

Science of Transgender

22nd January 2026

Jan Athis

Science of Transgender

Since taking on this topic in September 2025, “transgender” has become even more politicised, following the High Court ruling that **biological sex** is the only option

This talk is based on the biological **science of transgender**, not the socio-cultural or socio-political

NOT homosexuality

NOT cross dressing

NOT female impersonation



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Definitions

(Merriam-Webster.com/dictionary)

SEX

Biological
Male ♂

Biological
Female ♀

Allocated at birth from **phenotype**
– **NOT genotype** (*karyotype*)

GENDER

Behavioural, socio-cultural
and psychological



self representation as male or female

TRANS GENDER

Sense of self differs from
the sex assigned at birth



Concept of a “Third Gender”

Femminiello or
femmenielli embody
a **third gender** role in
traditional
Neapolitan culture.
(Wikipedia)



Muxe or muxe
(Mexico)



Hirji (India & Pakistan)

Legally recognised in India;
2011 census 490,000
[maybe >1.5 million]



Nat Kadaw (spirit medium)
(Myanmar)



Mahu (Hawaii & Tahiti)
= spiritual community,
valued by society – pre missionary

Kathoeys
(Thailand)



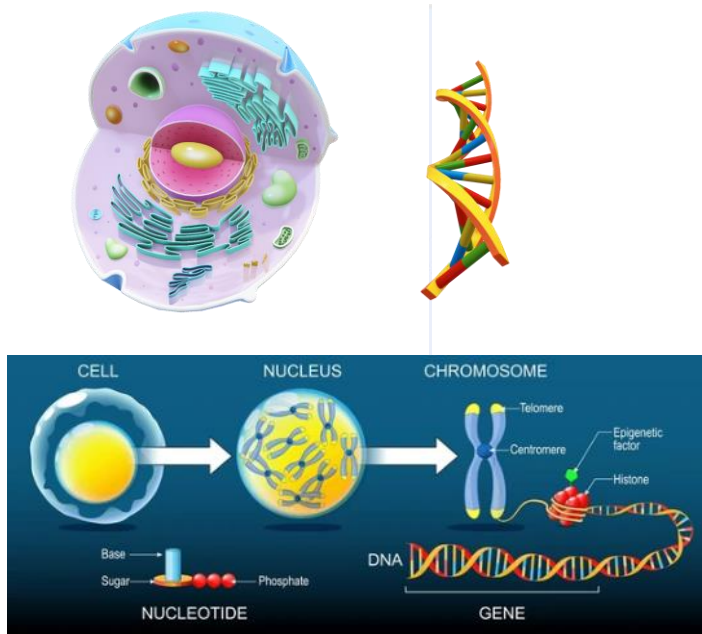
Brian Michael Smith, award-winning
trans actor



Barclays use androgenous
face on log in page

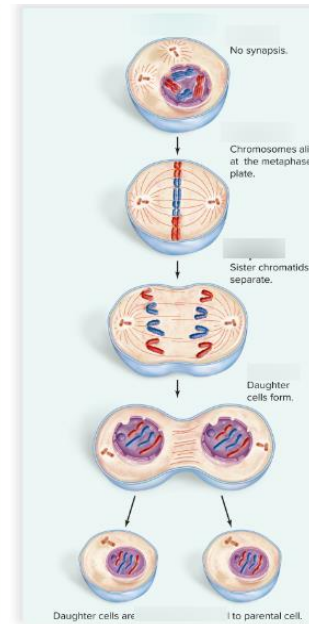
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Genetics



Epigenetic effects are random chance events that alter gene(s) expression, but not its sequence, and are NOT heritable

