Human Heredity
Chapter 4

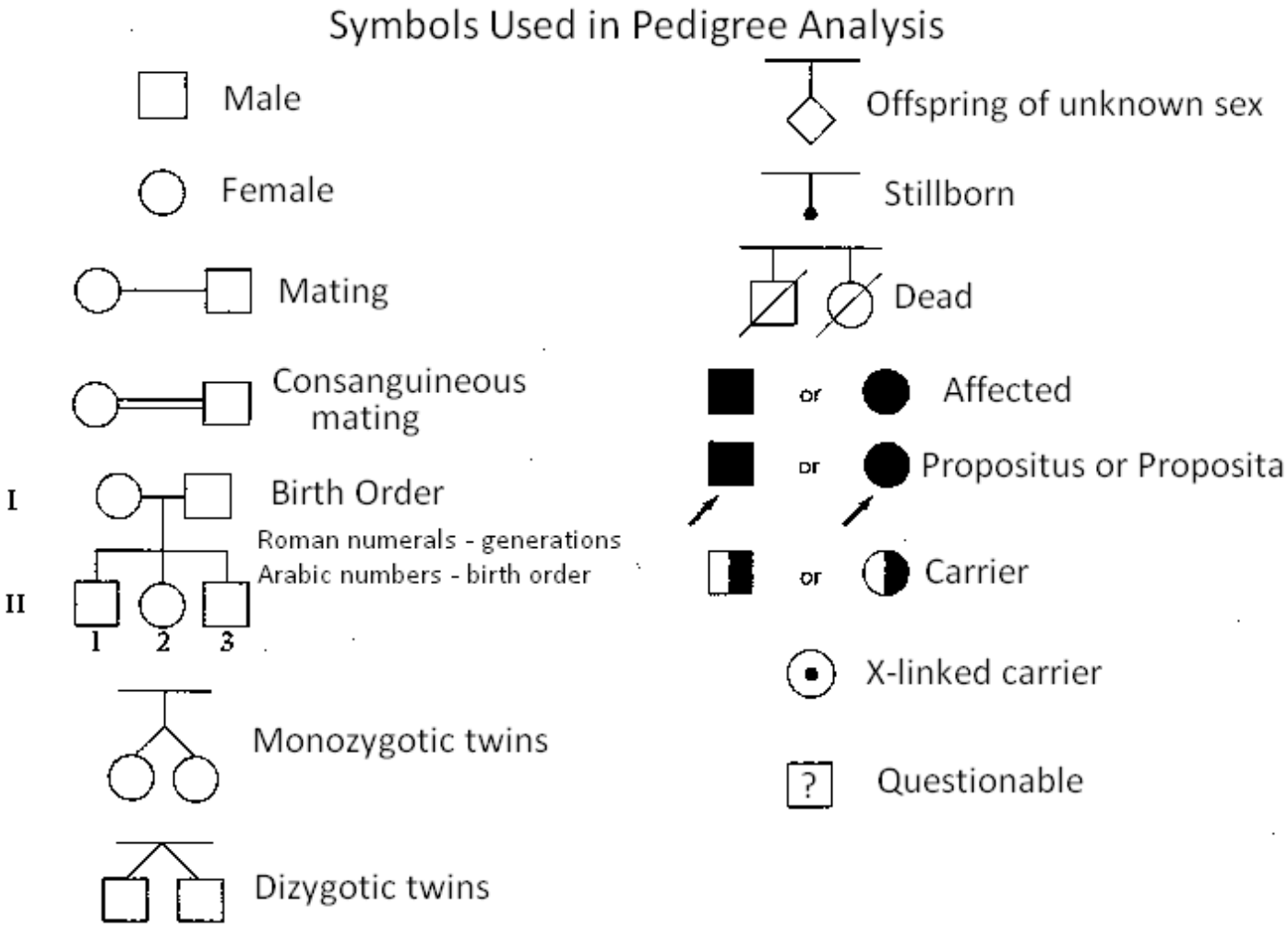
Inheritance Patterns

4:1 Pedigrees

PEDIGREE: family record that shows how a trait is inherited over several generations

Pedigrees may be used for:

- Revealing CARRIERS: a person who is heterozygous for a trait; usually used when referring to a genetic disease
- Determining if a trait is dominant, recessive, or sex linked.



