The BSPED is one of the affiliated speciality groups of the Royal College of Paediatrics and Child Health.

The society aims to improve the care of children and young people with endocrine disorders or diabetes mellitus, by bringing together professionals from a range of disciplines.

Please contact your local nurse or hospital doctor if you need more advice about your child's condition.

This leaflet has been written by Pauline Musson, CNS. Southampton Children's Hospital & reviewed by the Clinical Committee, with acknowledgement and thanks for the contribution of Dr Julie Alderson, Consultant Clinical Psychologist.

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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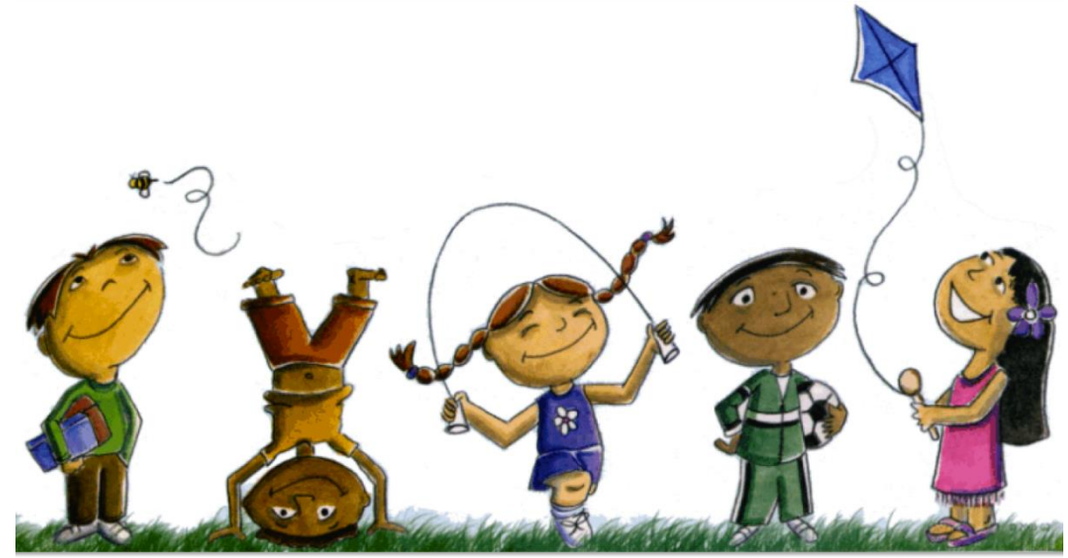
For more information, contact:

BSPED Office
Bioscientifica
Starling House
1600 Bristol Parkway North
Bristol
BS34 8YU
T: + 44 (0) 1454 642258
E: bsped@endocrinology.org

British Society for Paediatric Endocrinology and Diabetes

Complete Androgen Insensitivity Syndrome (CAIS)

Information for patients, parents and carers



What is complete androgen insensitivity syndrome (CAIS)?

Complete Androgen Insensitivity Syndrome (often shortened to CAIS), is the medical name used when a person's body cannot recognise or use a hormone it produces.

Hormones are chemical messengers. They are made in glands and travel round the body in the bloodstream.

In CAIS the body cannot recognise or use the hormones known as androgens, specifically testosterone. Testosterone is a sex hormone that affects how a baby's body develops in the womb and during puberty.

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, but not always, 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 a certain lock.
In CAIS the receptor doesn't work 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 people with XY chromosomes who have a change in the gene that is responsible for making the androgen receptor. In the same way that changing one letter can change the meaning of a word (such as fire and fare or wood and word) the change in the gene means the instruction for the androgen receptor no longer works.

In girls and women with XX chromosomes only one of the X chromosome has the change in the gene. 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 should be offered 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 androgens. However, the body is not able to respond to the androgens and so develops as a baby girl with labia and a clitoris.

The diagnosis of CAIS may be made soon after birth or in early childhood. Some girls are diagnosed because they develop a hernia or a swelling in the labia or groin. 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, but have very little under arm and pubic hair. She will not have ovaries or a uterus (womb) so will not have periods or be able to become pregnant.

How is it confirmed?

A scan (ultrasound and/or MRI) of the abdomen (tummy) will show that there are no ovaries or uterus but there will be internal testes. These are the parts of the body that produce most of the person's androgens.

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.

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

How often does this happen?

Complete androgen insensitivity syndrome happens 1-5 times in every in 100,000 births.

What care is offered to people with CAIS?

If a hernia is present it may need to be repaired by an operation. The testes will produce hormones that get changed to oestrogen, which

gives the girl breast development and also helps to keep her bones and heart healthy.
There is a very small chance (less than 5%) that the testes may form a tumour during adult life. This will be discussed with the young adult woman and she can decide whether she wishes to have an operation to have the testes removed.

If her testes are removed then the young woman will need to have oestrogen treatment as tablets or patches until she is in her early 50's.

A girl with CAIS will not have a womb (uterus). This means she will not have periods or be able to carry a pregnancy.

Having no womb also means that she will not have a cervix or the deeper part of the vagina. If the woman's sex life involves vaginal penetration then this will put pressure on the vagina, which may feel uncomfortable or painful.

A gynaecologist can examine the vagina or explain how an adult woman can do this herself to understand how her body has developed. Teenagers who wish to find out more about this may be offered an examination under anaesthetic.

Some women who find that their vagina is short may wish to make it deeper. The specialist gynaecology team will be able to support the young woman to do this once they have completed puberty.

Life with CAIS

Girls and women with CAIS have good health.

A girl or young woman with CAIS will learn that their body has developed in ways that were unexpected. For the girl learning about CAIS, infertility and sexual function should be a gradual process. Ideally it should start as early as possible and continue through to young adulthood.

The doctors, specialist nurses and psychology teams can help them and their parents understand their body and support them emotionally. They can be really helpful in helping parents decide when and how to explain the condition and any investigations/appointments to your daughter and other family members.

Some families find meeting (online or in person), or learning from other people with CAIS can be very helpful.
Suggested sites for further information:
www.dsdfamilies.org/charity
www.nhs.uk/conditions/androgen-insensitivity-syndrome/treatment