

Support Information for Parents: Thinking About Your Child's Sex Development



An informational guidebook that takes support of children born with differences in sex development, and their families, to heart

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This section consists of a series of information sheets. Providers should review the list of information sheets (see the [Information sheets](#) section below), select those related to the child's specific diagnosis or features of the condition, and provide them to parents.

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This section also includes a series of information sheets. Providers should review the list of information sheets (see the [Information sheets](#) section below), select those related to the child's specific diagnosis or features of the condition, and provide them to parents.

Information sheets

- Provider recommendations checklist
- From this list, parents should receive those related to the child's specific diagnosis or features of their child's condition.*

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More information

- Your well-being
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- **Introduction:** Welcome, How to use this guide, Questionnaire – My current understanding, Why does my baby’s body look different, and Sex development basics
- **Support:** Support for families, Questions to ask, and Questionnaire – Sharing information with others and reaching out for support
- **Early medical evaluation:** Genital/physical exams, Before your appointment, and Questions to ask

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- **Testing:** Genetic testing, Incidental findings, Hormones, Learning about your child's body, Questions to ask, and Questionnaire – Genetic testing
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- **My Child's body looks or works different:** What do I do if my baby's body looks different, What do I do if my baby's body works differently, Questions to ask, Questionnaire – Genital surgery, and Questionnaire – Gonadal surgery
- **More Information:** Your well-being, Parents' stories, and Questionnaire – What have I learned

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Disclaimer

The information contained in this guide is developed to help you make higher quality health decisions. The information is not intended or implied to be a substitute for professional medical advice, diagnosis, or treatment. All content, including text, graphics, images, and information in this guide is designed to be used in partnership with your child's healthcare providers for general information purposes only.

Welcome to the Support & Information Guide

Our work

We have created an information resource guide for parents and family members to explore. This guide provides basic information about conditions that affect how your baby's body looks or works and continues with more specifics about their condition and its management. Some specific topics include how you can share information with other people (including family, friends, and your child as he or she grows up) and whether you and your medical team may consider further testing (including genetic tests). Depending on your child's specific condition and the healthcare team who takes care of your child, parents can be asked to help make treatment decisions. When that happens, we want you to know you are not alone. Your child's medical team is there to help, and we are here to help by giving you information and helping you think carefully through different options. There are different sets of questions throughout this guide to help you.

We are glad you have found us. Having a child, whom you love and cherish, with a difference that you did not expect can bring many questions and concerns. We are here to help answer questions you might have.



Our goal

As you learn more about your child's condition, we hope you will have time to gather information, ask questions, and learn about possible options. Learning more about the condition, how it's managed, and considering other factors that are important to you and your family (like your beliefs and values) will help when you take part in medical discussions about your child's care. Each child is different, and you play an important role in deciding what is right for your child. Our goal, like yours as a parent, is to help set your child on a path towards becoming a well-adjusted adult in the best way we know how.

How to use this guide

This guide will take you on a journey and provide information about your child's condition and decisions that need to be made.



- You do not need to read or understand everything all at once. It is here for you to revisit.
- You don't need to do or decide anything now.
- Take your baby home and hold them close. Care for them, hold them, talk to them, play with them, read to them, and tell them often that you love them. That is the first step in raising confident and happy children.

And, when you're ready, you can start here:



"Why does my baby's body look different?" gives an overview of different development paths and how they can happen



"Early Medical evaluations" helps you learn about important parts of doctor visits



Your child's characteristics or diagnosis can provide clues about how to make decisions; see the **Information sheets** on diagnoses and clinical management



Families address differences in how their child's body looks or works in different ways

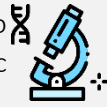


Parents who may be in situations like yours share their stories

The "My current understanding" questionnaire asks what you already know about your child's condition and how confident you feel



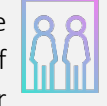
Tests can provide specific answers to questions that come up; "Genetic Testing" dives into specific concerns



"Support for families" explains the importance of having a support team



Some families and their healthcare teams may need to think about if their baby will be happier as a boy or a girl



Both your child's and your own well-being are very important



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Your Family Name: _____
Relationship to Patient: _____

Date: _____

My Current Understanding

Before you begin reading this guide, your child’s healthcare team has some questions designed to help us gain a better understanding of the kinds of things that may concern you. Please answer the following questions and bring them to your next clinic visit. You will be asked the same questions at the end of this guide to see how your understanding of your child’s condition and your confidence in its management may have changed over time.

What do you think about following statements?

1. Differences in Sex Development (DSD) are all conditions in which babies do not develop along the most common sex development pathways.

Strongly disagree

Strongly agree

2. Most DSD have a known genetic cause that can be found with genetic testing.

Strongly disagree

Strongly agree

3. DSD are usually not life-threatening conditions.

Strongly disagree

Strongly agree

4. In DSD, genital surgery is not essential for a child’s well-being.

Strongly disagree

Strongly agree

5. Genital surgery can be performed at any age, not just on babies or young children.

Strongly disagree

Strongly agree

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6. Genital surgery holds some risks.

Strongly disagree

Strongly agree

7. Children with DSD can question the feelings they have about themselves as boys or girls.

Strongly disagree

Strongly agree

8. People with DSD are just as likely as people without DSD to be attracted to members of the same sex.

Strongly disagree

Strongly agree

9. My child's DSD cannot be kept a secret from my child.

Strongly disagree

Strongly agree

10. Many people with DSD will be able to have biological children.

Strongly disagree

Strongly agree

11. When it is not clear if a baby should be reared as a boy or girl, the decision should be made through discussion between the family and the doctors.

Strongly disagree

Strongly agree

12. Discussion about a child's DSD spans over the course of their lifetime, regardless of if genital surgery is done early in life.

Strongly disagree

Strongly agree

My confidence in gathering information to make decisions for my child

As a parent, part of your job in caring for your child is working with medical professionals to make the best decisions for your child’s health. Please rate how certain or confident you are that you can do the things that we list below.

How certain are you that you can...

1. Face all the care decisions that might come along the way?
 Not at all A little Somewhat Quite a bit A great deal

2. Get all the information you need (e.g., searching for extra info on your own or with help) to make decisions?
 Not at all A little Somewhat Quite a bit A great deal

3. Learn all about the risks, harms, and benefits that matter to you most in these decisions?
 Not at all A little Somewhat Quite a bit A great deal

4. Figure out how involved you want to be in decisions?
 Not at all A little Somewhat Quite a bit A great deal

5. Work with your partner (your child’s other parent) to make decisions that you both agree on?
 Not at all A little Somewhat Quite a bit A great deal

6. Identify questions that you want to ask doctors?
 Not at all A little Somewhat Quite a bit A great deal

7. Talk to doctors about what matters most to you?
 Not at all A little Somewhat Quite a bit A great deal

8. Ask a doctor for more information if you don’t understand what he/she said?
 Not at all A little Somewhat Quite a bit A great deal

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9. Figure out what to do when you hear information that does not agree with the things you have already heard?

- Not at all A little Somewhat Quite a bit A great deal
-

10. Answer questions about how your child's body looks and works if family members ask you about it?

- Not at all A little Somewhat Quite a bit A great deal
-

11. Talk to your child about their body when your child grows up?

- Not at all A little Somewhat Quite a bit A great deal
-

12. Ask your doctor to put you in touch with families with the same or a similar diagnosis?

- Not at all A little Somewhat Quite a bit A great deal
-

13. Talk to your child about their body when your child grows up?

- Not at all A little Somewhat Quite a bit A great deal
-

Why does my baby's body look different?

Your child was born with a body that looks and works differently from other babies. This is because their sex development has been affected by one of many conditions, known as Differences of Sex Development (DSD). There are a lot of different words people use to describe these conditions. Some people will say “Disorder” of Sex Development. Others may say “Variation in Sex Characteristics (VSC)” or “Intersex.” Words like these are “umbrella terms” that can mean many different conditions. You may choose to use one term or another based on what you hear your child’s doctors use, or based on which you feel most comfortable with. Once you know the name of your child’s specific condition, you and your doctor will probably prefer to use that instead.

Babies can follow different sex development paths in the womb. Some paths are taken more often than others. All sex development paths lead to a combination of sex characteristics. These sex characteristics include, for example, sex chromosomes (X and Y) and how the genitals look.



Most Boys Typically Have

- XY Chromosomes
- A penis
- Testes and no uterus
- A lot of testosterone and a little estrogen

Most Girls Typically Have

- XX Chromosomes
- A vagina and clitoris
- Ovaries and a uterus
- A little testosterone and a lot of estrogen

When sex development takes a less common path, a person can have a different combination of sex characteristics.

Children with a condition called congenital adrenal hyperplasia (CAH) who have XX chromosomes typically have ovaries and a uterus. But because they produce a lot of testosterone, the clitoris may be larger and the vagina and urine tube may share an opening. Children with a condition called complete androgen insensitivity syndrome (CAIS) with XY chromosomes typically have testes. But because their bodies do not respond to testosterone, the genitals and reproductive structures develop as a clitoris and vagina. In general, the amount of testosterone that your baby produces, and can react to, will influence how the genitals look.

Variations of development

Sex development is a complex process and many combinations or variations are possible. Each of these variations can change the way your baby's body looks or works.

To clarify, sex development refers to the biological or physical sex characteristics of a person. These include the sex chromosomes, hormones, gonads (ovaries or testes), and genitals. These do not include gender or sexual orientation.



Why did my child take a different path?

There are many reasons why babies take different pathways in development. Sometimes, they involve genes. Genes are pieces of information in every cell of the body that serve as the instruction manual for how bodies develop. Changes in genes may sometimes cause unexpected combinations of sex characteristics to develop. They can also affect the way babies produce or respond to hormones. Sometimes, changes in genes are inherited from the biological parents. Other times, they occur randomly.

How common are these different paths?

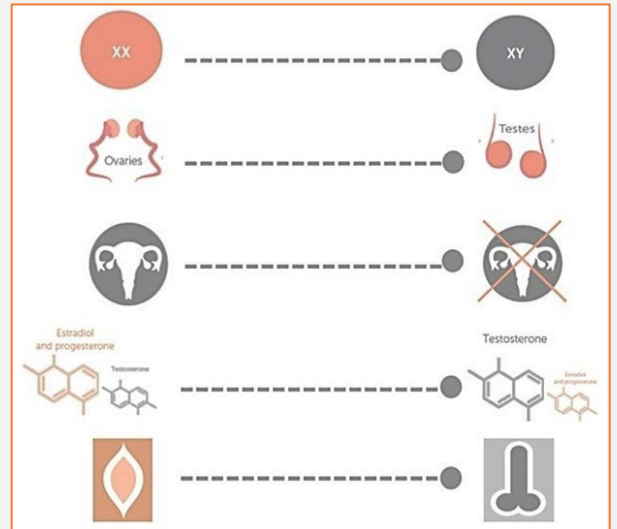
Some paths are relatively rare. Others are quite common. Although the changes these paths cause in a body are already present before birth, they can go unnoticed for months or years. Not all of these changes lead to visible differences in how a body looks.

Are these paths life-threatening?

Most changes in a baby's sex development do not affect general health, but some do. Children with one type of change may require different care than those who are affected by another type. That is why it is important to understand which condition (or "diagnosis") your child has.

Why are there so many different names for these conditions?

Terms like DSD or Intersex are general terms covering lots of different conditions. Some of these conditions have very different effects on health, and doctors use different words to describe this. Some doctors use the word "disorders" instead of "differences" of sex development. This is because differences that cause a change in the way a body works are often referred to as disorders.



Some people don't like the term "disorders" because it sounds like something is "wrong" with their child. Because of this, many people prefer the term Differences of Sex Development, or use the name of their child's specific condition instead.

Parent to Parent:

There are lots of different ways to describe these conditions. You can (and should!) ask your child's healthcare team to use the term that makes you feel most comfortable.

Sex development basics

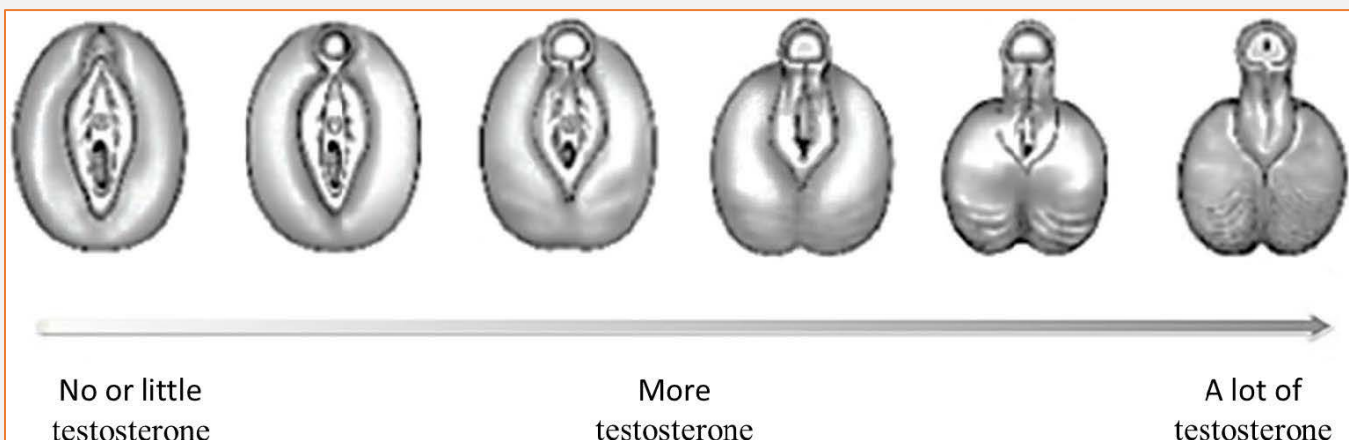
Why do some fetuses grow into typical girls, while some grow into typical boys?

1. Sex determination

The first step in the pathway of sex development is called “sex determination.” It lasts 5-6 weeks starting from when the egg (ovum) of the biological mother is fertilized by the sperm of the biological father.

Usually, the egg has one X chromosome and the sperm carries either an X or a Y chromosome. The fertilized egg (the embryo), can have either two Xs, or an X and Y. There could also be a combination (called “mosaicism”) – where some cells have XX while others have XY. Sometimes, there are 47 chromosomes (47, XXY) or only 45 chromosomes (45, X). Many combinations are possible.

During the first 5-6 weeks of growth in the uterus, all babies have the same internal and external sex structures. Part of the internal sex structures are called gonads. The gonads usually develop further into testes or ovaries which can produce sperm or eggs for reproduction later in life. The external sex structures are called the genitals. In some babies, they develop into a penis and scrotum (the pouch containing the testes). In other babies, they develop into a vulva, which includes the clitoris, the vagina, and the labia (any of the folds of skin bordering the vagina).



2. Sex differentiation

The next step is called “sex differentiation.” It starts at 6 weeks, when a cascade of genes found on the Y chromosome, including the SRY gene, signals the gonads to develop into testes. If these genes are missing, do not work, or there is no Y chromosome, then the baby will usually not grow testes, but will grow ovaries. Some gonads develop into ovotestes (where there are aspects of both testes and ovaries). Other gonads do not develop further (and are also called “streak gonads”).

Gonads differ in the hormones they produce in the womb. Testes produce a lot of testosterone, which typically guides the genitals to develop into a penis and scrotum. When there is no or little testosterone, or when testosterone cannot work, the genitals will develop into a clitoris, vagina, and labia. Remember, there is a big spectrum between “typically male” and “typically female,” and many variations of genitals are possible depending on the amount of testosterone to which the baby responds in the womb. Finally, testes also produce anti-Müllerian hormone (AMH). This AMH makes sure that there is no development of a uterus. Without AMH, a uterus usually develops.



Support for families

All children need love and support from their parents. In addition, children whose bodies work or look different need to be able to call on care from their doctors. A psychologist, counselor, psychiatrist, social worker, or nurse specialist can help you understand what the medical team is doing (diagnostic process) and how your child's biological and physical sex development affect his or her long-term care. It is essential to talk about your child's emotional well-being, as well as your own, now and in the future. There are many resources you can turn to who can give you support in dealing with tricky or stressful situations, such as how to manage information about your child's body, how to talk about it with other people and, most importantly, how you talk to your child about their body in the future.

The amount and type of information you may share with others (e.g., babysitters, childcare providers, and friends) will depend on many factors: your child's medical needs (e.g., if your child needs medication), your personal preferences and values, and what you and your child feel comfortable sharing. As your child grows older, it is important to let him/her decide as much as possible. One of the main challenges is respecting your child's privacy until he/she is ready to decide what can be shared. Waiting to share information can lead to feelings of isolation for new parents. For most parents, having trusted friends or family members who know about their child's condition can be helpful.

Although your own family and friends offer support in their own way, they, themselves, are not living with the same condition. Talking with other people who have a body like your child's or who are parenting a child with one may help with feelings that no one else understands what you are going through. If you feel like talking with other families, check with your doctors to see if they host family days, or if they can connect you with families or family groups that have had similar experiences. However, it is important to remember that every family may deal with things in very different ways.



There are lots of types of resources that can help provide you with support and education as you navigate your baby's health care. Some of these resources can include:

- Psychologists
 - These are professionals who may or may not have a specific focus on children whose bodies look and work like your baby's. If your baby receives care from a multidisciplinary team, there may be a psychologist on the team who you can meet with. The psychologist can help you with understanding your child's diagnosis and what it means for your baby. They can also help answer questions you may have about your baby's future, or questions that come up during meetings with the healthcare team.
- Support and resource organizations and support groups
 - There are many organizations available for the families of children with conditions like your baby's. Many of these organizations have in-person meetings, while others may be online or on social media. This can be a valuable way to connect with other parents who are having a similar experience, as well as parents of older children with your child's condition.
- Advocacy organizations
 - Advocacy organizations are groups that promote a specific cause. There are several advocacy organizations related to conditions like your baby's that focus on different issues. These groups can give you different perspectives on common experiences in people with bodies like your baby's. You may choose to become involved in an advocacy organization, or just use it as a way to get more information.
- Support and counseling from another source
 - Most families have their own set of values and beliefs that are very important to them. For some families, this involves their religion. In this case, you might benefit from talking to a religious figure who you trust, such as your pastor or rabbi. Even if they are not providing medical advice for your baby, it can help to have someone to talk to who prioritizes your family's values and who can work with you to feel ready to discuss them with your healthcare team.

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Sharing information with others – psychologists, friends, family, and support groups – may give you more time to get used to your child’s body and more confidence to share this information with your child later. How much you share is always up to you. Not all information has to be shared at once. Educating children about their bodies is a lifelong process and their questions should be answered in ways that they can understand during each period of development. This helps them grow up with a healthy understanding of their body and what to expect from it. If you are comfortable and confident talking about this, then your child might be, too.



**Check out “Questions to Ask” at the end of this section for a list of questions you can ask about Support or Advocacy Organizations, and sharing with others.*

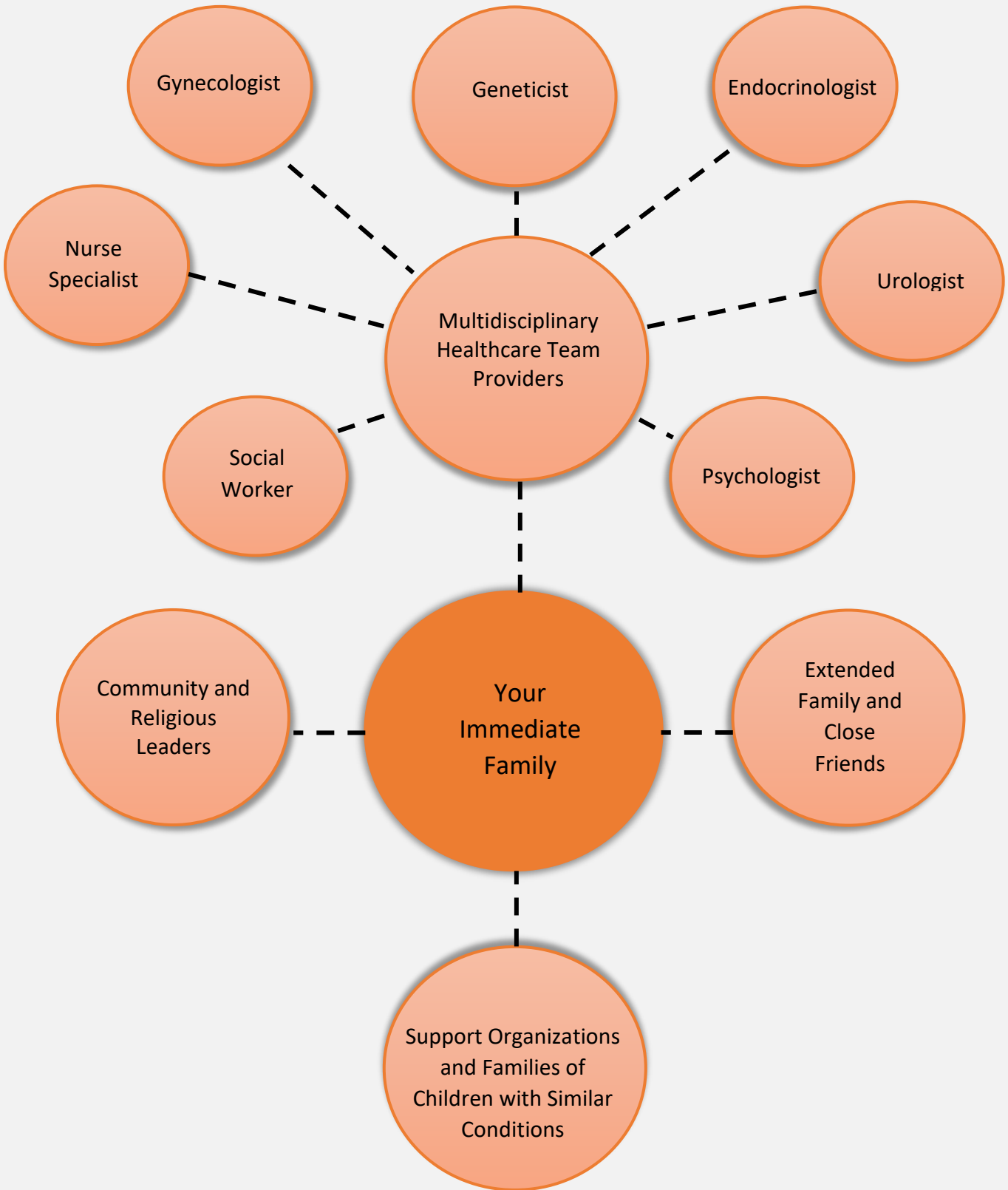


Figure 1: People who could be involved in your support

Questions to ask

Before going to your child's appointment, think about questions you might want to ask your child's healthcare team. The following are some questions parents have asked in the past about information and support, and that you may want to consider asking. Read through these and check which ones you do not have answers to yet. Then, bring this list with you to clinic so you can be sure to ask the team when you get there.

The answers to some of these questions and the questions you choose to ask may change over time as your child grows and develops.

About support groups...

- How do I find support groups?
- What is the purpose of support groups?
- Are clinicians involved in support groups? If so, how are they involved?

About talking to others with a similar diagnosis...

- What can I learn from the parents of children who have the same or a similar diagnosis as my child's?
- What can I learn from talking to adults with the same or a similar diagnosis?

About advocacy groups...

- What can advocacy groups do for my child or for me?
- Who is involved in advocacy groups?

About counseling...

- Do you recommend that my family or my child receive psychological counseling?

About privacy and sharing information...

- What should I do if I am uncomfortable with the way my child's condition is described during appointments or in the medical record?
- Are there some examples of how I can talk to family and friends about my child's condition?
- What is the right age to start telling my child about their condition?
- What should I tell my other children?

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Your Family Name: _____
Relationship to Patient: _____

Date: _____

Sharing information with others and reaching out for support

This next section is a set of questions to help you and your healthcare team think about how you usually like to share personal matters with people you are close to. After reading the “Support for Families” section, show how you feel about the following statements by filling in the bubble that best describes how you are feeling (closer to the statement means you agree more with what is being said). Please consider bringing this with you to your next clinic visit.

