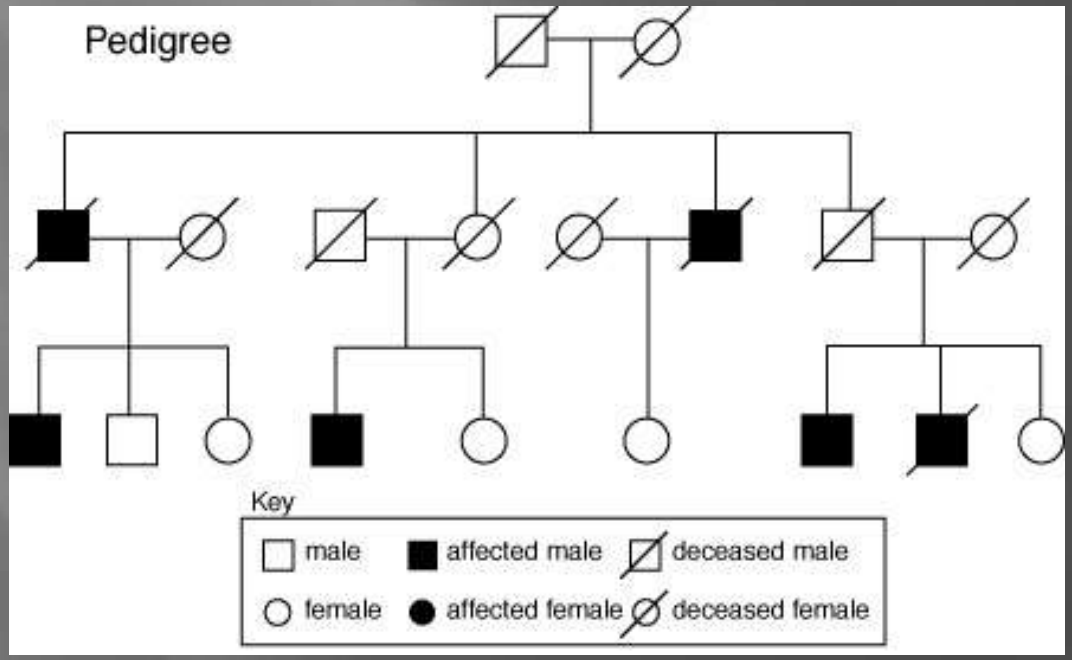
PATTERNS OF HEREDITY AND HUMAN GENETICS

Chapter 12

I. Pedigree

- Graphic representation of genetic inheritance. (Look at Fig 12.1 and 12.2)



