Blood won't clot

HEMOPHILIA

HEMOPHILIA IS AN SEX-LINKED RECESSIVE DISORDER. HEMOPHILIA IS A MEDICAL CONDITION IN WHICH THE ABILITY OF THE BLOOD TO CLOT IS SEVERELY REDUCED, CAUSING THE SUFFERER TO BLEED SEVERELY FROM EVEN THE SLIGHTEST INJURY. SYMPTOMS INCLUDE MANY LARGE OR DEEP BRUISES, JOINT PAIN, SWELLING, UNEXPLAINED BLEEDING, AND BLOOD IN URINE OR STOOL.

TREATMENTS: INJECTIONS OF CLOTTING FACTOR OR PLASMA.

MOST IMPACTED: MALE

GENOTYPES:

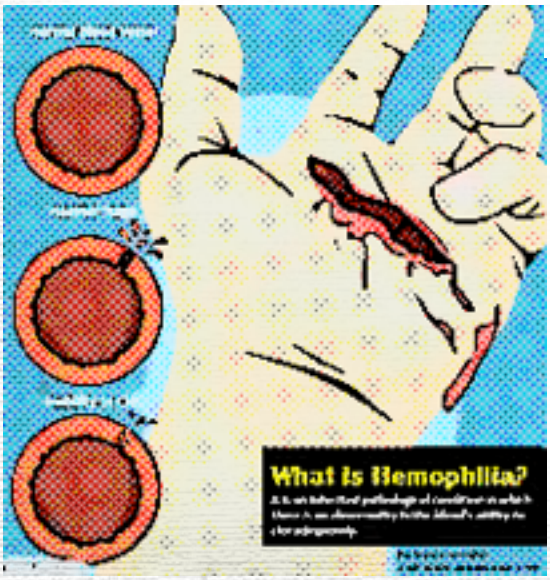
Allele Key X^H *Normal*

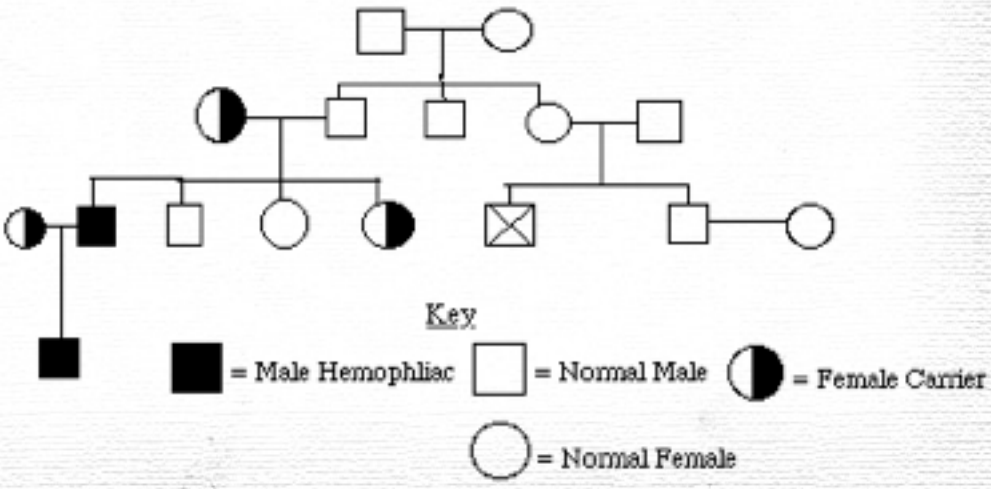
X^h *Haemophiliac*

Female Genotypes	*Male Genotypes*
$X^H X^H$	$X^H Y$
$X^H X^h$	$X^h Y$
* $X^h X^h$	

WE LOOK AT HEMOPHILIA WITHIN FAMILY PEDIGREES!

SINCE HEMOPHILIA IS A SEX-LINKED RECESSIVE DISORDER, WE ARE ABLE TO SEE IT PASSED DOWN THROUGH MANY GENERATIONS WITHIN A PEDIGREE. NOTE HOW THE DISORDER IS MORE COMMON IN MALES VERSUS FEMALES.





Can't see colors properly

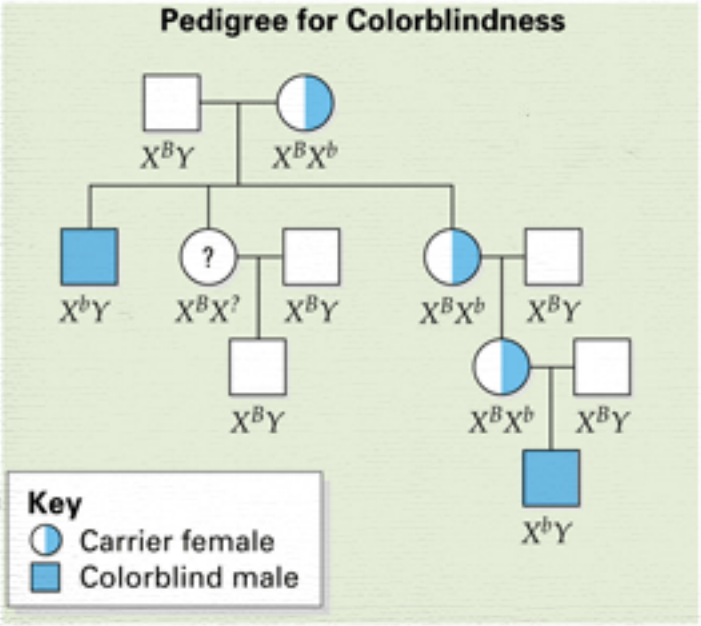
COLORBLINDNESS

COLORBLINDNESS IS AN SEX-LINKED RECESSIVE DISORDER.

