Inheritance of Gene Differences – non-Mendelian geneic interactions, part 2

I. Autosomes v. Sex chromosomes
   A. Morgan's experiments
   B. Pattern of Sex linked inheritance
II. Human Genetics & Pedigrees

II. Sex chromosomes v. Autosomes

Sex chromosomes: house some key genes responsible for determination of separate sexes

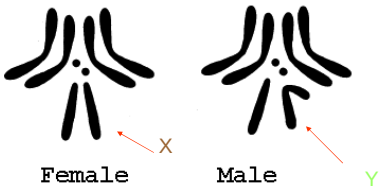
A. Heterogametic, XY or ZW
   – X & Y differ in size, but they do share a region of homology for proper pairing during cell division
   – Genes located on the differential part of the X are called X linked genes
B. Homogametic, XX or ZZ
   - X & Y differ in size, but they do share a region of homology for proper pairing during cell division
   - Genes located on the differential part of the X are called X linked genes
C. Autosomes = the remaining (non-sex) chromosomes

A. Morgan's *Drosophila* experiments

Some *Drosophila* have irregular white eye color. When a white eye female is crossed with a normal male, the F_1 generation consists of ½ normal, ½ white. The F_2 generation also consists of ½ red eye and ½ white eye. WHY?

Thomas Hunt Morgan

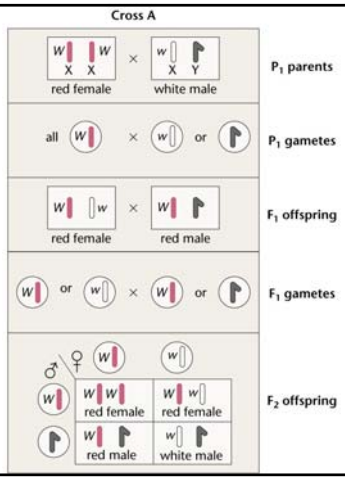
chromosome complements of male & female *Drosophila*



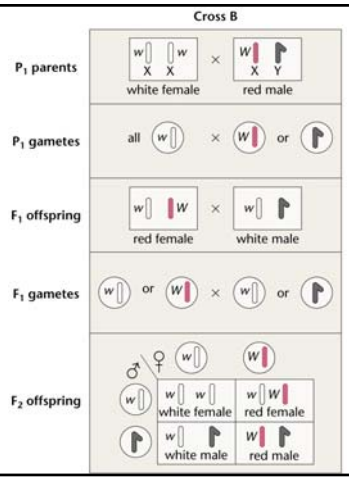
Through several crosses, Morgan and his students determined that the genes for eye color were located on the X chromosome.

Sex linked genes:

- members of the homogametic sex (XX) have two alleles per gene, members of the heterogametic sex (XY) have only one allele
- Cross A
  - P: Red eye female (X^+X^+) x white eyed male (X^wY)
  - F_1: All red eyes
  - F_2: All females red eyes, ½ males white eyes



Reciprocal cross:



Human karyotype – males are heterogametic, similar to *Drosophila*

Therefore, fathers pass along the genes on their X chromosome to their daughters, and they pass the Y onto their sons!

B. Pattern of Sex linked inheritance

- Genes located on the sex chromosomes are <u>not</u> equally inherited by sons & daughters

	sperm: X	sperm: Y
eggs: X	XX	XY
eggs: X	XX	XY

Female to male ratio is 1:1

Sex limited & sex influenced

- Autosomal genes responsible for the existence of contrasting phenotypes, but the expression of these genes is dependent upon the hormone constitution of the individual.
  - <u>Sex limited</u>: expression is limited to one gender
  - <u>Sex influenced</u>: phenotype expressed by a heterozygote is influenced by gender
    - Allele appears dominant in one gender and recessive in the other

	<u>men</u>	<u>women</u>
BB	bald	bald
Bb	bald	unaffected
bb	unaffected	unaffected

II. Human Genetics & Pedigrees

- Geneticists are primarily interested in humans to establish the pattern of transmission of inherited traits – specifically those associated with disease
  - Mendelian ratios do not apply in individual human families because of the small size
  - Controlled matings cannot be made as is possible in experimental genetics
  - Pedigrees useful in terms of understanding the transmission pattern of specific traits