I. Pedigree

Circle = female

Square = male

Shaded = shows trait being studied

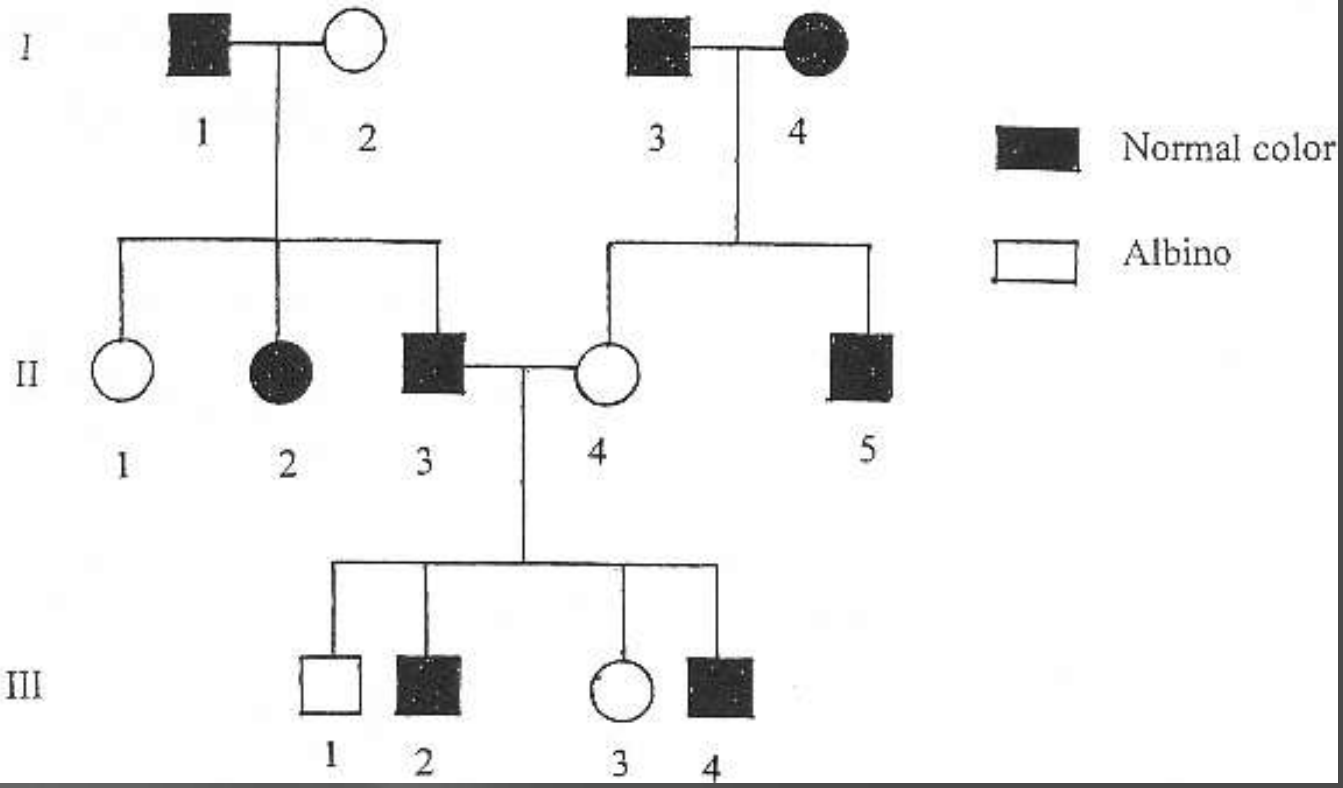
Half Shaded = Carrier

Carrier – heterozygous individual

I. Pedigree

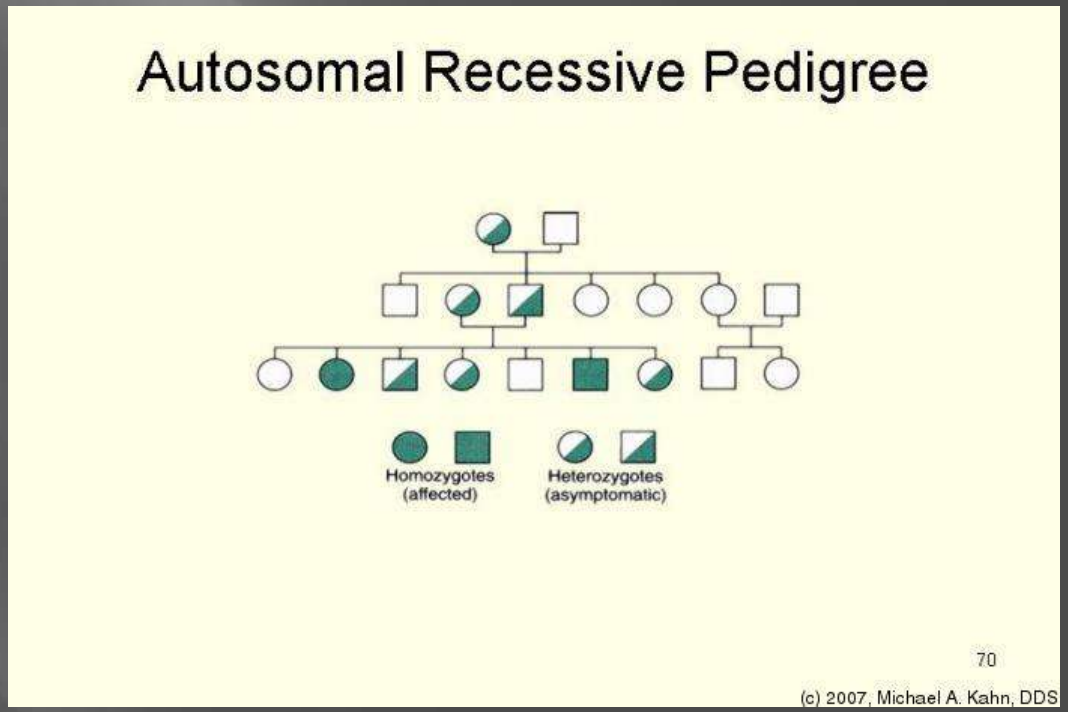
- Lets practice analyzing

Figure 14.3-5 Pedigree for skin color



II. Simple Recessive Heredity

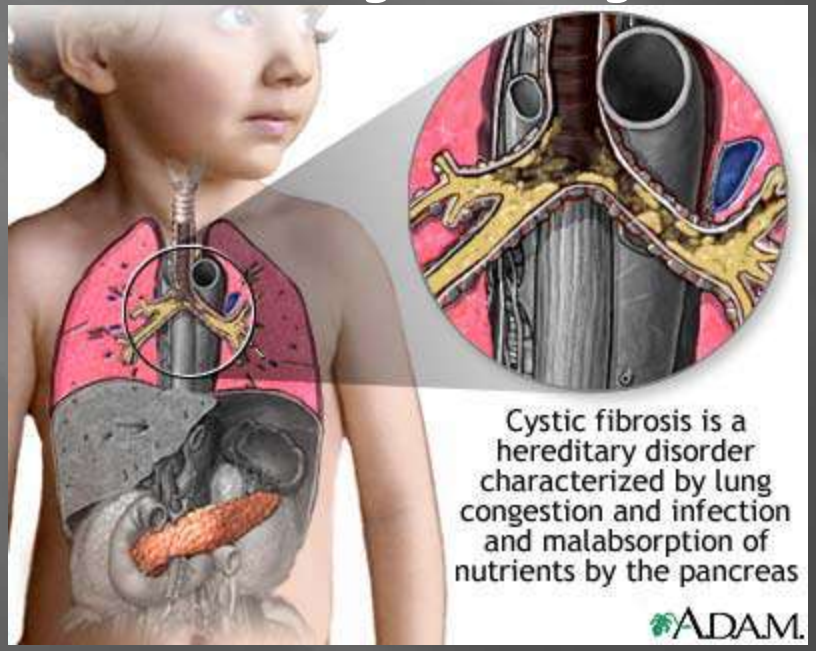
- Most disorders caused by recessive alleles
  - Cystic Fibrosis
