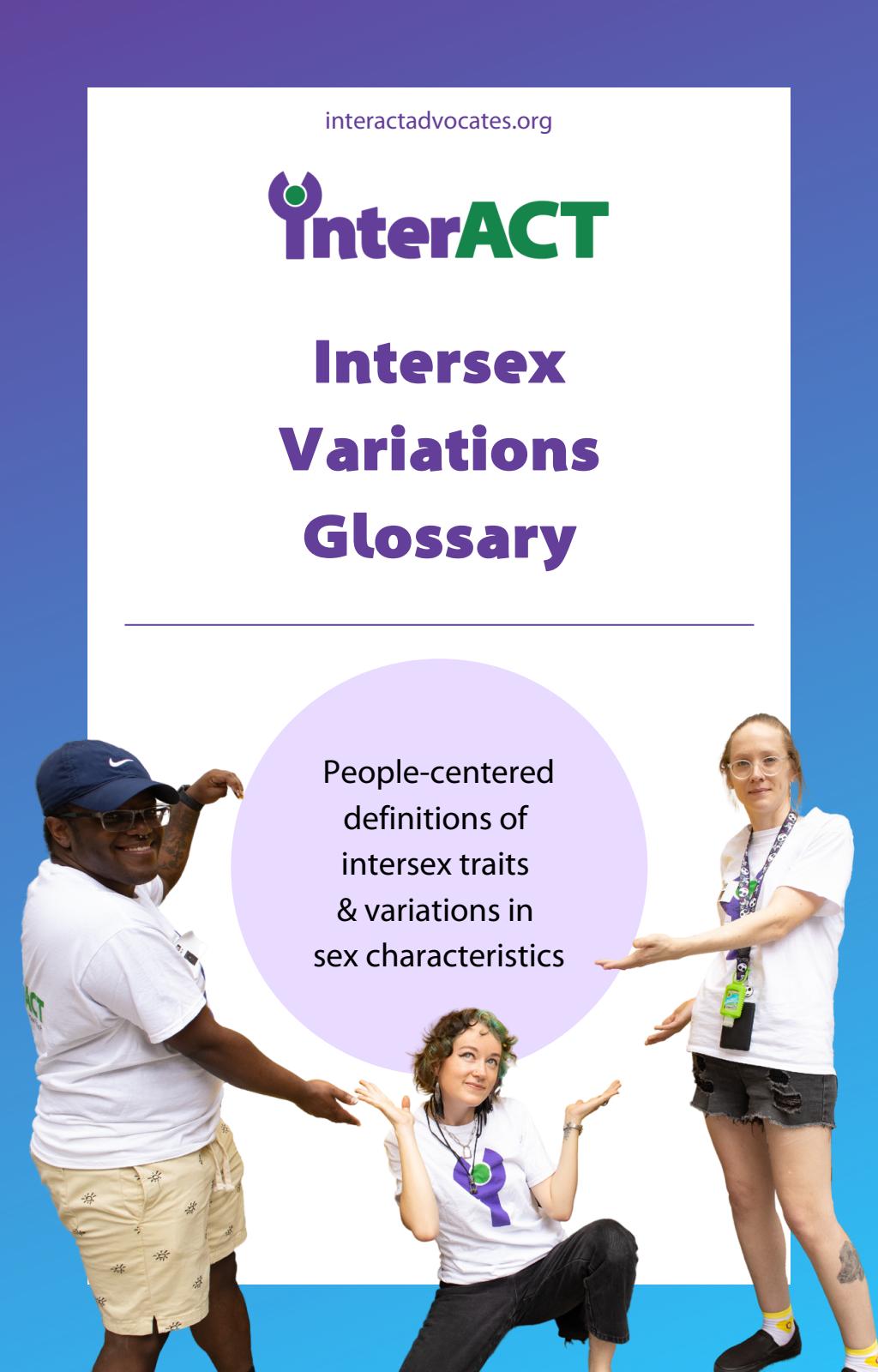




Intersex Variations Glossary

A photograph of three people of diverse backgrounds and expressions. A Black man on the left wears a blue baseball cap, glasses, and a white t-shirt with "ACT" printed on it. A woman in the center has green hair and is wearing a white t-shirt with a purple intersex ribbon graphic. A white woman on the right wears glasses, a white t-shirt, denim shorts, and a lanyard with various items. They are all smiling and gesturing towards each other in a circle.

People-centered
definitions of
intersex traits
& variations in
sex characteristics

INTRODUCTION

Welcome! This glossary was created to provide people-centered, educational definitions about a wide variety of intersex variations and how they can manifest in people's bodies.

Why make this glossary?

While there is information across the internet about these intersex variations, much of this information is:

- Pathologizing, reducing people to “defects;”
- Difficult to understand, written in medical journals or studies;
- Gendered, in a way that may or may not align with each person’s gender identity;
- Not representative of the full scope of how variations can look.