Mitosis

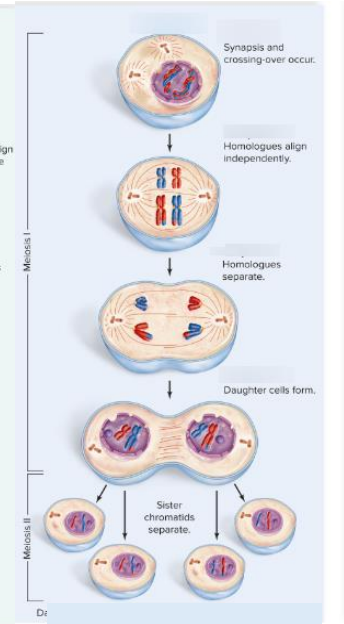


Reproduction
ie another of the same



DIPLOID
2 copies each chromosome

Meiosis

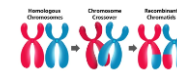
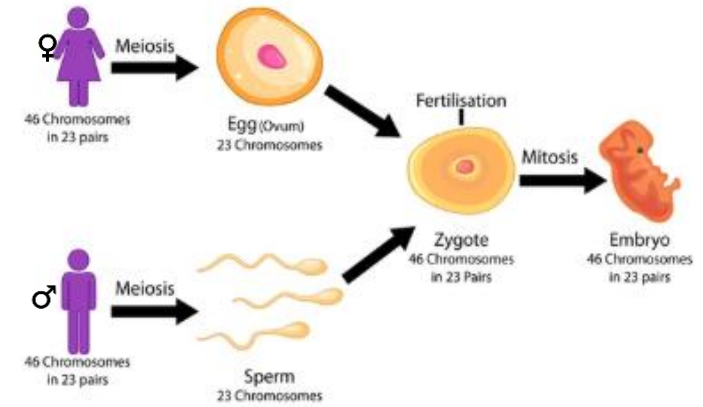


Recombination
ie product cells have half genetic material



HAPLOID
1 copy each chromosome

Each egg & each sperm has 22 autosomes and 1 sex chromosome



		Paternal	
		X	Y
Maternal	X	XX	XY
	x	XX	XY

One X chromosome in ♀ is usually [partially] inactivated
achieves parity in effect of sex chromosomes

Gene(s) for androgen [sex hormone] receptor occurs on
X-chromosome

Genes affecting sexual characteristics also occur on
some autosomes

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Genetics / cont

Knowledge of **The Human Genome** is NOT the same as knowing function, location etc of every gene!

It IS known that some CHROMOSOMES are associated with certain effects (*correlation*) & so it is likely they carry the GENES for the effects (*causation*)

Genes influence the production (nature, timing, quantity etc.) of **hormones** which in turn interact with genes, activating or deactivating them

X-chromosome (so-called because it was the “unknown” factor)

All nucleated human cells have at least **ONE X chromosome**, so unsurprisingly genes on X chromosomes affect more than sexual characteristics. Viz seem to be associated with immune function & inflammation

Y-chromosome (so-called because it was found after X!! Alphabetical, not appearance)

Y- chromosome is smaller & much simpler than X chromosome; has 70 – 200 genes – mostly related to ♂ sexual characteristics & fertility

SRY (Sex Regulating gene on **Y**) is the key gene which triggers a **cascade of effects** in the embryo and mother, including **enzymes & hormones** resulting in phenotypic ♂ development *(next slide)*.

Y-chromosome more prone to mutation than rest of genome.

From Scientific American February 2025

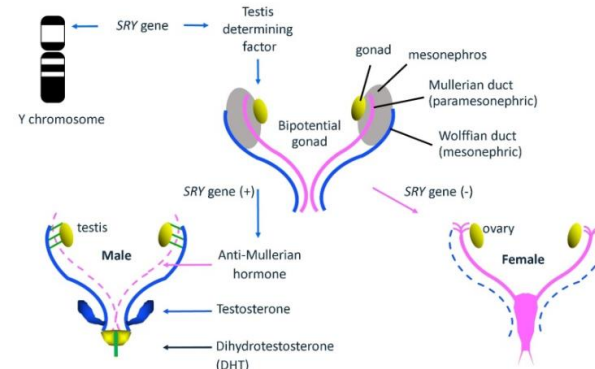
“Genetics is an extremely complicated field of study; although the human genome was sequenced more than 20 years ago, our understanding of molecular networks and how they influence organ development and function is still relatively rudimentary”

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Embryonic gonads are bi-potential

Presence of SRY initiates cascade of events leading to development of testes, which secrete hormones (*testosterone & anti-Mullerian*) which enhance Wolffian duct (spermatocord) and regresses Mullerian duct (Fallopian tube, cervix upper vagina) ; DHT derived from testosterone, has major role in external genitalia & prostate