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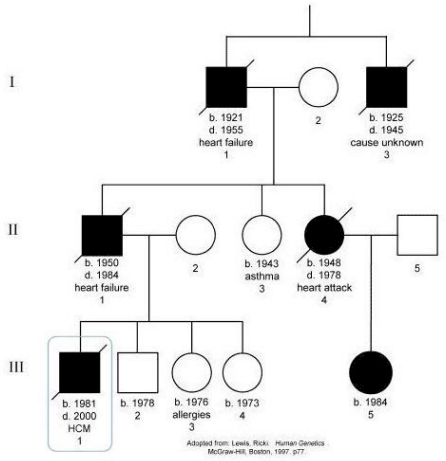
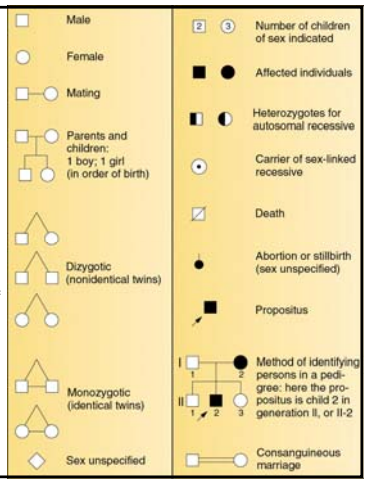
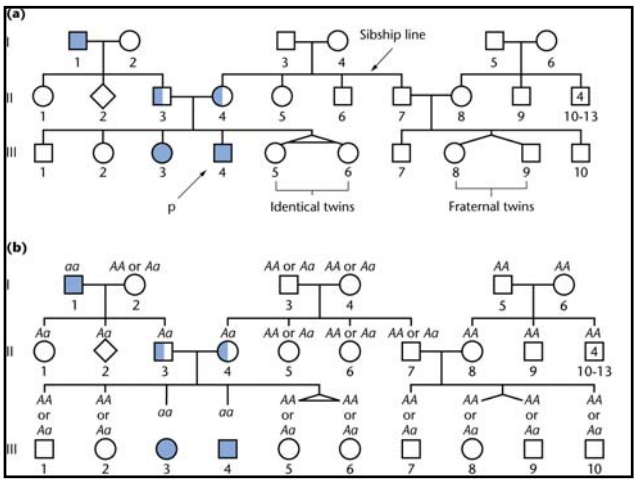


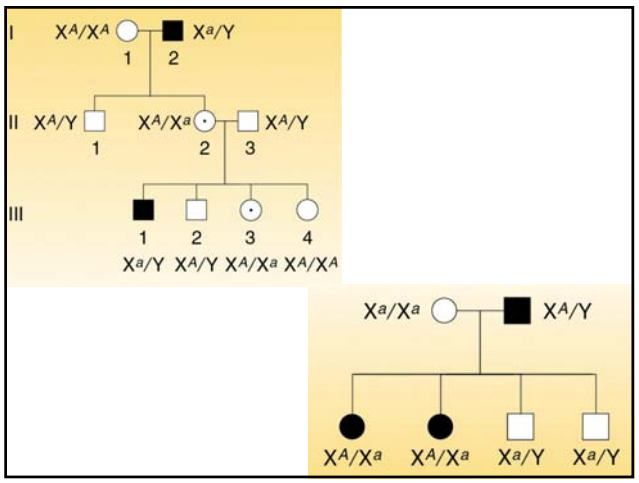
Pedigree:
Contains info about the individuals in a family, sex, phenotype, relatedness, who is alive, twins, inbreeding, etc.

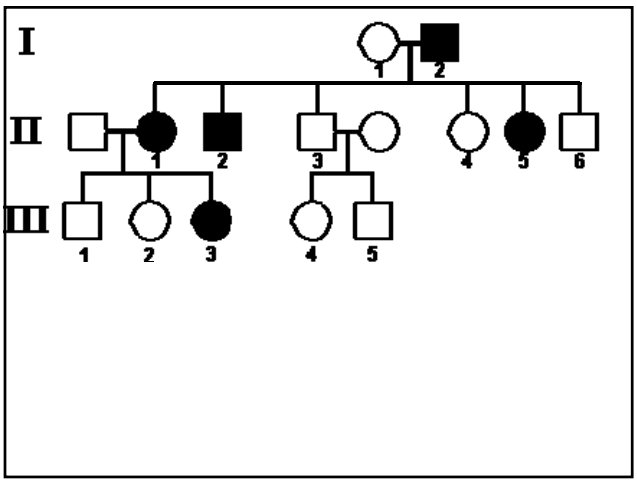
Generations in a pedigree diagram are **numbered**, by convention, using **Roman numerals**, starting with the parental generation, at the top of the diagram as generation I.

