

Androgen Insensitivity Syndrome (AIS)

What is a hormone?

Hormones are chemical messengers. They are made in glands and travel round the body in the bloodstream. Hormones affect how other organs in the body work.

What is androgen insensitivity syndrome? (AIS)

The testes in boys are responsible for making hormones called androgens. When a baby boy is developing in the womb it is the action of androgens that develop the genital area into a penis and scrotum. In AIS the body is unable to respond to these hormones. This can mean that the baby is born with genitals that look female. This is called Complete Androgen Insensitivity Syndrome (CAIS).

Why does CAIS happen?

Chromosomes are structures in our cells that carry our genes. Genes are like a set of instructions for our body. Usually males have one X and one Y chromosome and females have two X chromosomes. CAIS is a rare genetic condition carried on the X chromosome.

For any hormone to work it has to join on to a particular part of a cell called a receptor. Each hormone needs its own receptor –in the same way that keys are only designed to work in the correct lock.

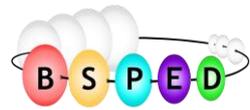
In CAIS the receptor is faulty so the androgen hormone cannot join with it. This means that even though the child can make lots of androgen hormone it cannot affect the body.

The condition only occurs in XY persons if they have a faulty X chromosome. In females with XX the other X chromosome works normally. This means that a woman will not be affected by the condition but can pass this on to her children. This will be talked about more in clinic and you will also have an appointment with a specialist doctor called a Geneticist to find out more.

How does it affect the child?

Babies with CAIS have XY chromosomes and so develop testes, which produce normal male amounts of androgens. However, the body is not able to respond to the androgens. Because androgens are needed to develop the male genitalia before birth, a baby born with CAIS looks like a girl.

The diagnosis of CAIS may be made soon after birth or in early childhood. The girl may present with a hernia or a swelling in the labia. A scan may show that



the swelling looks like a testis. Sometimes a testis may be found during an operation to repair the hernia.

CAIS may also be identified when the girl is a teenager because she does not start her periods. A girl with CAIS will develop breasts normally, but have very little under arm and pubic hair. However she will not have ovaries or a uterus (womb) so will not have periods or be able to become pregnant. Adult females are usually taller than average women.

How is it confirmed?

A scan (ultrasound and/or MRI) of the abdomen (tummy) will show that there are no ovaries or uterus.

A blood test is needed to check the chromosomes. If they are found to be 46XY then more detailed tests on the blood are done. The genetics doctors are able to look at the gene that is responsible for giving the instructions for the special receptor cells to see if this is faulty.

This information can also be used if other members of the family wish to be tested to see if they are carriers of CAIS.

How often does this happen?

Complete androgen insensitivity syndrome happens in 2 to 5 per 100,000 male births.

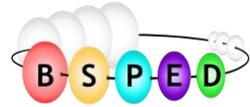
How is it treated?

If a hernia is present it may need to be repaired by an operation. Otherwise no treatment is needed until after puberty. The testis will produce hormones that get changed to oestrogens, which causes breast development.

Some parents want the testes removed early. If this is done, oestrogen needs to be given from about 11 years of age so that the girl develops breasts at around the same time as her friends.

There is a very small chance (less than 5%) that the testes may form a tumour during adult life. If the testes have not been removed during childhood this will be discussed with the young adult woman and she can decide if and when she wishes to have an operation to have the testes removed.

If the testes are removed then the young woman will need to have long-term oestrogen treatment as tablets or patches.



Because the uterus does not develop the vagina is often shorter than usual. This can make sexual intercourse difficult and uncomfortable. The teenager/young adult will need to have an examination under anaesthetic. It is important that this is done well before she is thinking about sexual relationships. If the vagina is too short the girl can stretch it using special dilators when she is ready to do this. More information about this will be discussed in clinic during teenage years.

Are there any long-term complications?

Girls and women with CAIS have good health. Psychological well-being is similar to other women with female identity.

Telling the young girl about CAIS, infertility and sexual function is a step-wise process. It should start as early as possible and continue through to young adulthood.

You will meet many health care professionals involved in your daughter's care and part of their role is to support the family throughout this. Many centres have a psychologist as part of their team – their role is to help you support your daughter. They can be really helpful in helping you decide when and how to explain the condition and any investigations/appointments to your daughter and other family members.

Many families find the AIS patient support groups very helpful.

Suggested sites for further information:

www.aissg.org

<https://www.nhs.uk/conditions/androgen-insensitivity-syndrome/treatment/>

This leaflet has been written by members of the BSPED & reviewed by the Clinical Committee. It is designed to give you some general information about your child's condition and treatment. Your child's doctor or specialist nurse will be able to answer any further questions you have about your child.

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