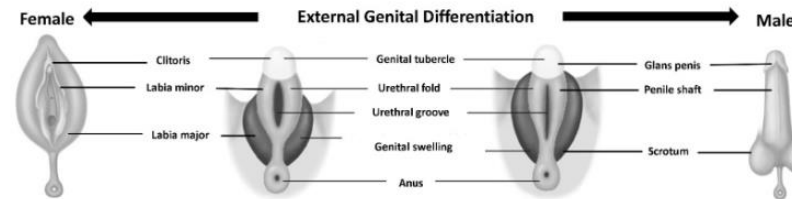
Absence of SRY means development of uterus, fallopian tubes, cervix & upper vagina



MALE is superimposed on “base state”
which is **FEMALE** (*sorry Adam ...Eve was first!*)

External genitalia : appearance is used to decide / declare baby's sex

Absence of androgens and **independent** of presence of ovaries **female genitalia develop**



Sometimes the appearance is ambiguous

(1 in 1000- 4500 births)

Primary sexual characteristics

Reproductive organs and structures present at birth including external genitalia

Secondary sexual characteristics

These develop during puberty and include

- ♀ breast development, hip widening, pubic hair
- ♂ facial & pubic hair, voice deepening

Presence of androgens* initiate changes of base state to create **male genitalia**

* Androgens = masculinising hormones produced by maternal & foetal adrenals
placenta [maternoembryonic cells – ♂ = Y-chromosome]
foetal testes

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Some effects of sex chromosome aberrations

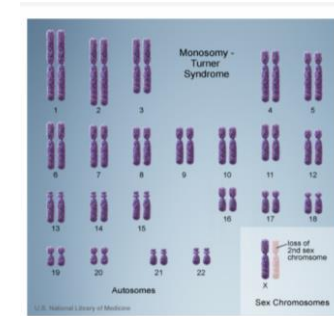
Single X chromosome (XO) = Turner's Syndrome (some phenotypical effects)



Mosaicism – not all cells in body have same karyotype
(10%-12% 46,XY – ie ♂)

Table 1. Types and Frequencies of Karyotypes Associated with Turner Syndrome

Frequency	Description
40%-50%	Monosomy X (45,X)
15%-25%	Mosaicism with 46,XX (45,X/46,XX)
3%	Mosaicism with 47,XXX (45,X/47,XXX; 45,X/46,XX/47,XXX)
10%-12%	Mosaicism with 46,XY (45,X/46,XY)
15%	Isochromosome Xq (46,X,i(Xq)) and isodicentric Xp (46,X,idic(Xp)) (two copies of the long arm connected head-to-head)
Rare	Ring X chromosome (45,X/46,Xr(X)), proximal deletion of Xp (46,XX,del(p11)), and unbalanced X-autosome translocation (a part of the ends of short and long arms of the X chromosome is missing)



Double X chromosome (XXY) = Klinefelter's Syndrome (commonest male sex chromosome aberration (~1 in 550 births) – frequently undiagnosed; impaired testosterone affecting 2nd sex characteristics)

Triple X chromosome syndrome (XXX) (~1 in 1000 births) some/ no phenotypical effects, increased risk of some diseases & behavioural issues



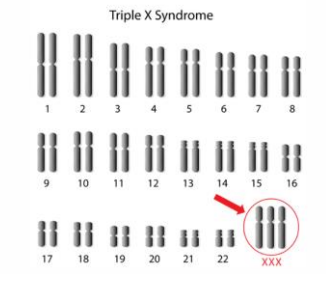
Hypertelorism & epicanthic folds



Hypertelorism



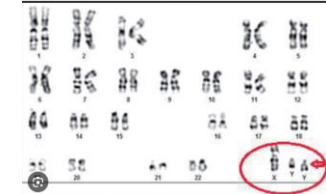
No facial dysmorphism



Double Y chromosome (XYY) = Jacob's Syndrome – Signs & symptoms variable in nature & degree – includes behavioural effects (~1 in 1000 births)



Very tall; long arms; large teeth; acne;



