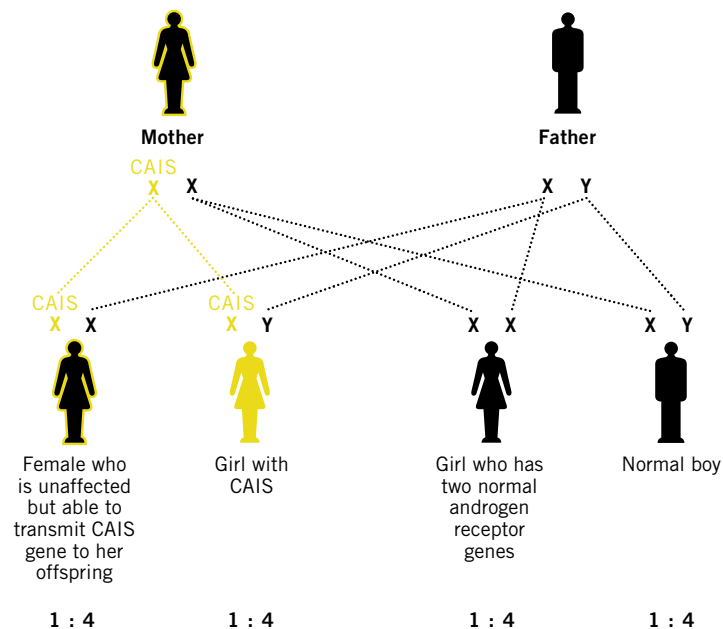


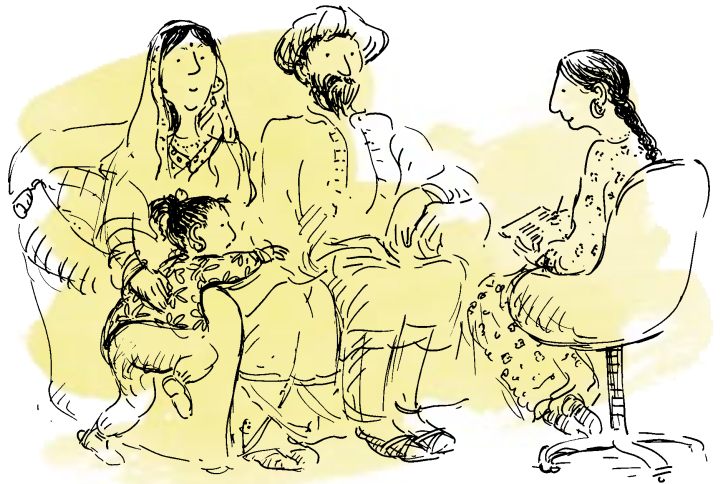
Figure 5 Genetic diagram showing how a mother who carries the CAIS gene on one of her X chromosomes can pass it on to her children. The risk of having a child with CAIS is 1 in 4 for every pregnancy.

THE DIAGNOSIS OF CAIS

The physical features that initially suggest the possibility of CAIS differ from person to person, and these differences influence the age at which the condition is usually diagnosed. The most common feature is a bulge (a hernia) in the groin area in an otherwise normal baby or little girl. When it is operated on, the hernia is found to contain a testis. If hernias go unnoticed or are not present, it may not be known that there is anything wrong until she is a teenager, when she has not begun to menstruate and neither pubic nor underarm hair has appeared, although her breast development will be normal.

To confirm the diagnosis, a blood test is done to identify the XY chromosome pattern. The ability of the testes to produce androgen can be measured in blood samples collected before, and 72 hours after, an injection of

a hormone called human chorionic gonadotrophin (hCG), which stimulates the testes. This proves that testes, rather than ovaries, are present. The hCG test is also useful for distinguishing CAIS from other medical conditions in which an XY female has testes, but they cannot produce androgen. An ultrasound examination will establish that there is no uterus.