It also can be difficult to find lists of intersex variations gathered in one place, and our hope in gathering them together here is to both raise awareness of intersex variations and counter the isolation people might otherwise feel by showing the wide range of traits that exist under the intersex umbrella.

What this glossary is NOT:

- Fully comprehensive—we are discovering new intersex variations every day.
- A tool to diagnose yourself—we are not doctors, and the information in this resource is not medical advice.
- A definitive guide of what is intersex and what is not intersex—people may disagree about whether certain variations should “count” as intersex, although interACT uses the terms “variations in sex characteristics” and “intersex variations” as synonyms.



Why include the variations we included?

This glossary takes the perspective that intersex is a broad category, including both traits classically thought of as intersex and those that may not be. In this resource, we aim to include definitions for any innate physical trait that falls under the umbrella of variations in sex characteristics, generally meaning that the variation:

- Shows up in a person's chromosomes, genitals, gonads or other internal reproductive organs, or how their body produces or responds to hormones;
- Differs from what society or medicine considers to be "typical" or "standard" for the development, appearance, or function of female bodies or male bodies; and
- Is present from birth or develops spontaneously later in life.



Why include the variations we included? (cont'd)

Some examples of things that would not be considered variations in sex characteristics under these guidelines would be:

- Having an uncircumcised penis (since it is typical to be born with foreskin)
- Experiencing changes related to a typical menopause
- Any alterations to sex characteristics caused by a medical procedure or injury.

Intersex is a spectrum, as is each variation. Not everyone represented by this list will feel that they fall under the intersex umbrella—although we encourage everyone here for whom the term resonates to use it.

If you are only now discovering you might be intersex, we encourage you to get in contact with our interACT Youth Group (for those under 29) or InterConnect Support Group (for anyone!).

We hope that anyone represented in this document, whether or not they identify as intersex, finds the information useful.

interACT: bit.ly/iSpace-intersex

InterConnect: interconnect.support

Who is interACT?

We are an advocacy organization. interACT uses innovative legal and other strategies to advocate for the human rights of children born with differences in their genitals, chromosomes, hormones, and reproductive anatomy.

How can I share this resource?

You might consider talking to staff at your local schools, medical centers, LGBTQIA centers, libraries, or anywhere else that could find a glossary useful, to ask if you can provide them with copies. This glossary is free to print and share.



interACT

A Note on "Typical"

When we are talking about variations in sex characteristics, we are by definition talking about traits that may not be considered “typical” since they do not align with the sex-related stereotypes created by medicine and society.

For clarity, we will use the words “typical” and “typically” in this glossary to mean that a characteristic, combination of characteristics, or a process of development is considered typical based on societal or medical standards.

We will use words like “often” or “generally” when describing the ways that traits tend to show up for a group of people with a specific variation.

No one’s value depends on whether or not their body conforms to sex or gender stereotypes. There is no one normal or “right” way to be a man, a woman, or a person outside of the gender binary.

interACT would like to thank InterConnect Support Group who began this project, and from whom some of our definitions originated.

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GLOSSARY

5-alpha reductase deficiency (5-ARD)

People with 5-alpha reductase deficiency have XY chromosomes, testes, and produce typical levels of testosterone, but their bodies do not have the enzyme responsible for converting testosterone to the more powerful androgen dihydrotestosterone (DHT).

This affects how their external sex characteristics develop, and people born with 5-ARD will often have genital differences that are noticed at birth. Some will have a penis that may be smaller than typical, and some will have genitals that do not look either like a typical penis or vulva. Some others will have a vulva and will be assigned female at birth (possibly without their variation being identified).

In adolescence, people with 5-ARD will likely develop some features associated with a typical testosterone puberty such as increased muscle mass and depth of voice, and they may experience genital growth at this point as well.

17-beta Hydroxysteroid Dehydrogenase 3 Deficiency (17 beta or 17-beta HSD)

People with 17-beta have XY chromosomes and testes, but their bodies do not have one of the enzymes needed to synthesize testosterone. Most people with 17-beta are born with a vulva and vagina, and their testes are most often undescended. Some people with 17-beta will be born with genitals that do not clearly resemble a typical penis or a typical vulva.

Some will have a penis that may be smaller than typical (known as a micropenis), and the urethra may open on the underside of the penis rather than at the tip (known as hypospadias). In adolescence, people with 17-beta will often develop some secondary sex characteristics associated with a typical testosterone puberty.

Androgen Insensitivity Syndrome

People born with AIS have XY chromosomes and testes, and their bodies have varying levels of insensitivity to androgens (including testosterone). This means that their cells do not respond typically to the testosterone that they produce. Like other variations in sex characteristics, AIS is a spectrum, with the subcategories of complete (CAIS), partial (PAIS), and mild androgen insensitivity (MAIS).

Complete AIS (CAIS)

People with complete AIS have no response to androgens and are born with a vulva and clitoris, usually a shorter-than-typical vagina, and undescended testes. In adolescence, they will develop breasts and other features associated with a typical estrogen puberty due to the body's natural conversion of testosterone to estrogen through a process known as aromatization. They will not menstruate since they do not have a uterus or ovaries.

