



First Days-
when your baby is born with genitals that look different

When your baby is born with genitals that look different and doctors are looking into the causes, it might feel like you are the only parents this has ever happened to.

That's not the case.

Each year, more than 100 families in the UK find themselves in this situation.

The following information has been put together by parents and specialists to guide you through the first days and weeks.

When your baby is born with genitals that look different... we need to look at the bigger picture

The development of your baby in the womb is likely to have been affected by one of many causes that doctors call DSD (Differences – or Disorders - of Sex Development).

Sex development is a complex process and the sex of a baby is not determined by what genitals look like. There's a much bigger picture, and many things together can influence the sex of a baby, such as:

how the genitals look like on the outside,
how the baby's reproductive organs developed on the inside,
hormones and how your baby's body responds to them, and
genetic information

By working together with health care professionals and by talking with other families parents can get the information they need to understand how their baby has developed and how to raise a happy, healthy child.

Why might DSD happen?

Sex development is a complex process, and there are many different variations that can happen on the way. A good way to think about this is to imagine how babies in the womb follow different paths to develop.

Some of the most common reasons for a DSD are congenital adrenal hyperplasia (CAH), peno-scrotal hypospadias, hormone imbalances or variations in the baby's chromosomes.

In most circumstances your expert team will be able to discuss a clear way forward with you quite quickly and you can get on with enjoying your baby at home. In the months that follow there may be follow-up visits with your team to talk further and think about the medium to long term care of your baby.



What happens next?

Diagnosis requires a team, takes time and involves various steps and tests

Healthcare for children with DSD involves a specialist or professional team which would ideally include specialists from endocrinology (hormones), urology/gynaecology (kidneys, bladders, genitals) and psychology.

Other team members may include nurses, geneticists, neonatologists, and people who work in the laboratories. These 'multi-disciplinary' teams are based in some larger hospitals.

When a baby with DSD is born, the medical/midwifery team in the local hospital will care for your baby safely and arrange for some initial tests be done (see the section entitled Tests your baby might need). More specialist tests are usually arranged by the multi-disciplinary team and often involve going to the hospital where they are based.

You, as the parents, play a key role in this whole process!

Once all the necessary results are to hand the health care team will discuss these, and the way forward, with you. Usually this takes a few days. Sometimes it can take longer.

Don't worry about how to fill in the baby's birth registration.

In England, Wales and Northern Ireland a birth should be registered within 42 days and in Scotland within 21 days. This is enough time to learn all you need to know before registration is required. Usually discussions about how to bring up your baby take place well before. Birth registration requirements can vary in other countries, so check with your team.

No one is expecting families to take in all the information at once. Spending time at this stage asking questions and for information to be repeated is important.

And just as important - for all parents - is being with your baby. You need time for recovering from the birth, establishing feeding, learning to bathe a new born, and most importantly trying to get some sleep.



A psychologist helps ordinary families in extraordinary circumstances

The psychologist can be very helpful in going over the information you have been told and in providing some perspective if the various tests and medical discussions seem overwhelming. They are there to help you think and to work out how you feel.

This can help you manage news about your baby's birth, how to discuss your child's condition with other people and, importantly, in the future how you might talk to your child about their condition.

Many families also really appreciate the support of a specialist endocrine or urology nurse to talk through and help them understand what they are learning about baby's development.



Tests your baby might need

Your baby's doctors will examine your baby and ask about your family's medical history.

Doctors will check your baby's chromosome pattern with a blood test. The chromosome pattern (also known as "karyotype") helps doctors understand the background to your child's development and helps them consider in which direction to take the further tests. The initial karyotype result usually takes 2-3 working days to come back.

Many babies are kept in hospital for the first few days so that salt levels in the blood can be monitored. Doctors will also measure hormone levels in the blood, and possibly in the urine to see how the adrenal glands and gonads are working.

Ultrasound tests and scans can sometimes show the internal sex organs (such as the womb/ uterus). These are best performed in specialist centres and can sometimes be difficult to interpret.

Doctors might rarely look at the bladder and opening of the vagina with a special 'telescope' (cystoscope) and very rarely they might look at the gonads with a laparoscope and examine tissue samples (biopsies) from them.



