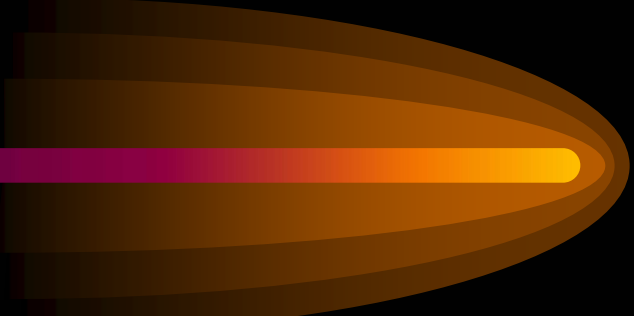


HUMAN HEREDITY



ONE OF THE MOST IMPORTANT
AND
USEFUL FORMS OF BIOLOGY

- 
- HUMANS CONTAIN DIPLOID CELLS
 - 46 CHROMOSOMES; 23 PAIRS
 - EACH CELL CONTAINS 6 BILLION NUCLEOTIDE PAIRS
 - IF WRITTEN IN A TEXT BOOK IT WOULD HAVE 1 MILLION PAGES
 - EACH PARENT CONTRIBUTES 1 COPY OF EACH GENE THEREFORE THE GAMETES (SEX CELLS) CONTAIN 1 COPY OF EACH. THEY ARE HAPLOID
 - THE SEX CELL CHROMOSOMES ARE X AND Y
 - TRAITS ARE CARRIED ON TO THE NEXT GENERATION BY SINGLE TRAITS, DOMINANT ALLELES, SEX-LINKED ALLELES, OR ANY NUMBER OF OTHER WAYS