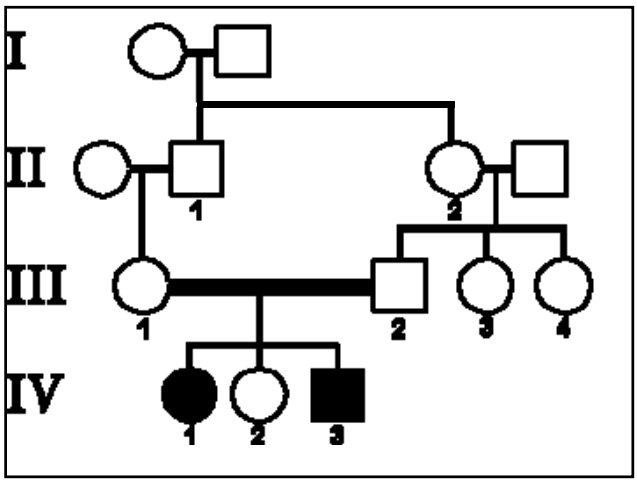
For convenience, the members of each generation are numbered across the line, from left to right, using normal numerals



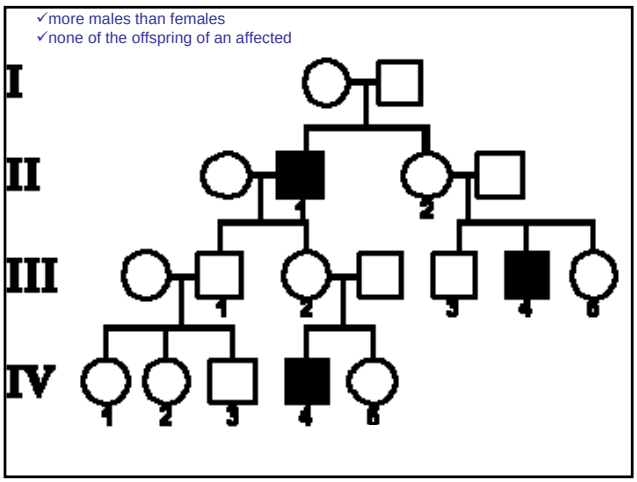








✓more males than females
✓none of the offspring of an affected



1) Autosomal recessive disorders

- oculocutaneous albinism
- alkaptonuria
- Bartter's syndrome
- cystic fibrosis
- Tay-Sachs disease
- galactosaemia
- Gaucher's disease
- glycogen storage disease
- phenylketonuria
- Wilson's disease
- xeroderma pigmentosa
- Sickle-cell anemia

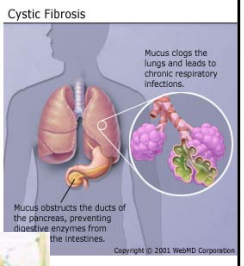
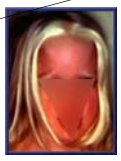


Fig. 255. Xeroderma pigmentosum (Freckles & photosensitivity)

2) Autosomal dominant disorders

- Huntington's disease
- Polydactyly & Brachydactyly
- Piebald spotting



3) X-linked recessive disorders

- ~290 disorders have been identified!
- Red-green color blindness
- Hemophilia
- Duchenne muscular dystrophy
- ichythyosis
- fragile X-linked mental retardation
- agammaglobulinaemia

4) X-linked dominant disorders

- Hypophosphatemia
- Xg blood group
- Rett's syndrome

Different types of inheritance patterns: a review

- Simple Mendelian –
- Incomplete Dominance – occurs when the heterozygote has an intermediate phenotype between either corresponding homozygote because it is haploinsufficient
- Codominance – occurs when the heterozygote expresses both alleles simultaneously and equally
- Lethal Alleles – an allele that has the potential for causing death
- Epistasis –

- X linked – involves the inheritance of genes located on the X chromosome. In mammals & fruit flies, males are hemizygous
- Sex influenced – refers to the impact of sex on the phenotype of the individual, some alleles are dominant in one sex and recessive in the other
- Sex limited – refers to traits that occur only in one of the two sexes