Example question:

Reasons to carpool to work

I enjoy talking to others during my commute

○ ○ ○ ○ ○ ○ ●

Reason not to carpool to work

I enjoy quiet and solitude during my commute

The example above shows that the person who answered this question usually enjoys quiet and solitude during their commute.

How do you feel about sharing information with others?

Reasons to not share with others

I generally do not like to share information with others about my family’s health.

○ ○ ○ ○ ○ ○ ○ ○

Reasons to share with others

I generally like to share information with others about my family’s health.

I am worried about how others will react if I share information with them.

○ ○ ○ ○ ○ ○ ○ ○

I am not worried about others’ reactions if I share information with them.

My culture has an influence on my decision to share information with others.

○ ○ ○ ○ ○ ○ ○ ○

My culture is not a factor in my decision to share information with others.

I worry that my child will be treated differently because of the appearance of his/her genitals

○ ○ ○ ○ ○ ○ ○ ○

I do not worry that my child will be treated differently because of the appearance of his/her genitals

How do you feel about sharing information with others?

Reasons to not share with others

Reasons to share with others

I generally keep things to myself and find information on my own.



I generally reach out to others for support and information.

The next questions will help show how you are feeling about sharing information now and will help your health team support you. The answers you pick now may change over time as your and your child's needs change.

Do you think it would be helpful to share information about how your child's body looks and works with the following people?

1. Close family

- Not at all A little Somewhat Very

2. Extended family

- Not at all A little Somewhat Very

3. Close friends

- Not at all A little Somewhat Very

4. Co-workers

- Not at all A little Somewhat Very

5. Daycare providers/babysitters/teachers

- Not at all A little Somewhat Very
-

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The following questions ask how you are feeling now. Your answers will probably change over time.

How much are you interested in...

1. Talking with parents of children who have a similar condition?

Not at all

A little

Somewhat

Very

2. Talking with adults who have a similar condition?

Not at all

A little

Somewhat

Very

3. Visiting a support group website?

Not at all

A little

Somewhat

Very

4. Visiting an advocacy organization's website?

Not at all

A little

Somewhat

Very

Early medical evaluation

A diagnosis requires a team of professionals and several tests

When doctors evaluate your child, their priority is the health of your child. Some babies will need to stay in the hospital for a few days to check for underlying health conditions that may have a life-long impact.

Having a team of doctors with experience caring for children and families affected by a condition like your baby's will help you feel confident that your family is receiving expert care. Usually, these teams are based in specialized centers and can arrange for more specific tests to find the cause of your baby's condition. These teams are also multidisciplinary, including specialists from various professional groups. Usually, there are specialists in endocrinology (hormones), urology/gynecology (kidneys, bladders, genitals, and internal reproductive organs), and psychology (emotional well-being). Other team members usually include geneticists and genetic counselors, nurses and nurse specialists, social workers (who can help with emotional, financial, and social matters), and child life specialists.

Genital/physical exams

Your baby should be examined only by medical personnel who are directly involved in your child's care. Whenever possible, all specialists involved in the assessment and management (e.g., pediatric endocrinologist, urologist, geneticist, and pediatrician) should be present at the same time and do the physical/genital exam together. This will minimize the number of exams required. In addition, the number of people present during the exam should be kept to a minimum, to avoid overwhelming



your child and your family. If any teaching of medical students or residents is planned, your permission should be sought before the exam. Similarly, if medical photography is planned, your informed consent must be obtained beforehand.

What can you do?



If you do not want teaching or medical students attending, before the exams, explain that you would prefer to have only the people who are directly involved in your child's care present.

You can be there during the exams and are encouraged to ask questions. The doctors should describe their findings to you and to other medical professionals in attendance (with the caveat that information and answers may be limited before a definitive diagnosis is made) in clear, easy-to-understand words. At the very least, privacy curtains or screens should be drawn and the exam should be performed in a closed room or other private space.

If you give permission for medical photography, let the doctor explain again the privacy and confidentiality rules related to these photos, for what purposes photographs will be used, and how they will be stored.

**Check out "Questions to Ask" at the end of this section to find questions you can ask your healthcare providers regarding Physical or Genital Exams.*

Thinking ahead...

1. Toddler and childhood years

For children who are old enough to be aware of an examination of their genitals, this can be perceived as an invasion of privacy and others not respecting their boundaries. Thus,

even greater care should be taken to minimize the number of exams performed and number of doctors present in the room

What can you do?

You can explain to your child in advance that the doctor may want to look at his/her genitals (use your preferred words and communicate them with the doctors), and that it will be okay because you (or another parent or family member) will be there the whole time. A child who is old enough to understand what is going on will also be asked by the doctor for his/her permission to participate in exams (called “assent”). Doctors will never force an exam against your child’s wishes. When the exam has been agreed to, you can help your child undress and put on an examining gown or perhaps a piece of clothing brought from home, such as a large T-shirt, so that your child is not fully naked during the exam. Some toddlers and young children may feel more comfortable if they are examined while seated on a parent’s lap, rather than on an examination table.

If you think this approach might make your child feel safer during the exam, suggest this to the doctor(s). Ask them to explain to your child what is about to happen before they touch your child, and provide reassurance to your child that the examination will stop if requested. As a parent, you can help your child feel positive about the experience by showing confidence in interactions with the doctors and providing reassurance to your child during the process.

Generally, the results of the exam will not be discussed with your child in the exam room because your child may not have a clear understanding of his/her condition. For this reason, it is often helpful to have someone in your family or a close friend attend the appointment. That way your child can play in the waiting area while you discuss findings and implications with the doctor.

2. Teenage years

Teens start to have an understanding of the associations between genitals and sexual function and may be experiencing their own increasing sense of sexuality. This can make the process of genital exams feel uncomfortable.

What can you do?

Ask the doctor before your upcoming appointment whether your teen needs a genital exam, so that he/she can discuss any fears or concerns with you and be

prepared. Some teens feel more comfortable being examined by a doctor of the same gender, so it may be helpful for you to communicate this with the healthcare team, if this is what your teen wishes. Your child's consent for examination is required, not just yours. Your child can also choose not to have you in the room. Another medical professional (generally of the same sex as your teen) might then be present as a chaperone. Your child should be encouraged to tell the physician if they feel discomfort, have questions, or wish the doctor to stop the examination. After the exam, the doctor should share the results with your teen. As parents, you may want to consider allowing your teenager to choose how much information he/she shares with you about the completed exams, as long as the information is not medically critical.

Before your appointment

Your child's healthcare team is likely to ask you a number of questions. It can help to have answers already prepared before you go to the appointment.

- What are your child's symptoms?
- When did you first begin noticing these symptoms?
- Are you experiencing any problems related to your child's DSD?
- Has anyone in your family been diagnosed with a DSD?
- Are you planning to have more children?

For appointments as your child gets older, there may also be questions directed to your child. It might help to talk about some of these things with your child before the appointment, so that they are prepared to answer. Some children might be shy and not feel comfortable answering questions themselves. In that case, talking with your child beforehand can help you get a sense of how your child is feeling so that you can share it with your healthcare team.

- Are you satisfied with your treatment?
- Are you happy with the changes to your body?
- Are you having any problems at school or with other kids because your body looks or works differently?

Questions to ask

Before going to your child's appointment, you should consider questions you might want to ask your child's healthcare team. The following are some questions parents have asked in the past, and that you may want to consider asking. Read through these and check which ones you do not have answers to yet. Then, bring this list with you to clinic so you can be sure to ask the team when you get there.

The answers to some of these questions and the questions you choose to ask may change over time as your child grows and develops.

About a diagnosis...

- What is likely causing my child's signs and symptoms?
- What is causing my child to look or act the way he or she does?
- Are there any other possible causes for these symptoms?
- Is my child at risk for other medical conditions because of the way their body looks or works?
- How do I get a second opinion?
- Do you recommend that our family meet with a genetic counselor?

About physical exams...

- How many people will be in the room for a physical exam?
- How often does my child need to have this kind of exam? How long should we wait between exams?
- Can this exam be postponed?
- Will the results of this genital exam change my child's treatment?
- How do I prepare my child?

About medical photography...

- Should I consent to medical photography of my child's genitals?
- If pictures are taken for treatment purposes, where are they stored? Who will have access?
- Will my child's genital photographs be used in case conferences or educational presentations? Will I be informed?

Testing

Medical care often involves different types of tests and evaluations. To diagnose and manage a medical condition that affects your baby's sex development, doctors use different types of tests including checking genetic information and what type and levels of hormones your child's body is producing. Others look at how the internal sex organs (such as the uterus and gonads) are formed and how the external genitals might continue to develop. Some of these assessments can be important for predicting your child's response to future hormone treatment. Talking to you will help the team learn how you think and feel about some of the challenges involved with raising a baby whose body looks or works differently.

Genetic testing

The genetic puzzle

Because there is no one genetic test that can explain everything that can cause a baby's body to look or work differently, you might be offered two or more genetic tests by your usual team or during a separate visit to a genetics team.

Things to consider

Genetic testing is helpful in diagnosing a specific condition. For some parents, learning about chromosome patterns that are less common can be difficult.

With any genetic testing, there is also the risk of finding other things about your child's genetic make-up that are not helpful to know. These are called incidental findings. There are lots of different types of incidental findings, and they may be related to your child's sex development or not. Some examples of these findings can be found in Appendix A.

Genetic tests

Usually, before any test, the team will ask you about your child's medical history. The doctor can use this family health history to become familiar with the conditions that may run in the family. The more people in the family there are with the same condition, the higher the chance that genes are involved in causing the condition. But, even if there is

no family history, there could still be a genetic cause. Sometimes, a different path in sex development is caused by changes in genetic material that are not inherited.

Your child's doctors might recommend and order one, a few, or many different genetic tests. Some of these can be complicated. As parents, you do not have to know all the details about every single one. And your child's healthcare team (especially the geneticists and genetic counselors) can explain each one to you. Also, we are providing more detailed information about some of the tests you might be offered for your child in this section starting with the more common ones. This way, you have access to the information if and when you need it.

1. Chromosome Pattern Test

In genetic testing, doctors will typically begin by checking your child's chromosomes (or karyotype). Each person usually has 46 chromosomes or 23 chromosome pairs, including one pair of sex chromosomes, XX or XY. This information is written down as 46, XX or 46, XY.

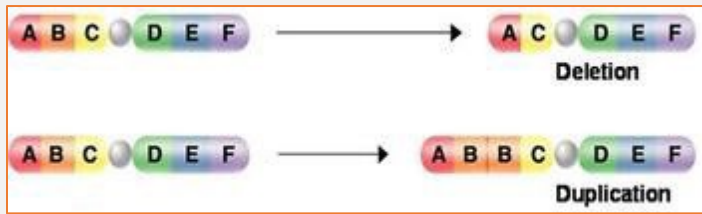
Sometimes, there is an atypical number of chromosomes. For instance, there could be 45 chromosomes (if there is only one X, this is written as 45, X). Sometimes, there are 47 chromosomes (if there is an extra X chromosome, this is written as 47, XXY). Other people have combinations of sex chromosomes that differ from cell to cell. This is called "mosaicism." For instance, some cells of the body might have 46, XX and others 46, XY or some cells may have 45, X and others 46, XY. Many combinations are possible.



2. FISH Test

A test that is commonly performed together with the chromosome pattern test is the FISH (Fluorescence in situ hybridization) test. It can be used to check whether a specific gene linked to sex determination (the SRY gene) is present and if it works.

3. Chromosomal Microarray

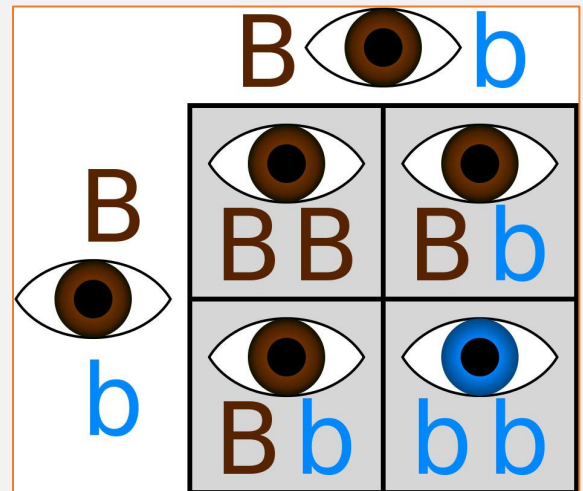


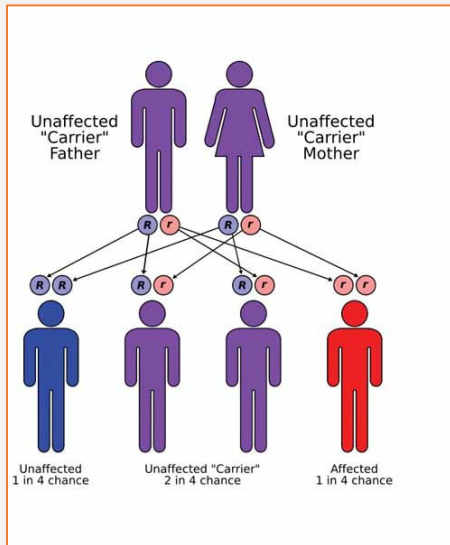
A Chromosomal Microarray Analysis (CMA) looks at the chromosomes in a more detailed way, via a blood sample, and the results from testing take roughly 3-4 weeks. CMA uses a gene chip (a

“microarray”) to look for imbalances in the amount of chromosomal material, by comparing your child with a “control” group (a group of individuals with no known genetic anomalies). If a difference is found, the test can show where the difference is located on the chromosomes and what the reason is for that difference. Sometimes, a small amount of material is missing (deleted) from a chromosome (called microdeletion). Sometimes, a small amount of material is present in an extra copy (duplicated) on a chromosome (called microduplication). Too much or too little chromosomal material can lead to differences in the overall development of a child, such as physical and mental differences, including developmental delay and autism.

4. Targeted Gene & Carrier Testing

Targeted gene testing helps determine if there are changes in certain specific “targeted” genes (and their sequences) that influence sex development (such as SRY, SOX9, SF1, WT1, and many more). We all carry two copies of each of these genes, one from our biological mother and one from our biological father. Some conditions require only one of these two gene copies to be affected by unusual changes in order for someone to have a different sex development path. This is called a “dominant” condition. Other conditions require both copies to be affected. This is called a “recessive” condition. For this to happen, both the biological mother and father must have an affected copy and each transmits that affected copy to their baby. It is also possible that only the mother or only the father passed on an affected copy. In this case, the biological mother or father is called a “carrier.”





Being a carrier means that people do not have a genetic condition themselves, but have the ability to pass on the affected gene to their offspring. Carrier testing can tell whether the child and other biological family members (e.g. brothers, sisters, parents) “carry” a certain targeted gene change. This may have medical consequences for these family members. If planning future pregnancies and both partners are carriers for the same recessive condition, they have a 1 in 4 or 25% chance of having an affected child in each pregnancy. The American College of Medical Genetics currently does not support carrier testing in children. If tests find changes in these targeted

genes, the results may not predict how mild or severe your child’s (or other biological family members’) health problems will be. If they did not find changes in targeted genes, they rule out some important diagnoses, but do not rule out all conditions affecting sex development caused by changes in other genes.

5. Whole Exome Sequencing (WES)

A person’s collection of genetic material (genome), is their own personal blueprint for life. Whole Exome Sequencing (WES) only screens a person’s exome (1% of the genome). Compared to other genetic tests, WES looks at more genes and has a higher chance of finding a genetic cause for your child’s condition if previous tests failed to find one. However, 75% of patients do not receive a specific diagnosis explaining why their body looks or works differently from WES. The test is usually done via a child’s blood sample, and other biological family members can also be tested. Results from testing take roughly 4-6 months. The WES test will compare your child’s exome (or that of other family members) with that of a “control” group (a group of individuals with no known genetic disorders or abnormalities), and try to detect differences. Sometimes, these differences can be the cause of your child’s body looking or working differently. It is possible that WES will not find gene differences that are actually present. WES is a new and expensive genetic test. Not all health insurance companies will pay for the cost of the test. All WES tests will be discussed with your insurance company before testing. If your insurance does not cover the test, it could cost several thousand dollars.

Incidental findings from genetic testing

Sometimes, genetic testing might reveal things about your child's genetic makeup that you did not want or expect to know. These are a few examples of situations that could come from genetic testing of you or your child:

- A test might reveal that your child carries a gene for a disorder. This can raise questions about how to manage your child's condition, who else in the family might have these genes, and how to share information with other family members.
- A test might pick up other changes in chromosomal material that we do not have much medical information about. This can make it difficult to predict what type of other problems may or may not develop in your child.
- If the child's parents are also undergoing genetic testing, this may reveal that the biological parents are related (called consanguinity), or reveal information about family relationships. For example, the test might show that the father is not the biological father of the child.
- Genetic testing may also not detect a cause for the way your child's body looks or works, or change the treatment for your child.

Hormones

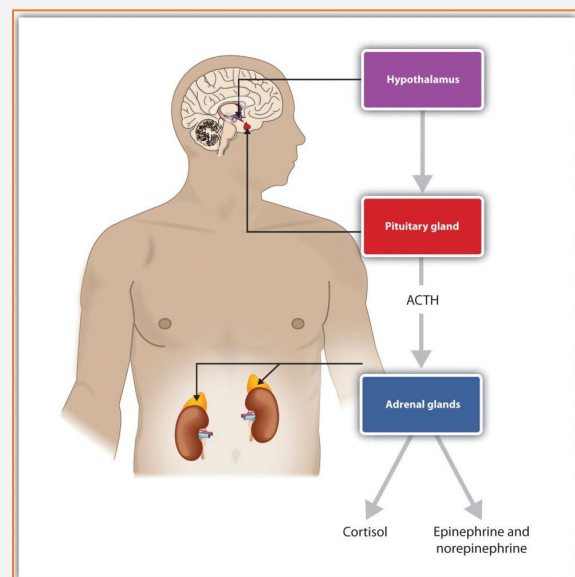
Hormones are chemical messengers in the blood that alert different parts of the body to perform certain actions. Your doctor will want to know what hormones your child's body is producing and how much of those hormones are present. Hormone evaluations can help your doctor come to a diagnosis, determine if your child is at risk of becoming unwell (such as due to salt wasting in certain types of CAH), and help make decisions about managing your child's health. The body's control of hormones is called the "endocrine system."

How does the endocrine system work?

Most hormone tests will assess a part of the hypothalamic-pituitary-adrenal (HPA) axis, which is a major part of the neuroendocrine system that controls reactions to stress and regulates many body processes. These include digestion, immune system functioning, mood and emotions, sexuality, energy storage, and energy use.

The HPA axis is named after its three components: the hypothalamus, the pituitary gland, and the adrenal glands. The hypothalamus regulates the body and brain and is considered the control center for the body's automatic responses. When excited, the hypothalamus activates the pituitary gland, which releases corticotrophin-releasing hormone (CRH) to activate the pituitary gland.

The pituitary gland, often referred to as the "master gland," is responsible for controlling the hormones in the body. This pea-sized part of the brain releases hormones to stimulate other areas of the body including gonadotropin and growth hormones that regulate growth, sexual development, and reproduction processes. The pituitary gland also releases the adrenocorticotropic hormone (ACTH) to trigger the adrenal glands. The adrenal glands sit on top of the kidneys, like little hats. The kidneys filter blood and are in the center of the body. This is an ideal location for a hormone that needs to affect the entire body, because all blood passes through the kidneys. The adrenal glands release several hormones including cortisol and aldosterone. These hormones are important for



regulating the body's physical stress response including heart rate, blood pressure, breathing rate, and salt balance.

Hormone tests: How are hormones evaluated?

Hormones are usually tested by taking a blood sample. Some hormone levels are looked at as part of a standard newborn screen. Depending on the test, your child may need to stop taking certain medications (e.g. if they contain steroids).

There is a small risk for a child of having blood taken. Veins and arteries vary in size and taking blood from some children may be more difficult than from others. Other risks of having blood drawn are small but may include: excessive bleeding, fainting or feeling light-headed (more frequent in adults than young children), blood accumulating under the skin (hematoma) and/or infection (a slight risk any time the skin is broken).

Just like genetic tests, as parents, you do not have to know all the details about every single hormone and endocrine test. Your child's healthcare team (especially the endocrinologists) can explain each one to you. We are providing more detailed information in this section so that you have access to the information when you need it.

1. Standard evaluation

There are several hormones that are important in sex development. Testing levels of these hormones early in treatment or evaluation can give doctors an idea of what conditions your child might have:

- 17-hydroxyprogesterone- a building block for cortisol, the body's "stress" hormone.
- Testosterone- a hormone commonly associated with male sex development but present in everyone. A person's level of testosterone is important in sex development.
- Gonadotropins- a term for several hormones that are produced in the pituitary gland and act on the gonads (ovaries or testes). The levels of these hormones can be used to predict how the body will develop at puberty.
- Anti-müllerian hormone- plays a role in inhibiting the development of the Müllerian ducts early in development. Later in development, the Müllerian ducts can develop into fallopian tubes, a uterus, the uterine cervix, and the upper regions of the vagina.

- Serum electrolytes- function in keeping water flowing in and out of cells and in nerve impulses. Sometimes referred to as salts, they include sodium, potassium, chloride, and bicarbonate.

By looking at the levels of these hormones (and any others that were looked at), your doctor will be able to make some determinations about how your child's body is functioning and developing. In some cases, it may be necessary to perform more intensive hormonal evaluation through stimulation tests. The three stimulation tests often performed as part of DSD care are: ACTH, hCG, and GnRH stimulation tests.

2. ACTH Stimulation Test

In this test, synthetic ACTH is given to your child to stimulate the adrenal glands to produce cortisol (the stress and energy hormone) and aldosterone (the salt balance and blood pressure hormone). If there is not enough cortisol and aldosterone, your child may show signs of weakness, poor feeding, poor weight gain, and low blood pressure. If not treated medically, this can be life-threatening.

How is the test performed?

Your child's blood will be drawn at the beginning of the test, and the synthetic ACTH will be given. About 90 minutes later, blood will be drawn again. Longer versions of the test are also available, but are rarely performed. Because of the small size of babies and children, sometimes blood cannot be obtained at all time points. In some babies, the synthetic ACTH may lead to a temporary rash, high blood pressure, and lower/higher heart rate. Usually, something called a "Heparin Lock" will be used to avoid repeated needle sticks. You will typically get the results for the cortisol level back in one day. It can take one to two weeks for the other results. From testing cortisol levels, the endocrinologist (hormone doctor) will be able to determine if the adrenal glands and pituitary gland are working properly. They can probably tell you if there is an appropriate increase in cortisol after the synthetic ACTH is given or if there is no increase in your child's cortisol level. If there is no increase, more testing may be needed to make a diagnosis and to specify what kind of hormone replacement is necessary.

3. hCG Stimulation Test

If your child has testes, during the first three months of life, your child's testes can produce large amounts of testosterone naturally as part of a "mini-puberty." After three months, testosterone levels begin to decrease to the low levels seen throughout

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childhood. If your baby is being evaluated after this early peak of testosterone has passed, specific stimulation tests may be used.

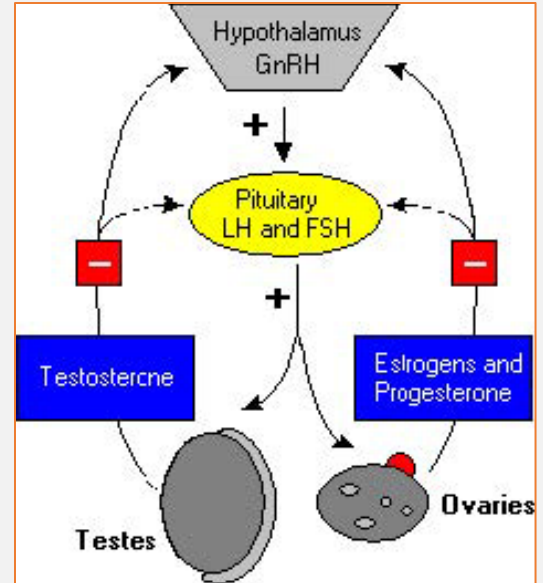
One way to stimulate the testes to produce testosterone is to give your child a synthetic hormone called hCG (short for Human Chorionic Gonadotropin), administered in a series of injections.