  - Tay-Sachs disease
  - Phenylketonuria



II. Simple Recessive Heredity

A. Cystic Fibrosis

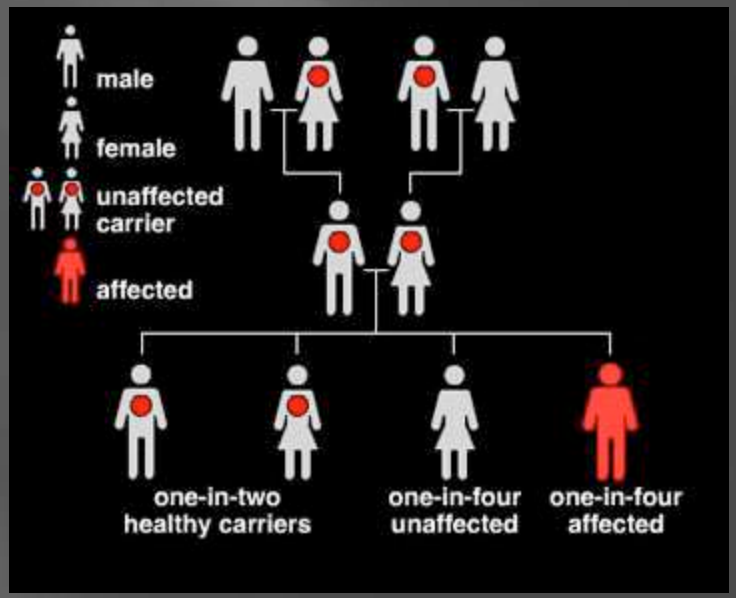
- 1 in 28 carriers
- 1 in 2500 born inherits disorder
- Thick mucus in lungs and digestive tract



II. Simple Recessive Heredity

B. Tay-Sachs

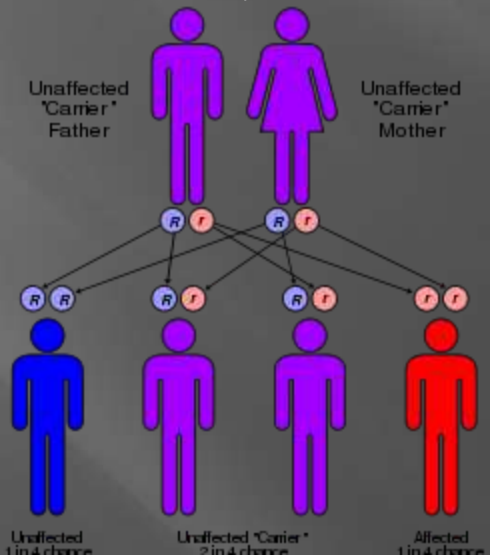
- Absence of enzyme that breaks down lipids stored in nervous system
- Common in those whose ancestors are from eastern Europe