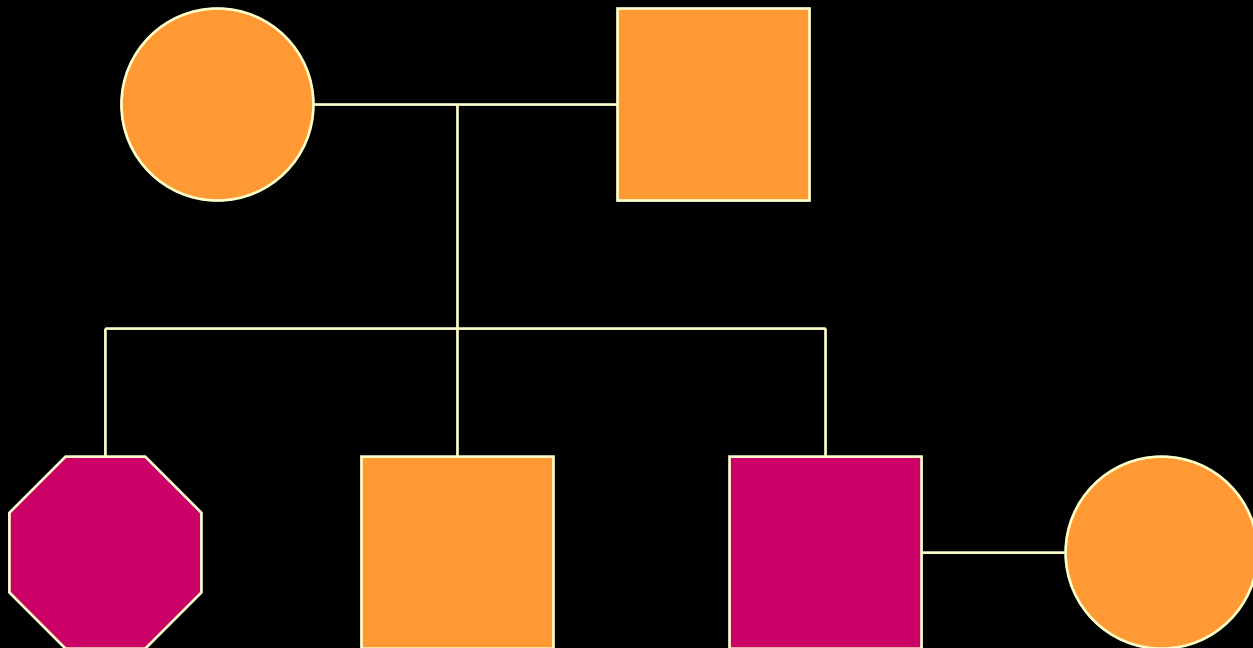
INHERITANCE OF HUMAN TRAITS

- THERE ARE OVER 1000 GENES THAT HAVE BEEN STUDIED AND DESCRIBED

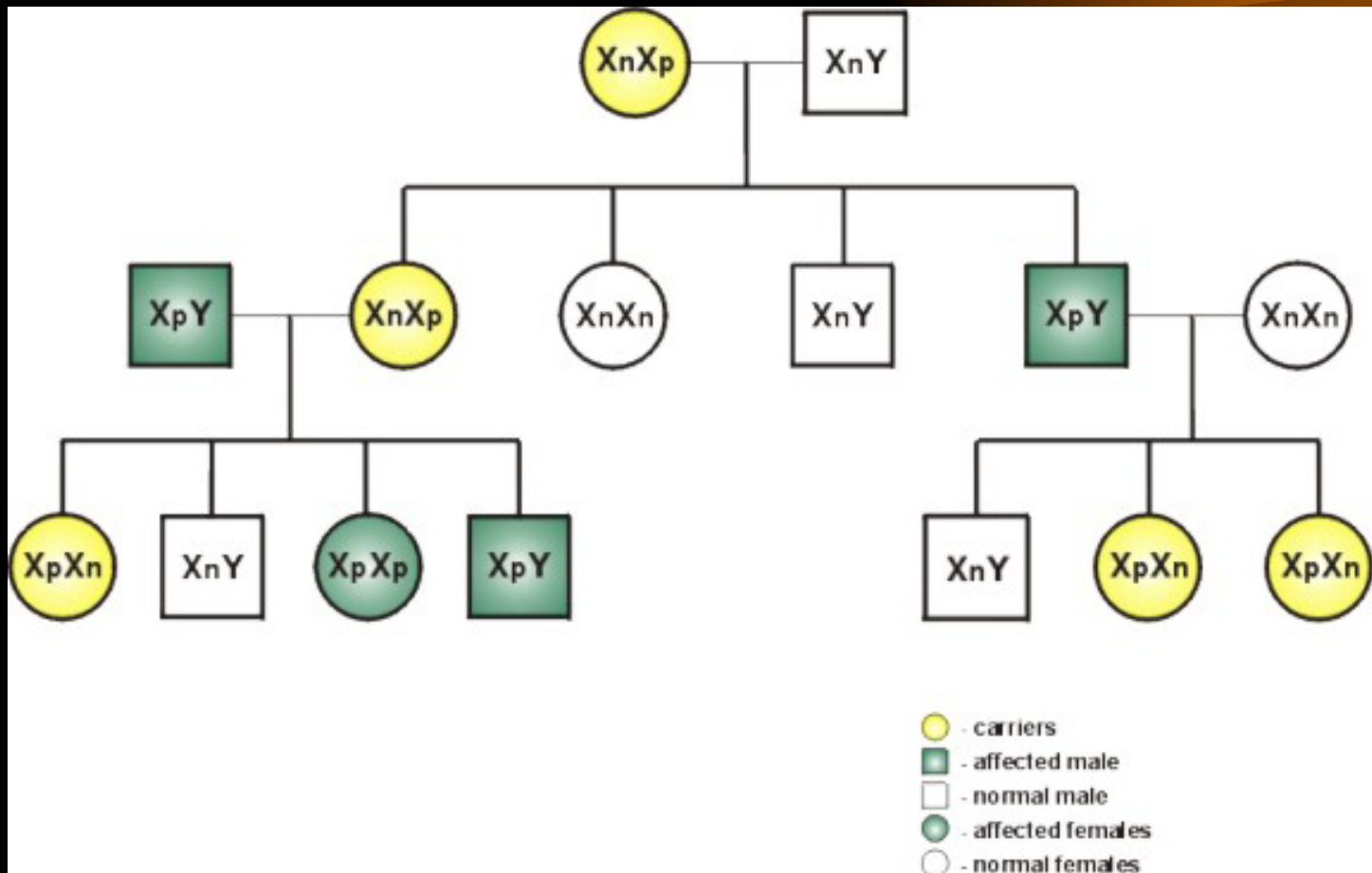


Making A Pedigree

- A map of genetic traits from generation to generation



Pedigree Example w/ Colorblindness

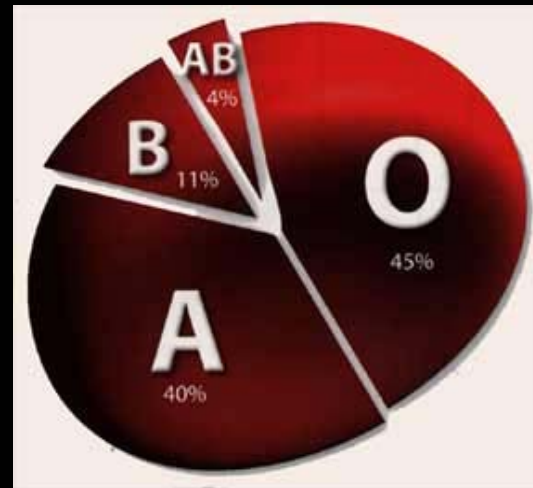


BLOOD GROUPS

- Blood groups are coded by multiple alleles (3 or more)
- ABO blood
 - blood types are determined by proteins and chemical substances
 - blood contains antigens (molecules in the immune system found in the blood)
 - antigen A and antigen B
 - 4 different blood types exist
 - blood type A: contains antigen A, but not B
 - blood type B: contains antigen B, but not A
 - blood type AB: contains both antigen A and B
 - blood type O: contains neither antigen A or B
 - Opposite blood types can't mix during transfusions
 - AB can receive any blood
 - A can receive O and A and give to A and AB
 - B can receive O and B and give to B and AB
 - O can receive from O and give to all

BLOOD GROUPS CONT.

- BLOOD TYPES ARE DETERMINED BY A SINGLE GENE WITH 3 ALLELES
 - I^A, I^B, i
- $ii = O$
- $I^A I^A, I^A i = A$
- $I^B I^B, I^B i = B$
- $I^A I^B = AB$



BLOOD GROUPS CONT.

- Rh factor:
 - another antigen present in blood which determines whether blood transfusions will agglutinate (clump)
 - Rh+ : antigen is present
 - Rh- : antigen is not present (recessive)
 - determined by multiple alleles (8 or more)
