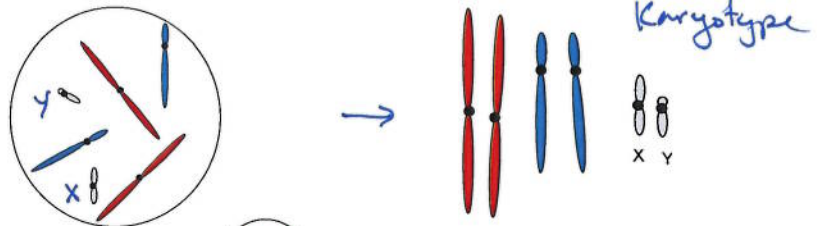
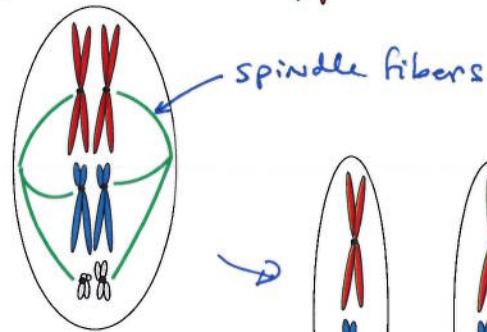


Lecture 34: Genetic Diseases Associated with Meiosis, Introduction to Mendel

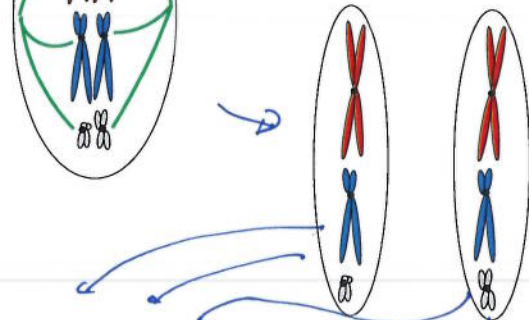
1. Given the following chromosomes in this cell, generate a karyotype. Is this individual male or female?



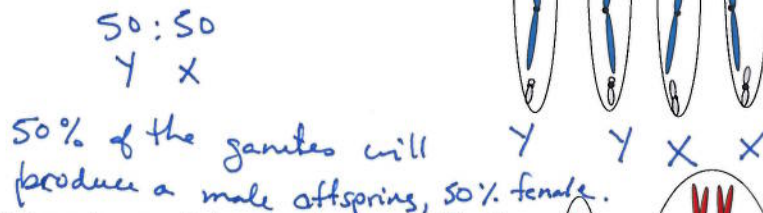
2. Draw the cell at metaphase 1 in meiosis.



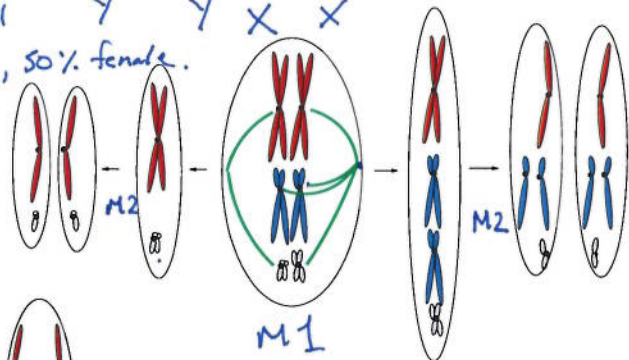
3. Draw the cells after cytokinesis in meiosis I.



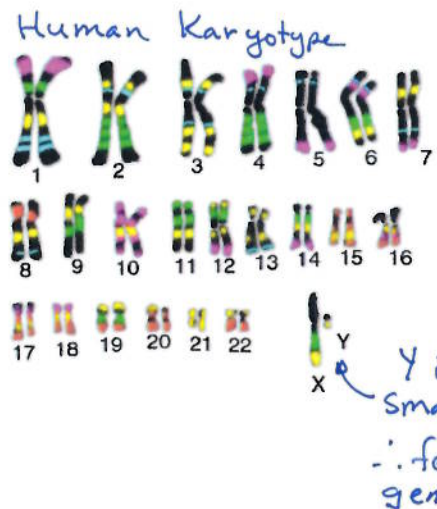
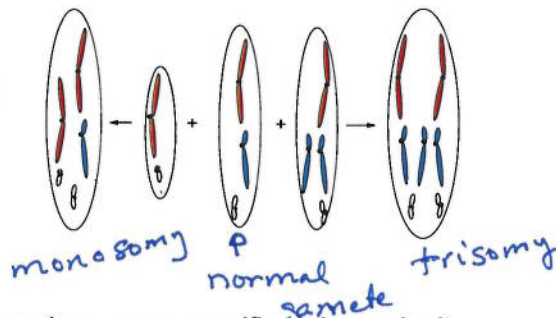
4. Draw the gametes (sperm, egg) produced after meiosis II. How do they differ?