II. Simple Recessive Heredity

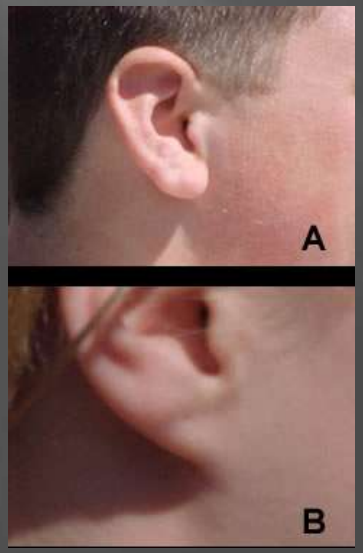
C. Phenylketonuria (PKU)

- Fails to convert AA phenylalanine to tyrosine
- Builds and damages CNS
- Babies tested positive put on strict diet
- Woman becomes pregnant- fetus damaged

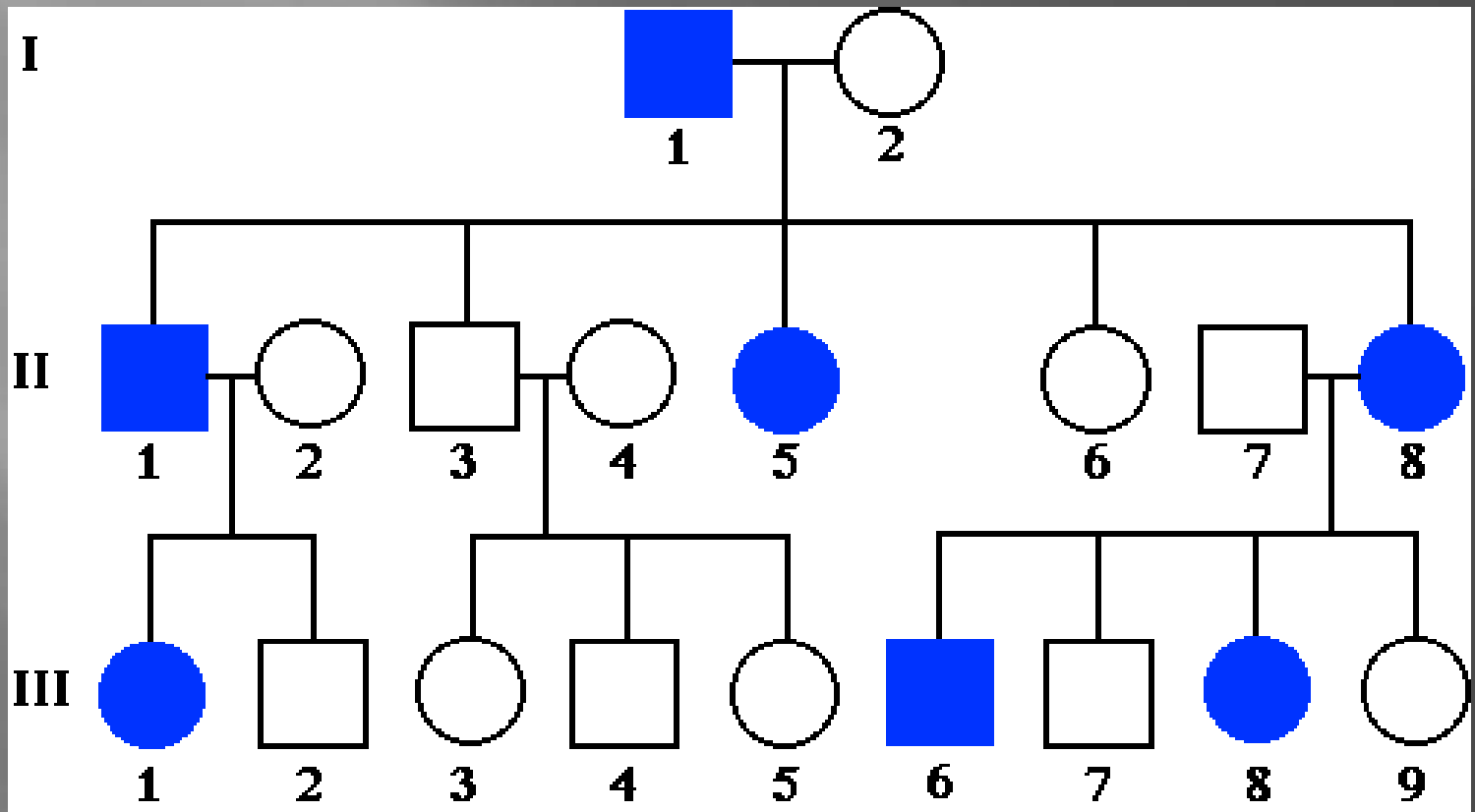


III. Simple Dominant Heredity

- Rule of Dominance
  - One dominant allele = displayed trait
  - Cleft Chin, widows peak, dangling earlobes, hitchhikers thumb, thick lips, hair on middle section of fingers