 - problems that occur are with pregnancies (1 in 300-400)
 - Rh- female x Rh+ male: child may inherit Rh+
 - if second Rh+ child occurs antibodies from mother will attack babies blood

Genetic disorders

- Huntington disease:
 - produced by a single dominant allele (H)
 - symptoms appear at age 30-40
 - damage to nervous system, loss of muscle control, mental disfunction, DEATH usually w/in 12 years



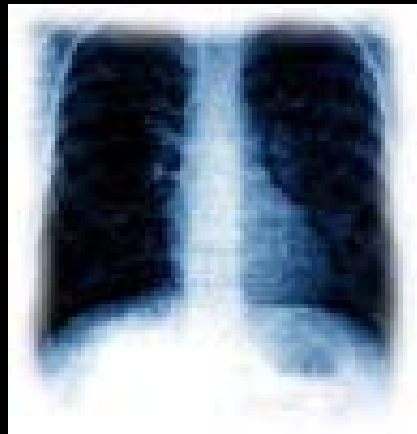
Sickle Cell Anemia:

- causes a deformity in shape of red blood cells
- changes protein structure in the cells
- cells lose their ability to carry oxygen
- single nucleotide mutation



Cystic fibrosis:

- defective protein in the plasma membrane that causes an accumulation of thick mucus in the lungs and digestive tract



Tay-Sachs disease:

- recessive disorder of the central nervous system, caused by an absence of an enzyme that normally breaks down a lipid produced and stored in tissues of the CNS
- Leads to neuronal swelling, and death

Phenylketonuria (PKU):

- A recessive disorder that results from the absence of an enzyme that converts one amino acid (phenylalanine) to a different amino acid (tyrosine)
 - Phenylalanine builds up in the body and damages the CNS



SEX LINKED INHERITANCE

- X chromosome is essential for survival
- Y chromosome determines the sex of the individual (all fetuses start out female, but the Y chromosome switches when the embryo develops)