Partial AIS (PAIS)

In partial AIS, the body has some response to testosterone, but not as much as is typical for a person with XY chromosomes without androgen insensitivity. People with PAIS may be born with testes that are undescended or partially descended, and with genital differences such as a vaginal opening which may be shallower than typical, a phallus that may be perceived as a large clitoris or a small penis, and variations affecting their urethra such as penile hypospadias or a common urogenital sinus (where the urethra and vagina merge into one external opening).

They might instead be born with genitals that look more like a penis or like a vulva and vagina. In adolescence, they may develop some features that are associated with a typical testosterone puberty and others that are associated with a typical estrogen puberty since their body responds to some of the testosterone that their gonads produce and the rest will be converted into estrogen.

Mild AIS (MAIS)

People with mild AIS respond to testosterone at a level that is only somewhat reduced compared to typical levels, which means someone with MAIS will usually be born with a penis (which may or may not be smaller than typical) and will later develop secondary sex characteristics associated with a typical testosterone puberty.

Because their bodies will convert a small amount of their naturally produced androgens into estrogen, they may have visible variations in features such as the amount of facial hair, body hair, or muscle tone they develop, and they may experience some potential breast development during puberty.

Anorchia

A person with anorchia has 46 XY chromosomes and is born without testes. As a result, they do not produce typical levels of testosterone. Someone with anorchia will usually have a typical-looking penis and scrotum because these organs are already formed by the stage of fetal development when the testes disappear.

Aphallia

People with aphallia have XY chromosomes, testes, and are born without a penis. This variation is also called penile agenesis. People with aphallia produce typical levels of testosterone and in adolescence they will develop secondary sex characteristics associated with a typical testosterone puberty.

Aromatase Deficiency

A person with aromatase deficiency does not have the enzyme responsible for converting androgens into estrogen (a process known as aromatization). Typically, most of the androgens that are produced by people with XX chromosomes are routinely aromatized into estrogen, but the androgens produced by someone with XX chromosomes and aromatase deficiency will influence the development of their external sex characteristics.

Aromatase Deficiency (cont'd)

At birth, someone with aromatase deficiency and XX chromosomes may have a larger-than-typical clitoris and their labia may be partially or completely fused (resembling the appearance of a scrotum).

At puberty, they may not develop breasts, may not begin menstruating, and may develop some secondary sex characteristics associated with a typical testosterone puberty such as facial hair and muscle mass. They may also develop multiple cysts on their ovaries.

People with XY chromosomes can also have aromatase deficiency, but it doesn't usually cause variations in their sex characteristics (although they may grow taller than typical since estrogen is usually responsible for closing the bone growth plates).

Aromatase Excess Syndrome (Hyperestrogenism)

People with aromatase excess have increased production of aromatase, which converts androgens (like testosterone) into estrogen.

People with XY chromosomes and aromatase excess therefore have more active estrogen in their bodies than people with XY chromosomes who have typical levels of aromatase.

They often stop growing at a shorter height because estrogen is responsible for closing the bones' growth plates, and they may develop breasts in adolescence along with features associated with a typical testosterone puberty. Because aromatase excess is genetic and breast growth in people with XY chromosomes is one of its most recognizable effects, it was formerly called familial gynecomastia.

People with XX chromosomes can also have aromatase excess, but it doesn't usually cause their sex characteristics to vary from what is considered typical for those assigned female at birth (although they may also experience breast enlargement and short stature, along with potential menstrual irregularities).

Bladder Exstrophy

People with bladder exstrophy are born with their urinary bladder exposed (through an open abdominal wall) or outside of the body. Along with an open abdominal wall, they usually have an open pelvis (resulting from the pubic bones not joining together), which also causes genital differences.

People with bladder exstrophy and XY chromosomes often have a shorter-than-typical penis with epispadias (meaning the urethra opens on the top surface of the penis instead of at the tip) and undescended testes.

People with XX chromosomes and bladder exstrophy often have a urethra that is placed higher than usual, widely spaced labia (due to the wide spacing of their pubic bones), and a clitoris that forms as two halves with a split in the middle.

Clitoromegaly

A person born with clitoromegaly has a clitoris that is larger than what society considers to be typical. Clitoromegaly is a trait that can be associated with several different variations, such as Congenital Adrenal Hyperplasia or Progestin-Induced Virilization.

Cloacal Exstrophy

A person born with cloacal exstrophy has an open abdominal wall exposing their colon, bladder, and sometimes other abdominal organs, or these organs may be positioned outside of the body at birth. The bladder is often split into two halves, and the colon and bladder may be connected. The anal opening may be blocked or may not have formed. The pubic bones do not join together (as with bladder exstrophy), resulting in an open pelvis with noticeable genital differences.