How is the test performed?

On day one, blood is drawn to measure the levels for testosterone and other androgens associated with testosterone. Then hCG is administered by a shot into a muscle in the arm, leg, or bottom. Two more injections, on days 2 and 3, are usually given to stimulate the testes. These shots can be done at the endocrine clinic or at your child's primary care provider's office. A second blood draw is done after the third shot, and again 24 hours later to see how much testosterone and other androgens have been made.

There are many variations of this test. This test may cause fussiness and an increased number of erections (if an erection lasts longer than 4 hours, the baby/child needs to be taken to the emergency department immediately). Usually, the test results will be back in a week and could show that your child's testes produce normal, low, or no levels of testosterone. The additional blood tests assessing other androgens may also help in determining the specific diagnosis. For example, some children produce normal testosterone, but have problems converting it to other androgens, which are also necessary for the development of the male external genitals.

Other children may produce normal testosterone, but the cells in their bodies (called receptors) are not able to respond to these hormones. There is a key, but it does not fit in the lock, because the lock has changed. Usually, doctors will recommend genetic testing to study the specific characteristics and structure of the lock.



4. GnRH Stimulation Test

GnRH (short for Gonadotropin-Releasing Hormone), is produced in the hypothalamus and signals the pituitary gland to produce Luteinizing Hormone (LH) and Follicle Stimulating Hormone (FSH). In males, LH stimulates the testes to produce testosterone, and FSH supports the maturation of sperm. In females, FSH and LH stimulate the ovaries to produce estrogen and progesterone and regulate the menstrual cycle.

How is the test performed?

In a GnRH stimulation test, synthetic GnRH is given as a shot to make the pituitary gland release LH and FSH. Before the test, your child may need to stop taking certain medications. Your child's blood will be drawn at the beginning of the test, and the synthetic GnRH will be given. About 90 minutes later, blood will be drawn again to measure LH and FSH. Longer versions of the test are also available but are rarely performed. Because of the small size of babies and children, sometimes blood cannot be obtained at all time points. Results are usually available in a couple of days. "Hypogonadism" occurs when the gonads (testes or ovaries) produce little or no hormones. This can be caused by some different sex developmental pathways, but it can also be caused by tumors. When hypogonadism centers on the testes or ovaries, it is called primary hypogonadism. When it centers in the pituitary gland and hypothalamus areas of the brain, it is called central or secondary hypogonadism. To treat hypogonadism, your doctor needs to know if it is primary or secondary.

When the LH response is higher than normal, it may indicate primary hypogonadism. When the response is too low, it may indicate secondary hypogonadism. Doctors may suggest an MRI scan of your child's brain and further genetic testing. Sometimes, your child can produce enough LH, but the body does not respond to it (there is a key, but it doesn't fit into the lock), and genetic tests can be used to find the problem with the LH "lock" (called a receptor). These and other tests may also be important to determine which type of hormone treatment your child may need to start puberty later in life.

Learning about your child's body, inside and out

External and internal sex anatomy

Here we explain some of the exams that doctors use when trying to define how your child's external genitals and internal sex organs have developed before birth and are developing over time with or without medical treatment and/or genital surgery. The results of these exams can be difficult to interpret.

Doctors will usually do an external genital exam first. They will measure how the genitals have developed, investigate whether there are testes and where they might be located (in the genital area, groin area, or in the belly), and check the location of the tube that carries urine from the bladder to the outside of the body (urethra). Sometimes in babies with a vagina, the urethra and vagina are together with one opening, called a urogenital sinus. In babies with a penis, but somewhere on the shaft or at the base of the penis. This is called hypospadias.

Doctors will also check for a uterus (womb) and whether your child has functioning ovaries, testes, or ovotestes (a combination of both ovaries and testes). Ultrasound tests and scans (e.g. MRI, genitogram) may sometimes show these internal sex organs, but the results of these scans are usually difficult to interpret. Another way of looking is through small incision in the skin of the belly, and passing a small "telescope" (laparoscope) through those incisions. Sometimes, a sample of tissue (biopsy) is taken for examination in a laboratory. Doctors may also suggest looking at the bladder and vagina with a special "telescope" (cystoscope). Your child will be anesthetized (medically asleep) for this procedure.

Checking the external genitals

Usually, the doctor will measure the length and width of your child's penis or clitoris, check how the labia/scrotum have developed, investigate whether there are testes and where they are located (in the genital area or groin area), and check the location of the urine tube and vaginal opening (if they are fused together, it is called a "urogenital sinus"). A genital exam can be important to predict the responses of your child's body to medical treatment (for instance, the size of the clitoris can sometimes become smaller, or the size of the penis larger, with hormone therapy) and will possibly also give you and

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the healthcare team the ability to measure any changes with treatment. Because some exams are more thorough (e.g. exams of the genitals and bladder/bowel), and can cause physical and psychological distress for babies and children (for instance if they need to lay still for some time), doctors may offer to do these exams under anesthesia (the medicine that prevents any immediate discomfort or sensation during the exam).

Anesthesia

The FDA warns against the use of repeated and/or lengthy anesthesia (over three hours), when not medically necessary, in children under 3 years old because it may affect brain development. If your child has previous anesthesia exposure and is under 3 years old, or if you are concerned, it is important to talk with your doctor about the harms and benefits of performing an exam under anesthesia. It may be helpful to ask if this exam can be combined with another procedure that your child may need anesthesia for (for example, a cystoscopy or laparoscopy).

When using anesthesia, there may be short term side-effects, such as drowsiness or disorientation after the exam, and your child may be restless or cry. Many parents worry that their child is in pain, but that is usually not the case. Generally, your child is reacting to the anesthesia wearing off. Crying may actually assist in removing the anesthesia from your child's system. The best thing you can do is try to comfort your child and wait. If your child is hungry, you may offer clear liquids. Breast fed infants may be given breast milk.



Checking the internal sex organs

1. Pelvic ultrasound

This imaging test is very similar to the ultrasounds that women receive during pregnancy. It produces pictures of the inside of the body using sound waves. An ultrasound is performed by first putting a gel on your child's lower belly and then using an ultrasound probe to touch your child's belly. Ultrasound images can show the structure and movement of your child's internal organs, as well as blood flowing through blood vessels. The key to getting useful information is to have this test performed and interpreted by a

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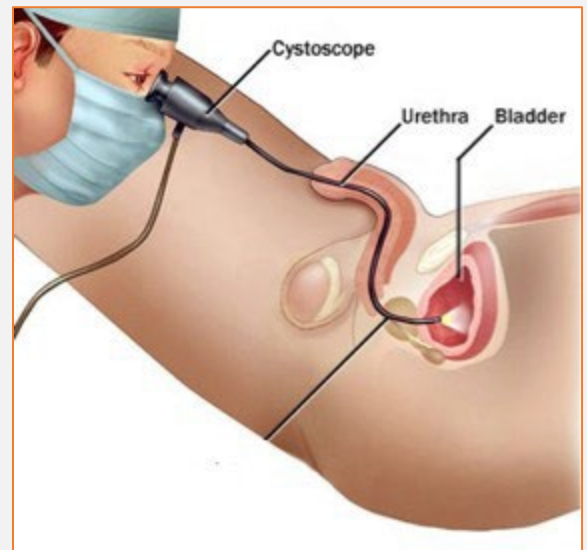
professional with experience in the evaluation of babies and children. Ultrasound examinations do not use radiation and are safe for use in children. Because the uterus is very small before puberty, it may be difficult to see on an ultrasound scan.

2. Magnetic Resonance Imaging (MRI)

MRIs give more information than pelvic ultrasounds about your child's internal (reproductive) organs and may better identify any problems that may exist. However, like in pelvic ultrasounds, MRI's can be difficult to 'read' when the child is very small. Because of this, MRI's are more often used in older children, but might be used on small children in certain circumstances. Sometimes, a contrast dye might be injected to get a better picture of some specific areas of your child's lower belly. This sometimes causes allergic reactions (such as rashes, hives, nausea, flushing, and dizziness). Some babies may have general anesthesia when they have an MRI so that they do not move during the test. In other babies, a sedative will help to calm them, but will not completely put them to sleep. To have an MRI, your child will be placed in a large scanning device and must be very still so that the MRI machine can make a good picture. The machine can be noisy, which might unsettle your child. The exam usually takes 20-30 minutes to complete. Your child's doctor will be able to explain why an MRI might be useful, and can help you prepare for the process.

3. Cystoscopy and vaginoscopy

Doctors can insert a special "telescope" into the bladder and opening of the vagina or urogenital sinus (cystoscope for the bladder, and vaginoscope for the vagina and urogenital sinus) to get a clear image of the insides of these structures. The "scopes" can also take pictures. Sometimes, there are under-developed sex structures with no real function (called remnants or remains), which might lead to infection or internal obstruction. Often, however, these structures are very small, will not cause problems, and can be left in place. Depending on their formation, your child's doctor may recommend having them removed if they risk developing problems later.



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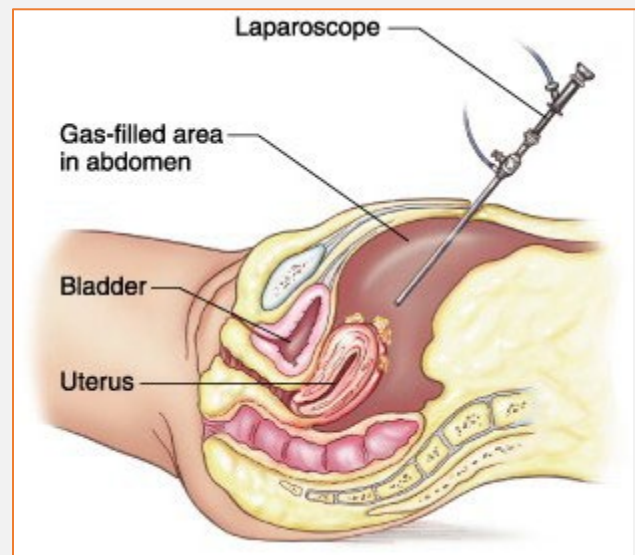
Your child will be given general anesthesia (which will help your child sleep during the procedure). Some children might have a reaction to the anesthesia. When performed by a pediatric urologist/gynecologist (or similarly trained surgeon), these procedures are usually relatively quick and harmless.

Usually, doctors will prescribe antibiotics for your child to take before and after the scopes to prevent infection developing. Very rarely, scopes may cause some bleeding because of damage to the blood vessels or pain because of damage to the bowel, bladder, and nerves.

4. Laparoscopy, including gonadal biopsy

Sometimes, it will be helpful to test a small sample of your child's gonad(s) (biopsy). Small incisions (cuts) into the belly or groin area are made to insert a laparoscope (a special telescope) and other instruments necessary to perform the procedure.

Your child will be given general anesthesia, and his/her belly will be inflated with a carbon dioxide (CO₂) gas. To allow ventilation with the CO₂ in the belly, your child will also be intubated. In other words, a tube will be inserted into the child's airway to help them breathe. This may cause a sore throat. After the surgery, the incisions are closed with a stitch or glue.



Laparoscopy can further help doctors to determine the specific diagnosis and provide some indication of future fertility of your child. The risks of laparoscopy are small, but include:

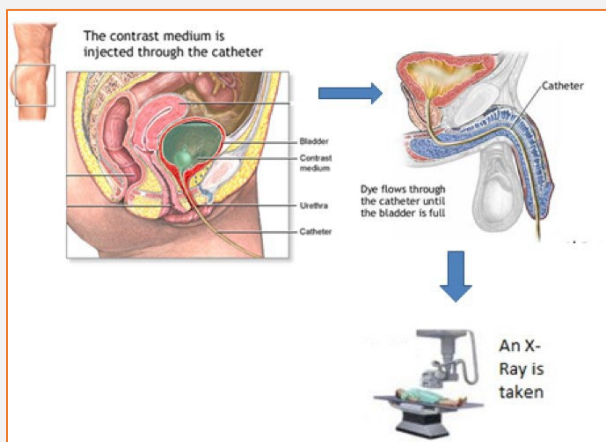
- Bleeding
- Infection
- Damage to the bowel, bladder, blood vessels, and nerves
- Reaction to anesthesia
- Rarely, a biopsy of the gonad can result in unintended damage to or loss of the gonad.

5. Genitograms

Genitograms use contrast dye injections and X-rays to get a clearer picture of your baby's bladder (a "cystogram"), vagina, urine tube, or urogenital sinus. Sometimes, your doctor may prescribe antibiotics for your child to take before and after the test to prevent urinary tract infections (UTIs). The test can only be done when your child has not had a UTI for at least 2 weeks before the test is scheduled.

During the test, a catheter will be inserted in the urinary tube or urogenital sinus and filled with a contrast fluid. A series of X-ray pictures are taken. Once the bladder is full, small babies and infants usually start to urinate (if not, the doctor might stimulate them to urinate). During urination, more X-ray pictures will be taken (called a "voiding cystourethrogram" (VCUG)), so that doctors have a clearer image of the bladder and lower urinary tract in motion, as well as the flow of the urine. While your child is urinating, the catheter will usually slide out spontaneously without your child feeling any discomfort. A few additional X-ray pictures will be obtained to complete the exam. The whole procedure takes approximately 20-30 minutes.

There can be a slight irritation from the catheter insertion, and there is a risk of infection (UTI) after the exam. Some babies might also have a slight allergic reaction to the contrast dye, such as rashes, hives, nausea, flushing, and dizziness. There is radiation exposure from the X-rays. While a small amount of radiation exposure is not harmful, the more X-rays and scans are performed, the higher the risk that the exposure becomes harmful. Therefore, you may want to ask your doctor about the amount of radiation used during the procedure and the risks related to your child's situation. It is always a good idea to keep a record of your child's past history of radiation exposure, such as previous scans and other types of X-rays, so that you can inform your doctor.



**Check out "Questions to Ask" at the end of this section for questions you can ask your team regarding Testing, Genetics, and Hormone Replacement Therapy.*

Your Family Name: _____
Relationship to Patient: _____

Date: _____

Genetic testing

After seeing the information about the different tests and what they entail, please let us know what you now think in relation to your child. Please bring your answers to your next clinic visit. Your answers will be part of a conversation. They show your thinking now. These are not your final decisions on testing.

Extra genetic testing for my child

Want testing results

I think I want to know all genetic test results, including those which are not related to my baby's sex development and won't change my child's management

○ ○ ○ ○ ○ ○ ○ ○

I think I only want genetic results that are not related to my baby's sex development if they have an impact on my child's management

○ ○ ○ ○ ○ ○ ○ ○

I would be comfortable getting genetic test results that are not related to my baby's sex development, even if doctors don't know if they'll change my child's management

○ ○ ○ ○ ○ ○ ○ ○

Do not want testing results

I think I don't want to get genetic test results that are not related to my baby's sex development and won't change my child's management

I think I don't want genetic results that are not related to my baby's sex development, even if they would have an impact on my child's management

I would not be comfortable getting genetic results that are not related to my baby's sex development, if doctors don't know if they'll change my child's management

Deciding what's next

Do you understand what all the different options or possibilities are for extra genetic testing?

- Yes No Unsure

Are you clear about the benefits and harms and what matters most to you?

Yes

No

Unsure

Do you have enough support and advice from others to make a choice?

Yes

No

Unsure

Certainty

How sure do you feel right now about a decision regarding extra genetic testing?

Not sure at all ○○○○○○○○ Very sure

Things I need to do before making a decision (check all that apply):

- I want to discuss options/possibilities with others
- I want to learn more about options/possibilities
- I'm ready to say what I think about additional testing

Genetic testing for research

Genetic testing for research is used to identify new genes that cause DSD and to further understand how those genes work. This information may not help your child directly, but can be important in the development of new tests that can be used in the future for other children and their families. The results of genetic testing for research may not be routinely shared with you, but this would be discussed at the time along with all other benefits and harms of genetic tests. It is always okay to say no to participating in research.

How do you feel right now about the possibility of participating in research?

I do not see the value in participating in research

○○○○○○○○

I see value in participating in research

Things I need to do before making a decision (check all that apply):

- I want to discuss options/possibilities with others
- I want to learn more about options/possibilities
- I'm ready to say what I think about participating in research

Carrier testing

Finally, some genetic tests may reveal that your child's condition is hereditary. If you are a biological parent, you may want to think about these tests, too.

- I am a biological parent
- I am not a biological parent

You may be a carrier for the condition, meaning that you don't have the condition yourself, but passed it on to your child. Other people in your family might also be a carrier and may pass it to their offspring. This information may challenge personal and family relationships.

Show how you feel about the following statements

I would blame myself if I learned that I passed on a genetic change to my child

○ ○ ○ ○ ○ ○ ○ ○

I feel as though I could not do anything to prevent passing on a genetic change to my child

I am worried about family members and/or partners finding out that I am a carrier for a genetic change

○ ○ ○ ○ ○ ○ ○ ○

I am comfortable sharing with other family members and/or partners that I am a carrier for a genetic change

I feel as though others will blame me if I share with them that I am a carrier for a genetic condition and ask them to consider testing themselves

○ ○ ○ ○ ○ ○ ○ ○

I feel as though others will not blame me if I share with them that I am a carrier for a genetic condition and ask them to consider testing themselves

Show how you feel about the following statements

If I plan on having more children, I want to know if they could be affected by a genetic condition

○ ○ ○ ○ ○ ○ ○ ○

If I plan on having more children, I am comfortable not knowing if they could be affected by a genetic condition

Now that you have thought about your feelings, you may have a general idea of where you stand. On the scales below, show which way you are leaning now.

What are you leaning towards?

I would like to know if I am a carrier of a genetic change

○ ○ ○ ○ ○ ○ ○ ○

I don't want to know if I am a carrier of a genetic change

I would tell other family members that I am a carrier of a genetic change

○ ○ ○ ○ ○ ○ ○ ○

I would not tell other family members that I am a carrier of a genetic change

Decide what's next

Do you understand what all of the different options or possibilities are for carrier testing?

Yes

No

Unsure

Are you clear about the benefits and harms and what matters most to you?

Yes

No

Unsure

Do you have enough support and advice from others to make a choice?

Yes

No

Unsure

Certainty

How sure do you feel right now about a decision regarding carrier testing?

Not sure at all ○ ○ ○ ○ ○ ○ ○ ○ Very sure

Things I need to do before making a decision (check all that apply):

- I want to discuss options/possibilities with others
- I want to learn more about options/possibilities
- I'm ready to say what I think about carrier testing

My child's condition

There are many different conditions present at birth that affect a child's genitals or reproductive structures.

This section consists of a series of information sheets on:

- Different diagnoses
 - Different clinical features
- AND
- What they mean
 - How they are managed

Your child's health care team will give you information sheets that are (or may be) relevant for your child. The reason we say "may be" relevant is that diagnoses aren't always clear early on. As you and the team progress along the journey of learning more about your child, you may find that the team gives you new information sheets that add to or replace the ones you first receive.

Gender of rearing

A person's biological sex includes all physical characteristics, chromosomes, genes, gonads (testes, ovaries, or ovotestes), internal reproductive structures (uterus), and external genitals. Not everyone views biological sex in exactly the same way. Sometimes, other factors can influence how we see it, like cultural values, the part of the world you live in, or the views of your child's healthcare team.

When a child is born with a condition that affects biological sex, it can be difficult to determine if that child should be brought up as a boy or a girl and marked as a male or female on their birth certificate. In some cases, the decision seems rather simple; in others it is more complex. Here are some things to know about gender:

- Gender is a way of thinking about the things people do and how they feel. Gender *identity* is how someone thinks and feels about him/herself. Gender *expression* is how he/she expresses those feelings using names, appearance, etc. Babies and children develop ideas about being male or female over time. Most families and societies relate to babies in gendered way based on their observed physical sex.
- As they grow older, some children might play and behave in a way that is not usually associated with their gender. If they have interests and enjoy activities that are thought to be more common for children of the other sex, that is okay. If someone suggests that a child's gender expression is atypical, this is not anything for parents to worry about, and how your child expresses their gender is not something that should be changed or discouraged. Holding on to expectations that are based on sex-role stereotypes will only limit children over time.

Parent to Parent:

For parents with babies whose bodies look different, seeing your child prefer toys or activities that are typical of the other sex can be stressful.

Remember that plenty of kids do the same and are perfectly happy with their assigned gender. Most likely, your baby is too.

Your child's healthcare team will explain the details of your child's sex characteristics and



will discuss with you whether you have a son or a daughter. Some parents may wish to try and raise their child in a gender neutral way. A child's gender identity will develop as they grow up. Specialists cannot be certain about the exact influence of hormones and chromosomes on a person's gender identity, with or without a different sex development pathway.

What we know:

- Most of the time, when a baby is born with a body that looks or works differently, there is no question about if they baby should be raised as a boy or a girl
 - Very rarely, a baby might be born with a condition that makes it harder to determine how best to rear the child
 - In this case, it can be difficult to anticipate whether your baby would be happier as a boy or a girl
 - Your child's healthcare team will take your family's beliefs and preferences into account, as well as your baby's specific diagnosis in order to make a recommendation
- For most people, with and without differences of sex development, gender identity will usually match their gender of rearing, but some people experience gender unhappiness (also called gender dysphoria or distress)
 - One in 2000 people in the general population experience this
- In some conditions impacting sex development, research shows a higher risk of gender dysphoria
- If your child grows up being uncomfortable with and questions his/her gender of rearing, it might be healthy thinking and self-questioning
 - It could also mean that he/she developed differently than expected from the initial observations and discussions
 - Additional specialist support should be put in place for your child at this point

When assigning a gender of rearing, a number of factors may influence the decision:

- Your child's physical sex characteristics
- Usual gender identity of those with the same diagnosis (if known)
- Development and potential for growth of the genitals
- Hormone production and action
- Potential to be fertile (possibility of biological children)
- Your family's cultural background and values
- Your spiritual beliefs or religion



** Check out the Questions to Ask section at the end of the Information Guide for questions you can ask about Gender of Rearing*

Questions to ask

Before going to your child's appointment, you should consider questions you might want to ask your child's healthcare team. The following are some questions parents have asked in the past about gender of rearing, and that you may want to consider asking. Read through these and check which ones you do not have answers to yet. Then, bring this list with you to clinic so you can be sure to ask the team when you get there.

The answers to some of these questions and the questions you choose to ask may change over time as your child grows and develops.

About gender...

- What is the difference between sex and gender?
- What is the difference between gender identity and gender role behavior?
- What is sexual orientation?

About gender of rearing...

- What does my child's genetics tell me/us about my child's gender?
- Are most babies with bodies who look like my baby's happier growing up as boys or as girls?
- What studies exist to suggest that my child is likely to identify as a girl/boy? Can I see those studies?
- What if I think my baby was assigned the wrong gender?
- What if my child later grows up and thinks he/she was assigned the wrong gender?

About anatomy...

- How might my child's internal anatomy work as a girl/boy?
- How might my child's external anatomy work as a girl/boy?
- What will my child's puberty be like as a girl/boy?
- What could my child's sex life be like as a woman/man?
- Will my child be able to have children of their own when they are an adult?

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Your Family Name: _____
Relationship to Patient: _____

Date: _____

My Child's Gender

The following questionnaire is for you to gather your thoughts about gender of rearing. Show how you feel about the following statements by filling in the bubble that best describes how you are feeling at the present time.

For each question, please read the two statements and show which one is closer to how you feel by filling in a bubble. Bubbles closer to one side statement means you agree more with what is being said on that side.

My culture is a factor in my wish to raise my child as a boy or a girl. ○○○○○○○○

My culture is not a factor in my wish to raise my child as a boy or a girl.

My religious/spiritual beliefs have an influence on my wish to raise my child as a boy or a girl. ○○○○○○○○

My religious/spiritual beliefs are not an influence on my wish to raise my child as a boy or a girl.

The idea that my child may choose a different gender later in life makes me uncomfortable. ○○○○○○○○

I am comfortable with the idea that my child may choose a different gender later in life.

My child's sex chromosomes play an important role in my wish to raise my child as a boy or a girl. ○○○○○○○○

My child's sex chromosomes play no role at all in my wish to raise my child as a boy or a girl.