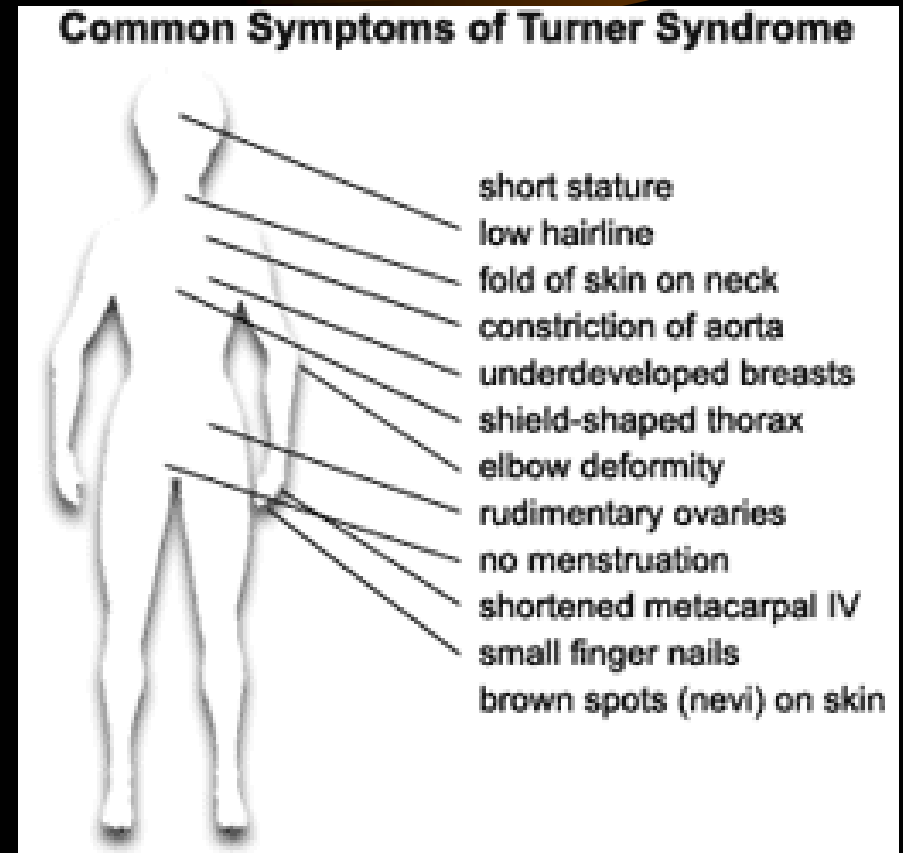
SEX LINKED INHERITANCE

- THE SEX OF A PERSON IS DETERMINED BY WHETHER THE MALE IS X OR Y CARRYING
 - NORMAL: XX = FEMALE (46XX)
 XY = MALE (46XY)
 - ABNORMAL: NONDISJUNCTION DURING MEIOSIS IN WHICH THE CHROMOSOMES FAIL TO SEPARATE COMPLETELY
 - 1 IN 1000 PROBABILITY: RESULTS IN OFFSPRING WITH EXTRA SEX CHROMOSOMES OR NOT ENOUGH
 - IF THEY DON'T HAVE ENOUGH THEY DIE
 - IF THEY HAVE TOO MANY THEY INHERIT A GENETIC DISEASE