People with XY chromosomes and cloacal exstrophy may have a smaller-than-typical penis that is flat or split into two halves, with an epispadic urethra (opening on the top of the penis rather than at the tip) and undescended testes. Someone with XX chromosomes and cloacal exstrophy usually has a clitoris that is split into two halves and may have two vaginal openings.

Chimerism

People that have two or more different sets of DNA are called chimeras. Someone can be born with chimerism when multiple zygotes (such as fraternal twins) combine and develop into one fetus. "Sex-chromosome discordant chimerism" is when at least one embryo with XX chromosomes fuses with at least one embryo with XY chromosomes. A person who develops this way would have sets of both XX and XY chromosomes, for a chromosome pattern of 46XX/46XY, and could have a wide variety of different possible sex characteristics.

Many people with chimerism and XX/XY chromosomes have both ovarian and testicular tissue, while others have only testes or only ovaries, and still others have gonadal dysgenesis. Some have noticeable genital differences, and others do not.

Complete Androgen Insensitivity Syndrome (CAIS)

See Androgen Insensitivity Syndrome (page 10)

Congenital Adrenal Hyperplasia (CAH)

CAH is a group of different but related genetic variations that affect the enzymes that allow the adrenal glands to make specific hormones that help regulate the body's functions. People with CAH do not consistently produce the needed amounts of cortisol (which regulates the response to stress or sickness), aldosterone (which regulate levels of potassium and sodium), or both, and may naturally produce higher-than-typical levels of androgens such as testosterone to help the body compensate.

Someone with CAH can be born with XX or XY chromosomes. People with CAH and XY chromosomes are usually not considered to have a variation in sex characteristics since an increased amount of androgens will not cause the development of any characteristics that are not considered typical for people assigned male at birth.

In people with XX chromosomes and CAH, the increase in androgen production can result in genital differences which may be noticed at birth, including a larger-than-typical clitoris, fused labia, and the fusion of the urethra and vaginal canal to form a single opening. They may also develop some characteristics like body and facial hair or increased muscle mass during childhood or puberty.

Classic CAH

The lower levels of cortisol and/or aldosterone production are more pronounced in a person and the variation is likely to be noticed at birth or shortly after.

Salt-wasting CAH

One subtype of classic CAH that causes “salt-wasting,” a dangerous scenario in which the low levels of aldosterone cause a person’s body to lose too much sodium. People with the salt-wasting form of CAH can experience life-threatening adrenal crises, especially when the body is under stress (for example, due to an illness or infection, or when undergoing a surgical procedure).

Non-classic CAH

Also called “late-onset CAH” and usually doesn’t become apparent in people until later in childhood, adolescence, or even young adulthood.

Congenital chordee

Chordee refers to a bend in the penis. Someone born with chordee has a penis that curves upward or downward, or bends to one side, usually due to bands of fibrous tissue that pull the penis in one direction. People with congenital chordee often have hypospadias as well (a variation where the urethral opening is not at the tip of the penis).

Cryptorchidism

Also referred to as “undescended testicles,” cryptorchidism is a variation in which one or both of the testes do not descend from a person’s abdomen into their scrotum. If both testes are undescended, this is known as bilateral cryptorchidism. If only one of a person’s testes does not descend, this is called unilateral cryptorchidism.

De la Chapelle Syndrome

Also known as “XX Male Syndrome,” this variation causes a person with XX chromosomes to be born with a penis and testes. This usually happens because a particular gene typically seen on Y chromosomes (known as the SRY gene) ends up on one of their X chromosomes and causes their genitals and internal reproductive organs to develop as they typically would in someone with XY chromosomes.

People with De la Chapelle might have testes that are smaller than typical or that are undescended, and they are often infertile. In adolescence, they may experience breast growth, and might not develop the characteristics that are usually associated with a typical testosterone puberty.

Disorders of Sex Development, Differences in Sex Development, or DSD

These terms have been used to refer to a wide range of variations in sex characteristics within medical contexts. Many people with variations consider these terms to be stigmatizing and pathologizing. People may still encounter these terms in their medical records, and many healthcare facilities with specialty centers that see intersex patients have chosen to include “DSD” terminology in the clinic name.

Epispadias

In epispadias, a person is born with a urinary opening located on the upper surface of their penis rather than at the tip (similar to the more common variation hypospadias). People with vulvas can also be born with epispadias, in which case the urethral opening is usually wider than typical and located higher than typical (toward or above the clitoris). The clitoris may also be split into two halves.

Epispadias is usually present whenever bladder exstrophy or cloacal exstrophy develops (where the abdominal wall and pelvis are not closed and the bladder and other organs are exposed or outside the body), but epispadias can also occur by itself.

Estrogen Insensitivity Syndrome (EIS)

People with estrogen insensitivity have a genetic variation where their bodies do not respond to estrogen.

People with XX chromosomes and estrogen insensitivity are usually born with a vulva and vagina, may have a small uterus and enlarged ovaries that later develop multiple cysts, and may produce very high levels of estrogen that their bodies do not recognize. In adolescence, they will most likely not experience the changes associated with a typical estrogen puberty such as breast development or menstruation, and they may develop pubic hair and acne due to the influence of androgens.