Talking about your baby in the first days or weeks ...with friends and family

It can be difficult to know how to explain/what to say to family and friends that your midwife thinks the sex of your baby is unclear. If your loved ones are waiting to hear if you and your baby are OK and whether you have a daughter or a son you could let them know that your baby is having tests because it is sometimes difficult to tell in a new-born.

What you choose to say to those who love you and what you share more widely can be different. Some parents will say: **'We can't tell you yet whether our baby is a boy or a girl, because our baby has been born with a condition that affects what the private parts look like. Doctors are doing further checks and are making sure our baby is healthy and well. We will get in touch soon with more news.'**

Other parents will inform family and friends in a general way and not provide any details, for instance, **'the doctors have some questions about how our baby's grown. It's complicated to talk about, and we will be in touch soon.'** You may be concerned that people might ask other questions, but just be really firm: **'I can't really explain it myself yet'**.

You can move the conversation on to something you do need to talk about, such as **'Thanks for picking up Sophie for us...'** If afterwards anyone whom you don't want to share information with asks: **'What was the problem? We were so worried'**, you can say: **'We just needed to make sure everything was ok, all is well.'**

Some parents will ask a friend or a member of their family to be their contact point. You can agree on a simple explanation that everyone sticks to and which can be as simple as **'mother and baby doing fine – a few tests need to be done and an update will follow'**.

Other parents will want to wait for the diagnosis before sharing information with families and friends. If you are not ready to talk about it you can say **'We have a lovely, beautiful baby. It was a really difficult birth and I/we just need a little family time, so don't be hurt if we seem to drop off the grid for a while.'** Don't be uncomfortable about protecting yourself a little once in a while.

...with people who are supporting you and your baby in the hospital and at home

DSDs are a large group of quite rare conditions. For some people involved in your post-natal care and in the care of your baby it will be the first time they come across these conditions. They will try to do their very best for you, and sometimes when trying to be helpful may say unhelpful things. You can say **'this is how I'd like to talk about my baby until we know more'**.

Other caregivers might have had some experience with another family and want to share this with you. However, there are many different reasons why a baby might have a DSD so it is important that any discussions are relevant to your little one.

Looking for information on the internet before having initial details on the reasons for your baby's DSD can add to confusion and sometimes misunderstanding. Use recommended websites (for English language resources go to www.dsdamilies.org with signposts to various related DSD conditions).



What do we call our baby in the meantime?

Doctors might initially advise you not to think of your baby as a boy or as a girl. Sometimes they ask you not to call your child by a name or say he or she. Some parents are happy with this while others find it more difficult. Don't worry, you won't harm your child. Call your baby whatever feels right.

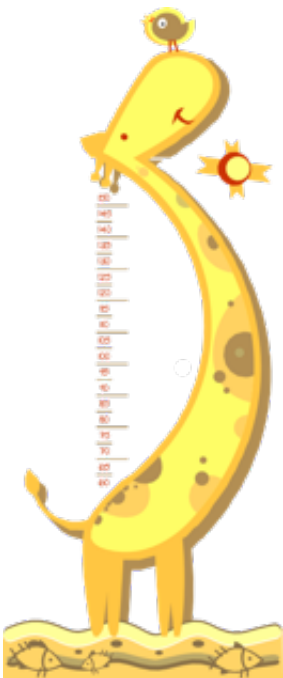
While you wait for test results you could use a simple endearing name, like sweetheart, honey, munchkin, or darling... A couple whose baby was born on Halloween called their baby 'pumpkin', until a few days later 'pumpkin' became Max. It is also common (in the UK) for parents to say that they just have not found the right name, and that they are waiting to see what name best fits their baby. If your faith requires your baby to be named within the first few days, and when this is not possible, talk to your religious leader about the best way forward.



What do we tell our other children?

If you have older children they too of course will ask whether they have got a brother or a sister. Depending on their age, you can tell them what is happening or –as they might be worried why baby needs various tests- try to reassure them without making up information. Remember that children accept things much easier than adults, and always try to use simple and honest language.

Make certain there are foundations of truth in what you say on which you can later build on what you tell any child. For example, you could say: **'The baby is so new, the doctors can't tell us yet.'** Or you could say **'The baby first needs to be examined by doctors. So we are going to wait for a few days to name the baby. When we're ready, we'd really like you to help choose the nicest name.'**



“But we've already told everybody”

In some rare cases, for example if baby's genitals look fairly typically male or female, doctors and midwives will say at the birth of a baby **'you have a boy'** or **'you have a girl'**. However, a few hours later - sometimes after you have texted family and friends - doctors pick up on a variation of genital development and advise you that the sex of your baby is uncertain. In some cases, after all the necessary investigations, they may advise you to raise your child in the other sex of what has been announced.

