

## Karyotype, mitosis and meiosis

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### (A) The Human Karyotype

- See figure (to be supplied)
  - 46 chromosomes (23 pairs) are present in all nucleated cells.
  - The autosomes are in pairs, numbered 1-22.
  - Females have two X, sex chromosomes, (46,XX) and men have one X and one Y, sex chromosomes (46,XY).
  - Chromosomes have a short arm (p) and long arm (q).
  - Euchromatin contains the active genes.
  - All chromosomes show normal variation in DNA content.
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### (B) Lyonisation

- Lyonisation is inactivation of one of X chromosomes in every female cell.
  - Inactivation only occurs in somatic cells.
  - Random process whether paternal or maternal X is inactivated, but is subsequently fixed for all descendants of that cell.
  - X inactivation affects most but not all genes on the X chromosome.
  - If cell has more than two X chromosomes then the extra X's are also inactivated.
  - The randomness of X inactivation accounts for some females being affected with X-linked recessive disorders.
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### (C) Cell Division

#### C(a) Mitosis

- Occurs in somatic cells.
  - One cell produces two identical daughter cells (see figure).
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#### C (b) Meiosis

- Occurs in gonads.
  - Two successive divisions.
  - DNA replicates only once, before the first division (S phase).
  - Somatic diploid chromosomal complement halved to a haploid number (see figure).
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### (D) Non-disjunction

- Failure of sister chromatids to disjoin at anaphase in either mitosis or meiosis.
  - Causes aneuploidy with two cells produced, one with extra copy (trisomy) and one with missing copy (monosomy) of a chromosome.
  - Related to increasing maternal age.
  - For example, Down syndrome (Trisomy 21), Edward Syndrome (Trisomy 18), Patau syndrome (Trisomy 13).
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### (E) Spermatogenesis

- Occurs from time of sexual maturity onwards.
  - In seminiferous tubules.
  - Primary spermatocyte undergoes 1<sup>st</sup> meiotic division to produce 2 secondary spermatocytes, each with 23 chromosomes.
  - Following the 2<sup>nd</sup> meiotic division, two spermatids are formed.
  - Spermatogenesis produces 4 sperm per meiotic division.
  - Production of a mature sperm takes 61 days.
  - Numerous replications increase chances for mutation, particularly in older men.
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### (f) Oogenesis

- Mostly complete by birth.
  - Primary oocytes form by the end of 1<sup>st</sup> trimester and remain in suspended prophase (dictyotene) until sexual maturity.
  - Oocyte released into fallopian tube after first meiotic division.
  - Completion of 1<sup>st</sup> meiotic division may take over 40 years.
  - First meiotic division results in formation of the 1<sup>st</sup> polar body.
  - Second meiotic division completed after fertilisation in fallopian tube, resulting in mature ovum and 2<sup>nd</sup> polar body.
  - Oogenesis produces only one ovum.
  - Long resting phase during the 1<sup>st</sup> meiotic division may be factor in increased risk of homologous chromosomes separation failure during meiosis (non-disjunction) in older mothers.
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### (g) Chromosomal Rearrangements

- Balanced rearrangements are common, 1 in 500.
- Unbalanced rearrangements have additional/missing genetic material, causing fetal loss or physical/mental handicap.