People with XY chromosomes can also be born with estrogen insensitivity, and may or may not experience variations in their other hormone production, testicular development, and development of secondary sex characteristics as a result.

Follicle-Stimulating Hormone (FSH) Insensitivity

People with FSH insensitivity have a genetic variation where their bodies do not respond to this hormone. People with XX chromosomes and FSH insensitivity will usually be born with a vulva, vagina, and ovaries. Their ovaries may produce lower amounts of estrogen than typical and may not produce fertile follicles (eggs).

In adolescence, they may not develop secondary sex characteristics associated with a typical estrogen puberty and may not menstruate. People with XY chromosomes and FSH insensitivity may develop smaller-than-typical testes, may produce fewer sperm than usual, and may not be fertile.

Fraser Syndrome

People with Fraser Syndrome are usually born with differences in the development of their eyelids, fingers, and toes, and can have variations in their sex characteristics as well (such as a larger-than-typical clitoris or undescended testes). They may also develop without one or both kidneys, and other parts of the urinary system can additionally be affected.

Goldberg-Maxwell Syndrome

This is another name for Androgen Insensitivity Syndrome (page 10)

Gonadal dysgenesis

Gonadal dysgenesis refers to a group of different variations that affect the development of the gonads before birth.

Complete gonadal dysgenesis

People with “complete” gonadal dysgenesis (also called “pure” gonadal dysgenesis) are born with XX or XY chromosomes and gonadal tissue that has not developed into testicles or ovaries (known as streak gonads).

People with complete gonadal dysgenesis are generally born with a vulva and vagina, and will usually not develop secondary sex characteristics at puberty as their streak gonads do not produce hormones.

Partial or mixed gonadal dysgenesis

A person with “partial” gonadal dysgenesis (also called “mixed” gonadal dysgenesis) is born with a mosaic (usually 45X/46XY) chromosome pattern and may develop some gonadal streak tissue and some testicular tissue.

People with partial or mixed gonadal dysgenesis may be born with a vulva or a penis, or with visible genital variations. They usually have a partially or completely developed uterus, and may have combinations of internal reproductive structures such as a fallopian tube and a vas deferens on opposite sides of the body.

In adolescence, they may develop some secondary sex characteristics associated with a typical testosterone puberty, depending on the amount of hormone-producing testicular tissue that they have.

Hermaphrodite

A term originating from Greek mythology referring to a being that is both fully female and fully male. The term was once adopted in the medical realm to describe people with intersex variations generally (and later to describe someone with both ovarian and testicular tissue specifically), but it is now recognized to be stigmatizing, imprecise, and misleading.

Many people in the intersex community consider the use of this term to be pathologizing and deeply offensive, while other intersex people have chosen to reclaim the term to describe themselves and their bodies. Similar to other reclaimed slurs, you should not use this term to refer to someone else unless you know that they use this term and that they would like for others to use this term to refer to them.

Hyperandrogenism

People with hyperandrogenism have higher-than-typical levels of testosterone and/or other androgens. In people with XX chromosomes, hyperandrogenism can result in the development of secondary sex characteristics that are associated with the influence of testosterone (such as facial and body hair) to varying degrees.

Hyperandrogenism may or may not be the result of an underlying intersex variation, but it is common in several variations (including CAH and PCOS). People with hyperandrogenism who aren't intersex will nonetheless often have many experiences in common with intersex people, such as encountering exclusion and discrimination in sex-segregated contexts like sports.

Hypogonadism

A person with hypogonadism produces lower-than-typical levels of hormones like testosterone and estrogen, or sometimes none at all. This can affect how a person's secondary sex characteristics develop, and can also affect their fertility. Hypogonadism can be a result of a difference in how someone's brain signals their gonads to produce (or not produce) hormones, or it can be a feature of their gonads directly. Hypogonadism is not always caused by an intersex variation, but can be related to Klinefelter Syndrome, Turner Syndrome, or several other variations.

Hypospadias

A person with hypospadias is born with their urinary opening located somewhere other than the tip of the penis. In "distal" hypospadias, the urinary opening is elsewhere on the glans (head) of the penis, and in "proximal" hypospadias, it is on the underside of the penis and is sometimes located further back near the scrotum.

Hypospadias is one of the most common and visible genital variations. People with hypospadias often have additional variations like chordee (a bend in the penis) or cryptorchidism (undescended testicles).

Isolated 17,20-lyase deficiency (ILD)

People with ILD have one of several specific variations in their genes that cause their bodies to make lower-than-typical amounts of testosterone. They may be born with genitals that do not appear quite like a typical penis, and their testes may be descended or undescended.

In adolescence, they may not experience all of the changes related to a typical testosterone puberty because their variation impacts their hormone production.