One effective way to approach this is first to learn about - and learn to talk about - the diagnosis and the causes of it. Practice explaining this with your partner and with some doctors; this is also where psychologists and specialist nurses can really be of great help. You can let friends and relatives know as much or as little as you want to.

Remember: if you are comfortable and confident about this, then your family and friends and your child will be too.

Another way to handle this is to simply let friends and family know that there has been a mistake. You can say: **'Her/his bits were small/ tucked away/swollen and we/they got it wrong'**.

In other cases, an **ante-natal scan** or tests might have suggested you are having a boy or a girl, and you have shared this information with friends before the birth. Then, when your baby is born, sex is not so clear. If these are your circumstances, there is no urgent reason to confirm or revisit this while you are getting more information. Use some of the strategies above, and concentrate on questions regarding health and feeding.

Another idea you might use if you have decided to raise your child a different sex from what you have announced, and you don't want to share much information with friends and possibly relatives, is just to say that the ultrasound was mistaken and you have had a lovely surprise.

'These things happen!'



What happens in the next weeks?

Having a newborn baby is a joyous time, but when there is some concern about the baby's health, joy can share a space with worry.

Privacy is not the same thing as secrecy!

One of the main challenges of DSD conditions is that many parents feel they must look after their child's privacy during childhood. This can leave parents feeling isolated.

Having friends who know something is worrying you can give you more time to get used to and more confident about discussing your baby's wellbeing. You can also ask your team of doctors and nurses whether they host family events, or to put you in touch with other local families who have gone through similar experiences; ask about family groups (such as a support group) or on line forums.

All questions are good questions!

Very few parents are able to take in all of the complexities around DSD in one go. And sometimes we worry about asking 'a stupid question'. There is no such thing as a stupid question and you should feel comfortable to ask your medical team to repeat information. Use drawings and notes to help you remember. If things are not clear ask your team to go over it all again.

After meeting your healthcare team, it is helpful to establish who your point of contact is, and how you can approach them when you have an urgent question (for example, by email or telephone). This could be a specialist nurse, or another member of the team. When you are at home with your baby, keep a notebook to jot down any questions that might come to you.



Timeline of care in the first days and weeks?

| What the health team does | What you can do |
|---|---|
| <p>Step 1</p> <p>The health team will look after the immediate health of your baby and make sure you recover well from giving birth.</p> | <p>Recover from birth and try to get some sleep!</p> <p>Concentrate on feeding; it's normal to feel worried and anxious after having a baby, and concentrating on something practical and important like feeding your little one is a good way to focus on and spend time with your baby.</p> <p>Like with any new-born, keep your beautiful new baby close if you can. Bonding is the first step in raising confident and happy children. Bonding does not just happen at birth, or just in the first few weeks – and you must not worry if you don't have a chance to focus on this straightaway.</p> <p>Children need bonding at any age: hold them close, play with them, listen to them, and make time for them. That way you make them feel that you really love them and always will.</p> |
| <p>Step 2</p> <p>The health team will look for the reasons behind your baby's DSD. They will discuss their findings with you and advise on bringing up your baby as a boy or a girl.</p> | <p>In this world of text, email and social media and with everyone wanting to know whether you gave birth to a boy or a girl... you may feel pressure to let people know about the sex of your baby.</p> <p>If people don't hear from you right away they will probably assume that you are tired or have just forgotten to say.</p> <p>Not posting anything is an option or you might let people know that you won't be online as much for a few days.</p> <p>Some parents talk openly about the development of their baby to family and friends, others don't – there is no right or wrong.</p> |
| <p>Step 3</p> <p>The health team will help you in looking after your child's overall health and wellbeing, and support your family in the long term.</p> | <p>Enjoy your lovely baby and do all the things new families do.</p> <p>Take time to recover from the birth and from the stress of the diagnostic process.</p> <p>Take time to inform yourself about DSD and the specific type of DSD your child is diagnosed with (if a diagnosis has been reached/if diagnosis is possible). Don't hesitate to ask the psychologist, or other doctors to explain again what the diagnosis is, and what this means for your child – and remember that all questions are good questions. Always!</p> <p>If you feel like talking with other families, check with your doctors if they host family days, or if they can connect you with a family or family group (e.g. support group or online forum). Psychologists who have expertise in DSD can help you understand and talk about the diagnosis and help you think about when and how to share information with your child.</p> |

Moving forward...What happens the next few months? And then?