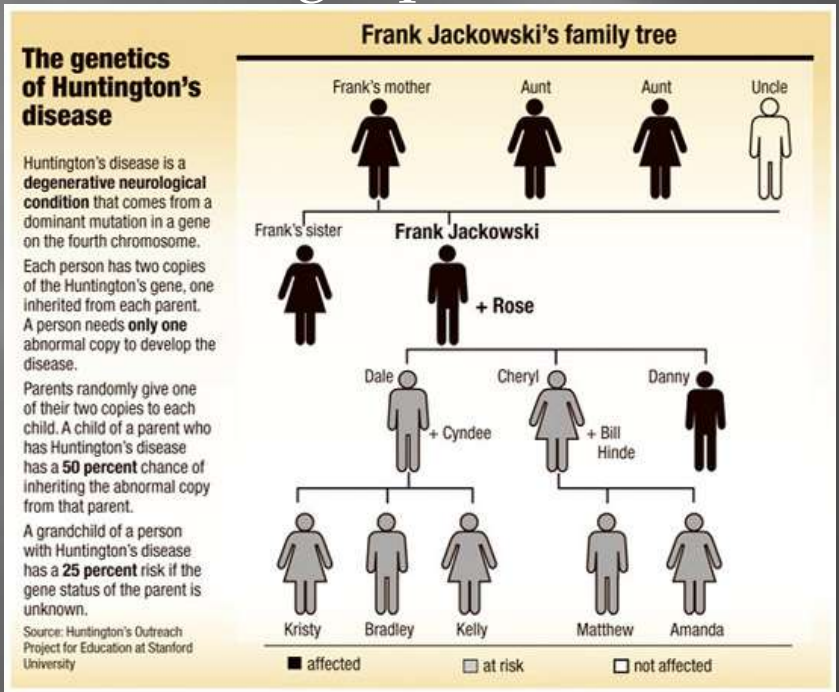
III. Simple Dominant Heredity



Pedigree 1. An idealized pedigree of a family with hypercholesterolemia, an autosomal dominant disease where the heterozygote has a reduced number of functional low density lipoprotein receptors.

III. Simple Dominant Heredity

- Huntington's Disease
  - Lethal and caused by dominant allele
  - Break down of parts of brain
  - How does this get passed if lethal?



IV. Complex Patterns of Inheritance

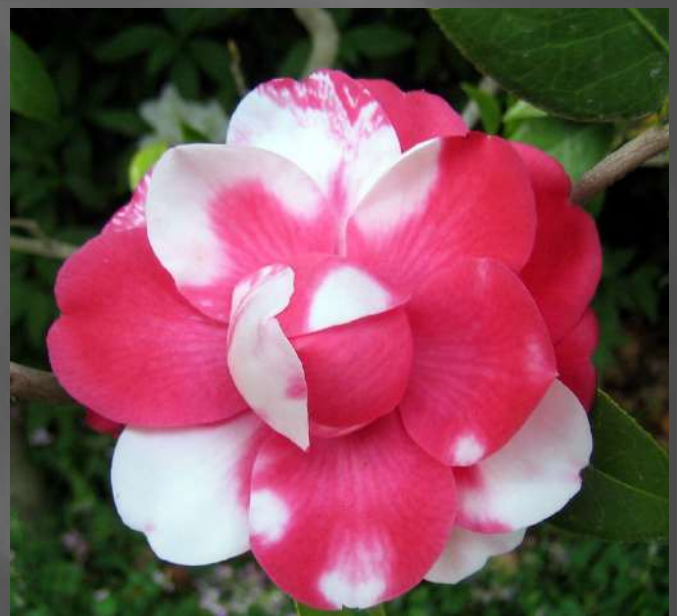
A. Incomplete Dominance

- Phenotype of heterozygous organisms are "between" parents' traits
  - Red flower (RR) with white flower (R'R') = Pink (RR')
  - R produces red pigment, R' produces no pigment