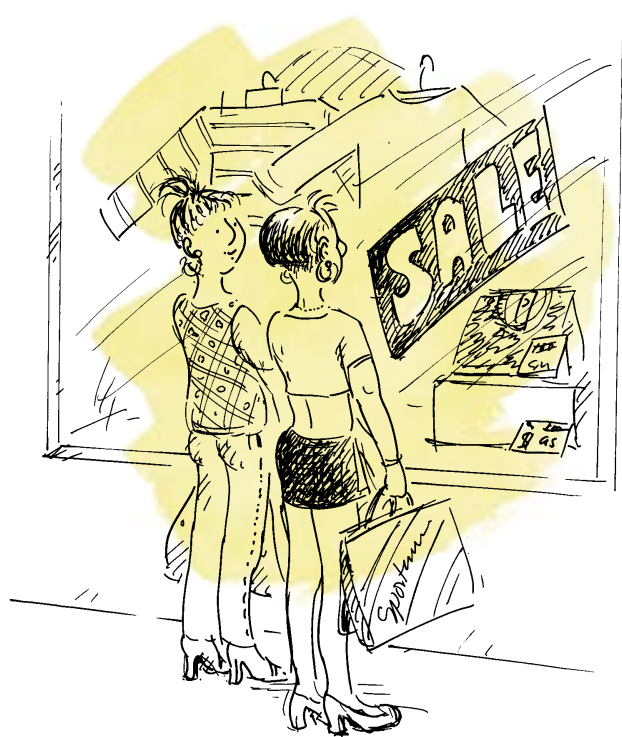
H O R M O N A L C H A N G E S D U R I N G C H I L D H O O D A N D A D O L E S C E N C E

During childhood, the testes are dormant. When the girl is about 11–12, they are stimulated to develop by hormones made in the pituitary gland, a part of the brain. The testes gradually enlarge and the level of androgen they produce progressively rises to adult male levels. At the same time, however, the body converts some of the androgen to oestrogen, and this causes breast development and widening of the hips, so the body shape becomes that of a woman.

The conversion of androgen to oestrogen is a normal process in the body, and is not confined to girls with CAIS. Many healthy

adolescent boys experience transient breast enlargement for the same reason. Boys, however, are fully responsive to androgen and this seems to inhibit further breast development.

Unlike many teenagers, girls with CAIS have very good skin with no acne (pimples). Acne occurs when oil-producing glands in the skin are stimulated by androgen, but this is impossible when the girl has AIS.



SURGERY FOR GIRLS WITH AIS

Removal of the testes

Testes that remain in the abdominal cavity, particularly those that are being over-stimulated by the pituitary gland, are prone to develop cancer. This develops in approximately 9% of women with AIS, but hardly ever before puberty. However, it is the opinion of most authorities that this risk of cancer after puberty is too high, and that removal of the testes before the age of 20 is advisable.

The timing of this operation is a matter for individual choice: some families decide that it should take place when the girl is small, while others (particularly in the US) are advised that the testes can be left in place until after the girl has gone through puberty. It is likely that removal after puberty is the better option in terms of the girl's self-esteem because, due to the conversion of androgen to oestrogen in the body, she will develop breasts and a female body shape without the need for hormonal treatment. The surgery would take place after the girl had been fully informed about her medical condition, and after she had been given the opportunity to discuss the feelings that arise under these circumstances. The

alternative approach – removal of the testes in early childhood – is chosen partly to eliminate the risk of cancer (which many parents worry about) and because parents and doctors may consider that the girl will suffer less distress if she does not have to be involved in the decision about the removal of her testes.

Early removal of the testes is essential in babies with partial AIS who are being raised as girls because failure to do so would result in progressive masculine development. In these girls, surgery to reduce the size of the clitoris and to separate the fused labia is also offered.

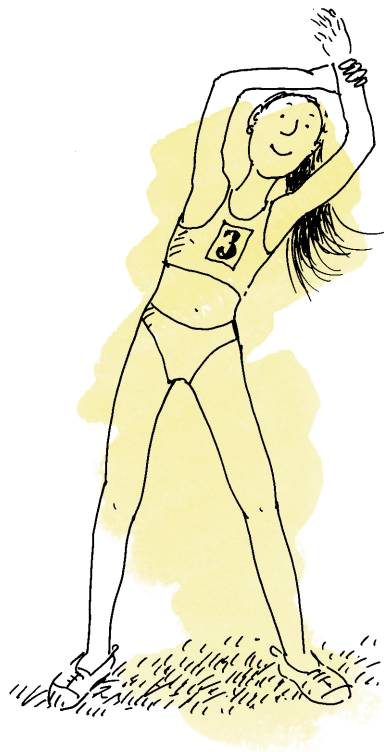
The vagina

Usually the vagina of a woman with AIS is about 6 cm, which is two-thirds the normal length, but it can be much shorter than this. Girls with AIS therefore need to have their vagina examined to assess its length. While surgery may sometimes be necessary to lengthen it, the vagina also grows with regular sexual intercourse. Before a girl becomes sexually active, she can use dilators to gradually bring about some extension.