My child's gonads (testes, ovaries, ovotestes, or streak gonads) play an important role in my wish to raise my child as a boy or a girl. ○○○○○○○○

My child's gonads (testes, ovaries, ovotestes, or streak gonads) do not play an important role in my wish to raise my child as a boy or a girl.

My child's potential to have children in the future (if any) is an important factor. ○○○○○○○○

My child's potential to have children in the future (if any) is not an important factor.

The appearance of my child's external genitals is important to their gender. ○○○○○○○○

The appearance of my child's external genitals is not very important to their gender.

The availability of cosmetic genital surgeries will influence my views.

○ ○ ○ ○ ○ ○ ○ ○

The availability of cosmetic genital surgeries will not influence my views.

The availability of hormone replacement therapy that my child can take is important for determining their gender.

○ ○ ○ ○ ○ ○ ○ ○

The availability of hormone replacement therapy that my child can take is not important for determining their gender.

I see my child as being a:

Boy ○ ○ ○ ○ ○ ○ ○ ○ Girl

Deciding what's next

Do you understand what all the different options or possibilities are for gender of rearing?

- Yes No Unsure

Are you clear about the benefits and harms and what matters most to you?

- Yes No Unsure

Do you have enough support and advice from others to make a choice?

- Yes No Unsure

Certainty

How sure do you feel right now about your child's gender of rearing?

Not sure at all ○ ○ ○ ○ ○ ○ ○ ○ Very sure

Things I need to do (check all that apply):

- I want to discuss options/possibilities with others
- I want to learn more about options/possibilities
- I'm ready to say what I think about my child's gender of rearing

My child's body looks or works differently

Some babies are born with bodies that may not look like a typical boy or a typical girl. For parents, this can lead to a lot of questions. The first step of healthcare support is to learn about how the baby's body has developed and the specific diagnosis. This can help your care team to provide you with more information about why your baby's body looks the way it does and how your baby's body works.

As your baby grows and develops, you may decide that you would like to address the differences in how your baby's body looks or works. There are lots of types of resources that can help you decide how you would like to do this and provide you with support and education as you navigate your baby's health care. Some of these resources can include psychologists, support groups, advocacy organizations, or other families with a child or adult with the same diagnosis as your baby.

What do I do if my baby's body looks different?

When a child's genitals look different than other children's genitals, it tells your child's healthcare team that there might be a medical problem that needs attention quickly. This is why your child's healthcare team will want to do a careful assessment of your child's body soon after the difference is noticed.

Usually, though, having genitals that look different than you expect them to does not cause medical problems all by themselves. Most children with these differences are healthy. Fortunately, this means that there is time to think about how you want to handle the fact that your child's body looks different from other children's. It also means that genital appearance can mostly be a matter for your child to address as a young adult when they begin to explore sexual function.

Having a new baby can be a very stressful experience, and worrying about your baby being different can make it much harder to focus on getting to know and love your new

baby. There are lots of different types of resources that you can use to learn more about your baby's body as they grow.

Learning more and getting support

Some families will choose to address the differences in how their baby's body looks by learning more about their baby. If your child gets their care from a multidisciplinary team, there might be a psychologist on the team. The psychologist can help you learn about and adjust to having a baby who looks different. Usually, they have experience working with other families who have babies who look like yours. They can help you think about what to expect as your baby grows up, and work with you to practice ways to tell important people like family members or daycare workers what to expect when helping care for your baby.

Surgery

Other families may choose to address the difference in how their baby's body looks with surgery.

Different healthcare teams might have different recommendations for when surgery should be done (if at all), for example:

- Some prefer to do it in the first few years of life when the baby won't remember it – but not right after a baby is born. Anesthesia has its own complications. And those complications can make it less safe for babies who are under one year of age.
- Others would only think about doing surgery to change how a person's genitals look when that person is old enough to want surgery and carefully think through the decisions for his or her own body.

Parent to Parent:

With a new baby whose body looks different, it's easy to think that you'll always see them or think of them differently. But, many parents with babies like yours find out:

After a while of getting to know your baby, you stop even noticing that your baby is different. Just like a birthmark or their hair or eye color, it's just part of who your baby is.

This means that you have time to consider all of your options before making a decision about surgery. There is no rush to do surgery right away, and it is okay to change your mind as your baby starts to get a little bit older.

Parent to Parent:

Some babies have differences in how their bodies both look and work. Sometimes, surgery to fix how the body works is needed, and changing how the body looks at the same time is an option. In this case, make sure that you're thinking about both surgeries separately, even if they might happen at the same time.

Would you decide to change how your baby's body looks even if they didn't need another medical procedure at the same time?

Make sure to talk about this with your child's healthcare team when thinking about surgery for your baby.

The type of surgery a baby might have to change how their body looks depends on what kind of difference they have. The two most common differences in how a baby's genitals look are severe hypospadias, where the urinary tract/tube opens somewhere on the shaft or base instead of at the tip of the penis, and urogenital sinus, when the vagina and urinary tract open into a single (common) channel, rather than separate openings. Other types of differences your baby might have include an enlarged clitoris or a very

small penis. Making changes to this type appearance of a baby's genitals is increasingly questioned because a baby or toddler is not bothered by genital appearance and the long term impact of infant surgery is hotly debated.

What do I do if my baby's body works differently?

Most of the time, the differences in how your baby's body looks and works because of their different sex development will not put your baby's health at risk. This means that you have time to ask questions and make decisions about what you may want to do for your baby. Usually, decisions do not need to be made right away. Unless your baby's doctors have told you that their condition is life threatening and they need to take action quickly, take time to enjoy and get to know your new baby.

When a child's body works differently than other children's, it is important to understand what needs to be taken care of immediately or sometime later.

Life-threatening situations

- In very rare (but not most) situations, babies are born with differences in the way their bodies work that can cause life-threatening problems that need to be addressed with surgery right away. Babies with a condition called “exstrophy” are born with some of their organs on the outside of their bodies, and will need early surgery to put them on the inside. A baby might also need surgery right away if their urine tube did not develop properly and their doctors need to create a way for the baby’s bladder to empty.
- You might hear words like “urgent” or “emergency” surgery to describe these situations where there is little time for parents to learn and act before their child receives treatment. Your child’s doctors will tell you if the differences in how your baby’s body works mean that they need urgent or emergency surgery early in life. Otherwise, there are other options depending on their condition.



For everything else, you can take time to decide how to address it.

Learning more and getting support

Learning about how a baby’s body has developed and how their condition will affect them as they grow up is the first step.

Some people might think that once you figure out if your baby is going to be brought up as a girl or as a boy, then you should make sure that everything looks and works in a way that matches their gender. It is important to understand that this may not always be helpful, necessary, or possible.

- While some people are glad that they had surgery during infancy or young childhood to change the way their genitals function; other people are not happy at all and wish that they had the opportunity to make decisions for themselves about surgery.

- Differences in the ways their genitals work affect people differently as they grow and develop. For example, babies will wet in their diapers, and it is not until adolescence or young adulthood that issues such as menstruation in girls or sexual intercourse are relevant.

This means that you have time to learn about and consider all of your options. Though surgery to “fix” the differences jumps out as a solution for many, there is no rush to do it right away. And it might not turn out to be the right thing for your child at all.

Non-surgical options

Depending on your child’s condition, there may be a number of medical options (including medication and non-surgical procedures) to pursue. You will want to talk with your child’s healthcare team to learn more about these since they will depend a lot on your child’s specific condition.

Options that do not depend on your child’s diagnosis include education, support, and counseling.

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body, their emotional health, and general psychological development as they grow. As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- Talking with other parents of children with the same condition or adults who have it is another good option. There are also a number of patient and family education and support organizations.
 - You can often get referrals to other families and recommendations on support organizations from your child’s healthcare team.
 - You can look at the section "Support for Families" for more information about these resources.

Parent to Parent:

It can be hard to imagine how your child can be happy and healthy if they grow up with genitals that work differently than others.

Talking with other parents of children or adults with conditions similar to your child’s can help you see possibilities you never dreamed of.

Non-urgent & elective surgery

In most situations, parents have time to think and talk about surgical possibilities for their children. Some conditions will require surgical intervention eventually, but not for years down the road. Other conditions do not necessarily require surgical intervention, but it remains an option for children to think about when they grow up.

Parent to Parent:

If your doctors tell you that the difference in how your baby's body works is not urgent, you don't need to worry about it for now.

We will need to act, but not until later

Some of these differences are things that will not affect your baby until he or she reaches puberty or even later. For now, enjoy being with your baby instead of worrying about the future.

Thinking ahead...

Surgical needs in the future: Gonads

In some conditions, the gonads (the tissue that can be ovaries or testes) do not work in the way that is expected. Depending on the baby's karyotype (genetic pattern), these gonads may need to be watched closely to make sure that they don't become a risk to the baby's health.

- Some babies with internal gonads are able to live well with them where they are
- Other babies have a risk of developing pre-cancerous changes in their gonads
 - This can happen to those who have Y chromosome material in their karyotype
 - The changes are called "germ cell cancer (GCC) precursor lesions." And if this does happen, a child could go on to develop a type of cancer known as GCC.
 - How doctors treat this:
 - If a child with internal gonads has a Y chromosome, doctors will suggest they monitor the gonads (testes, streak gonads, or ovotestes) closely or remove all or part of them if there is a risk, based on research into specific diagnoses or development paths.
 - There is usually plenty of time to decide about possible removal.
 - Many children will not need any surgery, and for others surgery will be discussed long after puberty and into adulthood.

Surgeries like these do not need to be considered during infancy, since there are many years before they might be necessary. You may choose to think about these surgeries now, while your child is a baby. Or, you might decide to wait until your child is nearing puberty, when their body will need to start to work differently and they are able to better understand and take part in decisions about surgery. Your healthcare team and resources like psychologists and support groups can help you decide what is right for your family.

Surgical considerations for the future: Changing how the body works

There are other reasons parents might consider surgery to change how their baby's body works, but most of these are not important until much later, when your child is a teenager or an adult. For example, babies born with a uterus are likely to have periods (menstruate) when they go through puberty. If a baby has a uterus but does not have a vagina, that is fine for babies, but one may need to be created to allow the girl to menstruate when she gets older.

There are also many conditions where there is no need for surgery unless your child grows up and decides that they want it. Some babies are born with a condition called "MRKH" that stops their uterus and the upper part of their vagina from developing properly. Because they have no uterus, parents of these babies will not need to worry about surgery to help with menstruation during puberty. However, some of these babies may grow into adults who decide that they would like to participate in vaginal sex. In this case, these women may choose to do surgery to create a vagina for this purpose.

In cases like these, there is no need to think about surgery now or even during childhood. Because the only purpose for a vagina for these babies is for vaginal sex in the future, the only reason to consider surgery to create one is if your child grows up and decides that she would like one.

Other babies born with a condition called "CAH" may have a urogenital sinus, where the vagina and the urinary tract open into a single, common channel. Some babies may require surgery to separate the vagina and the urinary tract during childhood if they get many urinary tract infections (UTIs).

For other babies, there are no health issues involved with having a urogenital sinus. These babies will not require surgery to change it during childhood.

Parent to Parent:

Most importantly, remember, you have time. Time to get to know your baby; time to learn about your baby; time to get used to everything.

When they get older, some of these women with a urogenital sinus may decide that they would like to participate in vaginal sex or to become pregnant. They may decide as adults to have surgery in order to create separate openings for the vagina and the urinary tract.

Women with CAH who have had surgery to change a urogenital sinus are often able to become pregnant naturally, but will likely need to deliver their baby via C-section instead of vaginally. For these women, there are no negative impacts on fertility if they wait to have surgery until they are adults and are considering vaginal sex or pregnancy. Therefore, there is no reason to have the surgery in childhood unless your healthcare team tells you it is needed for frequent UTIs.

In the meantime, your healthcare team can help you teach your child about their body as they grow up so that they can make health decisions like these in the future.

Decision-making

There are many decisions that your child's healthcare team and you will make on behalf of your baby. These include decisions that any new parent faces like breast-feeding vs formula, who gets to see the baby first (and when and where), and who your baby's pediatrician will be. There are also decisions specific to your child's medical condition, like how to handle differences in how their body looks or works.

Some of these decisions are complex and there is no one-best-answer that fits all children and families. The best decisions for any one child are based on:

- The child's specific condition
- The knowledge and expertise of the healthcare team
- The values and beliefs of the family in which the child lives

Your child's healthcare team will help guide you through all of the decisions that need to be made. Your team may practice something called "Shared Decision-Making (SDM)." This means that instead of the healthcare team telling you what to do at every stage of your child's treatment and care, you work together as partners to think about what is best for your child and your family. Your healthcare team will help you think about all of

Information Guide

the different options, and the risks, harms, and benefits that come with each option. Then, you can think about and discuss which approach you think will be right for your family.

The following worksheets will help you explore your values, beliefs, and preferences. You can bring them with you to your clinic appointments to help let your child's healthcare team know more about your family.

Questions to ask

There are many things for you and your child’s healthcare team to think about when you are considering surgery for your child. You can find answers to them by talking with the healthcare team, by talking with others who have been in a similar situation (other parents and affected adults), talking with people close to you (significant others, close friends, or family), and by thinking through your own values and beliefs. Not every question here will necessarily be important for every family, and you may have other questions not listed here.

About surgery in general...

- Why is the surgery performed? How is it performed?
- What will recovery be like?
- What are the short- and long-term risks or harms of the surgery? What are the benefits?
- Will my child need multiple surgeries? Will surgery in infancy meet adult needs, or will it set up the need for additional surgery later?
- How might the surgery affect my child’s quality of life?
- How might surgery affect or not affect a child's self-esteem and body image?
- How might performing or not performing genital surgery pose a risk for teasing or embarrassment?
- What are alternatives to surgery?
- How do my own beliefs, culture, and religion influence my decisions?

About surgery and gender...

- How might my child's gender identity and preferences develop?
- If our child grows up and identifies with another gender, what will this surgery mean for our child?
- Is this procedure “reversible”? Has anyone ever reported a successful reversal?

About timing of surgery...

- How much time do I have to make a decision? Can this procedure be done later?
- Is the decision to have surgery one my child could make for him/herself later?
- Will my child remember the surgery if it is done during childhood?
- How might my child remember the surgery?
- What is life like for people like my child if they don’t have early genital surgery?

Information Guide

- What are other possible time points when a young adult could make their own choice to alter their genitals?

About talking to your child about surgery...

- What might a child or adult think or feel when they learn that their parents decided to alter their sex organs during infancy or early childhood?
- How will I share the decision about surgery with my child later?
- How could my child be involved in decisions about his/her own body?

About surgery to change how the body looks...

- To what extent might genital appearance be changed by surgery?
- How might future sexual sensation and pleasure be affected by surgery?
- What will my child's sexual function and sensation be like without surgery?
- How might scars from the surgery change as my child's body grows? (i.e. could genital appearance and function could get worse as my child grows?)

About surgery to change how the body works...

- What do babies' genitals need to do now? How are they used (or not used)?
- What will a child's genitals be used for as they grow?
- How will a young adult's genitals will work for voiding, sexual pleasure, and possible parenthood?
- How might genital function be affected by surgery?
- How might future sexual sensation and pleasure be affected by surgery?
- What will my child's sexual function and sensation be like without surgery?
- How might scars from the surgery change as my child's body grows? (i.e. could genital appearance and function get worse as my child grows?)

Questions for your surgeon...

- How experienced are you in caring for children with conditions like my baby's?
- Is it likely that there will be improvements in surgical techniques before my child is an adult? What kinds of improvements may be possible in the future?
- How many times have you performed a certain procedure?
- How successful are your surgeries?
- Can you define what you mean by "successful outcome?"
- What are the complication rates of this surgery?
- Can I see pictures of the results?
- Do you have photos of grown-up patients who had this procedure in childhood?

Information Guide

- What do people who have had this surgery in the past say about it? Can I talk to some satisfied patients?
- Would you be willing to provide a referral for a second opinion?
- Are there other surgeons who would recommend a different technique, and what are their reasons?
- Is there research supporting your opinion?

Information Guide

Your Family Name: _____
Relationship to Patient: _____

Date: _____

Genital Surgery: My Values, Preferences, and Beliefs

Decision-making in complex conditions like your child’s requires joining knowledge and expertise with values, preferences, and beliefs. After reading about what to do if your child’s body looks or works differently, we would like to help you identify values, preferences, and beliefs important to the decisions made on behalf of your child.

Instructions: For each question, please read the two statements and show which one is closer to how you feel by filling in a bubble. Bubbles closer to one side statement means you agree more with what is being said on that side.

My culture has an influence on my thinking about childhood genital surgery

My culture has no influence on my thinking about childhood genital surgery

My religious/spiritual beliefs have an influence on my thinking about childhood genital surgery

My religious/spiritual beliefs are not an influence on my thinking about childhood genital surgery

Changing the appearance of my child’s genitals to look more sex-typical would motivate me in discussing childhood surgery

Changing the appearance of my child’s genitals to look more sex-typical would not motivate me in discussing childhood surgery

I worry that my child will be treated differently because of the appearance of his/her genitals

I do not worry that my child will be treated differently because of the appearance of his/her genitals

I worry that my child’s future sex life could be affected by childhood genital surgery

I am not worried that my child’s future sex life could be affected by childhood genital surgery

I am aware that there are possible complications of childhood genital surgery including the possibility of a child needing future surgeries

I am not aware of the possible complications of childhood genital surgery including the possibility of a child needing future surgeries

I think infancy/ babyhood surgery to change my child’s genitals might be right

I think not having / doing surgery during infancy / babyhood to change my child’s genitals might be right

I can raise my baby to be a happy child without surgery to change how their genitals looks



I am unsure if I can raise my baby to be a happy child if their genitals look the way they do now

Satisfaction and concerns about bringing up your child with their current appearance

How satisfied are you about these aspects of your child's appearance (if rearing your child as a girl is a possibility):

1. Length of the clitoris

Very satisfied Satisfied Dissatisfied Very Dissatisfied

2. Position and shape of the urine opening

Very satisfied Satisfied Dissatisfied Very Dissatisfied

3. Shape of the labia

Very satisfied Satisfied Dissatisfied Very Dissatisfied

4. General appearance of the vulva (clitoris, vulva, labia)

Very satisfied Satisfied Dissatisfied Very Dissatisfied

5. Length of the vagina

Very satisfied Satisfied Dissatisfied Very Dissatisfied

How satisfied are you about these aspects of your child's appearance (if rearing your child as a boy is a possibility):

1. Length of the penis

Very satisfied Satisfied Dissatisfied Very Dissatisfied

2. Position and shape of the urine opening

Very satisfied Satisfied Dissatisfied Very Dissatisfied

3. Shape of the glans (tip of the penis)

Very satisfied Satisfied Dissatisfied Very Dissatisfied

Information Guide

4. Shape of the penile skin

Very satisfied Satisfied Dissatisfied Very Dissatisfied

5. Curve / straightness of the penis upon erection

Very satisfied Satisfied Dissatisfied Very Dissatisfied

6. General appearance of the penis

Very satisfied Satisfied Dissatisfied Very Dissatisfied

What's next?

Do you understand what all the different options for how your child's genital development could be managed during childhood?

Yes No Unsure

Are you clear about the possible benefits and harms of changing a child's genitals using surgery and what matters most to you?

Yes No Unsure

Do you have enough support and advice from others to make a choice?

Yes No Unsure

Certainty

How sure do you feel right now about whether childhood genital surgery is right or wrong for your child?

Not sure at all Very sure

Things I need to do (check all that apply):

- I want to discuss options/possibilities with others
- I want to learn more about options/possibilities
- I'm ready to say what I think about genital surgery for my child

Information Guide

Your Family Name: _____
Relationship to Patient: _____

Date: _____

Gonadal Surgery: My Values, Preferences, and Beliefs

Decision-making in complex conditions like your child’s requires joining knowledge and expertise with values, preferences, and beliefs. After reading about what to do if your child’s body looks or works differently, we would like to help you identify values, preferences, and beliefs important to the decisions made on behalf of your child.

This assessment focuses on gonadal surgery in the case of babies with a Y chromosome and internal gonads.

Background: In babies with a Y chromosome (e.g. XY pattern, or XY in some cells, but not in all cells) and internal gonads, there is a small but increased risk (sometimes in later life) of changes in the gonad that can lead to cancer. The small risk of developing gonadal cancer varies greatly depending on the exact diagnosis and the position of the gonads, with some having no significant risk before adulthood. The main role of gonads during childhood is to make pubertal changes.

If people choose to have their gonads surgically removed (called gonadectomy), they will need to take sex hormones (medication) in order to make puberty happen. They will need to continue taking these hormones for life if they are a man, or until they reach a menopausal age if they are a woman. The body needs sex hormones for physical health and emotional well-being. Some people may be offered gonadectomy to remove gonads that don’t work, but others might have gonads that have a small chance of producing eggs or sperm (gametes) in the future and this is lost with gonadectomy. While some children with different sex development pathways have fertility potential, many children with differences in how their bodies look and work may require future medical interventions for conception or may not be able to conceive at all.

Instructions: For each question, please read the two statements and show which one is closer to how you feel by filling in a bubble. Bubbles closer to one side statement means you agree more with what is being said on that side.

It worries me that my child may not agree with my decision about gonadectomy when he/she is older

○ ○ ○ ○ ○ ○ ○

I feel comfortable making a decision about gonadectomy on behalf of my child

A small amount of cancer risk is too much.

○ ○ ○ ○ ○ ○ ○

Routine monitoring of gonads for risk factors is reassuring.

Information Guide

I worry that lifelong hormone replacement therapy would cause too much stress for me and/or my child	<input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/>	I am comfortable with lifelong hormone replacement therapy for my child
I worry my child's gonads will have unwanted effects on my child (e.g., at puberty)	<input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/>	I think that my child's gonads will have wanted effects on my child (e.g., at puberty)
The location of my child's gonad(s) worries me	<input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/>	I am comfortable with the location of my child's gonad(s)
Given the estimated level of cancer risk, I would like the gonads removed before puberty	<input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/>	Given the estimated level of cancer risk, I would want to leave the gonads in place for puberty
Have the gonads removed as soon as possible	<input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/>	Leave the gonads in place for as long as possible

Deciding what's next

Do you understand the different options for how your child's genital development could be managed during childhood?

Yes

No

Unsure

Are you clear about the possible benefits and harms of gonadectomy and what matters most to you?

Yes

No

Unsure

Do you have enough support and advice from others to make a choice?

Yes

No

Unsure

Certainty

How sure do you feel right now about whether childhood gonadectomy is right or wrong for your child?

Not sure at all Very sure

Things I need to do (check all that apply):

- I want to discuss options/possibilities with others
- I want to learn more about options/possibilities
- I'm ready to say what I think about gonadectomy for my child

Information sheets

There are many different conditions present at birth that affect a child's genitals or reproductive structures. And there are many ways of managing those conditions. Using this list, your child's health care team will check off which are relevant for your child at this time and provide you with copies.

Clinical features

- Atypical clitoris
- Enlarged clitoris
- Small penis
- Small vagina
- Undescended testes (cryptorchidism)
- _____
- _____

Clinical Management

- Management of the gonads
- Phalloplasty
- Surgery for a urogenital sinus
- _____
- _____

Diagnoses

- 5 Alpha Reductase Deficiency-2 (5 α RD-2)
- 17 β Hydroxysteroid Dehydrogenase Deficiency-3 (17 β HSD-3)
- 45,X/46,XY Mixed gonadal dysgenesis
- 46,XX Testicular DSD
- Cloacal and bladder exstrophy
- Congenital Adrenal Hyperplasia (CAH)
- Complete Androgen Insensitivity Syndrome (CAIS)
- Complete gonadal dysgenesis (Swyer Syndrome)
- Gonadal dysgenesis due to Denys-Drash Syndrome, Frasier Syndrome or WAGR Syndrome
- Hypospadias
- Klinefelter Syndrome
- Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome
- Ovotesticular DSD
- Partial Androgen Insensitivity Syndrome (PAIS)
- Partial Gonadal Dysgenesis
- Turner Syndrome
- _____
- _____

Clinical features

Atypical clitoris

What is the clitoris?