IV. Complex Patterns of Inheritance

B. Codominance

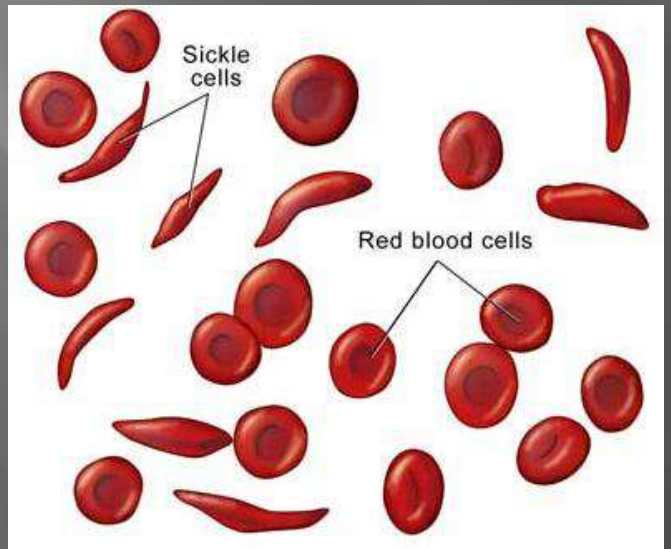
- Phenotypes of both parents to be produced in heterozygous individuals (expressed equally)
  - B and W (two different uppercase letters to express alleles)



Codominance in Humans

Sickle-cell Anemia

- African decent and countries around Mediterranean Sea
- Homozygous – red blood cells shaped like half moon
- Heterozygous- normal and sickle shapes



IV. Complex Patterns of Inheritance

C. Sex Determination

- Humans = 46 chromosomes (23 pairs)
- 22 pairs = **Autosomes**
- **Sex chromosomes** – male or female (XY, XX)
  - Abnormal numbers of sex chromosomes
  - XO, XXX or XXY, XYY

IV. Complex Patterns of Inheritance

Sex Chromosome Abnormalities

Genotype	Gender	Syndrome	Physical Traits
XXY, XXYY, XXXY	male	Klinefelter syndrome	sterility, small testicles, breast enlargement
XYY	male	XYY syndrome	normal male traits
XO	female	Turner syndrome	sex organs don't mature at adolescence, sterility, short stature
XXX	female	Trisomy X	tall stature, learning disabilities, limited fertility

IV. Complex Patterns of Inheritance

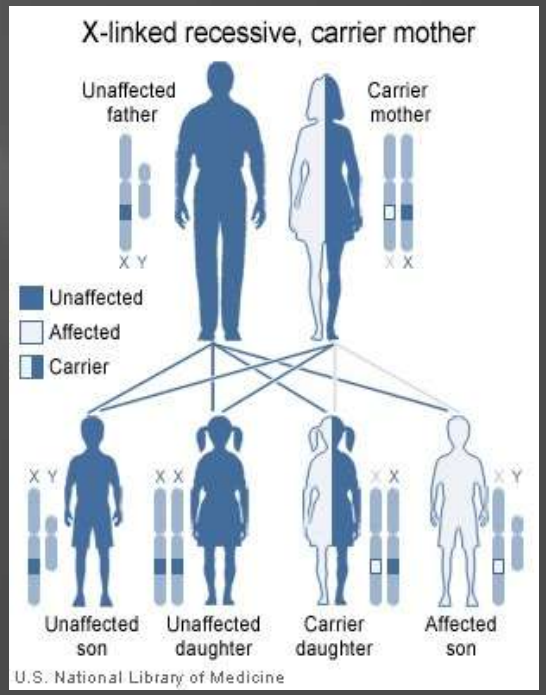
D. Sex Linked Inheritance

- Traits controlled by genes located on sex chromosomes
- Y has no corresponding allele
- Y linked only passed from male to male
- Females must have both recessive (xx)
- All X-linked are expressed in Males
  - No corresponding X to mask trait

IV. Complex Patterns of Inheritance

D. Sex Linked Inheritance

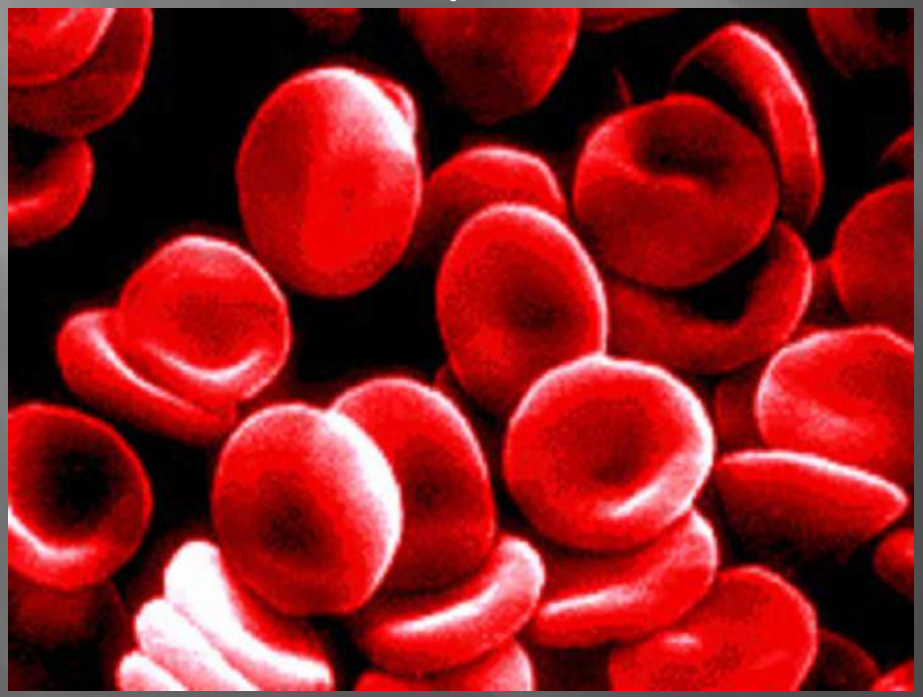
- Males pass X onto daughter, Y to son
- Son receives recessive allele on X chromosome = display of recessive trait (red-green color blindness, hemophilia)