What happens next depends for a large part on your baby as an individual. If your baby is diagnosed with CAH, your healthcare team will help and support you as you learn about timings and dosages of medication.

Once you and your baby are at home, further questions about how best to support your baby, how to talk with your child about his/her condition, how and when your child may be involved in care decision in the future, etc... will arise.

It can be helpful to have a detailed review meeting after a year or so with some members of your medical team to go over events after the birth, to talk about the diagnosis and what it means, and to prepare a long-term care plan.

Just like all kids, children with a DSD need love and support from their parents. Some children will need longer term support from a healthcare team that places the wellbeing of your child at the heart of all they do.

We asked other parents to share with you what they would have liked to know in those first days and weeks after their baby was born...

It's going to be ok. DSDs are actually quite common and there is more and more information and support available, for the parents and the child.

Don't rush into decisions. Very few issues having to do with a DSD need to be handled immediately.

Take time to bond and get to know your baby. The rest can be dealt with later. Find someone you trust to talk to face-to-face. There is a lot of online information but having one person for face-to-face conversations is very helpful.

You are not alone; whenever you are ready there are plenty of families to connect with via groups and on-line forums.

Honestly, it is good to talk with other families about what happened, about worries you might have and about your parenting successes.

Our babies, toddlers, pre-schoolers, pre-teens, teens... are not extra-ordinary – they are just as hard work as all the other kids!

The first days, maybe weeks, when all information seems behind closed doors, are the worst; then you start to understand and it all gets better. Now, we have decided to have another baby.

Bonding was difficult, at first. But from having had other babies, and, from speaking about this afterwards with other families, I understood that this is common outside of DSD, too...part of post-natal hormones and surprise (about my baby who was different than expected) and worry and tiredness.

You are not alone; whenever you are ready there are plenty of families to connect with via groups and on-line forums. Honestly, it is good to talk with other families about what happened, about worries you might have and about your parenting successes.

I suppose I could not imagine raising a kid with genitals that look different. To be honest, I was intimidated by what other people would think. I ended up having a confidential chat with the nursery and said 'this is how my baby is born, and he is all checked out and healthy. Any questions?'

I have another son, who does not have a DSD. He insists on wearing his Ben Ten hoodie towel to change into and out of his swimming trunks. 'Why do you do this?' I asked him. 'Because willies and bottoms are private' he tells me.

What makes the difference is how we choose to live with the things that happen to us. That is what I learned in those first weeks, and that is what I want to teach my daughter.

There's really nothing you need be doing now except enjoying your beautiful baby

If I were in the same situation again, I'd say to the doctors: Stop, I have no clue what you are talking about. Explain it again from the very beginning.

For far too many years I magnified and overanalysed my daughter's behaviour: did we make the right decision (She plays with a dolly today: yes!! She is so pretty: yes!! She does not want black shoes as too boyish: yes!!). Or was it the wrong decision (She does not play with a dolly today: no!! Today she plays with a train: no!!).

Then, I learned about the difference between sex and gender. I accepted that my child has mixed sex characteristics. Not 'had', assigning sex of rearing does not change that. But equally, that those sex characteristics don't affect 'who she is'.

In her case she is a smart and confident girl, who last spring bought a pink jacket (because she likes it) and this winter a black winter coat (because she likes it); she doesn't wear skirts (because she does not like them); she is good at maths (because she has a good teacher who makes it exciting) as well as Spanish (possibly genes inherited from me); and she is desperate to go to school with mascara (because her friends do, and she likes it). (And no she is not allowed to go to school with mascara – are you kidding!)

DSD Terminology for beginners

Sex: when we talk about sex we generally mean biological or physical features such as genitals, hormones, gonads and chromosomes.

Gender: when we talk about gender, we talk about how a child or person feels.

Hormones: hormones are chemical messengers that tell certain cells in different parts of your body what to do.

Hormones can influence many things, including when you feel hungry or full, when and how thirsty you are, how strong your bones are, whether you are big or small and a whole lot more.

