



The

Story of Sex Development

by dsdfamilies

Concept

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Reference

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Thanks also to Nina Callens, Naomi Crouch, Justin Davies and members of the Scottish Differences of Sex Development Network for input at various stages. EM

Dedication

Dedicated to my mum – who rooted for and adored her granddaughter and wished for openness and understanding among families. EM

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The Story of Sex Development

In this booklet we track the main factors that influence the sex development journey from embryo to birth.

Did you know...

About **150 babies** are born in the UK every year who travel along an unexpected sex development path.



Sex development occurs between week 8 to week 14-16 of pregnancy - when the foetus grows from the size of a raspberry to the size of a peach!

This story of sex development is for:

- People who want to learn more about their bodies
- Parents and others who want to understand sex development and the tests that doctors sometimes do

Throughout this story we deliberately use some of the words that doctors use, to help you link this story to the things they say about you or your child.

Sometimes...

Sometimes different sex development affects the internal reproductive organs only.

Sometimes someone will notice that a baby's genitals do not look as expected.

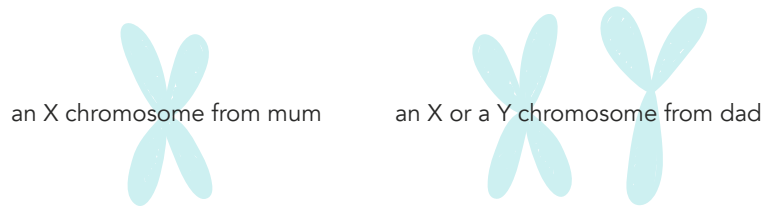
The very beginning

Sex development happens to everybody. And it starts with the information we get from our biological parents.

This information is the chromosomes and the genes that the chromosomes carry.

Most people get 23 chromosomes from mum and 23 chromosomes from dad, so we end up with 23 pairs.

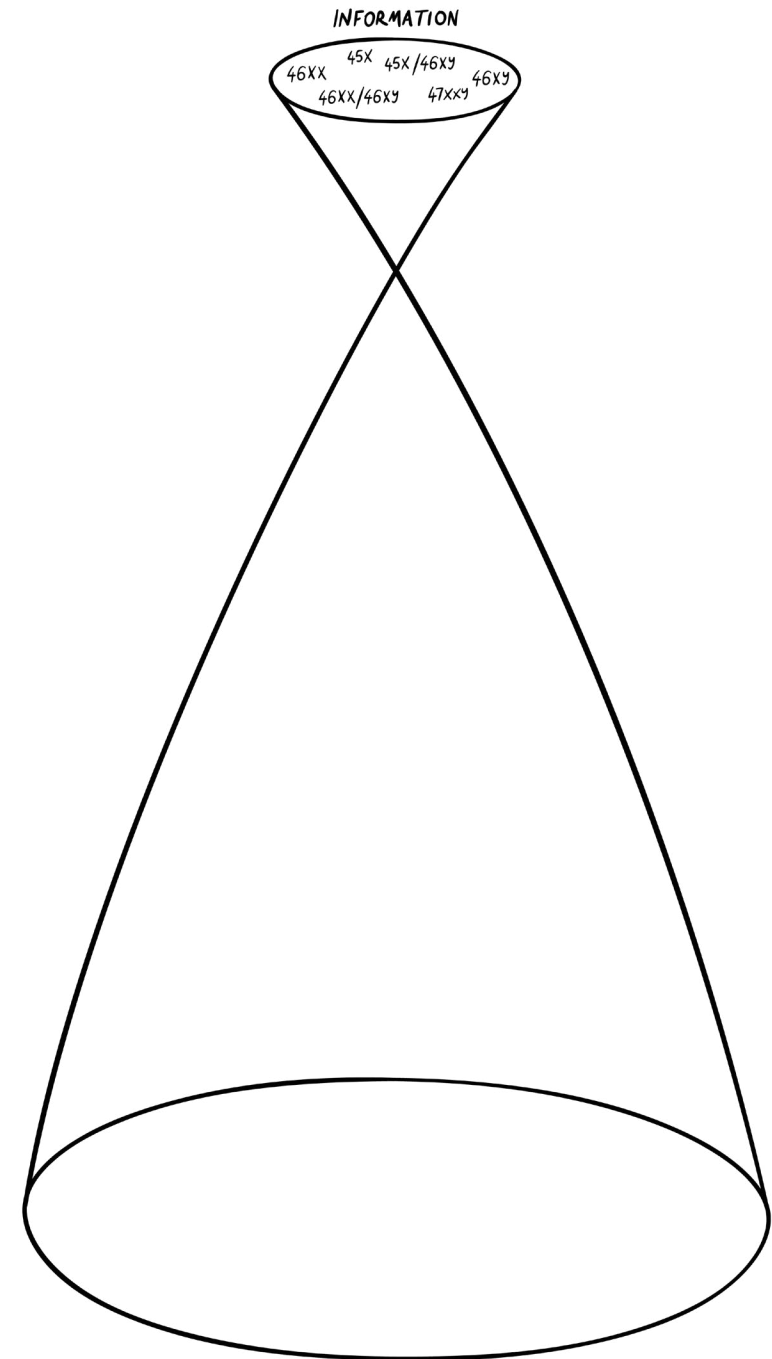
That usually includes:



This means that when we speak about chromosomes, we usually speak about 46,XX and 46,XY.

- Sometimes people will have fewer or more chromosomes, for example 45,X and 47,XXY
- And sometimes people will have unusual combinations, like 45,X/46,XY or 46,XX/46,XY

What are your / your child's chromosomes?



The first 8 weeks of pregnancy...

Up until week 8 of a pregnancy (this means: week 6 after conception) all babies develop in a similar way.

This means:

- A baby (the little fetus!) develops **gonads** that can become either an ovary or a testis, or a variant of these