Jacobs Syndrome

Also known as XYY Syndrome or 47XYY, people with Jacobs Syndrome develop with an extra Y chromosome.

People with Jacobs Syndrome might not have any obvious physical variations as a result, but they may be taller than usual or have other identifiable differences that do not relate to sex characteristics.

Some people with Jacobs Syndrome may also have a smaller-than-typical penis or scrotum, hypospadias, and/or undescended testicles.

Kallmann Syndrome

Kallmann Syndrome can occur in people with XX or XY chromosomes and is a form of hypogonadism. People with Kallmann Syndrome and XY chromosomes are often born with a smaller-than-typical penis and undescended testes.

In adolescence, most people with Kallmann Syndrome will either start puberty later than usual, or may not experience typical pubertal changes (such as developing facial hair and a deeper voice for people with XY chromosomes or developing breasts and starting to menstruate for people with XX chromosomes) unless they receive hormone therapy. Kallmann Syndrome also affects someone's sense of smell.

Klinefelter Syndrome

People with Klinefelter Syndrome develop with an extra copy of the X chromosome, resulting in a 47XXY pattern (instead of the typical 46XY). These individuals might have smaller-than-typical testes that produce lower amounts of testosterone and/or sperm, and they may start puberty later than typical (or may not go through puberty without hormone therapy).

They may grow tall, may develop breast tissue, and often experience infertility. This variation often does not cause obvious physical differences, and many people do not realize they have it.

Leydig Cell Hypoplasia

People with Leydig Cell Hypoplasia (LCH) have XY chromosomes and a genetic insensitivity to Luteinizing Hormone, which can be either complete (known as LCH Type 1) or partial (known as LCH Type 2).

Luteinizing Hormone is typically responsible for the development of Leydig cells in the testicles, and then signals those cells to produce androgens like testosterone. Someone with LCH will develop few or no Leydig cells, and therefore will produce less testosterone than typical.

Leydig Cell Hypoplasia (cont'd)

Type 1 LCH

People with Type 1 LCH produce almost no testosterone and will usually be born with a vulva and vagina and undescended testes.

They are usually assigned female at birth, often without their variation being recognized until adolescence, at which point they generally will not develop any secondary sex characteristics or experience other changes typically associated with puberty.

Type 2 LCH

People with Type 2 LCH have some response to Luteinizing Hormone and produce lower-than-typical amounts of testosterone, but more than people with Type 1 LCH.

At birth, their testes may be descended or undescended, and they may have a smaller-than-typical penis, often with other noticeable differences such as hypospadias. In adolescence, they will often develop some characteristics associated with a typical testosterone puberty.

Luteinizing Hormone Insensitivity

People with XY chromosomes and Luteinizing Hormone Insensitivity develop the variation Leydig Cell Hypoplasia, discussed above. Because people with XX chromosomes do not typically produce Leydig cells to begin with, someone with XX chromosomes who has a genetic insensitivity to Luteinizing Hormone will not be affected in the same way.

A person with XX chromosomes whose body does not respond to Luteinizing Hormone will not usually have any noticeable differences at birth, but their ovaries may not produce fertile follicles (eggs), and they may not menstruate.

Mayer-Rokitansky-Küster-Hauser Syndrome (MRKH)

MRKH is also called Müllerian agenesis because it results when someone's Müllerian ducts, which typically become the uterus and upper portion of the vagina during fetal development, do not develop in the typical way.

People with MRKH have XX chromosomes and are usually born with a vulva and either a vagina that is shorter than typical or with no vagina. They usually do not have a cervix, and may have a partial uterus (uterine remnant) or no uterus.

Type 1

In what is known as "Type 1" MRKH, these are usually the only differences someone will have.

Type 2

In "Type 2" MRKH, people have additional differences in other parts of their body such as their fallopian tubes, kidneys, spine, or heart. A specific example of Type 2 MRKH is known as Müllerian agenesis, Renal agenesis, Cervicothoracic Somite (MURCS) Association, where someone's kidneys do not develop typically and they may have scoliosis or fused vertebrae in addition to the usual features of MRKH.

People with either Type 1 or Type 2 MRKH usually have ovaries that produce estrogen, and in adolescence will generally develop secondary sex characteristics associated with a typical estrogen puberty. They often do not menstruate, but may experience cyclic pain if they have a uterine remnant with endometrial lining, and can develop menstruation-related conditions like endometriosis. (See also: Müllerian Duct Anomalies.)

Micropenis

A person with a micropenis has a penis that is smaller than the typical size range for an infant when a person is born. Someone may develop with a micropenis as a result of several different intersex variations. The size of a person's penis does not generally affect its functionality.

Mild Androgen Insensitivity Syndrome (MAIS)

See Androgen Insensitivity Syndrome (page 10)

Mosaicism

Typical chromosome patterns are either 46XX or 46XY. People with “mosaic” chromosomes have different chromosome patterns in some cells of their body than in others. Mosaicism results from random differences in how cells divide while an embryo is growing.

