



British Society for Paediatric Endocrinology and Diabetes

Disorder of sex development

When babies begin their development in the womb it is impossible to say whether they are male or female as their external genitalia as well as the internal reproductive organs look the same. Whether these physical features develop into those of a girl or a boy depends whether the developing baby has testes or ovaries and whether these organs make sex hormones which can work normally. In addition to these glands, there are also other important glands including the pituitary and the adrenal glands which also control and make sex hormones.

Whether a child has testes or ovaries will depend on the child's genetic make up. Although generally speaking, children who have 46 chromosomes including an X and a Y chromosome turn out to be a boy, in some cases these children may develop as girls. On the other hand, although most children who have two X chromosomes are girls, some may develop as boys. Sometimes, the appearance of the genitalia are such that the sex of the child is not immediately clear at birth. More commonly, and particularly in boys, the genitalia may be different in such a way that although sex assignment is not in question, there is some concern that the hormones controlling sex development may not be working normally.

Sex hormones are important not just in the developing baby but they are also important in the older child who is entering puberty. Sometimes, the sex may be clear at birth and it is when the child grows up that concerns about sex development are raised.

In many children with these different forms of sex development, there is a concern that there may be an associated problem with the child's immediate health or long-term health. Doctors, therefore, prefer to use the term '*disorder of sex development*' as it implies the possibility of a medical condition that needs to be investigated and treated. Identification of an underlying cause can help with treating any hormone deficiencies. It also helps with anticipating any other health concerns in the future. Knowing the cause also helps with explaining the condition to the parents and the growing child and allows them to plan for the future.

The timeline for medical care will vary from one condition to another as well as the age when the child first presents. In general, when it comes to DSD, there are four facets to medical care and although they are all linked to each other, these facets could be spaced out along an imaginary timeline for the child as well as the parents.

Sex Assignment

Newborn babies are usually given a sex at birth and sex assignment is, therefore, the first step on the timeline in the care of the child with suspected DSD. A delay in assigning sex usually occurs when the health care staff who are present at delivery are not clear. Sometimes, there is a concern about sex assignment when the baby is examined in the first few days by staff after delivery.

When the sex of the newborn baby is unclear, medical staff explain this to the parents and ask them to delay naming the baby. Clarification of the sex involves a thorough examination of the genitalia and often this is sufficient when performed by a more experienced doctor in the maternity hospital. Sometimes, the baby will need to be examined by a group of specialists in the field of DSD. A number of babies require tests and usually this involves a blood test to check the chromosomes and an ultrasound scan to check the internal reproductive organs.

These tests can be done immediately after birth and, if the technology and expertise exist, they are performed at the local hospital and the results can be back within one to two working days. Sometimes the baby needs to be transferred to another hospital for the tests as well as examination by a specialist. Over the first week, there may be a need to do other blood tests to check hormone levels and to ensure that the baby's health is unaffected.



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In most cases, the examination of the baby, the chromosome test and the ultrasound scan are enough to decide on the sex of the child, and a decision will be reached within 2-3 days. In a very small number of cases, there may be difficulties in deciding on the sex of the baby despite these tests and additional tests may be required. It may, therefore, be necessary to delay sex assignment for a few more days. However, a delay in sex assignment for over one week is exceptionally rare these days.

This is often a stressful period for parents. The professionals within the specialist DSD team will do their best to support the parents and routinely seek the help of psychologists who are part of this team. Parents are also provided with information about support groups and other information resources.

Looking After The Immediate Health

Glands such as the pituitary and the adrenals, that make sex hormones also make other hormones which are important for the health of the child. In newborns, where there are concerns about sex development, blood tests and urine tests may be performed to check these other glands. Some tests, such as checking the blood sugar level can be performed at the bedside and the results are available immediately. For other tests, samples have to be sent to special laboratories and the results may take anything from a few days to weeks to come back.

Generally, if the doctors are concerned about the baby's health, they will start the medicines after the tests are done and then these medicines will be stopped if the results turn out to be normal at a later stage.

Finding Out The Underlying Cause

Although in some cases, the actual cause of the condition is very clear in the first month of life, in most cases, finding out the actual cause of the problem can often take a long time ranging from months to years. In many cases, the actual cause may never become clear. Besides trying to find out the cause of the condition, tests also allow doctors to exclude those causes which they know may be associated with short-term or long-term problems of health. A short-list of possible causes also allows doctors to decide on future tests as well as care of the child. Some of the tests need to be performed in special centres and may require special skills such as scans or laparoscopy. Usually, it is better to wait and have a systematic plan to do these tests. The right plan of tests will be discussed by the specialist team with the parents, often at a special clinic where everybody is present. In most cases, the tests are performed within the first year of life and sometimes they are repeated at a later stage. Sometimes the samples are stored so that a new test which becomes available at a later stage can be performed without taking another sample. When children grow up and move to adult care it may be useful to repeat these tests.

Looking After The Long-Term Health

The effects on long-term health of the child will vary and depend on the underlying cause of the condition as well as how it has affected the child. The DSD team in specialist centres consists of a group of specialists who are experts in their own areas such as growth, puberty and development, surgery, urology, gynaecology, psychology and genetic counselling. Affected children and their parents may benefit from visiting such a clinic so that all the specialists can provide their input as and when necessary. Usually children with DSD attend the clinic of a paediatric endocrinologist who ensures that occasionally the child attends the special clinic run by the DSD team. At these clinics a plan will be made with the parents and the growing child about future tests and treatment. A plan can also be developed for how to explain the underlying cause of the condition to the child.

There are no fixed rules about how often a child should attend the paediatric endocrine or DSD clinic. Where there are concerns about long-term health, this author does not see any child at an interval greater than 2 years. The family should be able to change the appointment and move it earlier or later and should be able to contact the doctors by telephone or email.