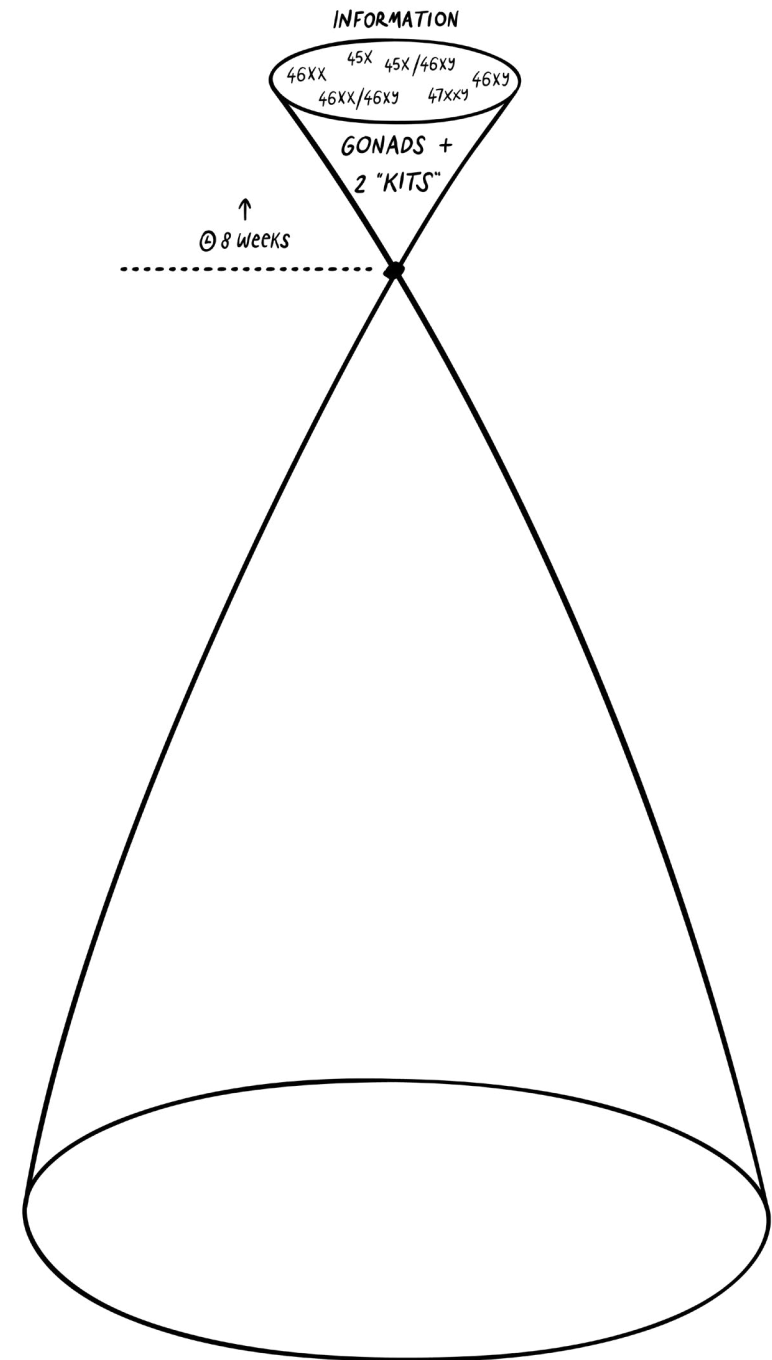
Gonads is a word that is used to describe both testes and ovaries. Gonads have an important role in producing hormones (see steps 5 and 6).

They also contain the **germ cells** which typically develop into sperm (testis) or eggs (ovary).

- At the start of development all babies develop 2 different sets of 'reproductive organs':

The **typical female kit** can become the womb, the fallopian tubes, and the upper part of the vagina.

The **typical male kit** can become the epididymis, vas deferens and seminal vesicle (the 'bits' that help store sperm, tubes that get sperm out and the bit that makes ejaculate).



Gonadal development - the instructions for the gonads to develop

There is one key gene that influences how the gonads develop.

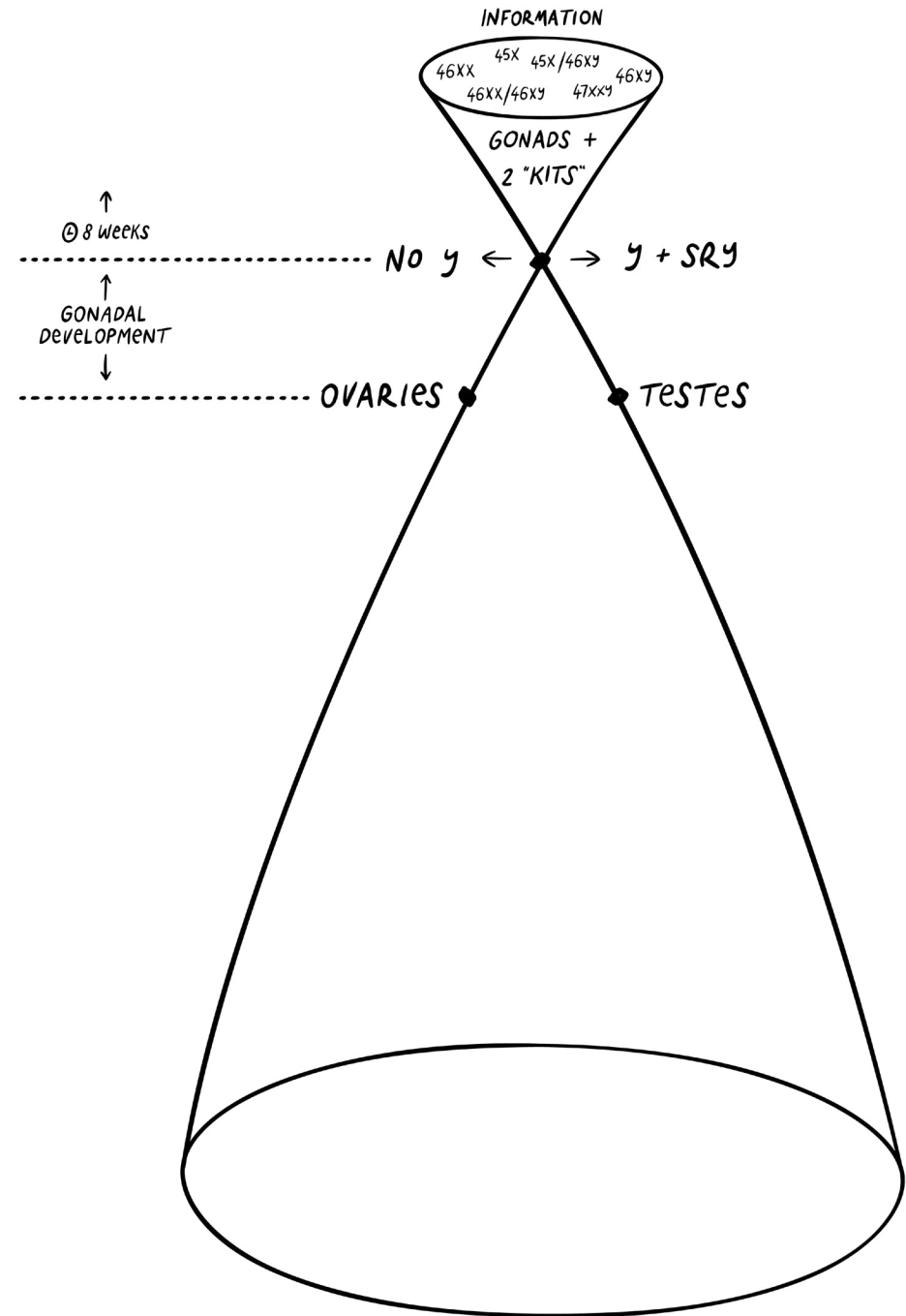
That gene is called the [SRY gene](#). And the SRY gene can almost always be found on the Y chromosome. So...