Because of how this process happens, someone with mosaicism will usually have different numbers of chromosomes in different cells – such as 45X/46XX or 45X/46XY, which are both variants of Turner Syndrome, or 46XY/47XXX or 46XX/47XXX, which are both variants of Klinefelter Syndrome.

(If someone specifically has 46XX chromosomes in some cells and 46XY in others, their variation is probably chimerism instead of mosaicism.)

Having mosaic chromosomes can cause someone to develop variations in their genitals, gonads, or hormone function. For example, if they have at least one Y chromosome, they may have both ovarian and testicular tissue, or genital differences like hypospadias, or they may develop some secondary sex characteristics at puberty that are not expected for the sex they were assigned.

People who have mosaicism without a Y chromosome (e.g., 45X/46XX, or 46XX/47XXX) may have “streak” gonads or may have ovaries that stop functioning at an earlier age than typical, which can cause someone not to experience the changes associated with a typical estrogen puberty and can cause infertility later in life.

Other people may not have any noticeable signs of their mosaicism at all.

Müllerian Duct Anomalies

People with XX chromosomes can have a broad range of variations in how their Müllerian ducts (which typically form the uterus, fallopian tubes, cervix, and upper portion of the vagina) develop. People with MRKH, also known as Müllerian agenesis, are at one end of this spectrum, and other people have variations related to their vaginal, cervical, and/or uterine development that may not be as noticeable. For example, someone might have a variation that causes their uterus to develop with a different shape than typical, or to be doubled.

Others can have vaginal variations such as a “hemivagina” (where one side of the vagina is obstructed, which can sometimes block menstrual blood from exiting), or a band of tissue dividing the vaginal canal (known as a vaginal septum). Someone else might have a doubled cervix or a cervix with a septum dividing it. Müllerian Duct variations usually don’t cause someone to have any variations in their ovaries or hormone production.

Ovotesticular DSD (Ovotestes)

People with this variation are born with both ovarian and testicular tissue. This can happen because of a chromosomal variation like chimerism or mosaicism, but most people who have both ovarian and testicular tissue have XX chromosomes, and some have XY.

A person can develop one testis and one ovary, or may develop one or more ovotestes, which means that a single gonad is made up of ovarian and testicular cells together. The levels of hormones they produce can vary, and how their genitals and secondary sex characteristics develop will depend partially on the levels of estrogen and testosterone that their bodies make.

They may be born with genitals that look more like a vulva, more like a penis, or that have visible variations. In adolescence they may develop features associated with either a typical estrogen puberty or a typical testosterone puberty, or they may develop some of each. Sometimes a person with an ovary and a testis or with ovotestes can produce viable sperm and viable eggs.

Partial Androgen Insensitivity Syndrome (PAIS)

See Androgen Insensitivity Syndrome (page 10)

Penoscrotal Transposition

People with penoscrotal transposition are born with a different genital configuration than typical, with their penis located below or in the middle of their scrotum instead of above it. Their scrotum might be bifid (split into two halves), and they may also have hypospadias and/or chordee.

Persistent Müllerian Duct Syndrome (PMDS)

People with PMDS have XY chromosomes, a penis and testes, and also may have a uterus, fallopian tubes, and/or upper vaginal canal. PMDS occurs when the Müllerian ducts—internal structures that typically break down in a fetus with XY chromosomes—remain and begin to develop as they would in a fetus with XX chromosomes.

PMDS is usually not noticed at birth, but someone may be discovered to have PMDS if they start experiencing pain or a hernia.

Progestin-Induced Virilization

A person born with progestin-induced “virilization” has XX chromosomes and may have visible genital variations at birth such as a larger-than-typical clitoris, sometimes also with fused or partially fused labia, and may or may not have differences in their vagina. They usually have typical ovaries, fallopian tubes, and a uterus.

This variation develops in utero as a result of their parent taking progestin medication while pregnant. Being exposed to these additional hormones before birth can change how someone’s genitals develop, but generally will not change how their own body will produce or respond to hormones later on.

This means someone born with progestin-induced virilization will usually develop features associated with a typical estrogen puberty.

Pseudohermaphroditism

This is a term that has previously been used in medical contexts to refer to different groups of variations, usually divided into categories of “male pseudohermaphroditism” and “female pseudohermaphroditism” on the basis of a person’s chromosomes. People may still find these terms used in their medical records or in medical journal articles when researching their variation, but they are often considered highly stigmatizing in addition to being vague and inaccurate.

Polycystic Ovary Syndrome (PCOS)

People with this variation have XX chromosomes and are generally born with a vulva and vagina, a uterus, and ovaries. Later in life, their ovaries often produce higher-than-typical levels of androgens like testosterone. (PCOS is a common cause of hyperandrogenism, but not everyone with PCOS has hyperandrogenism.)

This variation in hormone production can result in the development of some characteristics such as facial and body hair and can also interfere with the process of ovulation, which means that some people with PCOS will have irregular periods or no periods and may struggle to become pregnant if they want to.