- The clitoris is the key for sexual pleasure for most women. It is packed with nerve endings and is very sensitive. When a woman is feeling excited, the clitoris fills with blood and swells up.
- The clitoris has an outside part that you can see (the glans) and an inside part (the legs and bulbs) that extends behind the labia (folds of skin) and surrounds the vagina.
 - The outside part may be larger in some girls than typically seen in most other girls (e.g. because of androgen effects in the womb)
 - In girls with cloacal/bladder exstrophy, the clitoris may be split.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- In most children, an atypical clitoris causes no problems and genital surgery is not urgent or medically necessary.
- Some girls with a larger clitoris experience painful erections, which may cause discomfort and/or embarrassment. Other girls do not experience problems because of an atypical clitoris.
- Surgery to the clitoris is called a "clitoroplasty."
 - Doctors will try to avoid harming the tip ("glans") and sensitive nerves, so that the clitoris remains sensitive for sexual pleasure later in life.

- There is always a risk of irreversible damage to the nerves in the clitoris.
- Sometimes there is clitoris regrowth after surgery, and surgical revision may be necessary. With multiple operations, the risk of damage to the clitoris becomes higher.
- Some families choose to wait until their child is old enough to be involved in the decision of whether or not to have surgery.

When should surgery be done?

- It is important to take all the time that you need before deciding to have a surgery to reduce the size of your child's clitoris. There are many factors to consider, and decision making with your providers and support team can be very helpful.
- In the past, it was common to do this type of surgery in childhood. More recently, many parents and their healthcare teams have started making the decision to postpone any clitoral surgery to adolescence or young adulthood so girls can make their own decisions about whether or not to have surgery.
- Some parents who decide that they want their child to have surgery find it important that their child does not remember the procedure.
- Other parents decide to wait until their child is old enough to participate in the decision.

What should we consider when thinking about genital surgery?

- Before making decision about surgery, talk with your healthcare team, support groups, or other families who have faced similar decisions and can give you some extra insight into their experience (with and without surgery).
- You can also look at the "Questions to ask" section at the end of "My child's body looks or works differently" for a list of questions to consider or to ask your healthcare team.
- Working together with your doctors and others, you can make informed choices to help your child thrive.

What are the possible outcomes of surgery?

- Clitoral surgery is done to change the appearance of the genitals, but it may also change the function of the genitals.
- Some, but not all adults who had surgery as a child are happy about how their genitals look.

Information Guide

- Many adults who had clitoral surgery as a child have problems now with genital sensation and sexual pleasure (orgasm).
- Some, but not all, adults who had surgery as a child regret not being involved in a decision about their own body.
- Some young women find it helpful talking to a psychologist or counselor about possible concerns related to genital appearance and function, before making a decision about clitoral surgery.
- Keep in mind, the right approach for one child/family is not necessarily right for another child/family.

Enlarged clitoris

What is the clitoris?

- The clitoris is the key for sexual pleasure for most women. It is packed with nerve endings and is very sensitive. When a woman is feeling excited, the clitoris fills with blood and swells up.
- The clitoris has an outside part that you can see (the glans) and an inside part (the legs and bulbs) that extends behind the labia (folds of skin) and surrounds the vagina.
 - The outside part may be larger in some girls than typically seen in most other girls.
- Some girls with a larger clitoris may experience erections that can cause discomfort or embarrassment.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- In most children, a larger clitoris causes no problems and clitoral surgery is not urgent or medically necessary.
- Surgery to the clitoris to make it smaller is called a "clitoroplasty" or clitoral reduction surgery.
 - Doctors will try to avoid harming the tip ("glans") and sensitive nerves, so that the clitoris remains sensitive for sexual pleasure later in life.

- There is always a risk of irreversible damage to the nerves in the clitoris.
- Sometimes there is clitoris regrowth after surgery, and surgical revision may be necessary. With multiple operations, the risk of damage to the clitoris becomes higher.
- Some families choose to wait until their child is old enough to be involved in the decision of whether or not to have surgery.

When should surgery be done?

- It is important to take all the time that you need before deciding to have a surgery to reduce the size of your child's clitoris. There are many factors to consider, and decision making with your providers and support team can be very helpful.
 - Some parents who decide that they want their child to have surgery find it important that their child does not remember the procedure.
 - Other parents decide to wait until their child is old enough to participate in the decision.
 - Still other say that surgery should not be done at all during childhood, unless it prevents serious illness or death

What should we consider when thinking about genital surgery?

- Before making decision about surgery, talk with your healthcare team, support groups, or other families who have faced similar decisions and can give you some extra insight into their experience (with and without surgery).
- You can also look at the "Questions to ask" section at the end of "My child's body looks or works differently" for a list of questions to consider or to ask your healthcare team.
- Working together with your doctors and others, you can make informed choices to help your child thrive.

What are the possible outcomes of surgery?

- Clitoral surgery is done to change the appearance of the genitals, but it may also change the function of the genitals.
- Some, but not all adults who had surgery as a child are happy about how their genitals look.
- Many adults who had clitoral surgery as a child have problems now with genital sensation and sexual pleasure (orgasm).

Information Guide

- Some, but not all, adults who had surgery as a child regret not being involved in a decision about their own body.
- Some young women find it helpful talking to a psychologist or counselor about possible concerns related to genital appearance and function, before making a decision about clitoral surgery.
- Keep in mind, the right approach for one child/family is not necessarily right for another child/family.

Small penis

What is it?

Some boys are born with a small penis. This is sometimes called a micropenis or microphallus (when the penile length is smaller than 2 cm at birth).

What causes it?

- There are many reasons why this might happen.
- In some boys, there was not enough testosterone produced during development in the womb. This may have prevented the penis from fully growing. Doctors may check this with hormone tests.
- In other boys, the cause of a small penis remains unclear (also called “idiopathic” micropenis).

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby’s body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby’s condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section “Support for Families” for more information about these resources.

Genital surgery

- A smaller penis causes no medical problems.
 - Most boys with a small penis (without other genital differences such as hypospadias or undescended testes) have no medical problems and genital treatment in childhood is not medically necessary or urgent.
- Many boys with a small penis grow up to be happy and healthy men who can father children and experience sexual pleasure.

Information Guide

- The penis size can sometimes be increased by using testosterone during childhood.
 - Testosterone is usually given as three injections, 1 month apart, by endocrinologists.
- This testosterone will advance penis growth during childhood, but it will not influence the final penis length in adulthood. It seems there is a predetermined length for every boy.
 - This means that a child with a small penis will probably become an adult with a small penis.
- Surgery to make the penis larger (called phalloplasty) is very difficult, has a relatively high complication rate, and is not performed in childhood. It is only performed in late adolescence or adulthood, when your son is able to take part in the decision to undertake those risks.

Small vagina

What is it?

- Some girls have an underdeveloped vaginal opening (sometimes called a “dimple”), because the female internal sex organs did not fully develop.
 - These organs include the uterus, cervix, the Müllerian ducts, and the upper part of the vagina.

What causes it?

- Some girls have a condition called Mayer-Rokitansky-Küster-Hauser Syndrome.
- Other girls are insensitive to certain hormones (e.g. girls with partial or complete androgen insensitivity syndrome) or do not produce as much of certain hormones (e.g. girls with partial gonadal dysgenesis), causing the vagina to not fully develop.
- The cause sometimes remains unknown.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby’s body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby’s condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section “Support for Families” for more information about these resources.

Genital surgery

- A smaller vagina causes no medical problems.
- Girls with a small vagina have no medical problems and treatment in childhood is not medically necessary.

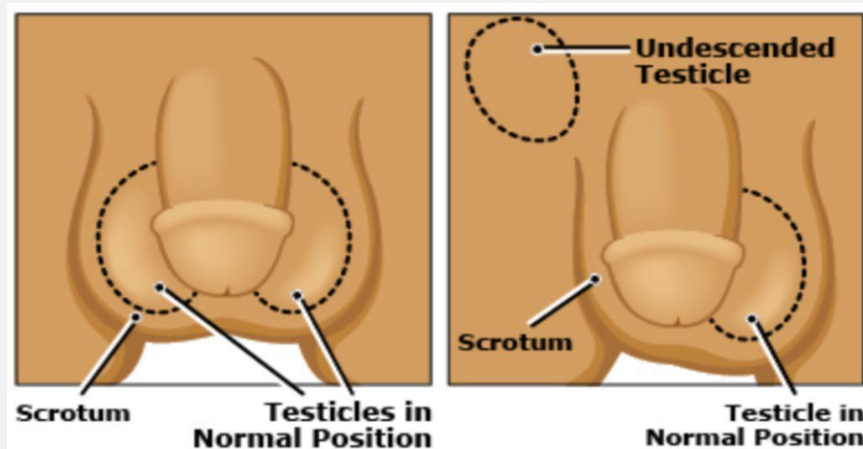
Information Guide

- Some young women might have to think about genital treatment when they are ready to have sex.
- For most women, the clitoris and labia (not the vagina) provide the most sexual pleasure. Women with small vaginas do not have to change their body in any way to be able to enjoy sex.
- To be able to have vaginal sex, the vagina may need to be deeper (or longer) and sometimes wider.
 - Usually, vaginal dilation is the first line of treatment, because vaginal tissue is very stretchy.
 - If vaginal dilation is not successful, vaginal surgery may be an option, but dilation will also be necessary after the surgery.

Undescended testes (cryptorchidism)

What is it?

- The testes typically form inside the belly and move down (descend) into the scrotum before birth. Sometimes, one or both testes do not descend.
- This occurs in 1 out of 3 premature babies and in approximately 1 out of 33 babies born to term.



What are the treatment options?

- In about half of babies, undescended testes will descend on their own by 6 months of age.
- If descent does not happen by then, it is important to get treatment because testes that remain undescended may not work properly. This could affect future fertility or lead to other medical problems.
 - When the testes are located in the belly or groin area instead of the scrotum, the cancer risk is also higher and it is difficult to keep an eye on them through examination and imaging tests.
- Doctors will suggest performing an operation called “orchidopexy” in childhood, to bring the testes into the scrotum.
- Regular follow-up of the testes by self-exam and doctor checkups (and sometimes ultrasound) helps to monitor the cancer risk.
 - Sometimes an additional testes biopsy after puberty may be necessary.

Diagnoses

5 Alpha Reductase Deficiency-2 (5 α RD-2)

What is it?

Children with 5 α RD-2 have XY chromosomes and gonads (testes). The gonads are usually located in the groin area or belly. These gonads produce hormones called androgens (e.g. testosterone), but in a smaller amount than is typically expected.

- If little testosterone is produced, children with 5 α RD-2 will develop physically like most other girls. If more testosterone is produced, children will develop more like boys.
- Children with 5 α RD-2 have no uterus or cervix, because other hormones produced did not make it possible for these structures to develop.
- Sudden production of androgens by the gonads at puberty may cause male typical sex characteristics to develop. Examples of these changes are lowering of the voice and growth of the genitals.

What causes it?

- The cause is a genetic change in the SRD5A2 gene.
- This genetic change prevents the enzyme 5 α reductase-2 from working properly and producing androgens.
- Usually, both biological parents have passed on a changed SRD5A2 gene to their child. This makes the biological parents “carriers.”
 - Being a carrier means that people do not have the condition themselves, but have the ability to pass on the affected gene to their biological children.

What are the effects?

- Depending on the amount of androgens produced before birth, there may be:
 - A larger clitoris with or without a urogenital sinus (the vagina and urinary tract open into one channel, rather than separately). The vagina may also be small.
 - A small penis with or without hypospadias (the urinary tract does not open at the tip of the penis, but somewhere on the shaft or base of the penis).

Is my baby a boy or a girl?

- Because the sex anatomy is variable from baby to baby with 5 α RD-2, doctors will usually consider how “masculine” or “feminine” the sex anatomy is, and what kind of fertility and sexual function can be expected in adulthood, in order to make a recommendation.
- Many children will grow up happy even if their genitals, chromosomes, and/or gonads are atypical, or if the combination of these biological sex characteristics is atypical.
- Gender role behavior in children with these syndromes is variable, but there is a tendency, even in children reared as girls, to prefer activities and hobbies typical of boys. This does not mean that these children are not happy as girls. You can look at the section “Gender of Rearing” for more information.
- If the gonads are left in place, production of androgens at puberty may cause male sex characteristics to develop. These can include growth of the genitals or lowering of the voice. If this happens, research tells us that 40 to 60% of these children are happier living in the male gender (even if they were reared as girls).

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby’s body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby’s condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section “Support for Families” for more information about these resources.

Genital surgery

- Sometimes, a urogenital sinus or severe hypospadias may cause urinary problems. In most children, however, atypical genitals cause no medical problems.
- Surgery to the vagina, clitoris, or penis in childhood may result in irreversible damage to the sensitive nerves.
 - This can lead to problems with sensation and sexual pleasure in adulthood.
 - Surgical revision is often necessary for complications of childhood surgery, such as narrowing of the urinary tract in boys or vaginal tightening in girls.

Gonadal management and removal with hormone replacement

- Gonadal cancer risk in children with 5 α RD-2 is low before puberty (<1%).
- Keep in mind, what is right for one child with 5 α RD-2 is not necessarily right for another child with 5 α RD-2.
- If gonads are removed, life-long hormone replacement is necessary for overall well-being and bone health.
 - Some people with 5 α RD-2 have a difficult time finding an appropriate hormone replacement that does not cause side-effects.
 - Assessments of overall well-being and bone health may be suggested by the healthcare team as a way to ensure that the right dose of hormone replacement is being used.
- Fertility potential is uncertain.

17 β Hydroxysteroid Dehydrogenase Deficiency-3 (17 β HSD-3)

What is it?

Children with 17 β HSD-3 have XY chromosomes and gonads (testes). The gonads are usually located in the groin area or belly. These gonads produce hormones called androgens (e.g. testosterone), but in a smaller amount than is typically expected. Many people shorten the condition's name to "seventeen beta H S D" when they say it out loud.

What causes it?

- The cause is a genetic change in the HSD17B3 gene.
- This genetic change stops an enzyme called "17 β hydroxysteroid dehydrogenase-3" from working properly and producing androgens like testosterone.
- In most cases, neither parent has this condition. But they both biological parents have passed on a changed HSD17B3 gene to their child. This makes the biological parents "carriers."
 - Being a "carrier" means that people do not have the condition themselves, but have the ability to pass on the affected gene to their biological children.



What are the effects of androgens?

- Androgens make children's bodies look more masculine on the outside.
 - If little testosterone is produced, children with 17 β HSD-3 develop physically like most other girls.
 - If more testosterone is produced, children will develop like most other boys.
- Children with 17 β HSD-3 have no uterus or cervix, because other hormones did not make it possible for these structures to develop.

- Depending on the amount of androgens produced before birth, there may be a:
 - Larger clitoris with or without a urogenital sinus (the vagina and urinary tract open into one channel, rather than separately). The vagina may also be small.
 - Smaller penis with or without hypospadias (the urinary tract does not open at the tip of the penis, but somewhere on the shaft or base of the penis).
- At puberty, the gonads can suddenly make more androgens.
 - More androgens at puberty may cause male typical sex characteristics to develop like lowering of the voice and growth of the genitals.

Is my baby a boy or a girl?

- Because the sex anatomy is different for every baby with 17 β HSD-3, doctors will usually consider how “masculine” or “feminine” the sex anatomy is, and what kind of fertility and sexual function in adulthood can be expected, to make a recommendation.
- Many children will grow up happy even if their genitals, chromosomes and/or gonads are atypical.
- Gender role behavior in children with these syndromes is variable, but there is a tendency, even in children reared as girls, to prefer activities and hobbies typical of boys. This does not necessarily mean that these children are not happy as girls. You can look at the section "Gender of Rearing" for more information about this.
- If the gonads are left in place, production of androgens at puberty may cause male sex characteristics to develop. If this happens, research tells that about half (40 to 60%) of these children are happier to live in a male gender role (even if they were brought up as girls before puberty).

What are the treatment options?

Support and Counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.

Information Guide

- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- Sometimes, a urogenital sinus or severe hypospadias may cause urinary problems. In most children, however, atypical genitals cause no medical problems.
 - A "urogenital sinus" means that there is a single opening for the vagina and urethra. This can happen in girls.
 - "Hypospadias" means that the opening to the urethra is located somewhere other than the tip of the penis. It can be on the underside of the penis, at the base of the penis, or in the scrotum. This can happen in boys.
- Surgery to your child's genitals during childhood may result in irreversible damage to the sensitive nerves.
 - Surgery can lead to problems with sensation and sexual pleasure in adulthood.
 - Surgical revision is often necessary for complications of childhood surgeries, such as narrowing of the urinary tract in boys or vaginal tightening in girls.

Gonadal (testes) monitoring and removal with hormone replacement

- Your doctors may talk to you about frequently monitoring your child's gonads or doing surgery to remove the gonads.
 - Many factors will influence a decision about the gonads. Keep in mind, what is right for one child with 17 β HSD-3 is not necessarily right for another child with 17 β HSD-3.
- The gonad cancer risk in children with 17 β HSD-3 is low before puberty (<1%), but becomes higher after puberty (around 17%).
- Fertility potential is uncertain.
- If gonads are removed, life-long hormone replacement is necessary for overall well-being and bone health.

Information Guide

- Some people with 17 β HSD-3 have a difficult time finding an appropriate hormone replacement that does not cause side-effects.
- Assessments of overall well-being and bone health may be suggested by the healthcare team as a way to ensure that the right dose of hormone replacement is being used.

46, XX Testicular DSD

What is it?

Children with testicular DSD have two X chromosomes and testes. The testes produce androgens, such as testosterone, but a smaller amount of androgens than children with XY chromosomes.

- Most children with 46, XX testicular DSD (about 80%) will physically develop as typical boys, but there may be:
 - A small penis
 - Undescended testes
 - Hypospadias (i.e., when the urinary tract does not open at the tip of the penis, but on the shaft or base of the penis)
- In 80% of children with testicular DSD, the diagnosis is made after puberty, when small testes are noted or breast development (called gynecomastia) begins.
- Because there are two X chromosomes, and no Y chromosomes, germ cells in the testes (the cells which usually grow into sperm) cannot function, resulting in a very low sperm count or no sperm at all.
 - This means fertility is affected and is often poor.

What causes it?

- Usually, 46, XX testicular DSD is not inherited. It results from a random new change in genes and occurs in people with no history of the condition in the family.
- In most children with 46, XX testicular DSD, a change in the SRY gene is the cause.
 - This gene controls the development of the testes.
- Changes in other genes can also cause 46, XX testicular DSD, such as SOX-9.

Is my baby a boy or a girl?

- Almost all children with 46, XX testicular DSD grow up to be happy as boys/men.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- Sometimes, severe hypospadias can cause urinary problems. In most children, however, atypical genitals, such as mild hypospadias or a small penis cause no medical problems.
- Surgery to the penis in childhood may result in irreversible damage to sensitive nerves.
 - This can lead to problems with spraying and/or dribbling and having to sit instead of stand to urinate, as well as appearance.
 - Surgical revision is often necessary for complications from childhood surgery, such as narrowing of the urinary tract or leaks.

Gonadal management and hormone replacement

- There is no increased risk of testicular cancer.
- If the testes are located in the abdomen or groin area instead of the scrotum, doctors may suggest performing an operation called "orchidopexy" in early childhood, to bring the testes down into the scrotum.
- Like all boys, doctors recommend regular follow-up of the testes with self-examination and annual checkups to examine for unusual lumps.
- Extra hormone therapy, usually testosterone, may be necessary for overall well-being and bone health.
 - Some men have a difficult time finding appropriate hormone therapy that does not cause side-effects.

Information Guide

- Assessments of well-being and bone health may be suggested by the healthcare team as a way to ensure that the right dose of hormone therapy is being used.

Congenital Adrenal Hyperplasia (CAH)

What is it?

The adrenal glands sit on top of the kidneys and produce three types of hormones (messengers in the blood):

- Cortisol: controls the stress response in the body.
- Aldosterone: controls blood pressure and the salt balance in the body.
- Androgens: influence the development of sex characteristics, such as underarm hair in puberty and growth of bones.

CAH is a collection of conditions which limit the ability of the adrenal glands to produce cortisol (and sometimes aldosterone).

What causes it?

- The cause of CAH is an inherited genetic change that both biological parents have passed on to their child.
- Often, there is a genetic change in the CYP21A2 gene.
 - Because of this genetic change, enzymes (certain chemicals in the body) that are necessary to make the hormones cortisol and aldosterone are not produced. One of these enzymes is 21-hydroxylase, which is related to the CYP21A2 gene.

What are the effects?

- In CAH, the adrenal glands are unable to produce more cortisol and aldosterone, but they can produce androgens.
 - Because the adrenal glands are continuously stimulated to produce cortisol and aldosterone, but cannot produce these hormones, they instead produce a lot of androgens.
- Because your child produced a lot of androgens before birth, there may be a:
 - Larger clitoris with or without a “urogenital sinus” (the vagina and urinary tract open into one channel, rather than separately). The vagina may also be narrow.
 - Larger penis with or without “hypospadias” (the urinary tract does not open at the tip of the penis, but somewhere on the shaft or base of the penis).

Information Guide

- If the adrenal glands cannot produce enough cortisol, “adrenal crisis” can occur, causing dehydration and vomiting, low blood pressure, and low salt levels. Although this can be life-threatening, it can be avoided with life-long medication.
 - Close follow-up is necessary to make sure your child has the right amount of the essential hormones.

Is my baby a boy or a girl?

- Because the sex anatomy is variable for every baby with CAH, doctors will usually consider how “masculine” or “feminine” the sex anatomy is, and what kind of fertility and sexual function can be expected in adulthood.
- Most children accept their gender of rearing and will grow up happy even if their genitals or chromosomes are not typical.
- Some children who have been exposed to a lot of androgens during development in the womb have been raised as boys and are happy as boys, even if they have XX chromosomes.
- Research tells us that of children with CAH raised as girls, around 95% of them will identify and be happy as girls.
- Gender role behavior in children with CAH is variable, but there is a tendency, even in children reared as girls, to prefer activities and hobbies typical to boys. This does not necessarily mean that these children are not happy as girls. You can look at the section “Gender of Rearing” for more information about this.
- Children with CAH and XX chromosomes have ovaries, and are usually able to conceive naturally in adulthood.



What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby’s body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby’s condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.

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- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section “Support for Families” for more information about these resources.

Genital surgery

- Sometimes, a urogenital sinus or severe hypospadias may cause urinary problems, when urine backs up to the kidney or bladder.
 - In most children, however, atypical genitals cause no medical problems, and surgery is not urgent or medically necessary.
- Surgery to the vagina, clitoris, or penis in childhood may result in irreversible damage to the sensitive nerves.
 - This can lead to problems with sexual pleasure and sensation in adulthood.
 - Surgical revision is often necessary for complications of childhood surgery, such as narrowing of the urinary tract or vaginal tightening.

Endocrine management

- Your child will need medications to replace the hormones that their body is not making.
 - This is not optional.

Complete Androgen Insensitivity Syndrome (CAIS)

What is it?

Children with CAIS have XY chromosomes and gonads (testes). The gonads are usually located in the groin area or belly. These gonads produce hormones called androgens (e.g. testosterone). The bodies of children with CAIS are completely insensitive to the androgens they produce. Therefore, their bodies cannot respond to androgens.

- Children with CAIS will develop with a vagina and clitoris like most other girls.
- The bodies of children with CAIS were sensitive to other hormones during development in the womb. Due to exposure to these hormones, it was not possible for a uterus or cervix to develop.
 - Children with CAIS will not be able to have biological children.

What causes it?

- The cause of CAIS is a change in the androgen receptor gene (AR gene).
- Like a key with a lock, hormones only fit certain receptors. Since there is a genetic change in the androgen hormone receptor (“the lock”), the hormone (“the key”) does not fit closely and will not turn the lock.
- The genetic change is usually inherited via the unaffected biological mother, who passes on an altered copy of the AR gene on one of her X chromosomes.
 - Sometimes, a new genetic change occurs in the biological mother’s egg cells before the child is conceived or during development in the womb.

What are the effects?