LACK OF PUBIC AND UNDERARM HAIR

The hair that XX women have in the genital and underarm areas is there because of the action of androgen produced in the adrenal glands and ovaries. Women with CAIS usually have either no hair at all in these areas, or very sparse hair. This lack of body hair is a physical characteristic that cannot be altered by medical treatment, but women may be able to develop positive ways of thinking about it. The amount of pubic hair that women generally have is very variable, and some women are relatively hairless. It has been reported that the mothers

of women with CAIS have somewhat less pubic hair than women who do not carry the CAIS gene, so it can honestly be said that a lack of pubic hair is 'in the family'. In addition, some women choose to shave off their pubic hair. Others have a medical condition called alopecia totalis that causes their body (and scalp) hair to fall out. In other words, there are a number of reasons other than CAIS for not having pubic hair – and for some people the absence of pubic hair is seen as erotic.



O E S T R O G E N R E P L A C E M E N T T H E R A P Y

Any girl over 12 years old with AIS who has had her testes removed needs to take a supplement of oestrogen, initially to induce development in her breasts, hips and genitalia, and to promote the deposition of calcium in her bones. Many girls with CAIS are tall because the Y chromosome carries genes for extra height. Oestrogen treatment, which accelerates the completion of growth in the growth plates (the zones of growing cartilage near the ends of children's bones), may be used to prevent excessive adult height. In addition, oestrogen replacement therapy is beneficial in reducing the risk of heart attack and stroke in post-menopausal women. For this reason, and because it prevents bone thinning (osteoporosis), women with AIS should take oestrogen all their lives.

Many different forms of oestrogen are available. Most commonly it is taken as a daily tablet, but skin patches that are changed every few days are also becoming popular. Small tablet-sized implants of hormone may be placed under the skin using local anaesthetic every 6 months, but these do not suit all

women. Millions of women take oestrogen every day as it is a component of most oral contraceptives.

Oestrogens may have side effects for some women, such as weight gain, fluid retention, nausea and migraines. Women on high-dose oestrogen are also at increased risk of deep vein thrombosis (blood clots in the calf veins), but high doses are not required for women with AIS.

Commercially available contraceptive pills all contain a second hormone (a progestogen) which is there to induce regular changes in the lining of the uterus so that periods occur. Women who have no uterus do not need to take progestogen, and it is preferable for them not to do so because progestogens may have side effects such as depression, bloating and abdominal pain.

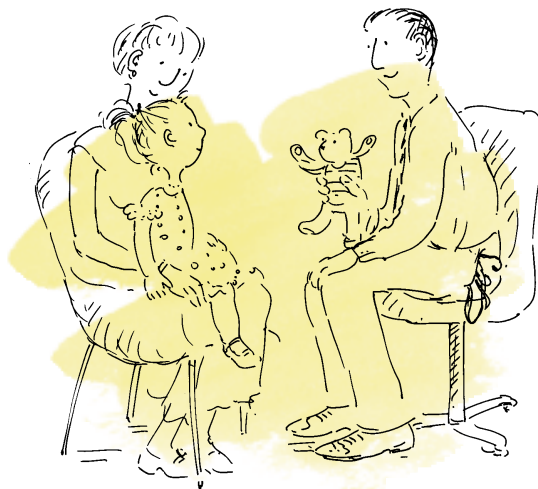
C O U N S E L L I N G A N D S U P P O R T

Better ways of understanding and working through the possible social and psychological effects of AIS are currently being developed, and many difficulties have been overcome. Nevertheless parents are anxious to maintain confidentiality, and affected women may feel uncertain about how others will respond to them. The general public is unaware of the existence of AIS because there is little media coverage of it. Any lack of community understanding is likely to be due to ignorance rather than prejudice.

Parents of newly diagnosed children

A medical diagnosis that affects the genital and reproductive organs is a particular challenge to understand, discuss and come to terms with in a helpful way. Since most families have never heard of anyone else with this kind of medical condition they usually ask their doctors for help in finding words and ideas. At the same time, they must find ways of dealing with their own powerful emotions, which often include great pain, sadness and anger. 'Why me?' and 'Why my daughter?' are natural reactions.