5. What happens if there is non-disjunction in meiosis I (both homologous chromosomes go to one pole of the cell), with normal separation of the chromatids in meiosis II? What do the gametes look like?



6. What happens when a normal gamete joins with the gametes produced in 5?



Modern Karyotype mapping – gene specific stains make it easier to identify homologous chromosomes.

Genetic Diseases caused by Non-disjunction:

Aneuploid fertilized egg/embryo may have:

1. one less chromosome – monosomy
2. one extra chromosome – trisomy

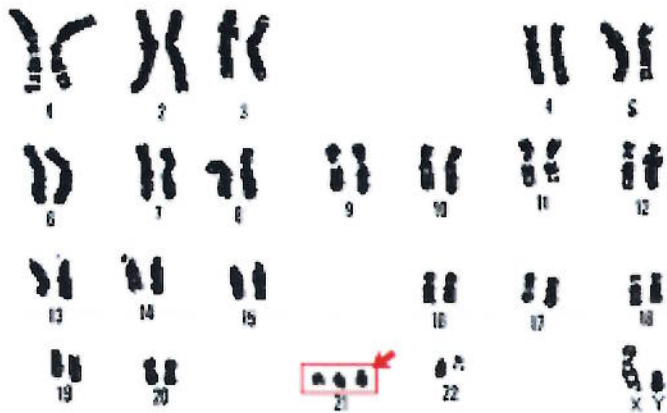
Most common diseases are associated with trisomy and involve smaller chromosomes:

Autosomal

- Down's syndrome – trisomy 21 (1/1000 for 30 yr old mothers, 1/12 for 50 yr old mothers)

Sex Chromosomes

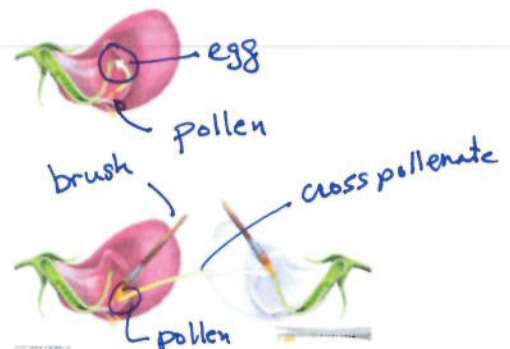
- Klinefelter syndrome – trisomy – XXY (1:1000)
- Turners syndrome – monosomy – XO (1:5000)



Mendelian Genetics – you can learn a lot from peas.

- Trait:** Any aspect of the appearance of an organism.
- Phenotype:** Observed characteristic, depends on genotype.
- Genotype:** Actual DNA composition of the organism.
- Dominant allele:** Single copy of allele gives rise to observed phenotype, usually symbolized with uppercase letters (A).
- Recessive allele:** both copies of alleles are required to observe phenotype, usually symbolized with lowercase letters (a).
- Codominance:** Both alleles contribute to phenotype.
- Homozygous:** Both alleles of a gene are the same (AA) *or aa*
- Heterozygous:** Two different alleles (Aa)
- Sex-linked:** trait associated with the X-chromosome

flowers color.



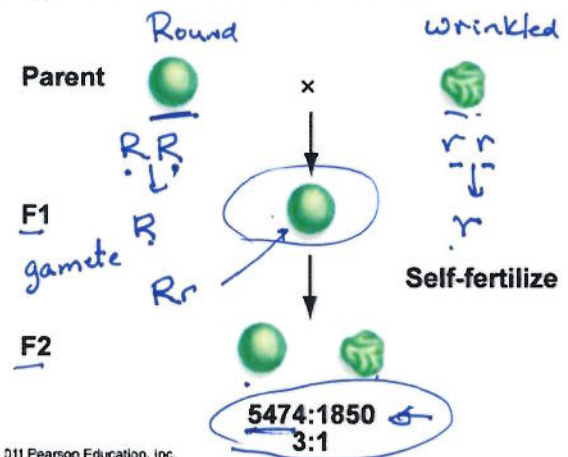
Peas were a very good model system, it was easy to breed pea plants using manual cross-pollination (instead of the more usual self-pollination), they grow quickly, and have easy to observe traits.

Monohybrid Cross:

- Crossed homozygous plants, one parent with round seeds, other with wrinkled seeds.
- First filial (F1) generation was all smooth.
- Second generation (F2) was 75% smooth, 25% wrinkled.

How to predict the phenotype and genotype of the F1 and F2 offspring:

- Assign symbols to genes
- Determine frequency of gametes
- Consider all possible combinations.



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