Examples of Aneuploidy in Human Sex Chromosomes

Sex chromosomes and chromosome condition	Apparent sex	Phenotype
XO, monosomic	Female	Turner syndrome
XX, disomic	Female	**Normal female**
XXX, trisomic	Female	Metafemale. Most appear normal; there may be developmental delays
XXXX, tetrasomic	Female	Rather like Down syndrome, low fertility and some intellectual disability
XY, disomic	Male	**Normal male**
XYY, trisomic	Male	Jacob syndrome, apparently normal male, tall, aggressive
XXY, trisomic	Male	Klinefelter syndrome (infertile). Incidence rate 1 in 1000 live male births, with a maternal age effect.
XXXY, tetrasomic	Male	Extreme Klinefelter syndrome, learning difficulties

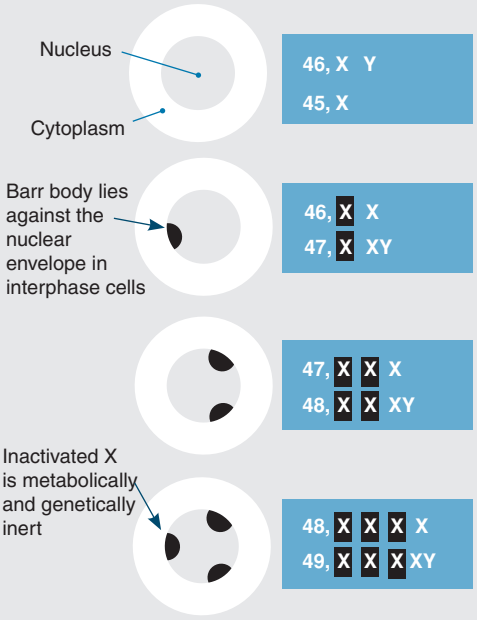
ABOVE: Features of selected aneuploidies in humans. Note that this list represents only a small sample of the possible sex chromosome aneuploidies in humans.

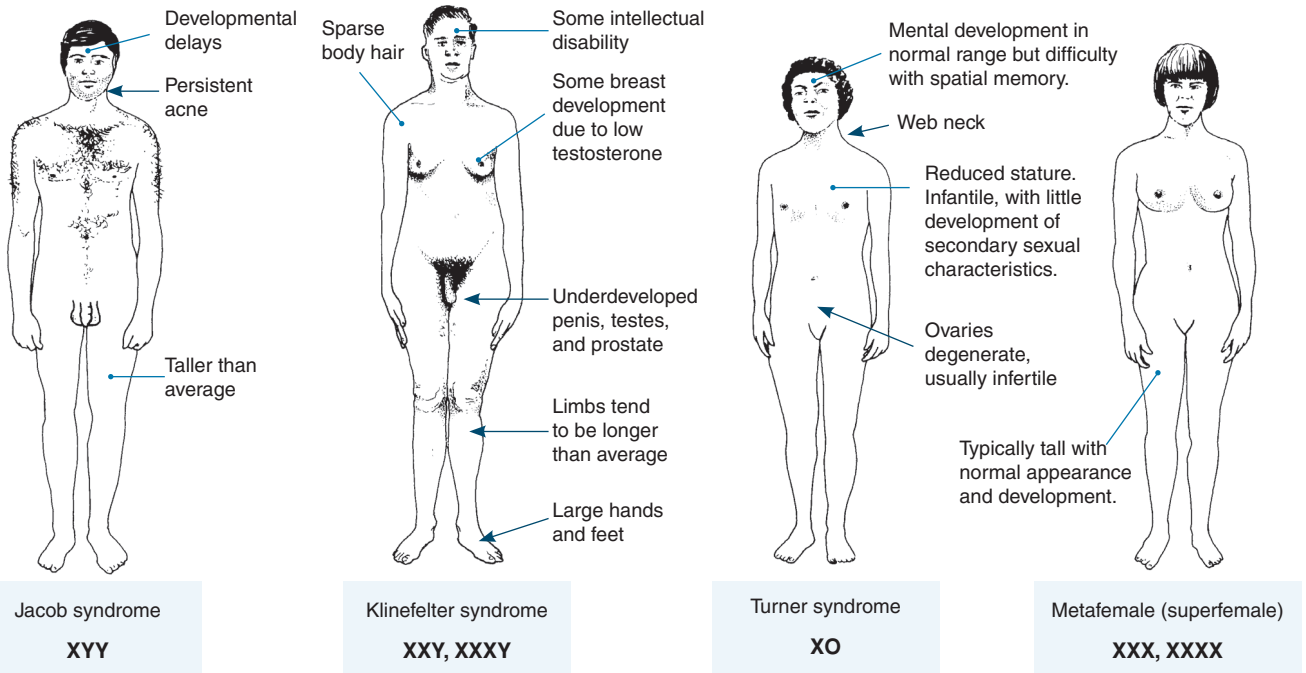
RIGHT: Symbolic representation of Barr body occurrence in various human karyotypes. The chromosome number is given first, and the inactive X chromosomes (Xi) are framed by a black box. Note that in aneuploid syndromes, such as those described here, all but one of the X chromosomes are inactivated, regardless of the number present.

If extra copies of X are inactivated, why do extra copies still produce the aneuploidy syndromes? This is because some of the genes on the Xi escape inactivation so the dosage of these non-silenced genes will differ as they escape inactivation.

Barr Bodies

In the nucleus of any non-dividing somatic cell, one of the X chromosomes condenses to form a visible piece of chromatin, called a **Barr body**. This chromosome is inactivated (Xi), so that only one X chromosome in a cell ever has its genes expressed. The inactivation is random, so Xi may be either the maternal homologue (from the mother) or the paternal homologue (from the father).





Jacob syndrome	Klinefelter syndrome	Turner syndrome	Metafemale (superfemale)
XYY	**XXY, XXXY**	**XO**	**XXX, XXXX**

5. State how many Barr bodies are present in each somatic cell for each of the following syndromes:

   (a) Jacob syndrome: _____ (b) Klinefelter syndrome: _____ (c) Turner syndrome: _____

6. Explain the consequence of X-chromosome inactivation in terms of the proteins encoded by the X chromosome genes:

   _____

   _____

7. State how many chromosomes for each set of homologues are present for the following forms of aneuploidy:

   (a) Nullisomy: _____ (c) Trisomy: _____

   (b) Monosomy: _____ (d) Polysomy: _____

ISBN: 978-1-927173-97-8