Terms used in Pedigree Analysis:

- CONSANGUINEOUS MATING: mating between close relatives, especially brother and sister
- PROPOSITUS (male)/PROPOSITA(female): first individual in family identified with the trait, lead to the construction of the pedigree
- MONOZYGOTIC: identical twins, arising from one egg and one sperm
- DIZYGOTIC: fraternal twins, arising from two eggs and two sperm

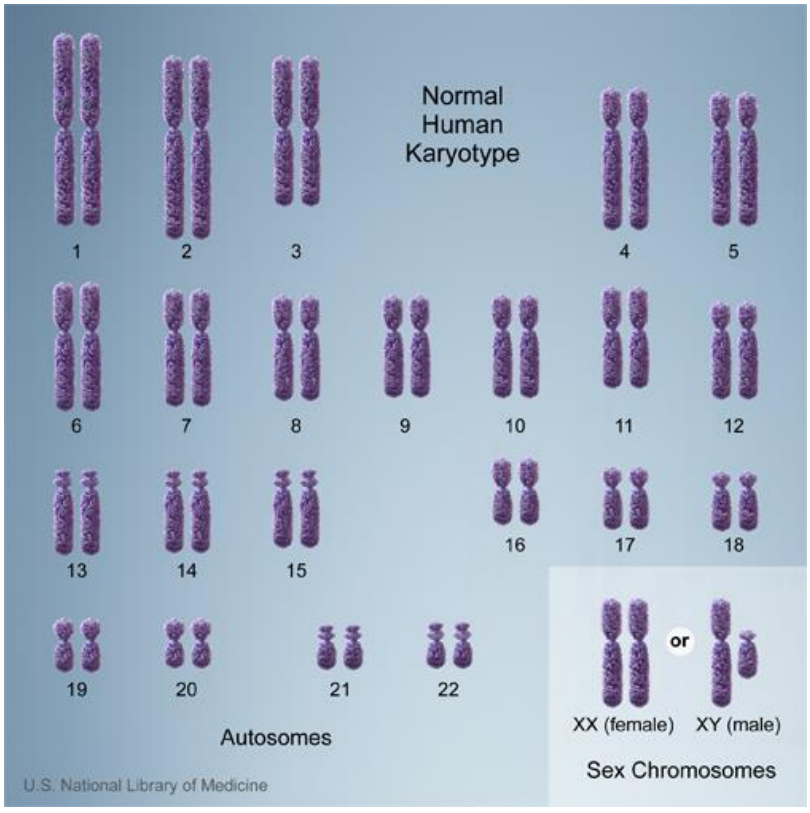
4:2 Sex Determination

The human KARYOTYPE is composed of 22 pairs of AUTOSOMES and one pair of SEX CHROMOSOMES.

KARYOTYPE: the chromosomes of an individual, photographed at metaphase and arranged in a standard sequence

AUTOSOMES: all homologous pairs of chromosomes, not sex chromosomes

SEX CHROMOSOMES: chromosomes involved in sex determination, X and Y in humans

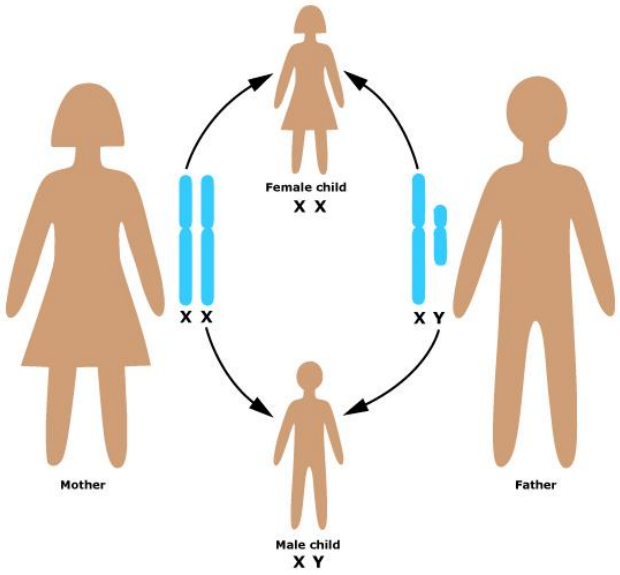


Female sex chromosomes are XX, females are HOMOGAMETIC: produce gametes that contain only one type of sex chromosome (XX). All eggs contain an X.