- Because the body’s cells are insensitive to androgens, there will be:
 - Female typical external genitals (clitoris, labia, vagina), but the vagina may be small.
 - Natural breast development in puberty, because the body changes some of the androgens into estrogens.
 - Little pubic or underarm hair in puberty.

Is my baby a boy or a girl?

- Most children with CAIS are raised as girls.
- Many children with CAIS are happy as girls/women.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- Some women with CAIS have a small vagina.
- For many women, the clitoris and labia (and not the vagina) give the most sexual pleasure).
- Women with CAIS do not have to change their body in any way to enjoy sex.
- Some women want vaginal sex and look for ways to make their vagina larger.
 - Vaginal dilation with the use of dilators is usually the first choice of treatment, because vaginal tissue is very stretchy and responsive to pressure.
 - If vaginal dilation does not work, vaginal surgery can be done, but dilation will also be necessary after the surgery.

Gonadal management and removal with hormone replacement

- The risk of cancer in the gonads in children with CAIS is low before puberty (<1%), but becomes higher after puberty (around 5-10%).
- Because the gonads naturally produce hormones which are necessary for overall well-being, bone health, and puberty, many doctors will recommend children keep them until puberty is complete.

- At that point, your daughter will also be old enough to be involved in decisions about her gonads.
- Some women want to keep their gonads (and have regular check-ups) and other women want to have them removed.
- If gonads are removed, life-long hormone replacement (usually estrogen) is necessary for overall well-being and bone health.
 - Some women have a difficult time finding an appropriate hormone replacement that does not cause side-effects.
 - Assessments of overall well-being and bone health may be suggested by the healthcare team as a way to ensure that the right dose of hormone replacement is being used.

Complete gonadal dysgenesis (Swyer Syndrome)

What is it?

Children with complete gonadal dysgenesis (CGD) have XY chromosomes and gonads. These gonads did not fully develop into testes, so they are called underdeveloped testes or “streak gonads.” As a consequence, they do not produce typical hormones, such as testosterone.

- Children with CGD will physically develop like most other girls, with a clitoris and vagina (which may be a bit smaller).
- At puberty, there will be limited natural breast development.
 - Children with CGD do not produce estrogens, which promote breast development.
- Women with CGD have a uterus and some women can successfully carry pregnancies after receiving donated, fertilized eggs.
 - Other women have a family by adopting children.

What causes it?

- Changes in a number of genes (e.g. SRY, SOX9, NR5A1, SF1, WT1, WNT4, NROB1 or DHH genes) can be the cause of complete gonadal dysgenesis.
- These genes usually provide instructions for the development and function of several endocrine (hormone-producing) tissues in the body, including the gonads.
- In most cases, complete gonadal dysgenesis is not inherited.
 - It results from new changes in genes and occurs in people with no history of the condition in their family.
- Sometimes, complete gonadal dysgenesis can be inherited, if both biological parents pass on the same gene change (e.g. DHH gene change) or because either parent passes on an affected copy of the gene (e.g. NR5A1 or WNT4 gene change). It can be passed on by the biological father (e.g. SRY gene change on the Y chromosome), or via the biological mother (e.g. NROB1 gene change on the X chromosome).

Is my baby a boy or a girl?

- Most children with CGD are happy as girls/women, even if they have XY chromosomes and gonads.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- Some women with CGD have a small vagina.
- For many women, the clitoris and labia (and not the vagina) give the most sexual pleasure.
- Women with CGD do not have to change their body in any way to enjoy sex.
- Some women want vaginal sex and look for ways to make their vagina larger.
 - Vaginal dilation with the use of dilators is usually the first line of treatment, because vaginal tissue is very stretchy and responsive to pressure.
 - If vaginal dilation does not work, vaginal surgery can be done, but dilation will also be necessary after the surgery.

Gonadal management and removal with hormone replacement

- Since ultrasound and blood tests cannot reliably detect cancer in an early stage, doctors might suggest taking a sample of tissue from the gonad (a biopsy) and look at it under the microscope to assess the risk.
- Many doctors will recommend removing streak gonads before puberty is complete.

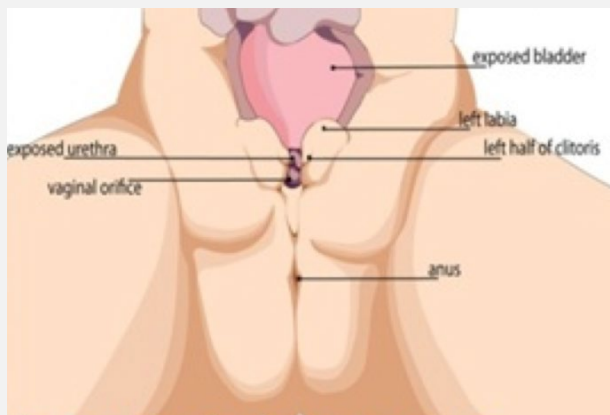
- If gonads are removed, life-long hormone replacement (usually estrogen) is necessary for overall well-being and bone health.
 - Some women have a difficult time finding an appropriate hormone replacement that does not cause side-effects.
 - Assessments of overall well-being and bone health may be suggested by the healthcare team as a way to ensure that the right dose of hormone replacement is being used.

Cloacal and bladder exstrophy

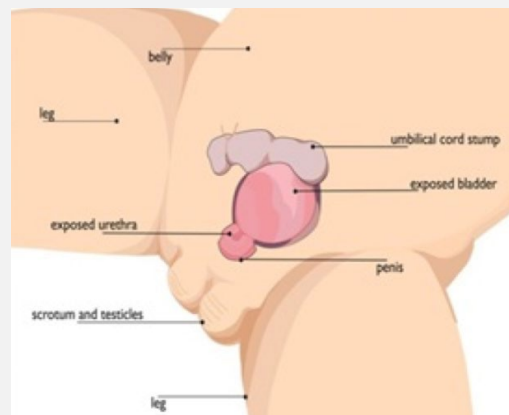
What is it?

- During a baby's development in the womb, the wall of the belly (abdomen) and underlying organs sometimes do not form completely and the baby is born with the bladder or intestines on the outside of the body.

The urinary tract (urethra) is usually shorter and its inner lining is visible on the top surface of the penis (in boys) or labia (in girls). This is also called epispadias.



Urinary tract of a girl with bladder exstrophy



Exposed inner bladder and urethra of a boy

What causes it?

- Bladder/cloacal exstrophy occurs in approximately 2.07 per 100,000 live births and is 5 times more common in boys than girls.
- The cause remains largely unknown, but may be inherited in some families.
- The risk of a family having more than one child with this condition is approximately 1 in 100. Children born to a biological parent with exstrophy have a risk of approximately 1 in 70 of having this condition.

Is my baby a boy or a girl?

- Children with exstrophy with XY chromosomes will typically grow up to be happy as boys/men, and children with exstrophy with XX chromosomes as girls/women.
- However, because exstrophy can affect how children appear and develop, some may have problems with body image or self-esteem.

- Emotional and psychological support can be valuable to help children manage their condition.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- Epispadias may be associated with urinary incontinence and back-up of urine from the bladder to the kidneys (called vesicoureteral reflux).
- Surgery in which the bladder and abdominal wall are closed is an important first step to manage urinary incontinence and vesicoureteral reflux.
 - Later, you and your child will also need to learn how to use a catheter to empty the bladder.
 - Many children need more than one surgery.
- Life-long follow-up care is important to make sure the kidneys and bladder are functioning well.
- Most boys with exstrophy have a small penis. In girls, there may be a small and narrow vagina.
- Different treatment options exist to make the penis or vagina larger.
 - Deciding between these treatment options or preferring not to have treatment is not a parent's decision to be made for their baby.
 - It will be a personal decision of the adolescent or young adult and will be discussed in private with members of the healthcare team.

Gonadal dysgenesis due to Denys-Drash, Frasier, or WAGR Syndromes

What is it?

Children with these syndromes have partial gonadal dysgenesis (PGD). The gonads (testes) are not typically or completely formed. They are also called “dysgenetic testes.” They do not produce as much of the typical hormones, such as testosterone.

- If only a small amount of testosterone is produced, children with PGD will develop physically like most other girls. If more testosterone is produced, children will develop like most other boys.
- Children with PGD do not have typical female internal sex organs, such as a uterus and cervix.
 - In some children, there are typical male internal sex organs, such as a prostate.

Denys-Drash Syndrome: The combination of gonadal dysgenesis, kidney problems, and Wilms’ tumor (kidney cancer).

- Children with XY chromosomes may have partial or complete gonadal dysgenesis.
- Children with XX chromosomes may have partial or complete gonadal dysgenesis.

Frasier Syndrome: Includes gonadal dysgenesis and kidney problems developing in late childhood, but no Wilms’ tumor (kidney cancer).

- Children with XY chromosomes usually have complete gonadal dysgenesis.
- Children with XX chromosomes have renal disease and have normal ovaries.

WAGR Syndrome: This syndrome combines gonadal dysgenesis and Wilms’ tumor (kidney cancer), with the absence of irises in the eyes and intellectual disability.

What causes it?

- Changes in the WT-1 gene can cause Denys-Drash Syndrome and Frasier Syndrome.
- These genes usually provide instructions for making proteins.

- Proteins play an important role in the development and function of several endocrine (hormone-producing) tissues in the body, including the gonads.
- This can be inherited or be the result of a new mutation.

What are the effects?

Depending on the amount of testosterone produced before birth, there may be:

- A larger clitoris with or without a “urogenital sinus.” The vagina may also be small.
 - A “urogenital sinus” means that the vagina and urinary tract open into one channel, rather than separately.
- A small penis with or without “hypospadias.”
 - “Hypospadias” means that the urinary tract does not end at the tip of the penis, but somewhere on the shaft or base of the penis.

Is my baby a boy or a girl?

- Because the sex anatomy is variable for every baby with these syndromes, doctors will usually consider how “masculine” or “feminine” the sex anatomy is, and what kind of fertility and sexual function can be expected in adulthood, to make a recommendation.
- Many children will grow up happy even if their genitals, chromosomes, and/or gonads are atypical.
- Gender role behavior in children with PAIS is variable, but there is a tendency, even in children reared as girls, to prefer activities and hobbies typical of boys. This does not necessarily mean that these children are not happy as girls. You can look at the section “Gender of Rearing” for more information about this.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby’s body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby’s condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.

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- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section “Support for Families” for more information about these resources.

Genital surgery

- Sometimes, a urogenital sinus or severe hypospadias may cause urinary problems. In most children, however, atypical genitals cause no medical problems.
- Surgery to the vagina, clitoris, or penis in childhood may result in irreversible damage to sensitive nerves.
 - This can lead to problems with sexual pleasure and sensation in adulthood.
 - Surgical revision is often necessary for complications of childhood surgery, such as narrowing of the urinary tract in boys or vaginal tightening in girls.

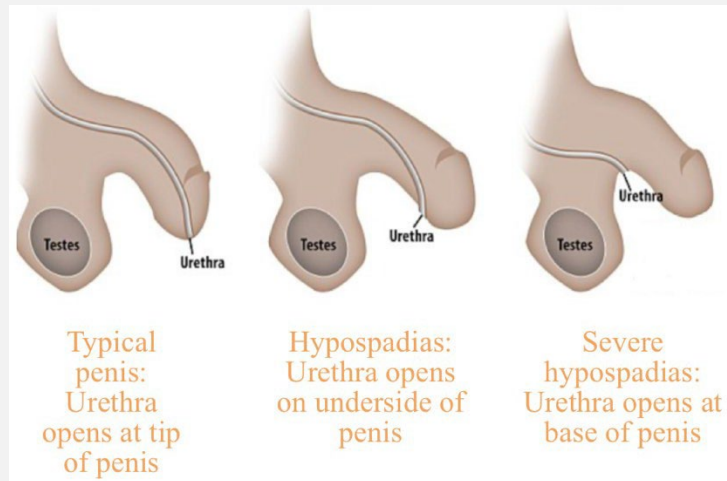
Gonadal management and removal with hormone replacement

- Gonadal cancer risk in children with gonadal dysgenesis is high before puberty (30-50%).
- Since ultrasound and blood tests cannot reliably detect cancer in an early stage, doctors might suggest taking a sample of tissue from the gonad (a biopsy) and look at it under the microscope to assess the risk.
- Many factors will influence a decision about the gonads.
 - Keep in mind, the right approach for one child is not necessarily right for another child.
- If gonads are removed, life-long hormone replacement is necessary for overall well-being and bone health.
 - Some adults have a difficult time finding an appropriate hormone replacement that does not cause side-effects.
 - Assessments of overall well-being and bone health may be suggested by the healthcare team to help ensure that the right dose of hormone replacement is being used.

Hypospadias

What is it?

- The urethra (urinary tract) opens somewhere on the shaft or the base of the penis instead of at the tip.
- Most boys with mild hypospadias have no difficulty urinating, even if the urinary opening is at a different place.
- If the penis is curved upwards or downwards (called chordee), it can be more difficult to urinate while standing.
- 1 in every 250-300 men have hypospadias.



What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- Most children with mild hypospadias will have no urinary problems.
 - Some boys with hypospadias learn to urinate while standing up and some men and boys prefer to urinate while sitting down.

- Doctors may suggest surgery to change the appearance of the penis to look more similar to other boys, or to allow your child to urinate while standing.
- If the hypospadias is severe and there is a problem with urinating, genital surgery may be needed to avoid urinary problems. If there is severe chordee (a curve of more than 60 degrees), erections may be painful.
 - Doctors will tell you if surgery is medically urgent or necessary. For example, when urine backs up into the bladder or kidney, it can cause urinary problems for your child.
- Surgery for severe hypospadias and chordee release can make the penis look longer, but it cannot actually make it bigger.
 - Surgery to the penis in childhood may result in irreversible damage to sensitive nerves. This can lead to problems with sexual pleasure and sensation in adulthood.
 - Surgical revision is often necessary for complications of surgery, such as narrowing of the urinary tract.

If surgery will be done, when?

- Some doctors recommend doing this surgery in childhood, before one year of age because the surgery is easier to perform at a younger age.
- Other doctors recommend waiting until your child is older if the surgery is not urgent or medically necessary, so he can be involved in this decision.

Klinefelter Syndrome

What is it?

Children with Klinefelter Syndrome (KS) have one extra X chromosome in addition to their 46, XY chromosomes (written as 47, XXY). Some children have the extra X chromosome only in some cells, while their other cells have 46, XY chromosomes. This is called “mosaic” KS (written as 46, XY/47, XXY).

- Most children with KS physically develop like most other boys.
- The testes are of a small to typical size at birth, but they do not grow further and do not produce as much testosterone, especially later in puberty. The penis, therefore, usually remains small. In addition, fathering children is usually not possible without surgical help.
- Children with mosaic KS may have milder signs and symptoms, depending on how many cells have an extra X chromosome.
- Some boys and men experience social anxiety or difficulties with social communication and interaction with peers. Others have difficulties with speech, spelling, reading, writing, and attention.
- Some men with KS also have sparse facial and body hair and may have enlarged breasts (called gynecomastia). They are usually tall and have less muscular bodies and weaker bones.

What causes it?

- The chromosomal change usually occurs as a random event during the formation of reproductive cells (eggs and sperm) in a biological parent. For example, an egg or sperm cell may gain one or more extra copies of the X chromosome as a result of an error in cell division.
- KS and its variants are not inherited.
- Biological parents who have a child with KS have a 1 in 100 chance that they will have another child with the condition.
- KS is quite common and occurs in 1 out of 500-1000 live births.

Is my baby a boy or a girl?

- Most boys with KS grow up to be happy as men, even if they have atypical chromosomes or smaller testes.
- Some boys with KS prefer less typical boys’ activities and tend to be more quiet, shy, and sensitive than boys without KS.

What are the treatment options?

Support and Counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- Extra speech therapy can help improve speaking, reading, and writing skills.
- Counselors and therapists can also give boys and men practical skills to help them feel more confident in social settings.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- Although uncommon, some boys with KS have “hypospadias.” If the hypospadias is more severe, it may cause urinary problems. In most children, however, atypical genitals, such as mild hypospadias or a small penis cause no medical problems and surgery is not medically necessary or urgent.
 - “Hypospadias” means that the opening to the urethra is located somewhere other than the tip of the penis. It can be on the underside of the penis, at the base of the penis, or in the scrotum. This can happen in boys.
- Surgery to your child’s genitals in childhood may result in irreversible damage to sensitive nerves.
 - This can lead to problems with sexual pleasure and sensation in adulthood.
 - Surgical revisions are often necessary to correct complications from childhood surgeries, such as narrowing of the urinary tract or leaks.

Gonadal monitoring

- There is usually no increased risk of cancer in the testes in men with KS.

Hormonal

- Because the testes are small and produce little testosterone, extra testosterone treatment will help bring on the usual changes of puberty, including a deeper voice, growth of the penis, and facial and body hair.
 - This hormone treatment is lifelong.
- Testosterone treatment may have additional benefits, including increased physical strength, improved bone density, and reduced breast growth.
 - However, some men have a difficult time finding an appropriate hormone replacement without experiencing side-effects.
- Testosterone therapy cannot increase size of testes or help with fertility.
- Although breast cancer in men is uncommon, there is an increased risk of breast cancer in men with KS.
 - Some men prefer to have cosmetic surgery to reduce their enlarged breasts.
- Physical therapy can build muscle strength and coordination.

45, X/46, XY Mixed gonadal dysgenesis

What is it?

The chromosome pattern in children with mixed gonadal dysgenesis (MGD) is usually 45, X/46, XY. This means some cells in the body have one X chromosome, while other cells in the body have X and Y chromosomes. This is also called mosaicism.

Children with MGD have gonads. The gonads did not fully develop into either testes or ovaries (they are also called “streak” gonads or dysgenetic gonads). Children can have a streak gonad on one side of the body, and a dysgenetic or well-developed gonad (ovary or testis) on the other side of the body. Streak gonads or dysgenetic gonads do not produce as much of the typical hormones, such as testosterone.

- If little testosterone is produced, children with MGD will develop physically like most other girls. If more testosterone is produced, children will develop like most other boys.
- Children with MGD do not have typical female internal sex organs, such as a uterus and cervix.

What causes it?

- Changes in a number of genes (e.g., SRY, SOX9, NR5A1, SF1, WT1, WNT4, NROB1, or DHH genes) can be the cause of mixed gonadal dysgenesis.
 - These genes usually provide instructions for the development and function of several endocrine (hormone-producing) tissues in the body, including the gonads.
- Usually, mixed gonadal dysgenesis results from new changes in genes and occurs in people with no history of the condition in their family.
- Sometimes, mixed gonadal dysgenesis can be inherited, if both biological parents pass on the same gene change (e.g., DHH gene change) or one parent passes on an affected copy of the gene (e.g. NR5A1 or WNT4 gene change).
- It may also be inherited via the biological father (e.g., SRY gene change on the Y chromosome), or via the biological mother (e.g., NROB1 gene change on X chromosome).

What are the effects?

Depending on the amount of testosterone produced before birth, there may be:

- A larger clitoris, with or without a “urogenital sinus.” The vagina may also be small.
 - A “urogenital sinus” is when the vagina and urinary tract share a single channel and single opening.
- A small penis, with or without “hypospadias.”
 - “Hypospadias” means that the urinary tract does not end at the tip of the penis, but on the shaft or base of the penis.

Is my baby a boy or a girl?

- Because the sex anatomy is variable from baby to baby with MGD, doctors will usually consider how “masculine” or “feminine” the sex anatomy is and what kind of fertility and sexual function in adulthood can be expected.
- Many children will grow up happy even if their genitals, chromosomes, and/or gonads are atypical.
- Gender role behavior in children with MGD is variable, but there is a tendency, even in children reared as girls, to prefer activities and hobbies typical for boys. This does not necessarily mean that these children are not happy as girls. You can look at the section “Gender of Rearing” for more information about this.

What are the treatment options?

Support and Counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- Sometimes, a urogenital sinus or severe hypospadias can cause urinary problems. In most children, however, atypical genitals cause no medical problems, and surgery is not urgent or medically necessary.
- Surgery to your child's genitals in childhood may result in irreversible damage to sensitive nerves.
 - This can lead to problems with sexual pleasure and sensation in adulthood.
 - Surgical revisions are often necessary to correct complications from childhood surgeries, such as narrowing of the urinary tract in boys or vaginal tightening in girls.

Gonadal monitoring and removal with hormone replacement

- Dysgenetic gonads may not make enough hormones, so your child may need supplementation.
- If the chromosome test shows Y chromosome material (as in 45, X/46, XY), the dysgenetic gonads may have a risk for cancer.
- The gonad cancer risk in children with MGD is high before puberty (30-50%).
- Since ultrasound and blood tests cannot reliably detect cancer in an early stage, doctors might suggest taking a sample of tissue from the gonad (a biopsy) and look at it under the microscope to assess the risk.
- Many factors will influence a decision about the gonads.
 - Keep in mind, the right approach for one child with MGD is not necessarily right for another child with MGD.
- Dysgenetic gonads may not have the capacity for adequate hormone production or fertility function, which limits the benefits of leaving them in place.
- If gonads are removed, life-long hormone replacement is necessary, for overall well-being and bone health.
 - Some people with MGD have a difficult time finding an appropriate hormone replacement that does not cause side-effects.
 - Assessments of overall well-being and bone health may be suggested by the healthcare team as to help ensure that the right dose of hormone replacement is being used.

Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome

What is it?

- Some girls only have a small vaginal opening (also called “dimple”) because the female internal sex organs (the Müllerian ducts) did not fully develop.
 - These include the uterus, cervix, and upper part of the vagina.
- These girls usually have normally functioning ovaries.
- Sometimes, the kidneys are atypically formed or positioned in unusual places in the abdomen.
 - Sometimes there is only one kidney that may also be unusually placed.
- Other problems may include skeletal problems, hearing loss, ringing in the ears (tinnitus), and/or heart problems.

What causes it?

- No specific genes have been associated with MRKH.
- Usually, MRKH is not inherited and there is no history of the disorder in the family.
 - In some families, it is passed through generations. However, this is difficult to determine because the signs of MRKH vary among affected women from the same family.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby’s body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby’s condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.

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- You can look at the section “Support for Families” for more information about these resources.

Genital surgery

- For most women, the clitoris and labia (and not the vagina) give the most sexual pleasure. Therefore, women with MRKH do not have to change their body in any way to be able to enjoy sex.
- Some women might have to think about genital treatment when they are ready to have sex.
- To be able to have vaginal sex, the vagina may need to be deeper (or longer) and sometimes wider.
 - Usually, vaginal dilation is the first line of treatment, because vaginal tissue is very stretchy.
 - If vaginal dilation is not successful, vaginal surgery may be an option, but dilation will also be necessary after surgery.

Fertility

- Most women with MRKH will not be able to carry children themselves.
- It is possible for some women without a uterus to have biological children via gestational surrogacy.
 - Gestational surrogacy is done by implanting a couple’s IVF (in vitro fertilization) embryos into another woman (the surrogate) who will deliver their baby.
- Uterus transplantation is a relatively new and experimental procedure and involves the risks of an invasive organ transplant surgery, such as organ rejection and infection.

Ovotesticular DSD

What is it?

Some children with ovotesticular DSD have XX chromosomes, while other children with ovotesticular DSD have XY chromosomes. There can also be a combination of XX and XY chromosomes (called mosaicism), meaning some cells in the body have XX chromosomes and other cells in the body have XY chromosomes.

The gonads can be a combination of ovaries, testes, or combined ovarian and testicular tissue (ovotestes). Around 2/3 of babies with ovotesticular DSD have ovotestes. In some babies, there is a testis on one side and an ovary on the other side. As a consequence, the hormones produced by the gonads, such as testosterone, vary from baby to baby with ovotesticular DSD.

- If little testosterone is produced, children with ovotesticular DSD will develop physically like most other girls. Some babies have a uterus, but the uterus is often small. The vagina may also be small.
- If more testosterone is produced, children with ovotesticular DSD will physically develop like most other boys. The penis is usually smaller, and often there is “hypospadias.” The gonads have usually not descended down into the scrotum, but are located in the belly or groin area.
 - “Hypospadias” means that the urinary tract does not end at the tip of the penis, but somewhere on the base or the shaft.