NO y y + SRY

If there is [no Y chromosome](#), a baby will begin to develop ovaries.

If there is a [Y chromosome](#) with the [SRY gene](#), a baby will begin to develop testes.

Doctors find out about chromosomes by using a [karyotype or microarray test](#). Initial results can take 2–3 days with a full report following later.



More on the development of the ovaries and testes

When there is no Y chromosome (and/or no SRY gene) the gonads usually develop into ovaries.

When there is a Y chromosome (and an SRY gene) the gonads are instructed to develop like testes.

For some babies with a DSD this process can result in:

- fully developed testes
- underdeveloped testes (sometimes called *dysgenetic*)
- streak gonads (this means just some tissue, without any function)
- ovotestes
- or a mix of the above

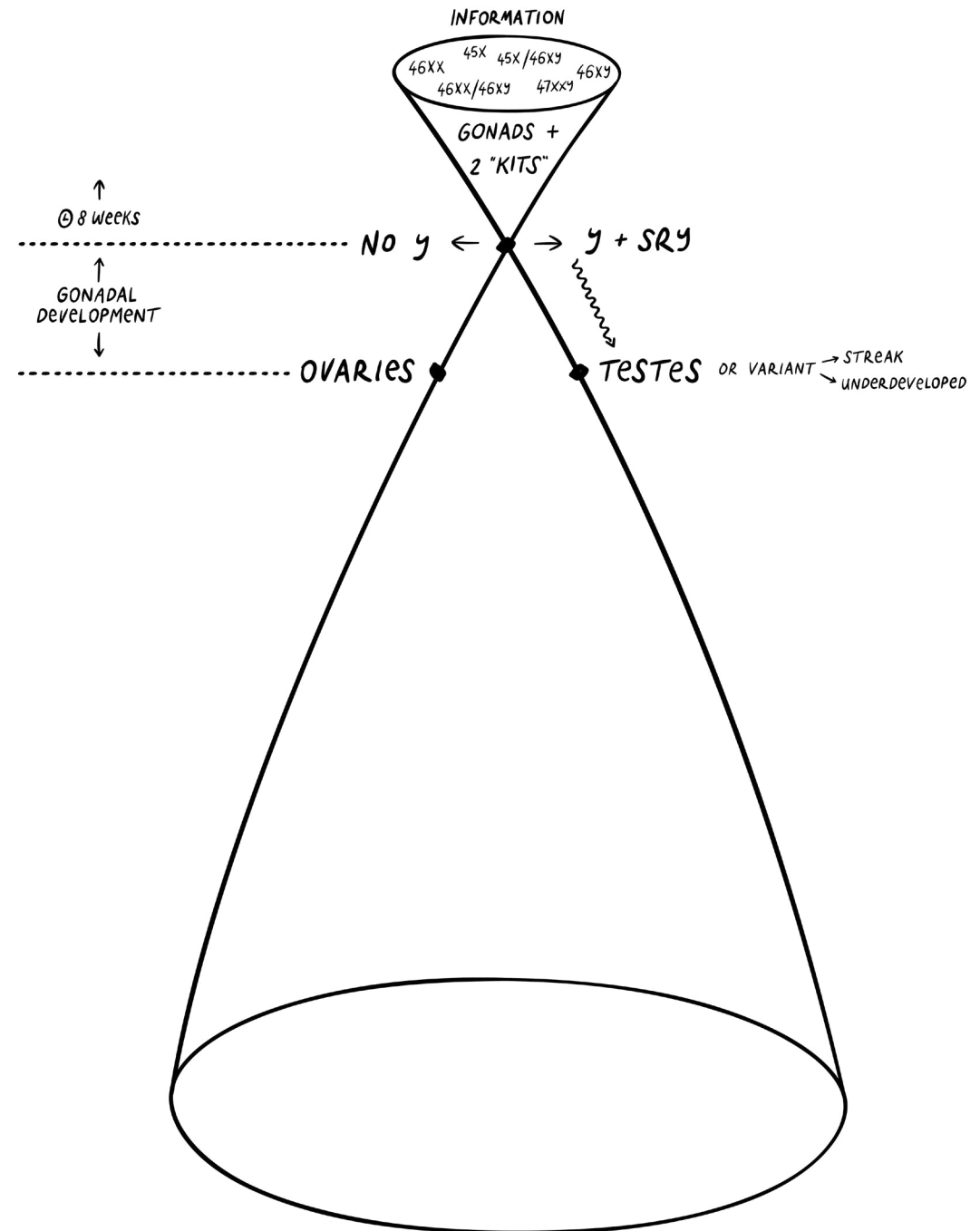
The presence or absence of the SRY gene triggers a cascade of developmental processes.

The outcome depends on numerous genes, enzymes and proteins working together in a coordinated way.

Proteins are like 'the minions', the workers of the body. And enzymes convert (change) hormones from one form to another.

Did you know...

Sometimes gonads may develop differently on either side of the body.



Hormone production and the development of the reproductive organs (inside the body)

How the gonads developed will impact on what type of hormones they produce and how much.

Hormones are chemical 'messengers' produced in one organ that have an effect elsewhere in the body.

OVARIES

The ovaries do not produce active hormones during development.

The 'typical female kit' (the womb, the fallopian tubes and the upper part of the vagina) simply grows as part of natural development of the fetus.