IV. Complex Patterns of Inheritance

E. Multiple Alleles

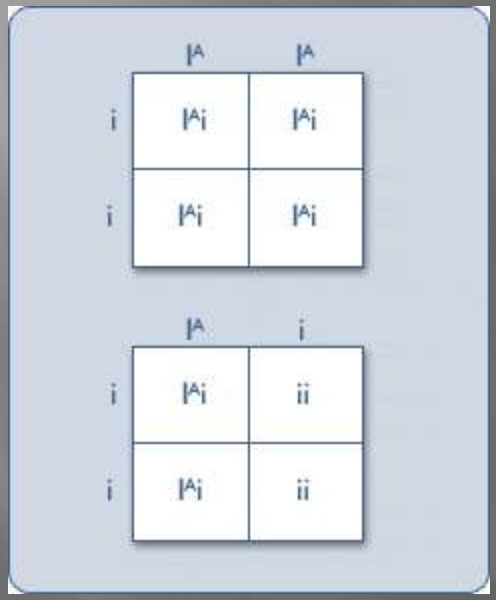
- Traits controlled by more than two alleles (same gene)
- Can be formed by mutations



Ex. Blood Types

Blood Type

- Blood types – A,B,AB, and O
- 3 Alleles – I^A,I^B, and i
- Child = AB, Mother = A, father could not be O



Blood Type	Genotype		Can Receive Blood From:
A	i^Ai i^Ai^A	AA AO	A or O
B	i^Bi i^Bi^B	BB BO	B or O
AB	i^Ai^B	AB	A, B, AB, O
O	ii	OO	O

Blood Type

- **Universal Donor** – O; can give blood to any individual
- **Universal receiver** – AB; can receive blood from any individual

IV. Complex Patterns of Inheritance

F. Polygenic Inheritance

- Inheritance pattern of trait controlled by two or more genes
  - Plant height – A,B, and C (3 genes on 3 chromosomes)
  - Ex in humans – eye color, skin color
  - How is this different than multiple allele?

V. Environmental Influences

- Internal factors can influence expression
  - Hormones, age

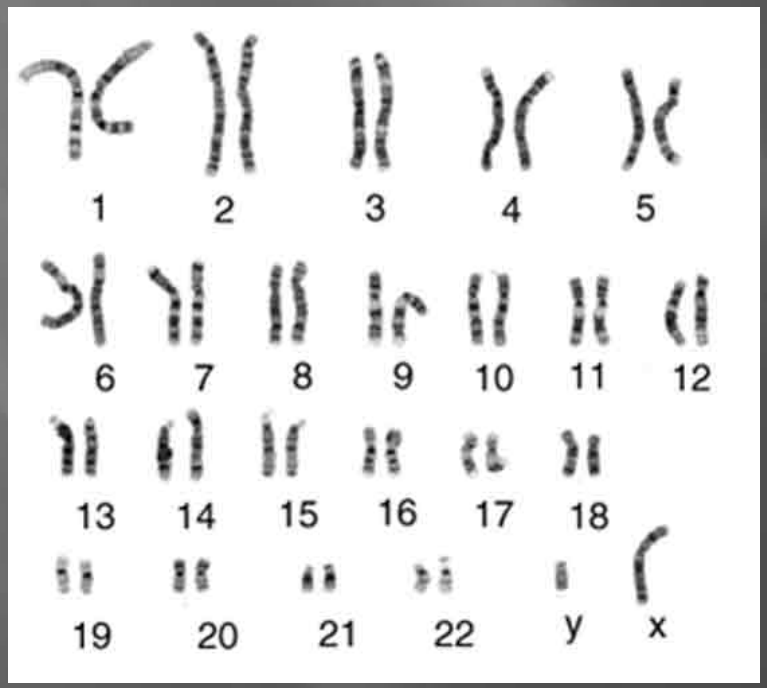
- External factors can influence expression
  - Temperature, nutrition, light, chemicals
  - Arctic foxes – temperature affects coat color