What causes it?

- The underlying cause of ovotesticular DSD remains unknown.
- It is not inherited.
- It usually results from a random new change in genes and occurs in people with no history of the condition in the family.
- In about 10% of children with 46,XX ovotesticular DSD, there is a genetic change in the SRY gene. This gene controls the development of the testes.
 - Changes in other genes, such as DMRT1, SOX9, or RSPO1 are also possible.

Is my baby a boy or a girl?

- Because the sex anatomy is different for every baby with ovotesticular DSD, doctors will usually consider how “masculine” or “feminine” the sex anatomy is, and what kind of fertility and sexual function can be expected in adulthood, to make a recommendation.
- Many children will grow up happy even if their genitals, chromosomes and/or gonads are atypical.
- Gender role behavior in children with ovotesticular DSD is variable, but there is a tendency, even in children reared as girls, to prefer activities and hobbies typical of boys. This does not necessarily mean that these children are not happy as girls. You can look at the section "Gender of Rearing" for more information about this.

What are the treatment options?

Support and Counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby's body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby's condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section "Support for Families" for more information about these resources.

Genital surgery

- Sometimes, severe hypospadias may cause urinary problems. In most children, however, mild hypospadias does not cause medical problems and surgery is not medically necessary or urgent.
- Surgery to the penis during childhood may result in irreversible damage to the sensitive nerves.
 - Surgery can lead to problems with sensation and sexual pleasure in adulthood.
 - Surgical revision is often necessary for complications of childhood surgeries, such as narrowing of the urinary tract or leaks.

Gonadal monitoring and removal with hormone replacement

- The gonadal cancer risk is low in 46, XX ovotesticular DSD.
- It is higher in 46, XY or 46, XX/46, XY ovotesticular DSD, due to the Y-chromosome.
- Since ultrasound and blood tests cannot reliably detect cancer in an early stage, doctors might suggest taking a sample of tissue from the gonad (a biopsy) and look at it under the microscope to assess the risk.
- If there is a testis on one side and an ovary on the other, both gonads can produce hormones. Some doctors will recommend removing at least one of them before puberty, to avoid changes that hormones can bring about.
 - Ideally, this decision will be based on the gender identity preferences of your child and should be postponed until late childhood, when your child can take part in this decision.
- In many children with ovotesticular DSD, one or both gonads have both testicular and ovarian tissue (ovotestes), and it may be more difficult to make a decision about which parts to remove, especially because it is difficult to separate these parts.
- If gonads are removed, life-long hormone replacement is necessary for overall well-being and bone health.

Fertility

- If there is a uterus, some women with ovotesticular DSD can get pregnant via egg donation. Not many women with ovotesticular DSD will be able to conceive naturally.
- Fathering a child biologically is more difficult because not all men with ovotesticular DSD can produce sperm or only produce sperm of a low quality.

Partial Androgen Insensitivity Syndrome (PAIS)

What is it?

Children with PAIS have XY chromosomes and gonads (testes). These gonads are sometimes located in the groin area or belly. They produce hormones called androgens (e.g., testosterone). The bodies of children with PAIS are partially insensitive to the androgens they produce, so these androgens cannot perform many actions in the body.

- If the body is somewhat more sensitive to these androgens, children with PAIS will develop physically like most other boys. If the body is somewhat less sensitive to androgens, children with PAIS will develop more like other girls.
- Because the bodies of children with PAIS are sensitive to other hormones, there is no development of internal sex organs, such as a uterus or cervix.

What causes it?

- The cause of PAIS is a change in the androgen receptor gene (AR gene).
- Like a key with a lock, hormones only fit certain receptors.
 - Since there is a genetic change in the androgen hormone receptor (“the lock”), the hormone (“the key”) doesn’t fit 100% and will not completely turn the lock.
- The genetic change is usually inherited via the unaffected biological mother, who passes on an altered copy on the AR gene on one of her X chromosomes.
 - Sometimes, a new genetic change occurs in the biological mother’s egg cells before the child is conceived or during development in the womb.



What are the effects?

If the body is not completely sensitive to androgens, there may be:

- A larger clitoris with or without a “urogenital sinus.” The vagina may also be small.

- A “urogenital sinus” means that the vagina and urinary tract open into one channel, rather than separately.
- A small penis with or without “hypospadias.”
 - “Hypospadias” means that the urinary tract does not end at the tip of the penis, but somewhere on the shaft or base of the penis.

Is my baby a boy or a girl?

- Because the sex anatomy is variable for every baby with PAIS, doctors will usually consider how “masculine” or “feminine” the sex anatomy is, and what kind of fertility and sexual function can be expected in adulthood, to make a recommendation.
- More than 90% of children with PAIS accept their gender of rearing and will grow up happy even if their genitals, chromosomes, and/or gonads are atypical.
- Gender role behavior in children with PAIS is variable, but there is a tendency, even in children reared as girls, to prefer activities and hobbies typical of boys. This does not necessarily mean that these children are not happy as girls. You can look at the section “Gender of Rearing” for more information about this.
- If the gonads are left in place, production of androgens at puberty may cause male sex characteristics to develop. These can include growth of the genitals or lowering of the voice.
 - If this happens, research tells us that 40 to 60% of these children are happier living in the male gender (even if they were reared as girls).

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby’s body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby’s condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.

- You can look at the section “Support for Families” for more information about these resources.

Genital surgery

- Sometimes, a urogenital sinus or severe hypospadias may cause urinary problems. In most children, however, atypical genitals cause no medical problems.
- Surgery to the vagina, clitoris, or penis in childhood may result in irreversible damage to sensitive nerves.
 - This can lead to problems with sexual pleasure and sensation in adulthood.
 - Surgical revision is often necessary for complications of childhood surgery, such as narrowing of the urinary tract in boys or vaginal tightening in girls.

Gonadal management and removal with hormone replacement

- The risk of cancer in the gonads in children with PAIS is low before puberty (<1%), but becomes higher after puberty (around 10-15%).
- Fertility potential is uncertain.
- Individuals being raised as girls may have gonads removed before puberty to avoid “masculine” pubertal development.
- Many factors will influence a decision about the gonads.
 - Keep in mind, the right approach for one child with PAIS is not necessarily right for another child with PAIS.
- If gonads are removed, life-long hormone replacement is necessary for overall well-being and bone health.
 - Some people with PAIS have a difficult time finding an appropriate hormone replacement that does not cause side-effects.
 - Assessments of overall well-being and bone health may be suggested by the healthcare team as a way to ensure that the right dose of hormone replacement is being used.

Partial Gonadal Dysgenesis

What is it?

Children with partial gonadal dysgenesis (PGD) have XY chromosomes and gonads (testes). These testes are not typically or completely formed, and are also known as “dysgenetic testes.” As a consequence, they do not produce as much of the typical hormones, such as testosterone.

- If little testosterone is produced, children with PGD will develop physically like most other girls. If more testosterone is produced, children will develop like most other boys.
- Children with PGD do not have female typical internal sex organs, such as a uterus and cervix. Sometimes, there may be male typical internal sex organs, such as a prostate.

What causes it?

- Changes in different genes (e.g., SRY, SOX9, NR5A1, SF1, WT1, WNT4, NROB1 or DHH genes) can be the cause of gonadal dysgenesis.
 - These genes usually provide instructions for making proteins. These proteins play an important role in the development and function of several endocrine (hormone-producing) tissues in the body, including the gonads.
- Usually, gonadal dysgenesis is NOT inherited. It results from new changes in genes and occurs in people with no history of the condition in their family.
- Sometimes, gonadal dysgenesis can be inherited if both biological parents pass on the same gene changes (e.g., DHH gene change) or if one parent passes on an affected copy of the gene (e.g., NR5A1 or WNT4 gene change). Sometimes it is passed on via the biological father (e.g., SRY gene change on the Y chromosome), or sometimes via the biological mother (e.g. NROB1 gene change on the X chromosome).

What are the effects?

Depending on the amount of testosterone produced before birth, there may be:

- A larger clitoris with or without a “urogenital sinus.” The vagina may also be small.
 - A “urogenital sinus” means that the vagina and urinary tract open into one channel, rather than separately.

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- A small penis with or without “hypospadias.”
 - “Hypospadias” means that the urinary tract does not end at the tip of the penis, but somewhere on the shaft or base of the penis.

Is my baby a boy or a girl?

- Because the sex anatomy is variable for every baby with PGD, doctors will usually consider how “masculine” or “feminine” the sex anatomy is, and what kind of fertility and sexual function can be expected in adulthood, to make a recommendation.
- Many children will grow up happy even if their genitals, chromosomes, and/or gonads are atypical.
- Gender role behavior in children with PGD is variable, but there is a tendency, even in children reared as girls, to prefer activities and hobbies typical of boys. This does not necessarily mean that these children are not happy as girls. You can look at the section “Gender of Rearing” for more information about this.

What are the treatment options?

Support and counseling

- Talking to a psychologist, counselor, or other professional can help you learn more about your baby’s body as they grow and develop.
- This can also help you become more familiar and comfortable with your baby’s condition and if or how you might choose to share information with others.
- As your child gets older, these same professionals can help you teach your child about their body and can help them answer questions they might have.
- You might also want to talk with other parents of children with the same condition or adults who have it.
- You can look at the section “Support for Families” for more information about these resources.

Genital surgery

- Sometimes, a urogenital sinus or severe hypospadias may cause urinary problems. In most children, however, atypical genitals cause no medical problems.
- Surgery to the vagina, clitoris, or penis in childhood may result in irreversible damage to sensitive nerves.

Clinical management

- This can lead to problems with sexual pleasure and sensation in adulthood.
- Surgical revision is often necessary for complications of childhood surgery, such as narrowing of the urinary tract in boys or vaginal tightening in girls.

Gonadal management and removal with hormone replacement

- The risk of cancer in the gonads in children with PGD is high before puberty (30-50%).
- Since ultrasound and blood tests cannot reliably detect cancer in an early stage, doctors might suggest taking a sample of tissue from the gonad (a biopsy) and look at it under the microscope to assess the risk.
- Many factors will influence a decision about the gonads.
 - Keep in mind, the right approach for one child with PGD is not necessarily right for another child with PGD.
- If gonads are removed, life-long hormone replacement is necessary for overall well-being and bone health.
 - Some people with PGD have a difficult time finding an appropriate hormone replacement that does not cause side-effects.
 - Assessments of overall well-being and bone health may be suggested by the healthcare team as a way to ensure that the right dose of hormone replacement is being used.

Management of the gonads

Why is it necessary?

- If your child has Y chromosomes and the gonads are not fully developed or typically working, cancer cells may develop. That is why these gonads need to be monitored or checked.
- Some diagnoses have a higher risk of cancer in the gonads than other diagnoses.
- In some diagnoses, the risk only becomes higher after puberty; in other diagnoses, the risk is already high during childhood.
- Gonads that are located in the belly or groin area have a higher risk than gonads located lower in the body (e.g., scrotum).

If your child is reared as a girl:

- The small amount of androgens produced by the gonads at puberty may lead to typical male body changes, such as lowering of the voices or growing of the clitoris. Whether these changes will occur and to what extent is difficult to predict.
 - Some girls may be unhappy with their assigned gender as girls. These girls might welcome these body changes.
- If the gonads are left in place until puberty, adolescent/young women can decide for themselves whether to proceed with removal or not.
- After puberty, some women prefer to keep their gonads. Others want to have them removed and will need lifelong hormone replacement therapy (usually estrogens) to maintain overall well-being and bone health.
 - Some women have a difficult time finding an appropriate hormone replacement that does not cause side-effects.
- Unfortunately, there is no evidence-based data regarding preservation of the gonads for future fertility.

If your child is reared as a boy:

- The small amount of androgens produced by the gonads at puberty may lead to male typical body changes, such as lowering of the voice and facial hair growth.

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- When the gonads are located in the belly or groin area instead of the scrotum, the cancer risk is higher and doctors will suggest performing an operation called an orchidopexy in childhood, to bring the gonads down.
- During the orchidopexy, a gonad biopsy (a small sample of tissue) may be collected and examined under the microscope.
 - If cancer cells are detected, removal of the gonads is usually necessary.
 - If no cancer cells are detected in the biopsy, doctors will often suggest regular follow-up of the gonads by self-exam and ultrasound, to monitor the cancer risk. Sometimes an additional biopsy after puberty is necessary.
- It is challenging (but not impossible) for some men whose testes have not been removed to father biological children.
- Extra hormone therapy (testosterone) is usually necessary to maintain overall well-being and bone health.
 - Some men have a difficult time finding an appropriate hormone replacement that does not cause side-effects.

Phalloplasty

What is a phalloplasty?

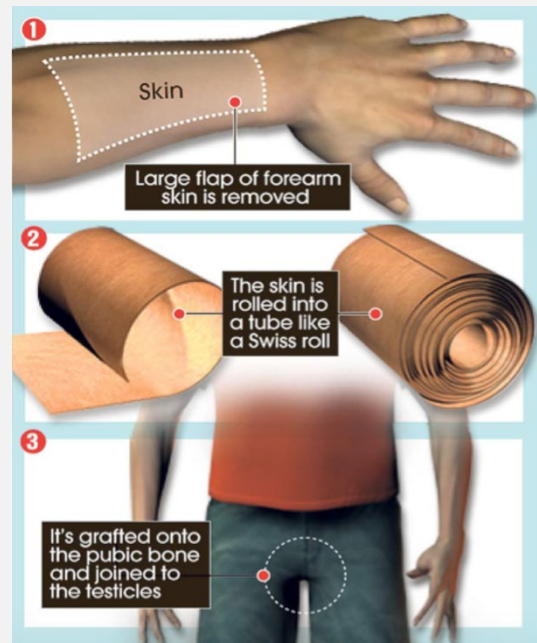
Phalloplasty is a surgery that reconstructs the penis. For instance, it could be used when a man has lost his penis because of an accident. Phalloplasty can also be done to make the penis larger in adolescent or adult men with a small penis. The surgery is not performed on children, as they are still growing.

Why should (or shouldn't) it be done?

- In most men, a smaller penis causes no medical problems and genital surgery is not necessary.
 - Surgery might be suggested by a surgeon when an adolescent/adult man indicates that he is very unhappy with his genital appearance and/or has problems with sex.
- Some boys with a small penis grow up to be happy, healthy men and experience sexual pleasure with no problems.
- Talking with a psychologist or counselor may help boys explore questions and possible concerns about genital appearance and future sexual function.

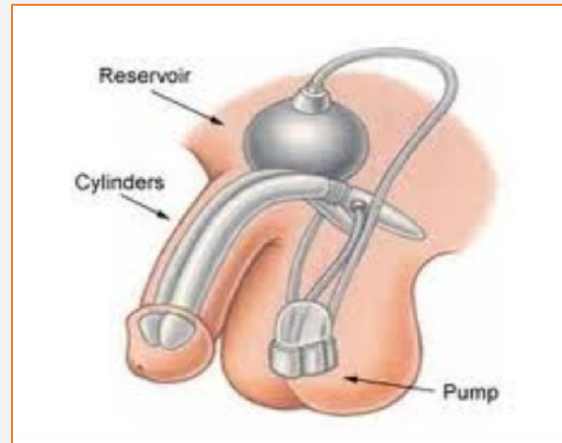
What happens during the surgery?

- Tissue with blood vessels from another part of the body (such as the forearm, leg, chest, or belly) is removed and rolled into a tube.
 - It is then attached using the blood vessels to the existing penis or penile tissue.
- The urinary tract will be relocated to end at the tip of the penis.
- Tissue taken from another part of the body and used for the surgery (the donor site) may take a while to heal and be less sensitive to touch, pressure, and temperature than before.
 - There will also be scar tissue.



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- Later, tissue will be taken to rebuild the donor site (called a skin graft), which will cause a second scar.
- The penis will require an erectile prosthetic (a flexible rod or pump) to become erect.
 - This will be implanted in the scrotum (the pouch containing the testes) during a second surgery.
 - The second surgery has to wait until the penis has healed, which usually takes up to a year.



What are the outcomes of surgery?

- At least 1 out of 3 men will need extra surgery because of complications, such as narrowing of the urinary tract (“stricture”) and/or leaks (“fistulas”).
- Other risks include reduced blood flow to the penis, which can result in loss of all or part of the penis, or rejection of the erectile prosthetic.
- Every surgery comes with the risk of damage to the sensitive nerves of the penis.
 - Some men say that their penis is not as sensitive as they hoped it to be, and experience problems during sex.
- Some, but not all, men are still not happy with how their genitals look after surgery or are bothered by the scars.
- Keep in mind, the approach for one man is not necessarily right for another.
 - Before making decisions about surgery, a man should talk with his healthcare team, support groups, or other men who have faced similar decisions and can give some extra insight into their experiences (with and without surgery).

Surgery for a urogenital sinus

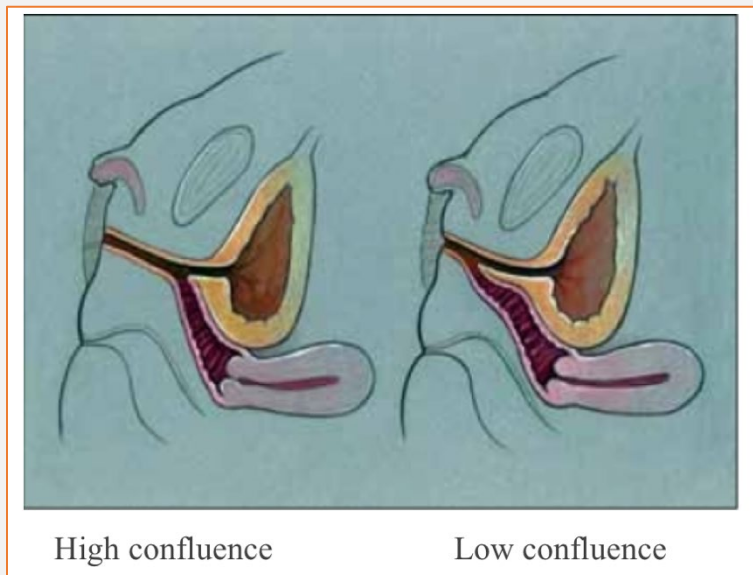
What is it?

Typically, there are separate openings for the urethra and vagina

- The urethra connects the bladder to the outside and lets urine pass through
- The vagina connects the uterus to the outside and, starting in adolescence and adulthood, lets menstrual blood pass through and allows for vaginal sex

A urogenital sinus happens when the vagina and urethra join into a common channel.

- “High confluence” means that the vagina and urethra join higher up in the body and then share one long common channel.
- “Low confluence” means that the vagina and urethra join together closer to the outside of the body and then share one short common channel.



Why might surgery be done?

- There are several reasons surgery might be done, including to:
 - Avoid urinary tract infections (if the urogenital sinus causes urinary problems)
 - Allow for menstrual blood to pass through the vagina (if there is a uterus)
 - Allow for vaginal sex
 - Change the appearance of the genitals
- Doctors will tell you if surgery is medically urgent, for instance if urine backs up into the bladder or kidney or your child has urinary problems.
 - Other times, a urogenital sinus causes no medical problems and surgery is not medically urgent or necessary.

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- If a urogenital sinus causes no medical problems, some doctors will recommend doing this kind of surgery before one year of age, because it is easier to perform in childhood.
 - Other doctors will recommend waiting until after puberty, to allow your child to be involved in the decision.

What kind of surgery would be done?

- A “vaginoplasty” will be performed to reposition the vagina and urinary tract so that they have separate pathways out of the body.
- After surgery, vaginal dilation is usually necessary to keep the vagina open.
 - Many girls will need more operations during puberty because of scar tissue tightening the vagina.

More information

For many conditions, support from others (family, friends, etc.) can be very helpful in dealing with many of the challenges that may arise. The challenges experienced at each stage in life for children whose bodies look or work differently are similar to those with other chronic conditions. Sometimes, families feel that they cannot really share the information they have been told with important people in their lives. It might be helpful to talk to other families who have been through similar experiences (see the “Additional Resources” section), or to a psychologist or behavioral health specialist who has experience working with patients with conditions like your child’s and their families.

Your psychological well-being

In order for your child to thrive, it is essential to think about your own psychological well-being.

The psychologist on the team may ask you to fill out questionnaires to survey your emotions and thoughts about what you have been through or what you are dealing with. These are to help provide the best care for you and your family.

The psychologist will also give you support and advice for dealing with tricky or stressful situations, such as how to manage news about your baby’s birth and how to discuss your child’s condition with other people, including your other children. One effective way to approach this is to first learn about and discuss the diagnosis and what caused it. Practice explaining this with someone close to you and with your child’s doctors.



Psychologists and nurse specialists can be of great help. If you are comfortable and confident sharing information, then your family and friends can be, too. More importantly, practicing these conversations might help you feel more comfortable talking to your child about his

or her body and health in the future. If you feel okay with something, your child is more likely to feel comfortable with it as well.

Thinking ahead: Your child's psychological well-being

When your child is old enough (usually starting at around 8 years of age), the psychologist might start exploring what your child knows about their medical diagnosis and how your child processes what is going on. This can help ensure that your child is feeling happy and healthy. Psychologists can answer some of the questions that your child might have.

Typical topics that are covered include:

- How your child is managing their health at school
- Friend and family relationships
- Your child's hobbies and activities
- Future dreams

Sometimes, questionnaires can be used to survey your child's emotions and psychological well-being.

The psychologist might also help you and your child:

- Explain more about his/her condition in an age-appropriate way
- Explore the benefits and risks of medical treatments (and what it all really means)
- Prepare a long-term care plan

Parents' stories

Sharing information with your child

This may sound challenging now, but raising a child who understands their body is a great principle for all parents. Look at dsdfamilies.org' "Top Tips for Talking" to learn how.

Setting the simple foundations for understanding body differences is easier than waiting to find the "right time" to explain something complicated. Sometimes, if you wait, the information that your child learns later might suddenly change the picture he/she has of him/herself, and this is almost always difficult to manage. Another risk of waiting is that your child may get information through other means, perhaps by overhearing conversations or directly from people who are less cautious. Inaccurate information or snippets of information on their own may alarm a child. Some teens and children wish they had been told everything when they were younger.

When talking with your child about the condition and its management, simplify it for your child's age. Choose your words carefully, based on reality and facts. Your child's healthcare team can work with you to find the right balance for your child and family. Parents sometimes worry about their children telling other people outside your family. A psychologist can help you discuss your child's knowledge and privacy, and help them work out how to decide what information about themselves they share with others.

In general, the best approach is to keep conversation open by mentioning related topics when they come up in other ways (see "Top Tips for Talking"). Follow any questions your child brings up him/herself, but do not wait for your child to initiate conversations about how their body looks or works differently. These questions should be answered in ways that they can understand, but don't worry if things are difficult to explain. Just as with other subjects, parents and children muddle through to reach a shared understanding. The explanations will become more sophisticated and complex as your child ages. This helps them grow up with a healthy understanding of their bodies and what to expect. If you are comfortable and confident about this, then your child might be, too.

Infants, toddlers, and young children

Most babies and toddlers below the age of 3 years are interested in exploring their own bodies. They enjoy positive sensory experiences, and love skin contact and snuggling. We encourage you to allow your child to explore his or her different body parts.



When your child becomes a toddler and learns to talk, you can use dressing and bath time to mention words for the different body parts and genitals. Later, you can tell the healthcare team what you and your child's preferred words are, so that there is a common language. This can help your child learn to participate in discussions and exams, even if they do not understand everything yet. The goal is to make your child feel good about or proud of his/her body and all its parts. It can help to make exams more positive, and really involve the child. If any natural resistance expressed by your child towards exams is respected by you and his/her doctors and handled sensitively, it will help your child feel safe and protected.



As children grow (between age 2 and 5 years), they become aware of gender. They learn that some people are girls and some are boys, and they learn to say "he" or "she." They can use these words to label friends, family, and themselves as a boy or girl. Children also begin to learn "gender stereotypes" – narrow understandings of what males and females are like based on the common images they see and people they know (e.g., a 3-year-old may think that someone with long hair must be female). Stereotypes can be a problem, because many times they are not accurate and do not allow for the range and diversity of people. Stereotypes might make girls who do not like the feeling of long hair worry that there is something wrong with them. Boys and girls who play nurturing roles in games can be encouraged equally. As a parent, you can help your child to explore lots of things and have many different experiences, whatever his/her sex and gender.