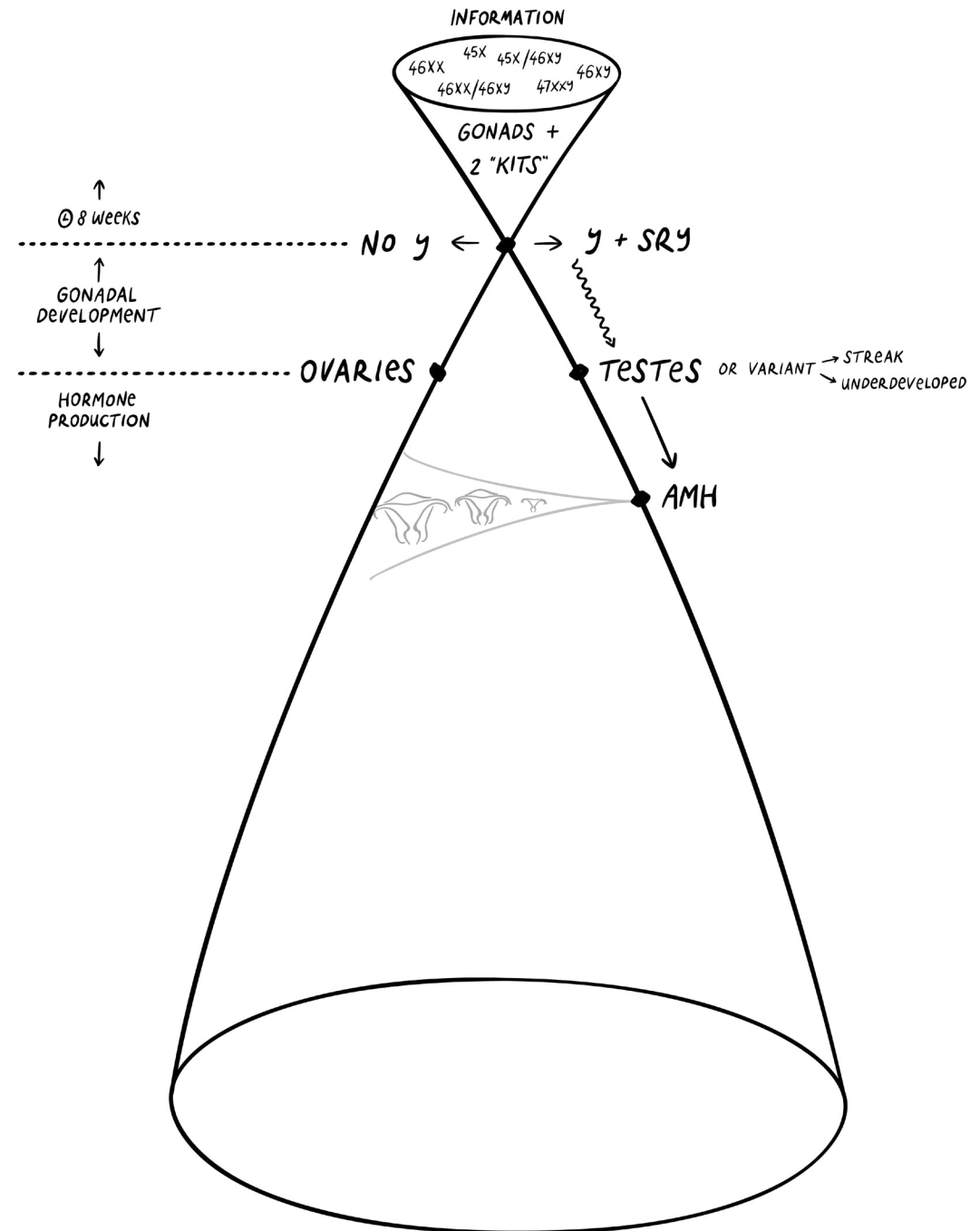
The ovaries do not produce the testosterone needed to grow the typical male kit, and it gradually disappears.

TESTES and VARIANTS of testes

The testes produce two main hormones: Anti-Mullerian Hormone (AMH) and Testosterone (T).

AMH 'gets rid of the typical female kit' (womb, fallopian tubes and upper part of the vagina).

- If the testes have fully developed, then the amount of AMH will make all these structures disappear.
- If the testes have not fully developed there will be less AMH and the typical female kit might not completely disappear. This means there may be some remnants of the typical female kit. Sometimes this occurs on one side of the body and not the other.
- If no testes have developed (streak gonads) then there will be no AMH and the typical female kit will simply remain in the body.



What else do testes produce? Testosterone!

Testes also produce [Testosterone \(T\)](#).

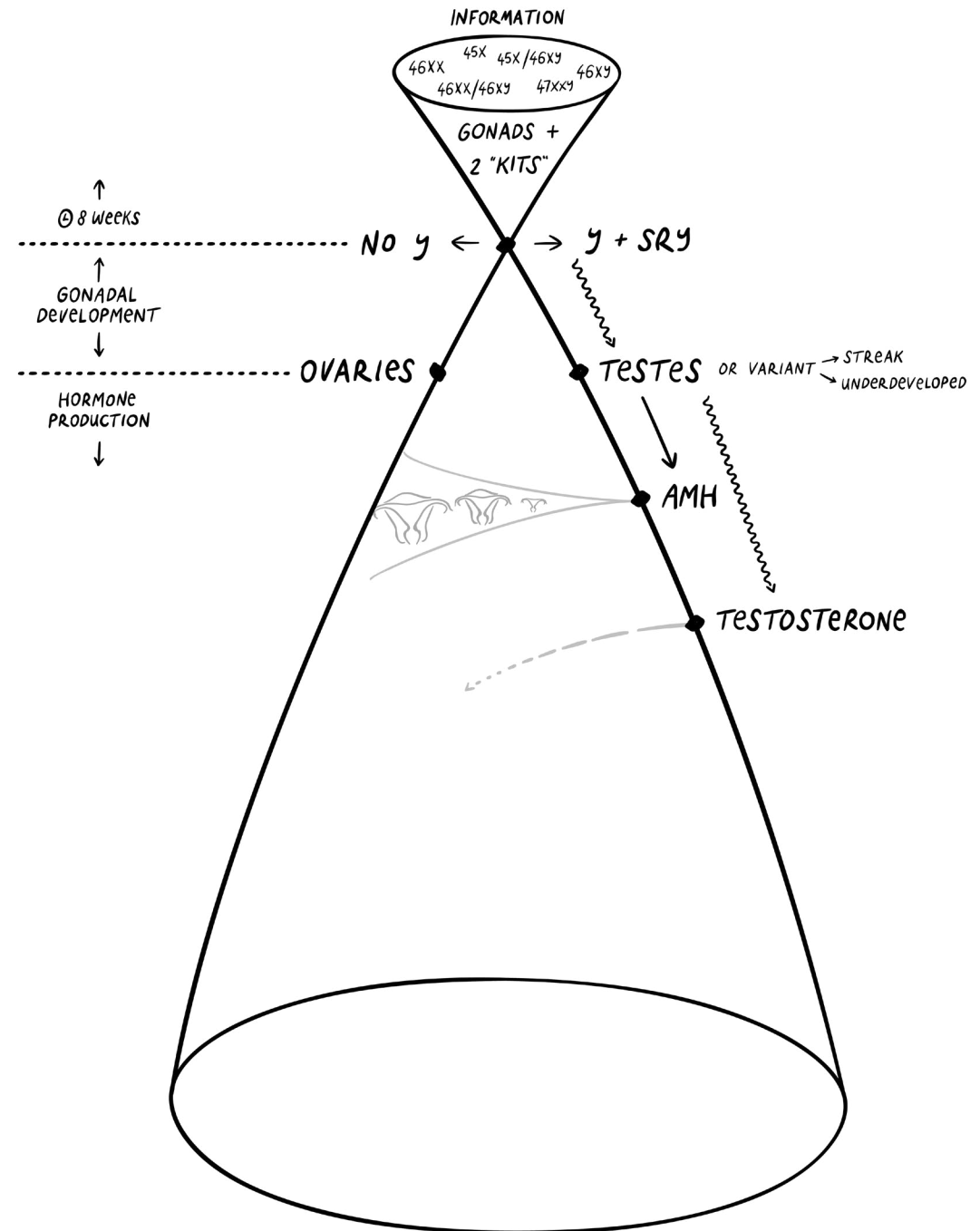
Testosterone is also a hormone, and it is a type of [androgen](#).

