Meiosis

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Meiosis

- Diploid cells give rise to haploid cells (gametes)
- Unique to “germ cells”
  - Spermatocytes
  - Oocytes
- Two steps: Meiosis I and Meiosis II
**Meiosis I**

- Diploid $\rightarrow$ Haploid ("reductive division")
- Separates homologous chromosomes

**Diploid Cell (2n)**
- Paired Chromosome "Homologous"

**Replicated Chromosomes**
- Crossover/Recombination

**Cell Division**
- Haploid Cells (1n)

**Blue = Paternal**
**Red = Maternal**

Meiosis I
Meiosis II

- Chromatids separate
- Four daughter cells
Meiosis

Blue = Paternal
Red = Maternal

Meiosis I

Meiosis II
Spermatogenesis

Begins at puberty
Oogenesis

• “Primary oocytes” form in utero
  • Diploid cells
  • Just beginning meiosis I
  • Arrested in prophase of meiosis I until puberty

• At puberty
  • A few primary oocytes complete meiosis 1 each cycle
  • Some form polar bodies → degenerate
  • Some form secondary oocytes (haploid)
  • Meiosis II begins → arrests in metaphase

• Fertilization → completion of meiosis II
Aneuploidy

- Abnormal chromosome number
  - Extra or missing chromosome
- Disomy = two copies of a chromosome (normal)
- Monosomy = one copy
- Trisomy = three copies
Meiotic Nondisjunction

- Failure of chromosome pairs to separate
- Most common mechanism of aneuploidy
- Can occur in meiosis I or II
Meiosis I Nondisjunction

- Blue = Paternal
- Red = Maternal

Meiosis I

- Homologous Chromosomes Fail to Separate

Meiosis II

Diploid Mixture Genes

- No chromosomes
Meiosis II Nondisjunction

Blue = Paternal
Red = Maternal

Meiosis I

Sister Chromatids Fail to Separate

Meiosis II

Haploid
No mixture genes

Diploid
No mixture genes

No genes
Nondisjunction

Blue = Paternal
Red = Maternal

Meiosis I NDJ

Normal

Meiosis II NDJ

Normal
Monosomy

• Fertilization of 1n (normal) and 0n gamete
• Usually not viable
• Turner syndrome (45,X)
  • Only one sex chromosome
Trisomy

- Fertilization of 1n (normal) and 2n gametes
- Not compatible with life for most chromosomes
- Exceptions:
  - **Trisomy 21** = Down syndrome (95% cases due to NDJ)
  - Trisomy 18 = Edward syndrome
  - Trisomy 13 = Patau syndrome
Trisomy

• Maternal **meiosis I** NDJ errors are a common cause
  • Meiosis I protracted in females
  • Begins prenatally, completed at ovulation years later
  • Advanced maternal age $\rightarrow$ ↑ risk trisomy
Trisomy

• Cause of NJD suggested by trisomy genotype
  • Father = 21A and 21B; Mother = 21C and 21D
  • Trisomy 21 ACD = Meiosis I nondisjunction in mother
  • Trisomy 21 ACC = Meiosis II nondisjunction in mother
Uniparental Disomy

- Child has **two copies** of one parent’s chromosomes
- No copies of other parent’s chromosomes
- Father = **21A** and **21B**; Mother = **21C** and **21D**
- Child **AA** (isodisomy) = Meiosis II error (father)
- Child **CD** (heterodisomy) = Meiosis I error (mother)
Uniparental Disomy

- Child is euploid
  - Normal number of chromosomes
  - No aneuploidy
- Usually normal phenotype
- Can lead to phenotype of **recessive disease**
  - Father = Aa (recessive gene for disease)
  - Child = aa (two copies of a from father)
Robertsonian Translocation

- Fusion of long arms of two chromosomes
- Occurs in **acrocentric** chromosomes
  - Chromosomes with centromere near end (13, 14, 21, 22)

Diagram:

- 14;21 chromosome fusion
- Lost chromosomes
Robertsonian Translocation

- Father
  - 14
  - 21

- Mother
  - 14
  - 21

Zygotes
- Normal
- Carrier
- Trisomy 21 (Down)
- Monosomy 14
- Monosomy 21
- Trisomy 14
Robertsonian Translocation

- Carrier has only 45 chromosomes (one translocated)
- Loss of short arms $\rightarrow$ normal phenotype (no disease)
- 13-14 and 14-21 are most common
- Main clinical consequences
  - Many monosomy and trisomy gametes
  - Frequent *spontaneous abortions*
  - Carrier may have child with *Down syndrome* (trisomy 21)
Karyotype

- Can be done in couples with **recurrent fetal losses**
- Used to diagnose chromosomal imbalances