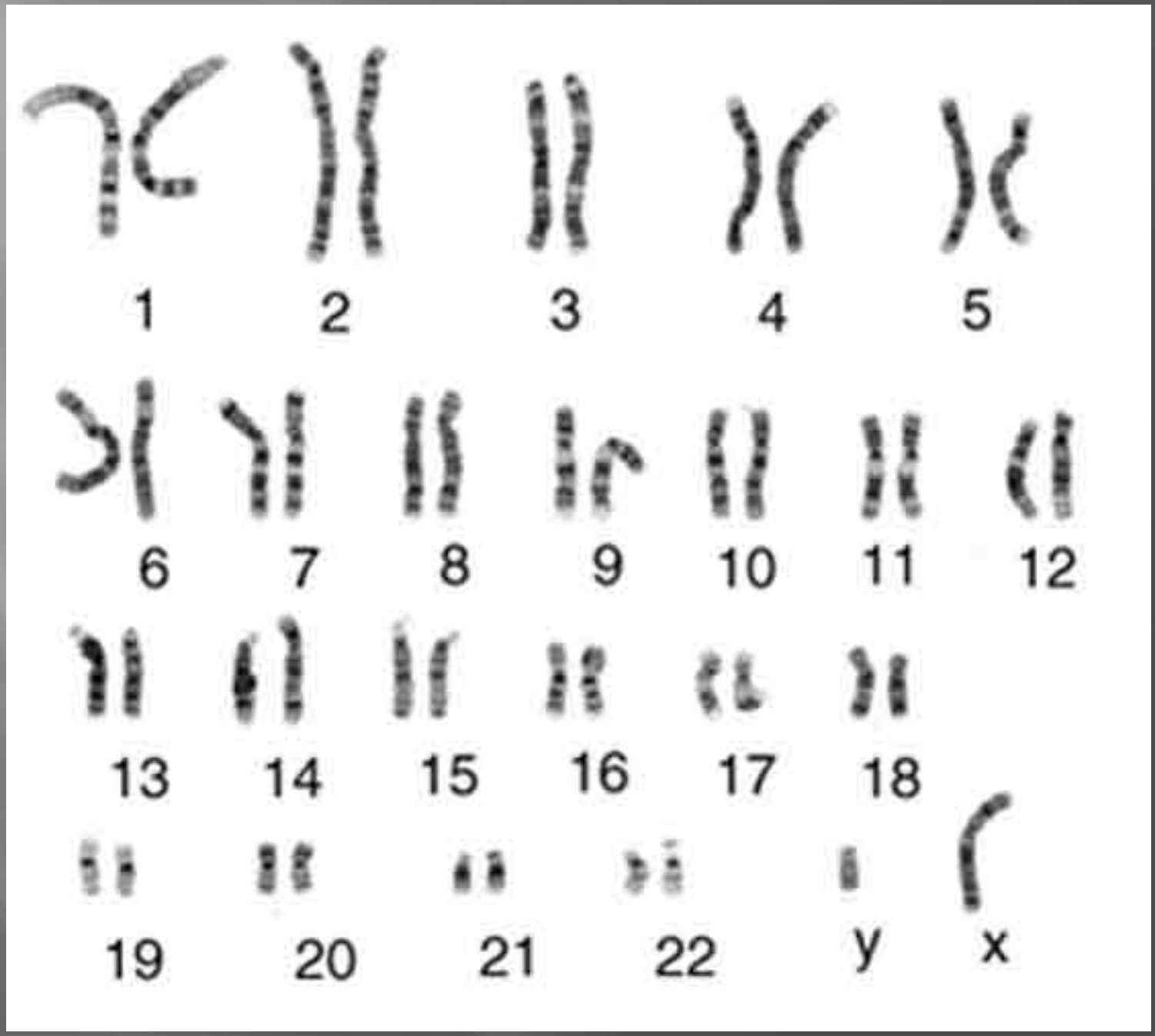
V. Environmental Influences

- Identical twins –
  - Same DNA
  - May look different because of environment
    - Nutrition
    - Exercise
    - Accidents (scars, etc)

VI. Chromosome #'s

- **Karyotype** – chart of chromosome pairs
- Down syndrome: chromosome 21 Trisomy
  - Higher chance with older mothers

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