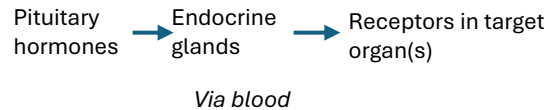
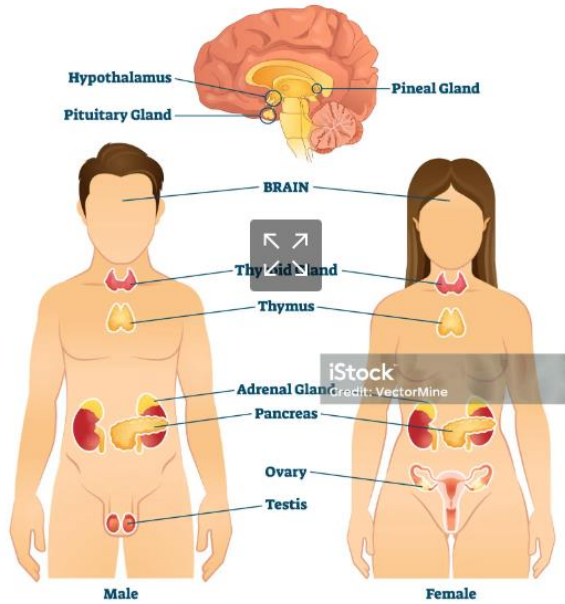
46,XX Difference of Sex Development **SRY gene** can be transferred from Y-chromosome to X-chromosome during meiosis resulting in a phenotypical ♂ with XX

The baby will be designated ♂ on basis of external genitalia but will be XX (ie NOT have Y chromosome). This occurs in 1 in 20000 births so around 1700 ♂ in UK

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Hormones (aka endocrine system)

(=chemical messengers usually circulated in blood to target tissue which has receptors)



Hormones are involved in all aspects of life - from conception onwards!

The main endocrine organs for sexual development are the **gonads** (ie ovaries & testes) with contributions from the **adrenals** and the **pituitary** (both of which produce /control many other hormones & thereby other aspects of life)

Hormonal influence on foetal sexual development

The hormonal environment *in utero* exerts an **epigenetic effect** on foetal development

Androgens

Some are solely **masculinising** hormones and are produced by

- maternal & foetal adrenals
- placenta [*maternoembryonic cells i.e has Y chromosome with ♂ foetus*]
- foetal testes

Mother needs "protection" from excess masculinising androgen, in part achieved by SHBG (sex hormone binding globulin) from liver

Specific androgens are essential precursors to oestrogens in both ♂ and ♀

Foetal exposure to **sex hormones (testosterone and oestrogens)** can affect some behaviour in later life

Foetal exposure to **excess sex hormones** due to Congenital Adrenal Hyperplasia (CAH) associated with ambiguous genitalia (*virilization of ♀*)

Foetal brain development (*at critical times*) susceptible ⇕ maternal THYROID & CORTICOSTEROID hormones – consequences can include increased anxiety & reactivity to stress

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How this science could
affect perception of own
gender – ie TRANSGENDER

Maternal hormone levels affect foetal brain development

- with subsequent effects on behaviour etc.
- ↓ androgen levels could alter the brain of a ♂ foetus → more stereotypical **feminine** thinking / behaviour
 - ↑ androgen levels could alter the brain of a ♀ foetus → more stereotypical of **masculine** thinking / behaviour



SHB globulin (*mother's protection from androgens*) production might over diminish sex-hormone levels – viz testosterone

Dysfunction of **hypothalamic –pituitary- adrenal** axis
(*many potential causes*)

Androgen Insensitivity Syndrome (AIS)

1:20 000 to 1: 64 000 births
Varies from negligible to complete feminisation of genitalia in XY karyotype;
no clinically significant effect in XX karyotype

46 XX, DSD (Difference in Sex Development)

SRY gene relocated from Y-chromosome to X-chromosome
→ **phenotypical ♂ ie external genitalia** but (XX)
double X chromosomes could exert feminising influence

CAH (Congenital Adrenal Hyperplasia) –

foetal adrenals produce excess androgens
→ **phenotypical ♂ ie external genitalia** with ovaries & uterus

More than just androgens affected – so can be fatal

Incidence in S. India 1 in 2800 births; 1 in 6334 in N India

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Thank you

I will try to answer any questions – but I make no promises!

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References

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[Author Information and Affiliations](#)

Last Update: November 13, 2023.

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