Male sex chromosomes are XY, males are HETEROGAMETIC: produce gametes that contain different kinds of sex chromosomes. Half of sperm contain X, the other half contain Y.

HOW IS SEX DETERMINED?

- Egg (X) fertilized by sperm with X results in female (XX) offspring.
- Egg (X) fertilized by sperm with Y results in male (XY) offspring.



Father determines sex of the children!

	X	Y
X	XX	XY
X	XX	XY

Predicted ratio → 1 male : 1 female

Actual ratio in live births → 1.05 male : 1 female

Why are more males born?

✓Sperm containing Y chromosome are slightly smaller and swim faster.

Sex ratio at age 25 → 1 male:1 female

Why do more males die at a young age?

Young males die of X-linked disease and accidents more than females.

Sperm containing X live longer in the female reproductive system. If intercourse occurs prior to ovulation then you have a greater chance of a girl because the sperm containing Y are dead.

4:3 Aneuploidy of Sex Chromosomes

ANEUPLOIDY: possessing a chromosome number that is not an exact multiple of the haploid
MONOSOMY: possessing only one chromosome of a homologous pair
TRISOMY: possessing three homologous chromosomes instead of a homologous pair

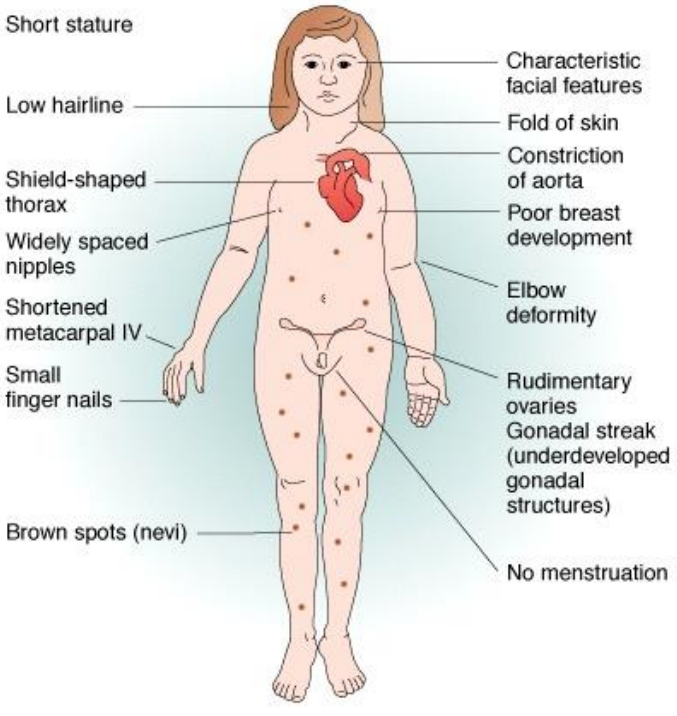
Aneuploidy is more common in sex chromosomes than in autosomes and most affected individuals are viable.
MONOSOMY Y (0Y) is always LETHAL.

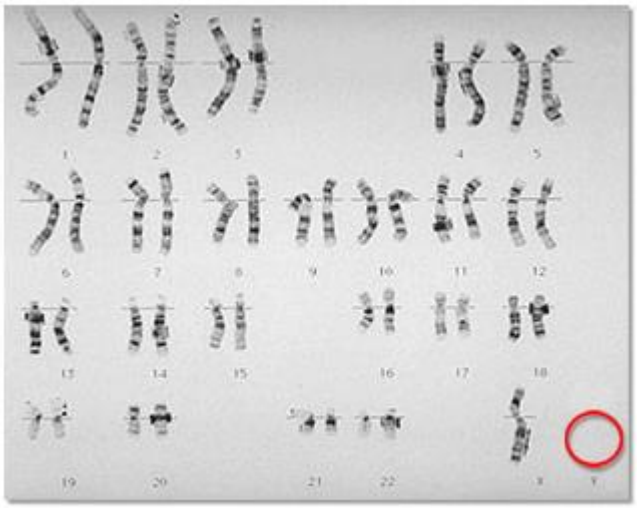
Diseases caused by XY aneuploidy

1. TURNER SYNDROME: Monosomy X (45,X)

Genotype: X0
Phenotypic Characteristics:

- Short
- Wide chested with under-developed breasts
- Lack of ovarian development
- Narrowing of the aorta





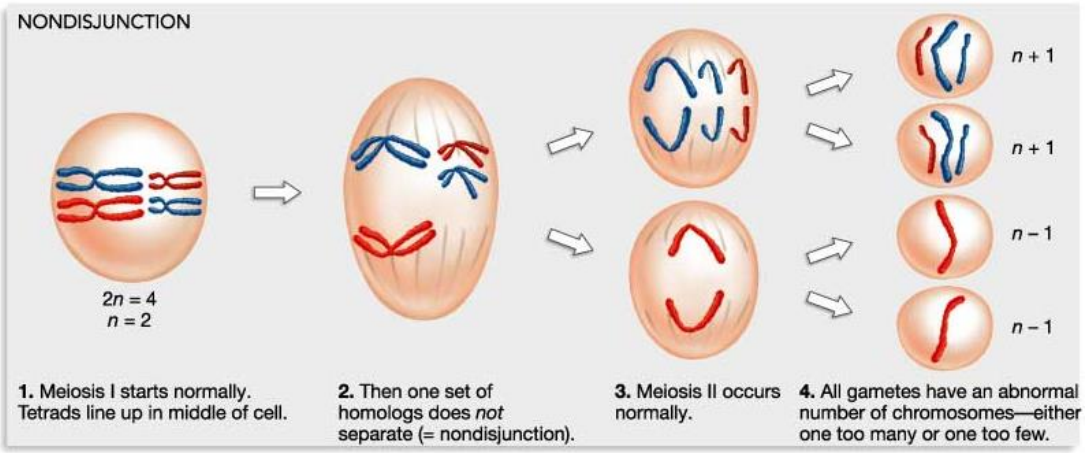
Rate of Occurrence:

- 1% of all conceptions, 95-99% of all 45,X embryos die before birth
- 1 in 10,000 live births

Cause:

- Nondisjunction, 75% of time in sperm

NONDISJUNCTION: when chromosomes fail to properly separate at anaphase

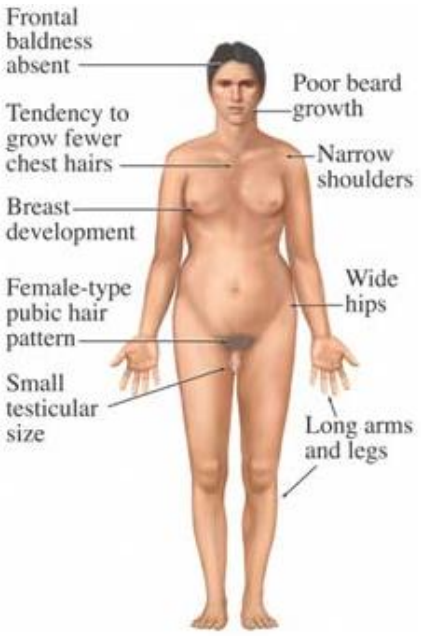


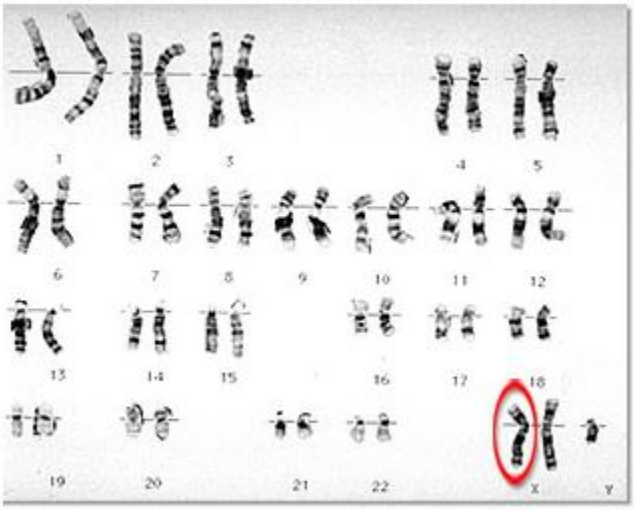
2. KLINEFELTER SYNDROME: 47,XXY

Genotype: XXY

Phenotypic Characteristics:

- Do not have problems until puberty
- Poor sexual development
- Lowered fertility
- Breast development in 50% of cases
- Often experience language deficits and academic difficulties



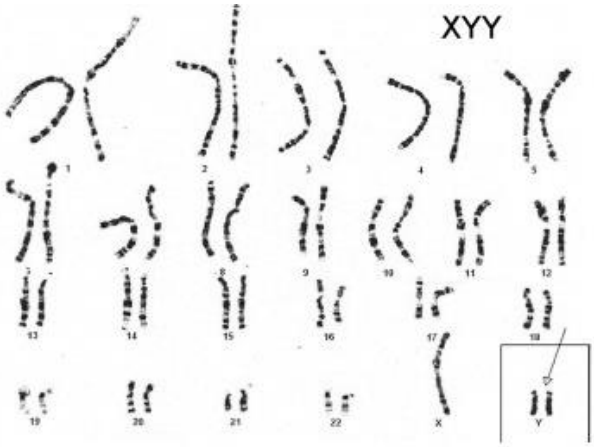


- Rate of Occurrence:
  - 1 in 1000 male births

Cause:

- Nondisjunction, 60% of time in egg
- Rate increases with advanced maternal age

3. XYY SYNDROME: 47,XYY



Genotype: XYY

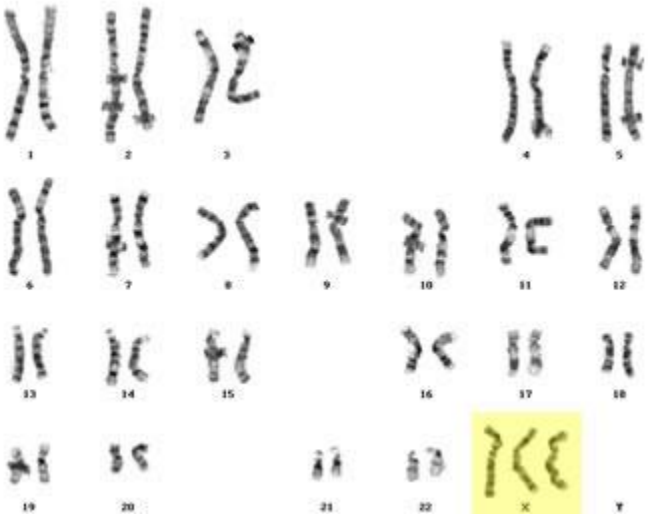
Phenotypic Characteristics:

- Above average height
- Personality disorders
- Increased risk of learning disabilities, delayed language skills
- Hyperactivity, impulsive behavior, lack of emotional control, and outbursts of temper
- Increased convictions for sexual abuse, burglary, arson as compared to “normal” men (2012)

Rate of Occurrence:

- 1 in 1000 male births

4. XXX SYNDROME: 47,XXX



Genotype: XXX

Phenotypic characteristics:

- Most perfectly normal
- Slight increase in sterility and learning disabilities

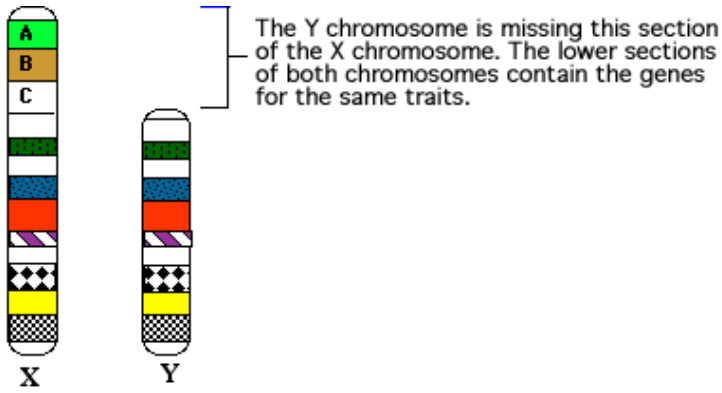
Rate of Occurrence:

- 1 in 1000 female births

4:4 Sex Linked/X-Linked Recessive Inheritance

Some traits are X-Linked because:

- X chromosome carries alleles for some characteristics, the Y chromosome may not carry those alleles.
- Females have two alleles for those characteristics, males have only one allele.