Testosterone 'stabilises' the typical male internal structures. Remember: these are the epididymis, vas deferens and seminal vesicle (the 'bits' that help store sperm, tubes that get sperm out and the bit that makes ejaculate).

Usually, testes produce a certain amount of T.

But when testes are underdeveloped or not developed at all, less T or no T will be produced.

And sometimes the enzymes needed to make T just don't function or don't work, and less T or no T is produced.



Changing T to SuperT!

The next step is converting T into **SuperT** or *dihydrotestosterone* (DHT). SuperT, like T, is a hormone that is sometimes called an **androgen**.

SuperT is a very powerful hormone that changes the genital appearance on the outside of a baby. This means: the growth of the penis or the clitoris, fusion of the labia and development of the scrotum.

How does this happen?

Testosterone is usually **converted** into SuperT by enzymes that are present in the developing genitals.

When underdeveloped testes produce a smaller quantity of testosterone or T, conversion to SuperT is also reduced, and there will be less SuperT.

Sometimes, the enzymes (controlled by the 5 Alpha gene) don't convert (work) properly or at all – so less or no SuperT will be produced.

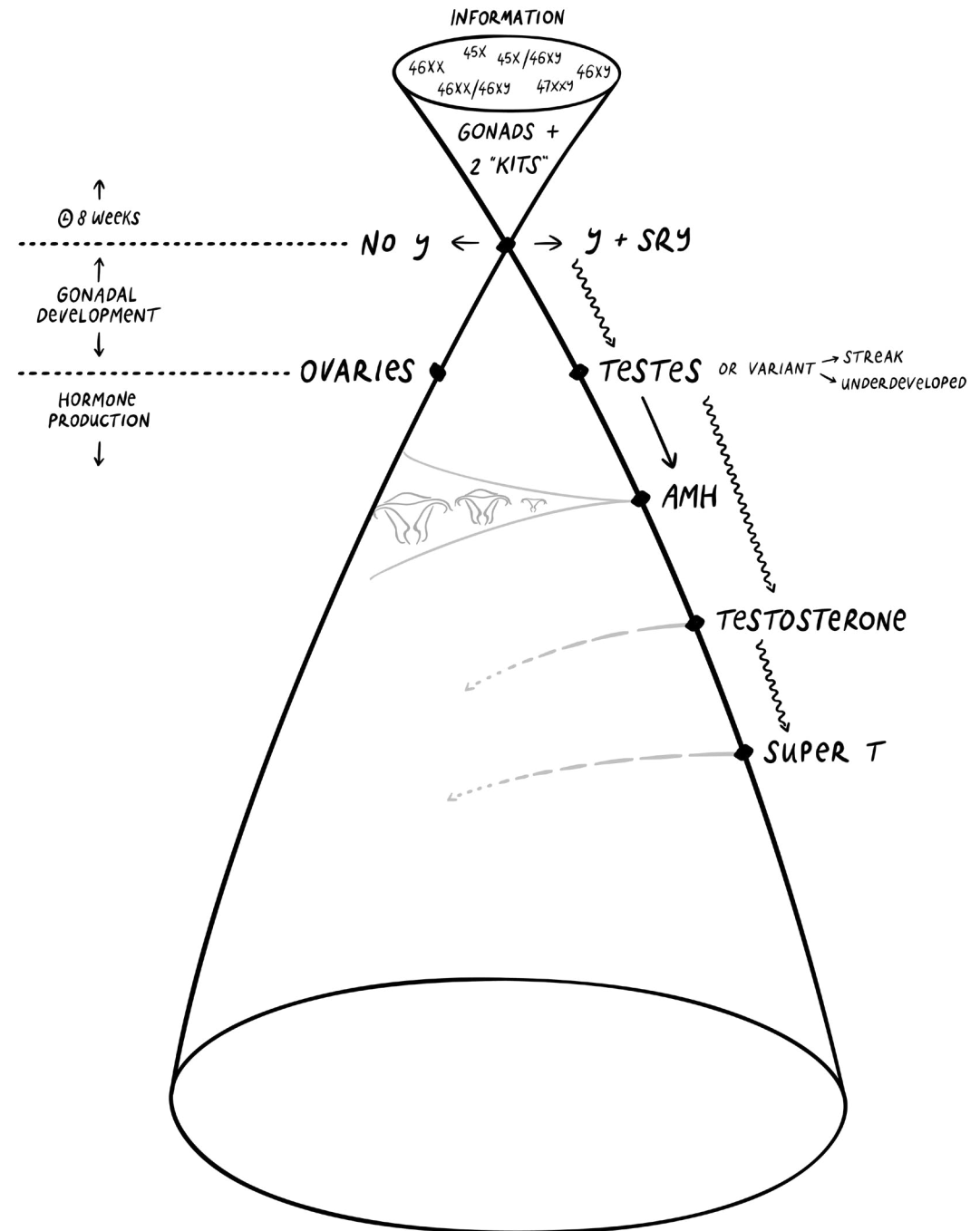
The baby's genital appearance depends a great deal on **how much** SuperT is converted.

Need to know more...

Children with some specific forms of DSD going into puberty may be able to produce T and convert to SuperT, even if as babies in the womb they did not produce T or convert to SuperT. Their bodies and individual organs find exceptional ways to do this.

This means that children might unexpectedly become exposed to more androgens as they go into puberty. This relates to children with 17betaHSD, with 5 Alpha, some with SF-1 and possibly some with PAIS – and only if they still have their gonads.

Going through puberty is a long process, and changes take place over many years. Good care involves children in all discussions and their agreement with every step.