Sex hormones, such as **oestrogen** and **testosterone**, are chemicals made by the body that influence the development of the genitals.

Androgens: a general name for hormones involved in typical male development. Testosterone is an androgen. Both men and women have androgens, but in different amounts.

Endocrine/endocrinology/endocrinologist: related to hormones; a doctor who specialises in hormones and how they affect changes in our bodies.



Image copyright courtesy of Cristina Vadillo, image from: 'Pipo and Pepo, two tiny explorers'.

Chromosomes: each person usually has 46 chromosomes, including two sex chromosomes. A person's chromosome pattern can also be called their "karyotype" (e.g. 46,XY or 46,XX).

We usually get one X from our mothers, and an X or Y from our fathers. There are various combinations of sex chromosomes:



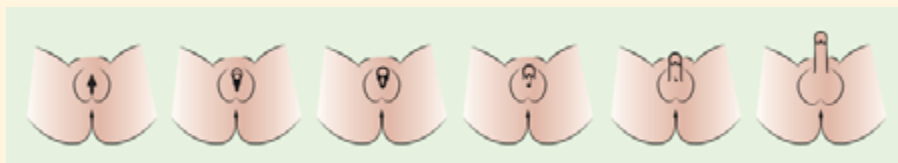
Genes/ Genetic testing: chromosomes are made up of many segments called genes which carry genetic codes (DNA) that influence many things, such as the colour of the eyes. Genes can influence sex development, and changes in specific genes can sometimes be the reason for a DSD.

Genitals: genitals are the external sex organs. This means: **the penis** (sometimes called 'willy'), **the scrotum** (the bag of skin containing the testes/testicles/balls) and the **vulva**, which includes all the external female sex organs including **the clitoris** (a female sexual organ that is small, sensitive, and located on the outside of the body above the opening of the vagina) and **the labia** (any of the folds of skin bordering the vulva and vaginal opening).

Your doctor might use the word phallus or phallis structure for either a penis or a clitoris.

All genitals develop from the same basic structures.

Genitals can have a range of appearance depending on the amount of androgens in the body at certain times.



Gonads: A general name for testes and ovaries. In DSD these can also be underdeveloped gonads (streak) or occasionally a mixture of ovarian and testicular tissue (ovotestes).

Ovaries: organ where eggs and sex hormones are typically produced

Testes: organ where sperm and sex hormones are typically produced

Urethra: the pathway from the bladder used to expel liquids (urine) usually opens at the tip of the penis in boys or between the clitoris and vagina in girls.

Uterus: the uterus is the space inside the abdomen in which a baby grows (also called: womb). Just like the genitals, the development of the uterus is controlled by hormones.

Urologist/ Urology: a urologist is a doctor who specialises in the kidneys, bladder & genitals.

Hypospadias: a fairly common condition (1 in every 300 baby boys) where the urethra does not open at the tip of the penis. There are different degrees of hypospadias. Many babies investigated for DSD have penoscrotal hypospadias.

Further Resources

dsdfamilies.org is an on-line information and support resource for families of children, teens and young adults who have a DSD - www.dsdfamilies.org

If your baby is diagnosed with Congenital Adrenal Hyperplasia, you will find lots of information on the website of the family group 'Living with CAH' - www.livingwithcah.com via the website you can also join an online Forum for families.

Notes

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....



This brochure has been developed with the kind support of Jeans for Genes

This brochure was produced by Ellie (dsdfamilies) and John Achermann (UCL/GOSH, London)* with lots of input and advice from parents, various members of multidisciplinary healthcare teams and family groups: Amber, Siobhan, Charlie, Elizabeth, Laurie, Joanne, Lizzy, Arlene, Alexander, Jenn, and Angela (parents whose children have a DSD); Polly Carmichael, Julie Alderson, Caroline Sanders, Sarah Creighton, Martine Cools, Santiago Vallasciani, Nils Krone, Katy Auckland and Sten Drop (specialists in DSD); Sue ('Living with CAH'), John (Hypospadias Forum) and Margaret (AISSG UK).

This information has been produced to guide you through the first days. All families' and children's circumstances are individual and your doctors and healthcare professionals can provide you with exact information for your circumstances.

If you would like to take part in developing this brochure further, and/or adapt it to local needs, please contact info@dsdfamilies.org

***reviewed June 2018 Co-edited with Julie Alderson (Bristol Royal Hospital for Children)**