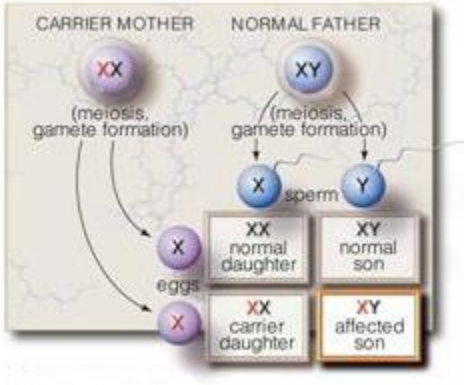


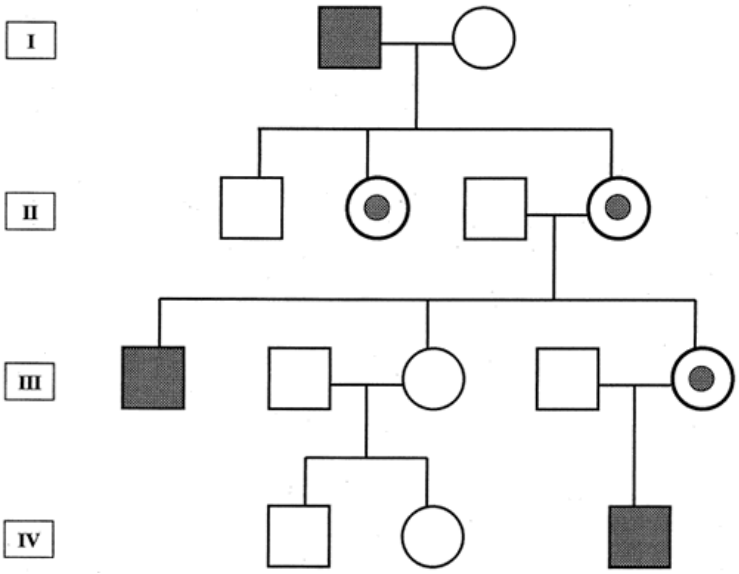
X-LINKED TRAIT: trait related to sex, allele for which is found on the X chromosome but not on the Y

HEMIZYGOUS: when an individual (usually male) has only one allele controlling a trait, single dominant allele causes dominant trait, single recessive allele causes recessive trait

Patterns of X-Linked Recessive Inheritance

1. Hemizygous males (with trait) pass the allele to all daughters, who then are carriers or have the trait.
2. Carriers pass the allele to 50% of their sons.
3. Fathers cannot pass the trait to their sons, only to the sons of their daughters, their grandsons.
4. Phenotype will be present at a much higher rate in males than females.





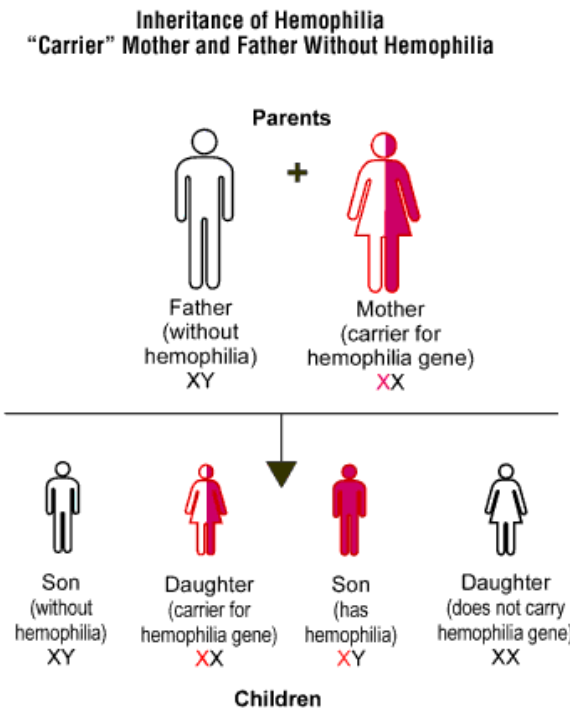
X-Linked Recessive Diseases

HEMOPHILIA A: disease in which blood lacks one of the clotting factors (proteins that interact to form clots)

Genotype: X^hY, X^hX^h, carrier - X^HX^h

Phenotypic Characteristics:

- Blood clots very slowly
- Affected individuals may bleed to death from small cuts (e.g. circumcision)
- Rare in females because most hemophiliac males did not live to reproductive age until recent advancements in treatment
- May be treated by the injection of clotting factor obtained from normal blood

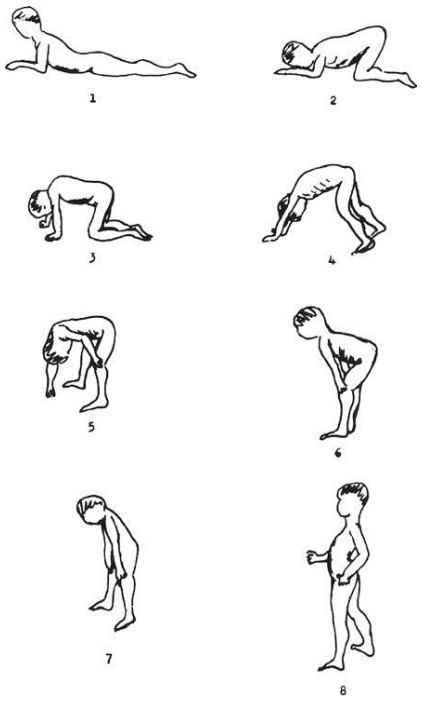


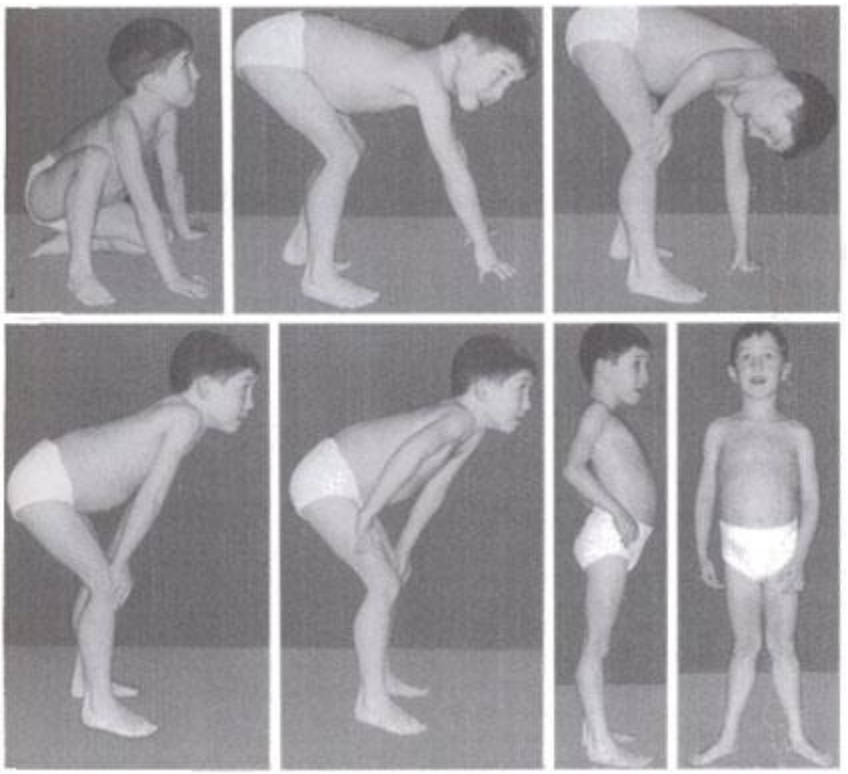
Rate of Occurrence:

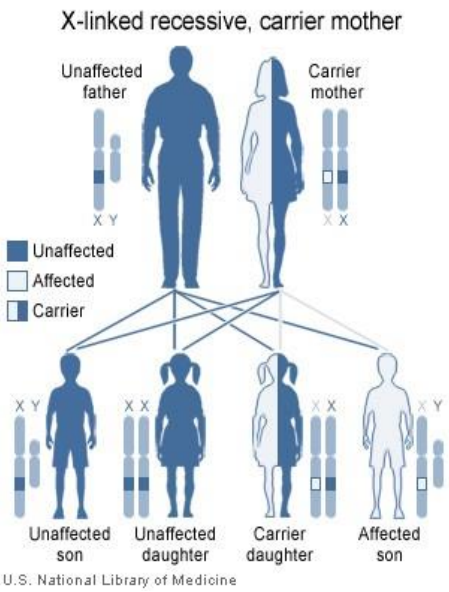
- Males 1:10,000
- Females 1:100,000

DUCHENNE'S MUSCULAR DYSTROPHY: X-linked form of the disease that produces muscular weakness and progressive muscle degeneration