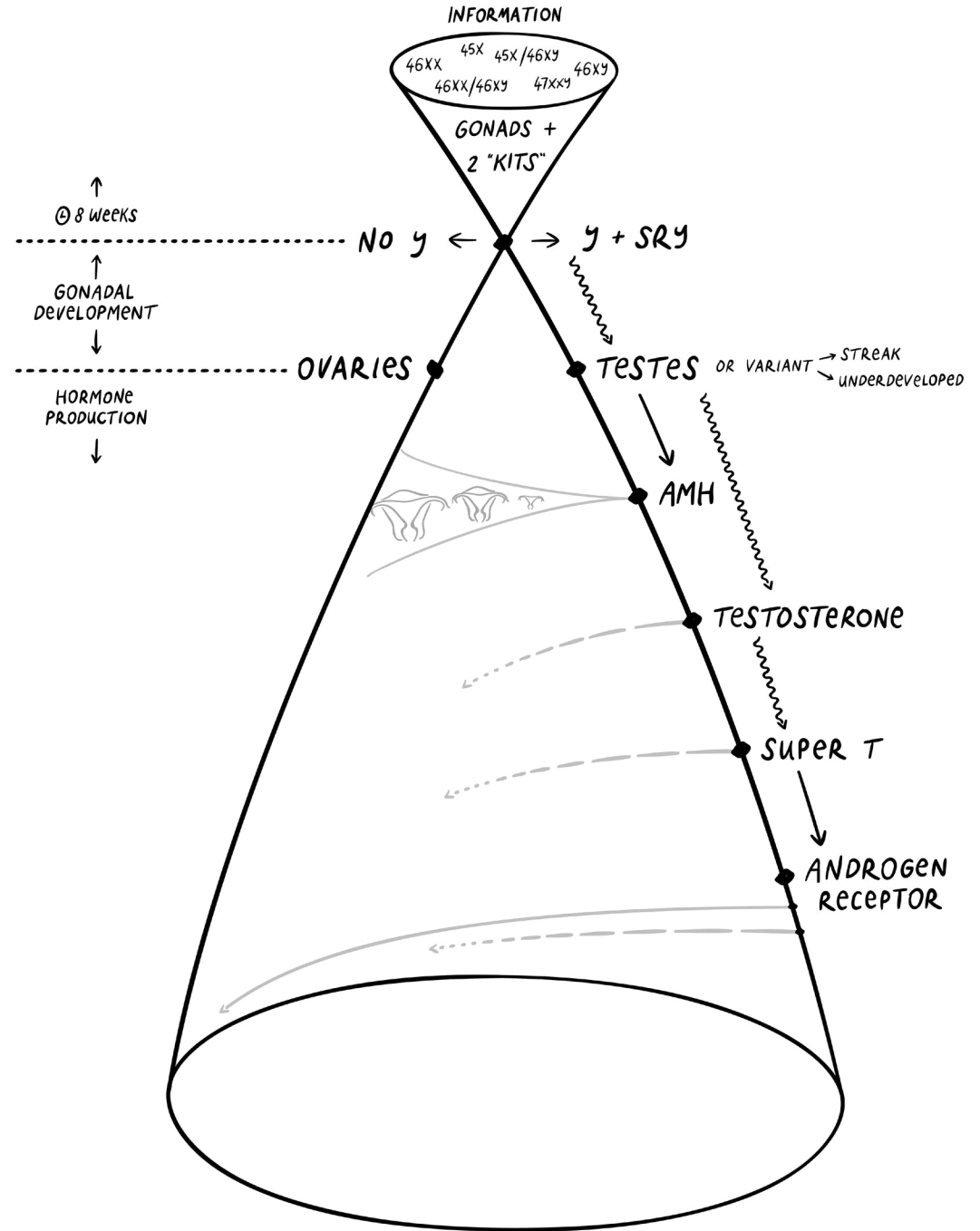
The Androgen Receptor 'doors'

Before SuperT can change genital appearance, it must face the androgen receptors!

Imagine that the androgen receptor is a door that SuperT usually goes through. For a small group of people the door can be partially closed or completely shut.

This relates to:

- girls with Complete Androgen Insensitivity Syndrome
- girls and boys with Partial Androgen Insensitivity Syndrome



Meanwhile, what happens in baby girls who developed ovaries?

When gonads develop into ovaries, the typical female kit develops with the natural growth of the baby. And usually the vulva will develop. This means the labia, clitoris and a vaginal opening.

In babies with Congenital Adrenal Hyperplasia (CAH), the body is missing an enzyme that helps the adrenal gland to make a hormone called cortisol. In other words, the body misses the enzyme that usually converts steroid hormones from one form to another.

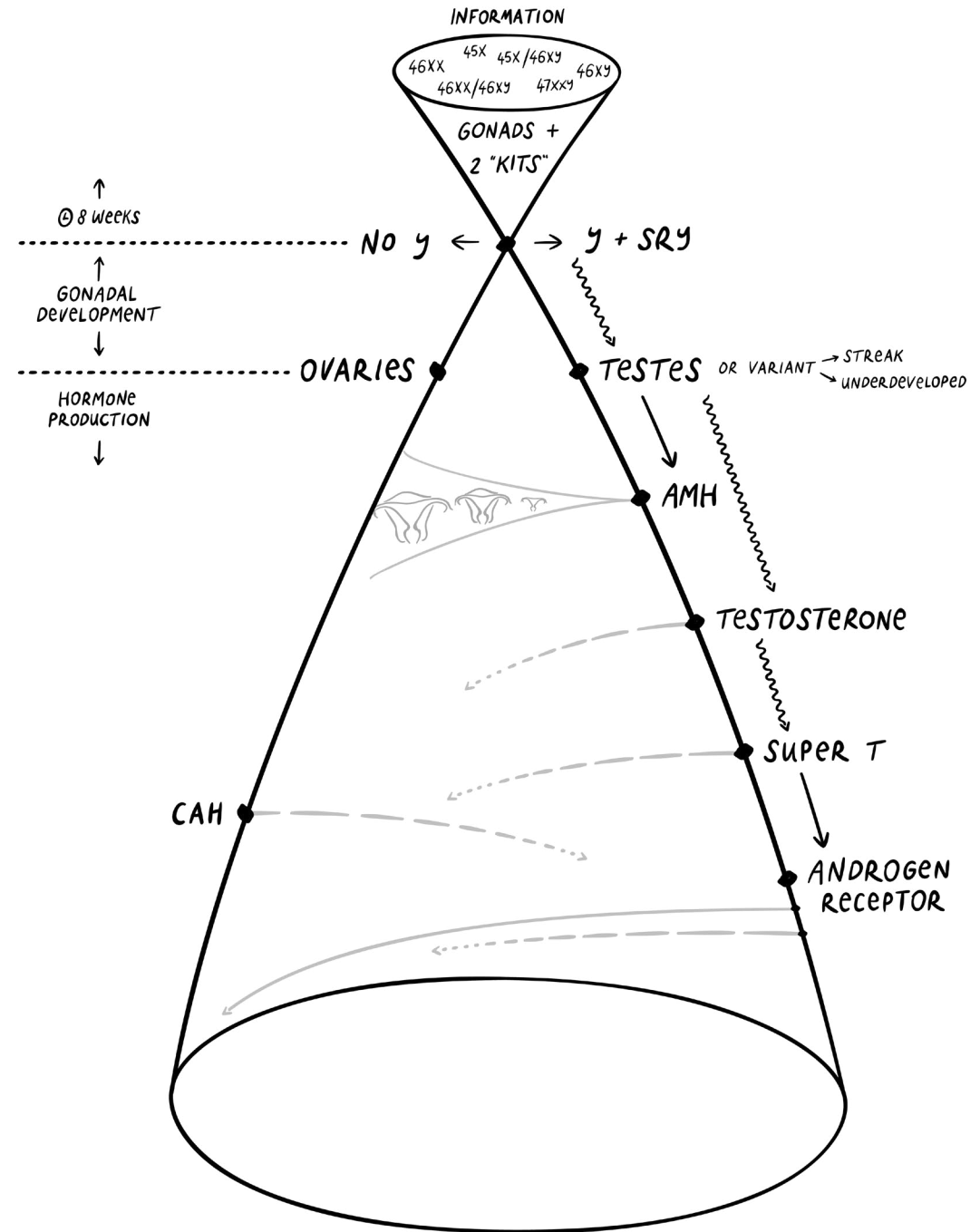
There is also an overproduction of androgens in the adrenal gland, and so more T/androgens than usual for XX babies is produced. This often affects internal and external genitals.