SEX LINKED INHERITANCE

GENETIC DISORDERS

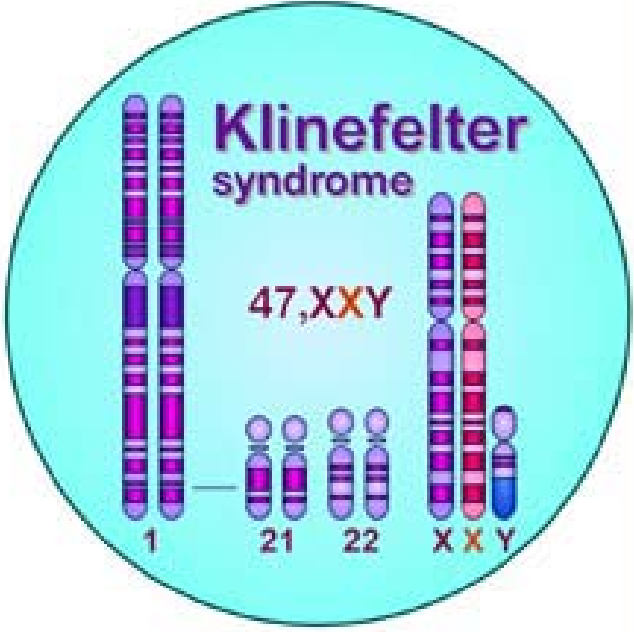
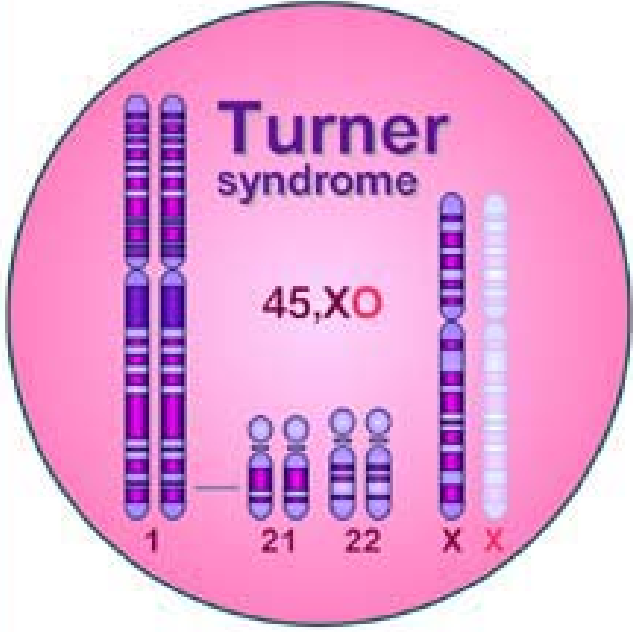
- **TURNER SYNDROME:**
 - female in appearance but lacks developed sex organs; therefore they are sterile
 - 45X or 45XO
 - lacks second sex chromosome



KLINFELTER SYNDROME:



- male in appearance, but sterile
- 47XXY
- has an extra X chromosome



SEX LINKED INHERITANCE

GENETIC DISORDERS

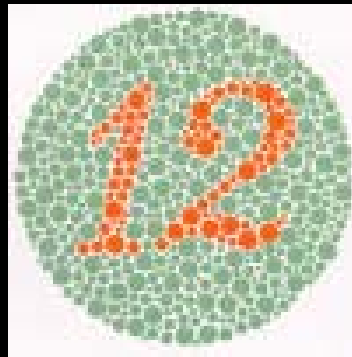
- Sex linked - genes carried on the sex chromosomes
 - Y: small, few genes carried
 - X: larger, more genes carried
- Recessive defects located on the X chromosome are more common in males
- All X-linked genes are expressed in males even if they are recessive

COLOR BLINDED CONDITIONS

- Recessive disorder located on the X chromosome
- most common: red-green colors
- most affected: males (8 to 1)
- X^C = dominant (normal)
- X^c = recessive (colorblind)
- $X^C X^C$ = normal female $X^c X^c$ = colorblind female
- $X^C X^c$ = normal female but a carrier
- $X^C Y$ = normal male $X^c Y$ = colorblind male



Are you colorblind?





HEMOPHILIA

- Recessive gene disorder - causes bleeding disorders
- very rare: 1 in 10,000 males, 1 in 100,000,000 females
- very serious disease
- treatable by adding missing AHF factor (antihemophilic)

MUSCULAR DYSTROPHY

- Progressive wasting away of skeletal tissue
- most often carried on the X chromosome
- kills in early adulthood



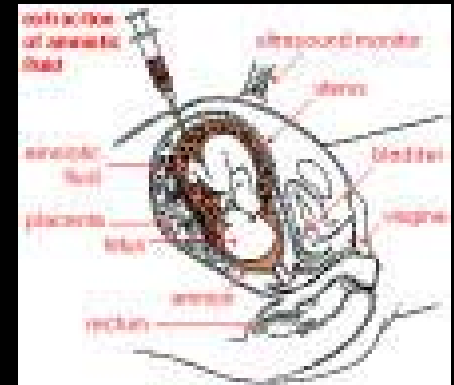


DOWN SYNDROME

- Non-disjunction of an autosome (non sex cells)
 - extra copy of chromosome #21
- causes mental retardation
- increased susceptibility to many diseases
- 1 in 800 in the U.S.
- can diagnose with a microscope

DIAGNOSIS OF GENETIC DISORDERS

- AMNIOCENTESIS:
 - TAKING OF FLUID FROM THE SAC AROUND THE EMBRYO AND GROWING A CULTURE



- MICROSCOPES:
 - VIEWING SAMPLES UNDER A MICROSCOPE CAN IDENTIFY SOME DISEASES