Some people with PCOS have many small cysts (fluid-filled sacs) on their ovaries, but some do not. (Despite the name of the variation, ovarian cysts are not a requirement for someone to be diagnosed with PCOS.) Signs of PCOS generally will not show up until a person starts menstruating, or potentially later into adolescence or adulthood.

Reifenstein Syndrome

An older, alternative term for Partial Androgen Insensitivity Syndrome (PAIS).

See Androgen Insensitivity Syndrome (page 10).

Sex Reversal Syndrome

This term is sometimes used in medical contexts to refer to different variations in which a person is born with XY chromosomes and a vulva and vagina (such as Complete Androgen Insensitivity Syndrome or Swyer Syndrome), or XX chromosomes and a penis (such as de la Chapelle Syndrome).

Swyer Syndrome (Complete Gonadal Dysgenesis)

Swyer Syndrome occurs when a person is born with XY chromosomes, but their gonads have not developed into testes.

Swyer Syndrome is a form of gonadal dysgenesis, meaning that a person with this variation will have “streak” gonads (fibrous tissue that is neither testicular tissue nor ovarian tissue) that do not produce hormones.

Since they do not produce testosterone, their body does not develop a penis and usually develops a vulva and vagina. Since they do not produce another hormone known as anti-Müllerian hormone (AMH), they often develop a uterus and fallopian tubes as well.

They will not usually start puberty or begin menstruating, but some people with Swyer Syndrome do menstruate (without ovulating) if they receive estrogen therapy.

Trisomy X (Triple XXX Syndrome)

In this variation, a person is born with 47XXX chromosomes instead of the typical 46XX. If they get the additional X chromosome from their parent's original sperm or egg cell, they will have 47XXX chromosomes in all of their cells.

If the additional chromosome appears early during the embryo's development process instead, only some cells will have 47XXX chromosomes and the person will be born with a mosaic chromosome pattern.

(Examples of mosaicism involving Trisomy X would be 46XX/47XXX or 45X/47XXX chromosomes.)

Most people with 47XXX chromosomes do not develop any variations in their other sex characteristics, but some may have ovaries that stop producing hormones at an earlier than typical age, and some may not menstruate.

Turner Syndrome (XO Syndrome)

Turner Syndrome is a variation in which a person is born with a 45X chromosome pattern (sometimes called 45XO) instead of the typical 46XX.

Turner Syndrome can also occur in combination with mosaicism, where some of a person's cells have 45X chromosomes and other cells have a different pattern, such as 46XX or 46XY. This is known as Mosaic Turner Syndrome.

People with Turner Syndrome may have some specific physical features such as short stature, a webbed neck and broad chest, and medical problems with their heart.

They can also develop variations in their other sex characteristics as a result of their chromosome pattern, such as smaller-than-typical ovaries that may stop producing hormones at a younger age than typical, and they might not menstruate or experience pubertal changes.

People with Mosaic Turner Syndrome who have 45X/46XX chromosomes may have fewer noticeable signs than people who have a 45X chromosome pattern in all of their cells. For example, someone with Turner Syndrome is more likely to go through a typical estrogen puberty and to start menstruating if they have a 45X/46XX chromosome pattern.

Someone with a 45X/46XY chromosome pattern may be born with testicular tissue that produces hormones, and they often go through a typical testosterone puberty if so.

People with this form of Turner Syndrome may be born with a penis and scrotum without any visible variations, or they may have genital differences such as a smaller-than-typical penis or hypospadias, or else they may have a typical-appearing vulva and clitoris.

Vaginal Atresia

People with this variation may be born without a vaginal opening, sometimes with a shallow “dimple” where an opening would typically be, or they may have a vaginal opening that is fused or blocked by fibrous tissue.

Someone with vaginal atresia will often have a typical upper vaginal canal (along with a typical uterus, cervix, fallopian tubes, and ovaries) with differences in the lower portion of the vagina only.

XXYY Syndrome

In this variation, someone is born with an extra copy of both an X and Y chromosome, resulting in an 48XXYY chromosome pattern instead of the typical 46XY.

People with XXYY chromosomes may often have undescended testes and produce lower-than-typical amounts of testosterone, which can cause them to start puberty at a later age than typical and to develop less pronounced secondary sex characteristics like muscle tone and facial hair. They may also experience breast growth.

They often have other physical features that are similar to those developed by people with Klinefelter Syndrome (47XXY).

CONCLUSION

interACT hopes you found this resource valuable—and if you are a person with one of the variations listed here, we hope this glossary helps you feel less alone.

If you have any questions, please feel welcome to explore our website at interactadvocates.org or reach out at info@interactadvocates.org.

We are a small organization and create resources, advocate for legal change, and lift intersex people up in the media on a budget that is partially fueled by generous donations from individuals.

If you can be one of our supporters, consider making a recurring donation at bit.ly/donate-IAD. Thank you for your support of all people with intersex variations!