Ideally, AIS should be correctly and promptly diagnosed, and an expert should be available immediately to provide counselling for the parents. This, however, is not always the case and parents may encounter hesitation and delays along the way. Not surprisingly, this generates confusion and anxiety. An opportunity to discuss these feelings with a professional who is familiar with the condition is often beneficial. Counselling takes the form of assistance to put words and ideas to the many powerful emotions that may be aroused. It is inevitable that parents will have stages of vulnerability as they adjust to a diagnosis with reproductive implications, and the child's future development will need to be discussed.



Counselling and support

Parents of older children and teenagers

When parents feel they can talk to their daughter about her medical condition and the feelings that arise they can gradually help her to understand something of the nature of AIS. In the end this should strengthen their relationship with her. Parents face a dilemma about how and when to tell their daughter about AIS. Should they provide all the information they have as early as possible, or delay telling her until in their judgement she is ready to hear the truth? The emerging view is that she has a right to know everything, but that information should be given in stages that take into account her level of development and ability to understand what is being said.

Children aged 6–11 years

Up to the age of 11, most children have little ability to think about long-term consequences, and a simple explanation about the reason for seeing the doctor will generally suffice. If parents have concerns about the questions or behaviours their child is presenting to them, they could have a discussion about her with a mental health professional interested and experienced in assisting parents and children with AIS-related issues.



Counselling and support

Adolescents with AIS

Girls usually develop the ability to reason and to think in philosophical terms about the age of 12, although the timing of this varies from person to person. It is only at this stage that a girl with AIS will be able to understand a discussion about the complex nature of her condition, and even then, she will be quite unfamiliar with the internal workings of the body.

Adolescence is a stage associated with peer pressures to conform. It is preferable for girls with CAIS to learn about their differences compared to others *before* this pressure to conform becomes dominant. Children learn about genes and chromosomes in school at around the age of 15–16, and it is best for them to be informed about their own diagnosis before this time.



Counselling and support

Adults

It is understandably difficult for many women with CAIS to accept the ways in which the disorder affects their lives and relationships. All medical conditions arouse complex feelings in people and their families, and relationships may at times come under strain as different family members adjust to the issues in their own way. Because it is a genetic condition, a woman with CAIS may have affected sisters, nieces or aunts who can provide mutual understanding and support.

Many women with AIS marry and adopt children. They and their partners know from the beginning that there will be no pregnancies and can plan their lives and careers accordingly.

This may be less painful than the experience of the 10% of all married couples who find after years of marriage that one or other partner is infertile. Women with CAIS are found in all walks of life.

When required, counselling services are available and a referral from the doctor to a mental health professional or social worker will be arranged on request. Many women find it very helpful to meet other women with AIS.



Counselling and support

Support groups

An international support group for people with AIS and their families has recently been formed, and much invaluable information has come from affected adults who are assisting the medical profession to find better ways of helping younger girls and women deal with their diagnosis. Regular newsletters are published, enabling a free exchange of views and experiences.

Australasian residents

For a fact sheet, publications list, sample newsletter and subscription details, please send \$A5 payable to 'AIS Support Group Australia' to PO Box 3371, Logan Hyperdrome, Loganholme, Queensland 4129. Tel (07) 3807 2712.
E-mail: pelican@gil.com.au

UK residents

For a fact sheet, publications list, sample newsletter and subscription details, please send £UK1.50 (cheque or postal order) payable to AISG (or £3 with request for sample newsletter) and enclosing a large, stamped self-addressed envelope to PO Box 269, Banbury, Oxon, OX15 6YT. Tel (0) 1295 670353.

The group's magazine A^HS and meetings are free to UK-resident members (subscription £10 per annum). A parent who is medically qualified is available to answer questions from clinicians on behalf of the group (Tel (0) 1487 773442.
E-mail: higgins@easynet.co.uk).

US residents

For a fact sheet and publications list please send \$US2 (payable to Sherri Groveman) and a stamped self-addressed envelope to 4203 Genesee Ave, Box 103-436, San Diego, CA 92117-4950. Tel (619) 569-5254.
E-mail: aissg@aol.com

Canadian residents

Please request details from PO Box 425, Postal Station C, 1117 Queen Street West, Toronto, MJ6 3P5. Tel (416) 535-0948.

Internet address

The AIS Support Group has recently established its own website on the Internet. The address is:
<http://www.medhelp.org/www/ais/>

Counselling and support

Contacts

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Additional material

Goodall J.

How children think.