Genotype: X^mY, X^mX^m, carrier X^MX^m





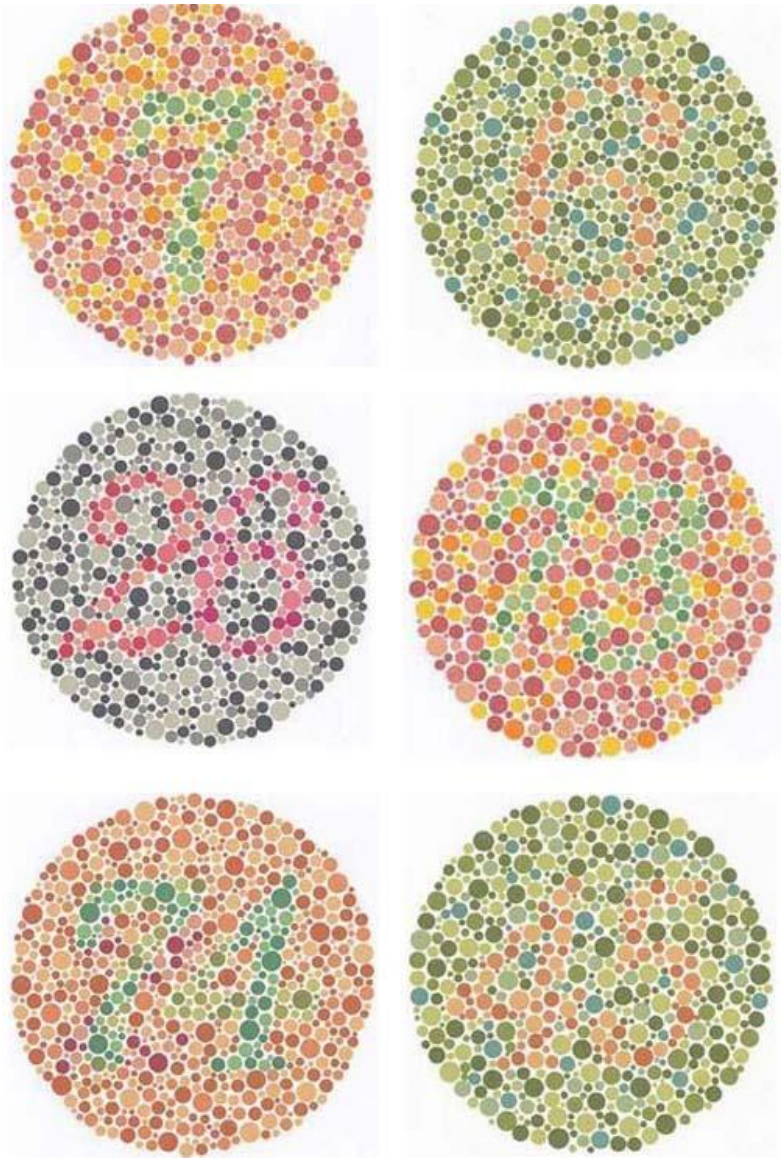


Phenotypic Characteristics:

- Gower Sign – affected child "climbs" from prone to standing position
- Onset 1-6 years
- Usually confined to wheelchair by age 12
- Death by age 20 due to respiratory infection or cardiac failure
- Most affected males die before they can reproduce, passed on by carriers

Rate of Occurrence:

- Males 3:10,000
- Females have almost no chance of being affected



COLORBLINDNESS: X-linked disorder in which an individual cannot distinguish between certain colors, usually red and green

Genotype: X^nY, X^nX^n, carrier X^NX^n

Phenotypic Characteristics:

- Colors appear as shades of gray

Rate of Occurrence:

- Males 8:100

CAN TWO NORMAL PARENTS HAVE A SON WITH AN X-LINKED DISORDER?

- Yes, if Mother is a carrier she passes the recessive gene to half her sons.

	X^N	Y
X^N	X^NX^N	X^NY
X^n	X^NX^n	X^nY

- Normal Father cannot have a daughter affected with an X-linked recessive disorder.