Early childhood play that does not fit some societies' gender-role stereotypes does not necessarily mean that your child is experiencing gender unhappiness, and does not relate to sexuality. Often, young children will casually or earnestly say that they are the other sex and will see themselves as growing up as the opposite sex. Sometimes, this is their

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way of saying that they are interested in something that appears available to the other gender, such as being a mother or a fire fighter. By age 6 or 7 years, most children understand and believe that a person's sex stays the same as they become an adult.

If your child expresses the wish to live in the other gender at this point, you can be given advice from an experienced counselor to help you keep options open for your child and minimize his or her distress.

When young children (age 4-6 years) see other children naked, they begin to notice differences and may comment in a nonjudgmental way. They have no standard expectations for how genitals should look, and this is a chance for you to introduce body diversity and range in a calm and accepting way. Children at this age are eager to understand differences and to make sense of them, so they might ask "why" questions such as, "Why does that girl not have a bit sticking out?" or "Why is my baby brother's willy bigger than mine?" If you can prepare for questions by having answers ready, this will help you show your child that his/her body can be talked about without distress or discomfort. This can help them understand and be happy with their body.



A parent's story

Until he was about 4 years old, my son [with a tiny penis and only one visible testicle] bathed with his cousins and played naked in the sprinkler and wading pool in the backyard. If there were any kids there who had not seen him before, I would hover a bit, and listen for any questions. A couple of times children asked things like: "Why does he look like that?" or "What's wrong with him?" I made sure to be close enough to answer... "Oh, that's how he was born, and it's normal for him." Then I would shift to... "We're all different from each other in lots of ways, right?" And then point out something like hair or height or glasses/no glasses differences in the group. In answer to the "what's wrong with him," I said "Nothing's wrong with him. That's how he was born, and it's normal for him. There are other children with privates like his, too."

Children aged 6-10 years old

At this age, children are very busy and eager to find their social roles and to comply with rules and expectations within their friend groups and with their parents. They will meet other people in their social surroundings, who are all different, and some with special health care needs. This is a good time to start talking about long-term health issues, because children at this age are able to have “islands of understanding,” and may begin to be able to explain their development differences or health issues to others.

You can explain differences in body shapes and sizes, and talk about the way your child’s body has developed. For instance, one different path of sex development might mean that a girl might not have a womb, or another will have made a boy’s penis develop differently. You might also give your child some tips about how to talk to others about having differences without giving him/her a feeling that this is something they need to be ashamed of. Children have different ways of handling the body diversity that comes with having a body that looks or works differently: some children see this as private and to be kept in the family or only shared with close friends; other children wish to proudly explain their body to their close friends.

By age 8-10 years old, children may ask questions or make references to parenthood, such as “Where do babies come from?” or “When I have a baby...” Having even a most basic understanding of the reproductive functions of the body will help a child to further understand their different sex development and the potential consequences. Educating children from a young age about the variation in families (e.g., some have adopted children, some couples have no children, some children live with their grandparents, or in foster families) allows them to develop ideas and questions about how these situations arise and to re-think the idea that all adults can have children if they want to.

A story from a mother of a 5-year-old child with AIS

Every once in a while, I find the opportunity to throw another small pebble in my daughter’s “pond” of understanding. “Pebbling” is a way to give information, without causing a huge shocking splash. It allows the body of “water,” or understanding, to expand naturally, growing with each small movement. This was evident when we read books, like “Katy No Pocket,” and discussed how some people were born without a pocket and that it was perfectly okay. It was there when we talked about adoption as a way to have a family and it was there when we discussed how everyone is special. Each

time I take these moments with my daughter, I feel like the load of information I am carrying for her lessens and that she is that much closer to developing an understanding of herself with positivity and acceptance. Last night was one of those nights. While washing her hair in the bath, I began. "Do you know that boys and girls are made with different types of juice?" I asked. "They are? What kind of juice do I have?" So I began. It was that simple. The door was open and all I had to do was put it in words she could understand. "Well, that is a neat story," I began. "Do you remember I told you God made you special?" to which she replied "Yes, I know." I started again. "You see, most boys are born with blue juice that makes them a boy, and girls have pink juice... but- there was one special bottle of blue juice left over, so God gave it to you because he knew you liked blue too." "Really?" she asked, "special blue juice?" "Yes. It is what makes you special," I told her. Minutes passed, and I thought maybe she didn't think it was important, or maybe it was too soon to talk about it. Hormones, after all, are a pretty heavy subject for 5-year-olds. I decided not to push it. When it was time to get out, I wrapped her in a warm towel. "Mommy," she said, "I am so glad you told me that. I just love that I have blue instead of pink juice! You just made my day!" There it was again. She had listened. It may not have been true understanding, but it was there to grow on, another pebble in the pond. The next one will be able to build from there, and she will have grown with the information all along.

Tweens (10-12 years)

Children at this age learn more about complex math, grammar, and biology in school. Now would be a good time to explain in a simple way (perhaps with animations or drawings) the function of the hormones and chromosomes in body development. It is important for your child to understand the basic information because he/she will now be participating more and more in his/her health care. While younger children cannot consent but are asked to participate and are being heard (called "assent"), older children have the privilege to truly participate in decision-making.



A story by a mother of an 11-year-old child with PAIS

When my children were little (ages 3 to 4 years old and onward), I started by always pointing out the ways that we are all different from each other (and pointed out specifics, including how some men are very different from other men, and the same for some women being very different from other women), as well as the ways we are similar. Then I started introducing the term DSD and said that part of their body – their genitals or private parts – had developed a bit differently from many other children. I told them that there were lots of other children whose bodies developed differently in this way too. I mostly wanted them to feel that their differences were a natural part of all the differences that make us human and explain to them something about why we went to see special doctors every 6-12 months who sometimes needed to see their private parts. When they were 5-6 years old (and developing more of an understanding of what makes a boy or girl physically different), I talked with the kids about the fact that there are some “juices” (hormones) in their bodies, and that we all – boys and girls, men and women – have some of each kind of juice “green juice” (testosterone) and “purple juice” (estrogen) that helps us grow and develop. I explained that many boys have more green juice, and only some purple juice, but some kids have different mixtures of the juice and they may develop differently because of that. In terms of explaining the gonads and XY chromosomes, I waited until my daughter was 9 or 10 or so...and the conversation came as a natural extension of earlier conversations about the ways her body is different and similar to girls who don't have a DSD...She understood chromosomes VERY basically as a part of “the map” that gives our body guidance about how to grow. I described to them how everyone's body has a map or plan that it follows when it grows in the uterus and afterwards...The map (chromosomes) can have different roads on it to get to the same place. We talked about how her map (“XY chromosomes”) has a path that can lead to someone being a boy, but it also has a path that many girls with DSDs go on that leads to being a girl. The gonads we explained as things that can become ovaries or testicles in a baby...and that in many children with DSDs they don't develop fully into either...so they can be helpful in growing, but at some point they can stop being helpful and we may need to take them out.

Adolescents

For many young people, it is important that they develop physically at the same time as their friends. The start of puberty is an important topic for children to be aware of. While some may want to be unique, they might also want to be just like they perceive everyone else to be. This means that young people will often agree to medical procedures that aim to make their bodies fit with their image of what is “normal” or usual.

The healthcare team will discuss what health care is available to your child at each stage of development and into adulthood. They can prepare you and your child for what might be offered in the future, and at the same time counsel your child to continue to understand his/her body and developing sexuality. These consultations will involve an increasing amount of time with the professional talking to your child on their own. This is important for their privacy, but it also helps develop the young person’s ability to handle conversations with healthcare providers in adulthood.

You can help by focusing on the emotional well-being of your child and supporting their relationships with friends, perhaps including early romance. Being as body-confident as possible is a challenge for all adolescents, and those whose bodies look or work differently may need extra support from parents to understand why their body might be different, in visible features like height or hidden ones like genital appearance. Girls who do not have periods may need help in deciding whether they let friends know this or not.

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Your Family Name: _____
Relationship to Patient: _____

Date: _____

What I have Learned

Now that you have learned more from this information guide, we would like to ask you the same questions we did at the beginning. You may be surprised by which answers change and which stay the same.

What do you think about following statements?

1. Differences in Sex Development (DSD) are all conditions in which babies do not develop along the most common sex development pathways.

Strongly disagree

Strongly agree

2. Most DSD have a known genetic cause that can be found with genetic testing.

Strongly disagree

Strongly agree

3. DSD are usually not life-threatening conditions.

Strongly disagree

Strongly agree

4. In DSD, genital surgery is not essential for a child's well-being.

Strongly disagree

Strongly agree

5. Genital surgery can be performed at any age, not just on babies or young children.

Strongly disagree

Strongly agree

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6. Genital surgery holds some risks.

Strongly disagree

Strongly agree

7. Children with DSD can question the feelings they have about themselves as boys or girls.

Strongly disagree

Strongly agree

8. People with DSD are just as likely as people without DSD to be attracted to members of the same sex.

Strongly disagree

Strongly agree

9. My child's DSD cannot be kept a secret from my child.

Strongly disagree

Strongly agree

10. Many people with DSD will be able to have biological children.

Strongly disagree

Strongly agree

11. When it is not clear if a baby should be reared as a boy or girl, the decision should be made through discussion between the family and the doctors.

Strongly disagree

Strongly agree

12. Discussion about a child's DSD spans over the course of their lifetime, regardless of if genital surgery is done early in life.

Strongly disagree

Strongly agree

My confidence in gathering information to make decisions for my child

As a parent, part of your job in caring for your child is working with medical professionals to make the best decisions for your child's health. Please rate how certain or confident you are that you can do the things that we list below.

How certain are you that you can...

1. Face all the care decisions that might come along the way?

Not at all A little Somewhat Quite a bit A great deal

2. Get all the information you need (e.g., searching for extra info on your own or with help) to make decisions?

Not at all A little Somewhat Quite a bit A great deal

3. Learn all about the risks, harms, and benefits that matter to you most in these decisions?

Not at all A little Somewhat Quite a bit A great deal

4. Figure out how involved you want to be in decisions?

Not at all A little Somewhat Quite a bit A great deal

5. Work with your partner (your child's other parent) to make decisions that you both agree on?

Not at all A little Somewhat Quite a bit A great deal

6. Identify questions that you want to ask doctors?

Not at all A little Somewhat Quite a bit A great deal

7. Talk to doctors about what matters most to you?

Not at all A little Somewhat Quite a bit A great deal

8. Ask a doctor for more information if you don't understand what he/she said?

Not at all A little Somewhat Quite a bit A great deal

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9. Figure out what to do when you hear information that does not agree with the things you have already heard?

- Not at all A little Somewhat Quite a bit A great deal
-

10. Answer questions about how your child's body looks and works if family members ask you about it?

- Not at all A little Somewhat Quite a bit A great deal
-

11. Talk to your child about their body when your child grows up?

- Not at all A little Somewhat Quite a bit A great deal
-

12. Ask your doctor to put you in touch with families with the same or a similar diagnosis?

- Not at all A little Somewhat Quite a bit A great deal
-

13. Talk to your child about their body when your child grows up?

- Not at all A little Somewhat Quite a bit A great deal
-

Glossary

#

17 β hydroxysteroid dehydrogenase deficiency (17 β HSD) and 5 α reductase deficiency (5 α RD): A genetic condition that results in the inability of the body to make a certain type of androgen (dihydrotestosterone, or DHT for short) in children with 46, XY chromosomes. As a result, the genitals may not have grown much. Usually, children have more female or ambiguous looking private parts at birth, but may become more masculine-looking at puberty if the testes are left in place. Because of the Y chromosome, there is no uterus and no menstruation.

A

Ambiguous: Of doubtful or uncertain nature; difficult to comprehend, distinguish, or classify.

Androgens: Any substance that promotes male characteristics.

Androgen insensitivity syndrome (AIS): A genetic condition that results in the decreased ability of the body to respond to androgens, so that the androgens cannot function properly in children with 46, XY chromosomes. This problem can be partial (PAIS) or complete (CAIS). In children with CAIS, the external genitals are feminine. However, because of the Y chromosome, there is no womb and no menstruation. In children with PAIS, there is a wide range of variation possible in

how the genitals look on the outside, depending on how much the body can respond to the androgens. There is also no womb in these children, because of the Y chromosome.

B

Biochemical: Relating to the chemical processes and substances which occur within living organisms.

C

Cervix: The lower part of the uterus that leads into the vagina.

Chromosome: A threadlike structure of nucleic acids and protein found in the nucleus of most living cells, carrying genetic information in the form of genes.

Cloaca: The cavity at the end of the digestive tract into which the intestinal, genital, and urinary tracts open.

Cloacal and bladder exstrophy: Exstrophy literally means “turned inside out.” In cloacal and bladder exstrophy, a portion of the large intestine and bladder lies outside of the body. In girls, the clitoris is split and there may be one or two vaginal openings. In boys, the penis is usually flat and short, with the exposed inner surface of the urine tube on top. The penis is sometimes split into a right and left half.

Congenital: Relating to a condition present at birth, whether inherited or

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caused by the environment, especially the uterine environment.

Congenital adrenal hyperplasia (CAH): A family of genetic conditions caused by defects in the production of certain steroids by the adrenal glands (*cortisol, the stress hormone and/or aldosterone, the salt balance hormone*). This may lead to an overproduction of androgens, which can cause more masculine-looking genitals at birth (such as an enlarged clitoris) in girls.

Cryptorchidism: One or both testes do not move into the scrotum.

D

Disorder/Difference of Sex Development (DSD): An inborn condition in which development of chromosomes, gonads (ovaries or testes), and internal reproductive and external genitalia are not in line with each other. All DSD are present at birth, but they sometimes go unnoticed for months or years. That is because some DSD only affect how the reproductive organs look on the inside, while others might also affect how the genitals look on the outside, making them more noticeable at birth or soon after birth.

Duct: Any tube, canal, pipe, or conduit by which a fluid, air, or other substance is conducted or conveyed.

Dysgenesis: Defective development, especially of the gonads.

Dysphoria: A state of feeling unwell or unhappy.

E

Epididymis: The cordlike structure along the border of the testes. It provides for storage, movement, and maturation of the sperm cells.

Estrogen: Any of a group of steroid hormones that promote the development and maintenance of female characteristics of the body.

F

Feminization: To cause feminine characteristics (as by implantation of ovaries or administration of estrogenic substances).

G

Gender identity: The sense of oneself as a boy or girl.

Gender role behavior (sex-typical behavior): Behaviors that are culturally associated with boys or girls.

Gender dysphoria: Unhappiness with one's gender and gender role, with the desire for the body and role of the other gender.

Gene: The basic physical unit of heredity; a linear sequence of nucleotides along a segment of DNA that provides the coded instructions for synthesis of RNA, which, when translated into protein, leads to the expression of hereditary character.

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Genotype: The entire genetic constitution of an individual.

Gonad: An ovary, testis, or ovotestis (combined ovary and testis).

Gonadal dysgenesis: The gonads did not fully differentiate on the pathway to becoming testes (also called “streak gonads”) and as a consequence, do not produce any or enough hormones, such as testosterone.

Gonadectomy: Surgical removal of one or both gonads.

Gynecomastia: Breast growth in boys/men.

H

Hormones: A regulatory substance produced in an organism and transported in tissue fluids such as blood to stimulate specific cells or tissues into action.

Hormone replacement therapy (HRT): Involves replacing hormones that the body is not making in “normal” amounts. Some children need hormones that are necessary for the body to survive – such as the stress hormone cortisol. Other children need sex hormone replacement – replacing the hormones estrogen and testosterone, which are hormones responsible for breast development, pubic hair, and development of the genitals in puberty, but also bone growth and general health.

Hypogonadism: A condition resulting from decreased function of the gonads

(ovaries, testes), with delayed growth and sexual development.

Hypospadias: The urethral opening is not on the tip of the penis, but somewhere on the shaft or base.

I

Intersex: One having both male and female sexual characteristics and organs. Now called Disorder/Difference of Sex Development (DSD).

K

Karyotype: Chromosome pattern. Each person usually has 46 chromosomes, including two sex chromosomes, XX or XY. We usually get one X from our biological mother, and an X or Y from our biological father. But there are various combinations of sex chromosomes possible. Sometimes, there is an atypical number of chromosomes, for instance 45 chromosomes (only one X) or 47 chromosomes (XXXY). Other people have combinations of sex chromosomes that differ from cell to cell (called “mosaicism”), with some cells having XX and others XY. Other differences in number or structure of the chromosomes are also possible.

Klinefelter Syndrome: Babies with Klinefelter syndrome have an extra X chromosome (47, XXY), but have male external and internal sex anatomy. However, they have very small testes. When puberty starts and boys’ bodies begin to make sex hormones, boys with

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Klinefelter syndrome usually do not produce as much testosterone, which can affect their penis and testicle growth.

L

Libido: Sexual desire, drive, or interest.

M

Masculinization: Cause to appear or seem masculine.

Menstruation: The periodic discharge of blood and mucosal tissue from the uterus, occurring approximately monthly from puberty to menopause in non-pregnant women.

Micropenis: A penis significantly smaller than the average male's.

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH): In girls with MRKH, the female internal sex organs have not fully developed. Some girls might have ovaries that function, but there is usually no womb or cervix, and no upper vaginal canal. There is a very small vaginal opening (also called a “dimple”). MRKH can also lead to other symptoms. In some women with MRKH, the kidneys are abnormally formed or positioned in different places, and other women have only one kidney that can be positioned in various places throughout the abdomen/belly. Other symptoms include skeletal abnormalities, hearing loss and/or ringing in the ears (tinnitus) and heart defects.

Müllerian ducts: Either of the pair of ducts that are present in the human embryo (alongside the pair of Wolffian ducts). The Müllerian ducts develop into the fallopian tubes and the uterus in typical female development, and disappear in typical male development.

O

Ovary: A female reproductive organ in which ova or eggs are produced, present in humans and other vertebrates as a pair.

Ovotesticular DSD: Both ovarian and testicular tissue are present.

P

Pelvis: Large bony structure near the base of the spine, to which the hind limbs or legs are attached in humans and many other vertebrates.

Perineum: The pelvic floor and the associated structures occupying the pelvic outlet.

Phenotype: What an individual looks like as a result of the interaction of its genotype and the environment – the entire physical, biochemical, and physiological make-up of an individual.

Physiology: The way in which a living organism or bodily part functions.

Primary sex characteristics: The physical characteristics directly involved in reproduction.

Puberty: The period during which adolescents reach sexual maturity and become capable of reproduction.

R

Rough-and-tumble play: Play behavior characterized by overall body contact or playful aggression.

S

Scrotum: The pouch that contains the testes in boys.

Secondary sex characteristics: Any physical characteristic developing at puberty which distinguishes between the sexes but is not directly involved in reproduction.

Sex: Biological or physical features such as genitals, hormones, reproductive organs, and chromosomes.

Sex differentiation: The process of development of the differences between males and females from an undifferentiated zygote.

Sexual dimorphism: Technically meaning “two forms,” mostly interchangeable with “sex difference.” Any psychological or behavioral characteristic that differs on average for males and females.

Sexual orientation: Erotic attraction to/interest in sexual partners of the same versus the other sex.

Sperm: A male reproductive cell.

Surgical reassignment: Surgery to change the sex of a person.

T

Tomboy: (slang) A girl who likes toys, clothes, and activities associated with or usually preferred by boys and who also likes to play with boys.

Transgenderism: A person whose gender identity is opposite the sex the person had or was identified as having at birth.

Turner syndrome: Girls with Turner syndrome are born with only one X chromosome (45 X), or they are missing part of one X chromosome. The effects vary widely among girls with Turner syndrome, although they are usually short in height. In addition to growth problems, Turner syndrome prevents the ovaries from developing properly, which affects a girl’s sexual development and the ability to have children. Because the ovaries are responsible for making the hormones that control breast growth and menstruation, most girls with Turner syndrome will not go through all of the changes associated with puberty until they get estrogen treatment for the condition. Nearly all women with Turner syndrome will need medical assistance to become pregnant.

U

Urethra: The canal conveying urine from the bladder to the exterior of the body.

Urogenital sinus: The vagina and urethra open into a common channel, rather than separately.

Uterus: The reproductive organ in women that is responsible for fetal development.

V

Vanishing testes syndrome: A condition in boys characterized by the absence of the testes at birth. When both testes are absent, the individual will not undergo puberty without testosterone supplements. The testes are thought to have been present in the embryo, but to have “vanished” before completion of

male sexual differentiation in the womb (also called “embryonic testicular regression syndrome”).

Virilization: Masculinization; the development of male physical characteristics in a female or precociously in a boy, typically as a result of excess androgen production.

W

Wolffian ducts: A pair of ducts that are present in the human embryo (alongside the pair of Müllerian ducts). They develop into the male internal reproductive tract if testes are present.

Additional resources

Condition-specific support and resource organizations

Accord Alliance

The Accord Alliance is a US-based organization that brings together patients and families, healthcare administrators, clinicians, support groups, and researchers to facilitate open communication and collaboration among all persons working together to improve care of those affected by DSD. Their website contains a link to an excellent book written by experts, including families and adults living with DSD (Handbook for Parents), other educational materials, information from DSD specialists, and updates on recent research.

dsdfamilies

An extensive information and support resources created by parents, for parents of children, teens, and young adults with DSD. It contains information about medical care and decision-making, as well as peer support. A very useful brochure for parents is the “Early Days” Brochure, for when your child is born with a genital difference.

The AIS-DSD Support Group for Women and Families

This group (formerly AISSG USA) provides access to a teen email group, as well as to the excellent AIS-DSD parent email group.

CARES Foundation: CAH

The CARES Foundation reaches out to families and individuals with Congenital Adrenal Hyperplasia (CAH) via one-on-one support and local support groups across the USA.

Informed Medical Decisions Foundation

An organization that advocates for public policy to help people to set and reach health behavior goals and to improve the care they receive. Includes videos on shared medical decision making and videos on shared decision making in practice.

HEA: Hypospadias and Epispadias Association (USA)

HEA provides support and education of people born with hypospadias or epispadias and their families, loved ones, and medical care given.

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Turner Syndrome Society of the United States

The official website of the Turner Syndrome Society of the United States provides a family guide and clinical practice guidelines, as well as extra information and support from others with TS who have dealt with similar issues.

Beautiful You MRKH

An informative website, providing up-to-date medical information, a link to a MRKH Support and Awareness Facebook page, sister-to-sister mentoring program, and various types of support for all who are affected by MRKH.

Klinefelter's Syndrome Association

Website providing more information and support for individuals with one or more X or extra Y chromosomes and their families.

Sex development in kids

Hospital for Sick Children: About Kids Health- Sex Development Animations

Created by healthcare providers at Toronto's Hospital for Sick Children, it has interactive animations containing detailed pictures and text about sex development and its variations. It also has information about puberty, genetics, and some specific conditions like Congenital Adrenal Hyperplasia (CAH), Androgen Insensitivity Syndrome (AIS), 5-Alpha-Reductase Deficiency syndrome (5-ARD), and hypospadias.

Your Child

Created by healthcare providers at the University of Michigan Health System. It provides information about typical development as well as the development of children with a variety of medical conditions, including DSD.

Disorders of Sex Development: A Guide for Parents and Physicians

A book created by experts, including families and adults living with DSD and providers at the University of Oklahoma Health Sciences Center.

Online medical information resources

HealthFinder

This site is supported by the US Department of Health and Human Services and serves as a gateway to selected consumer health and human services by providing basic information on health topics. You can find health information, and health library, a medical dictionary and encyclopedia, and information on how to locate physicians, support, and self-help groups. Information is available in English and Spanish.

National Human Genome Project

A website that gives you tips on how to find reliable health information online and directs you to some good websites.

Medline Plus

This site is supported by the US National Library of Medicine and the National Institutes of Health (NIH) and provides up-to-date information on a number of different health topics, a medical encyclopedia and a dictionary, drug and medication information, current health news, and links to other related resources. Information is available in English and Spanish.

Information Guide

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