This is because the tissue in that area is sensitive to T/androgens, so it grows in a way that any body would with more T around. It keeps growing beyond the point at which most girls develop.

Did you know...

All people have adrenal glands resting on the top of their kidneys. These produce hormones relating to salt levels, sugar levels and stress. They also produce androgens. This means that all boys and girls produce androgens/T in varying degrees.

Need to know more?
Occasionally/rarely a child with a 46,XX DSD can have more T than expected when part of the ovary contains some testicular tissue (ovotestis).



The range of genital appearance

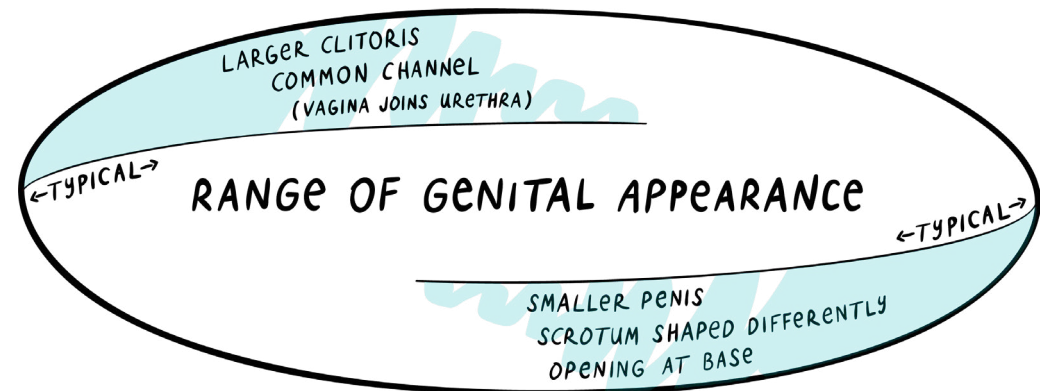
Whatever your story of sex development, every vulva and every penis looks different.

For some girls with CAH and for some girls with XY chromosomes this can mean:

- The clitoris will appear typical or it will be larger than usual.
- There is a vaginal opening or the lower part of the vagina joins the urethra (inside the body) to form a *common channel*. The urethra is the channel (the tube) down which the urine flows from the bladder. This means there is no separate vaginal opening. Sometimes that happens quite high up alongside the urethra and sometimes it happens lower down.

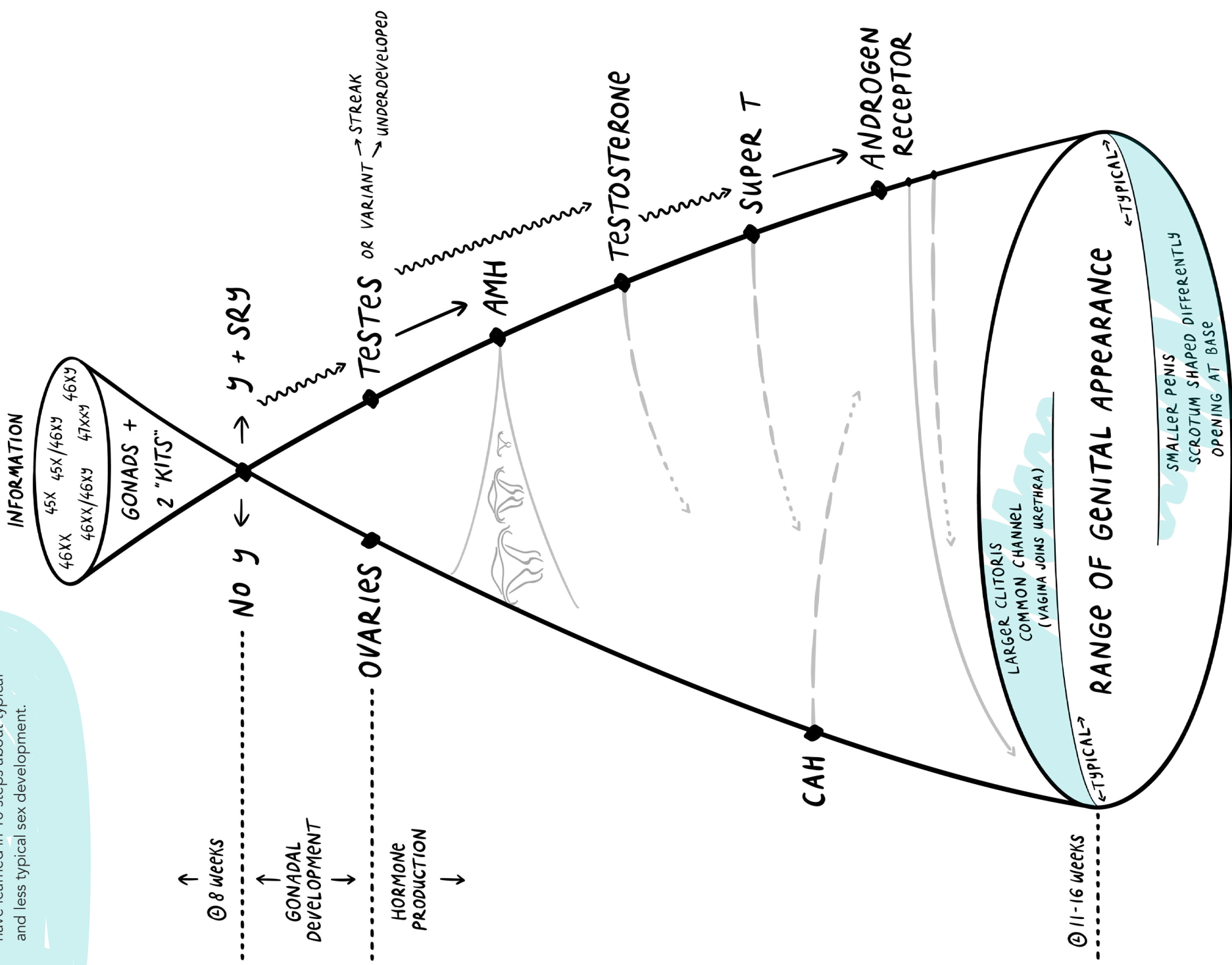
For boys with underdeveloped testes, or enzymes/proteins and genes that didn't work together during testicular development, or boys who are partially insensitive to T, and some boys with typical testes:

- The penis might be smaller, and the opening might not be at the tip but quite low along the bottom of the shaft. The scrotum might be underdeveloped or appear to be separated. The penis is often curved down (*chordee*). This is usually called *peno-scrotal* (or *proximal hypospadias*).
- One or both testes might not descend into the scrotum.