Journal of the Royal Society of Health 1987;

107: 224-26

Goodall J.

Helping a child to understand her own testicular feminisation.

The Lancet 1991; **337:** 33-35.

(Note: 'testicular feminisation' is the term formerly used for AIS)

Video

Dark Secrets: XY Women.

BBC Television 1996

(Hilary Clarke, Producer)



S H E R R I ' S S T O R Y

In August 1997, the author was privileged to meet Sherri Groveman, US representative of the AIS Support Group, when she visited Sydney as a guest speaker at the Annual Scientific Meeting of the Australasian Paediatric Endocrine Group. Here, in Sherri's own words, is her story.

My arrival into the world 38 years ago was a joyous occasion for my family because I was the first girl after two sons. However, ten days after my birth I developed hernias. Surgery performed at that time revealed that I had testes. Further investigations confirmed the diagnosis. I had AIS.

Like many women with Complete AIS who were raised in less enlightened times, I was not told the truth about my condition. Instead, I was left to figure this out for myself in a medical school library at age 20. I think this is unfortunate because I have experienced more emotional pain about the fact that my family didn't tell me the truth than about either my gonads or my chromosomes. I believe that with appropriate family support and counselling there is no reason any young woman with AIS should be destroyed by this knowledge. Having said this, however, I think it is only reasonable to assume that any young woman who is told the truth will go through a period of grieving. To my mind, this is a natural and healthy part of coming to terms with having any disorder.

It is unfortunate that doctors (and sometimes parents) overlook the difficulties faced by a young woman who does not have a menstrual cycle (which is a coming of age rite on the road to womanhood in this society) or pubic hair. These seem like such minor losses at one level, and yet they are very real sources of anxiety for those of us who have to face them. I remember all of my friends talking about getting their periods and feeling excluded from the discussion, terrified that they would know that I was different. To this day I have trepidation about anyone seeing me naked from the waist down, for fear that they will be shocked at my lack of pubic hair.

I also hope that doctors will investigate a range of options for dealing with the issue of vaginal shortening. This has been a real problem in my life and I am dismayed that the medical community expends so much money and energy developing new infertility treatments but so little effort at developing new techniques for dealing with the critical issue of vaginal length. While this fortunately is not a problem for many of our members, for those of us who have shortened vaginal length, it is of utmost importance.

Being unable to have biological children has, of course, also caused me some sadness. But I have chosen to invest my energies into my career and friendships . . . I do, however, have several friends

Sherri's story

who have AIS who have adopted children, or have had children through surrogacy. Having met their own personal challenges, I believe these women are especially well equipped to help their children come to grips with the emotional challenges of being adopted.

The road to healing the emotional scars experienced as a result of having AIS has been a long one for me. But the most critical element has been my involvement with the AIS Support Group. Meeting other women who have shared my experiences, and being able for the first time to have a community of friends who know what it is like to deal with the issues I face, has provided me with countless benefits. I feel so much more at ease with the whole concept of AIS.

I have now told several close friends that I have AIS, and am continuously amazed that by sharing my secret my friendships have only been strengthened. Indeed, I have found that sharing my most intimate secret with friends has freed them to share their deepest, darkest secrets with me.

It is wonderful that there is a support group to offer the latest information about AIS as well as emotional support and encouragement. I now have friends around the world who have AIS – this has been a wonderful 'fringe benefit' of being involved with the group. We talk, laugh, cry, and bond together. Meeting bright, interesting women with AIS, and not feeling at all put off by the fact that they have AIS, has allowed me to understand how others could see me in a similar light. What a huge burden this has lifted from my shoulders!

As odd as it may seem, in the final analysis I feel that AIS has been a blessing in my life. I think I am a more compassionate and intuitive person because of the personal pain I have experienced. AIS has forced me to base my self-identity as a woman on strength of character and warmth of spirit rather than more superficial physical attributes. And as I often say, if I didn't have AIS I would have gone through life like my two older XY brothers – instead I believe God intended me to be female; AIS was the route it took to get me there.

If you're reading this and either have AIS or are the parent or relative of someone with AIS, I extend my hand in welcome. I really look forward to meeting you!

Sherri Groveman

A N D F I N A L L Y . . .

In case readers of this book think that women with AIS never laugh about their condition, here is a verse that Sherri Groveman recited to the author during a stroll along a beach in Sydney:

I am Woman

Hear me roar

With testes too big to ignore!