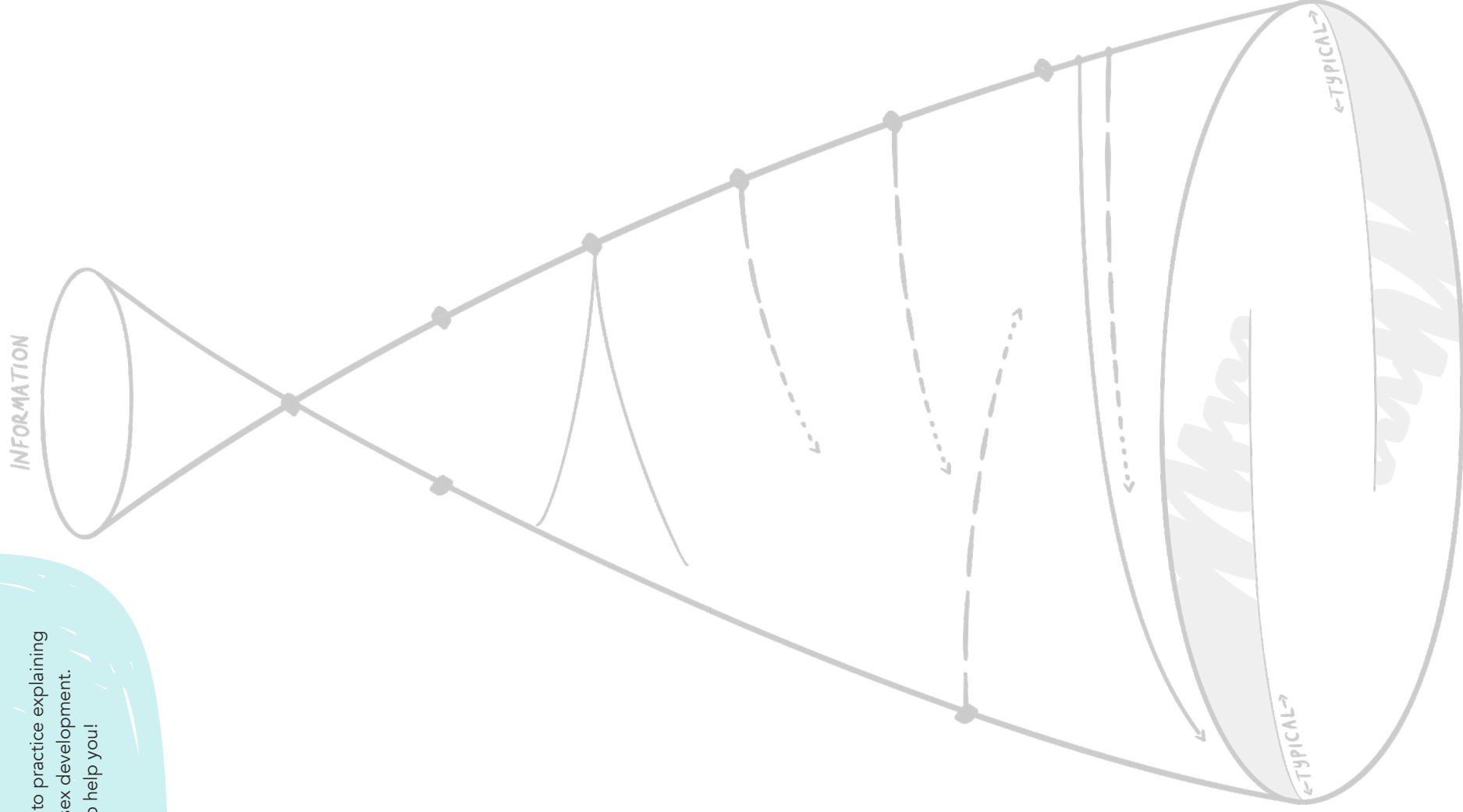
Bringing it all together

On this page we bring together all we have learned in 10 steps about typical and less typical sex development.



Template for parents and young people to draw sex development

Use this template to practice explaining your/your baby's sex development. Ask your doctor to help you!



Other resources by dsdfamilies

First days – when your baby is born with genitals that look different

Available in: English, Urdu, Polish, Arabic, Dutch, Spanish, German, Swedish, Turkish, Norwegian, Bulgarian, French, Russian, Catalan, Portuguese and Moldovan.

Top Tips for Talking

Tips and examples to help parents/carers talk to children about differences of sex development.

Timeline for Talking

For talking to children aged 0-6, 7-10, 11-14, 15+

Online only at www.dsdfamilies.org

Listen to Us

Consultations with children, young people and their families living with Different Sex Development, Final Report, 2019

Top Ten Tips for (Vaginal) Dilation

Distributed with support of the British, European and International Associations for Adolescent Gynaecology

www.dsdteens.org

Friendly, evidence-based and can-do information for pre-teens and teens about puberty, friendship, communication, school, medical care, relationships and sex.

The end

To draw genitals or not to draw genitals? That is the question...

While producing this Story we thought long and hard about including drawings of variations in genital appearance. Instead, we decided this booklet would focus on the biological process of sex development. Also, we are aware that every little baby will look different internally and externally and we don't want to confuse new parents.

Let us know what you think and what we would find most useful by sending us a message at info@dsdfamilies.org

A note about dsdfamilies

dsdfamilies provides educational, practical and peer support to children, young people and families living with differences of sex development*.

We work to ensure that the experiences of children, young people and their families inform best-care practice and professional training. We do this in collaboration with health care providers and peer support networks for adults and families.

**including Androgen Insensitivity, CAH, penoscrotal Hypospadias, Swyers, 5-Ard, some forms of Turner, Klinefelter's, Gonadal Dysgenesis, Ovotestes, 3-Beta, 17-Beta, SF1, Frasier and many others.*

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With thanks