MORE MEN ARE IMPACTED VERSUS WOMEN.

CAUSES THE INDIVIDUAL THE INABILITY TO DISTINGUISH BETWEEN SHADES OF RED AND GREEN.

Genotype	Phenotype
$X^C X^C$	Normal female
$X^C X^{\bar{c}}$	Normal female (carrier)
$X^{\bar{c}} X^{\bar{c}}$	Red-green color-blind female (homozygous)
$X^C Y$	Normal male
$X^{\bar{c}} Y$	Red-green color-blind male



WE LOOK AT COLORBLINDNESS WITHIN FAMILY PEDIGREES!

SINCE COLORBLINDNESS IS A SEX-LINKED RECESSIVE DISORDER, WE ARE ABLE TO SEE IT PASSED DOWN THROUGH MANY GENERATIONS WITHIN A PEDIGREE. NOTE HOW THE DISORDER IS MORE COMMON IN

Trisomy 21

Down's Syndrome

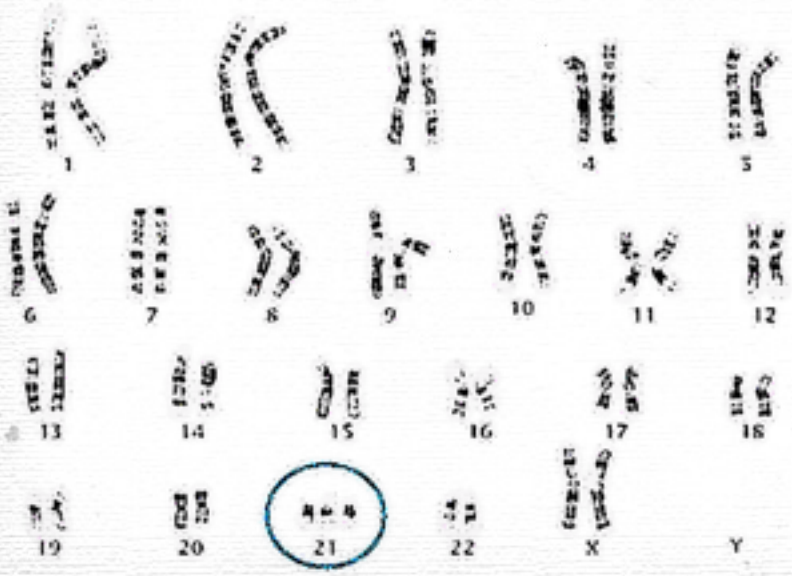
Down's syndrome is a chromosomal anueploidy on the 21st chromosome.

The genotype of the affected individual would read: 47XX +21 for a female and 47XY + 21 for a male

Impacts of Downs Syndrome:

- Distinct facial appearance
- Intellectual disability
- Developmental delays
- Short stature
- Delay in speech

Most affected? Down's syndrome is chromosomal aneuploidy so it occurs at random. Factors that increase risk are advancing maternal age.



We observe Down's syndrome on a karyotype!

Since Downs syndrome is a chromosomal aneuploidy we can observe the disorder using a karyotype. Karyotypes can be obtained by a processes called amniocentesis where doctors take a small sample of embryonic fluid in order to sample the chromosomes.

Observe: chromosome 21 has an extra

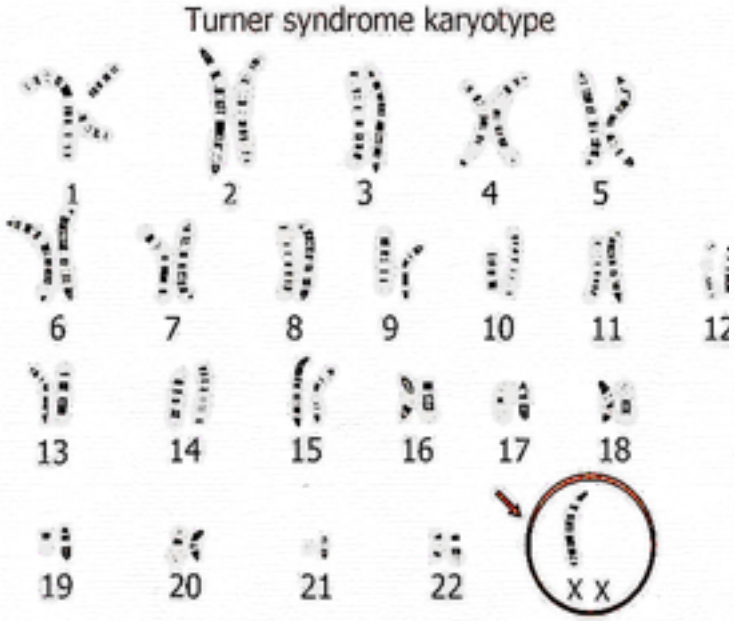
This is a female since she has two X chromsomes.

Monosomy 23

Turner's Syndrome

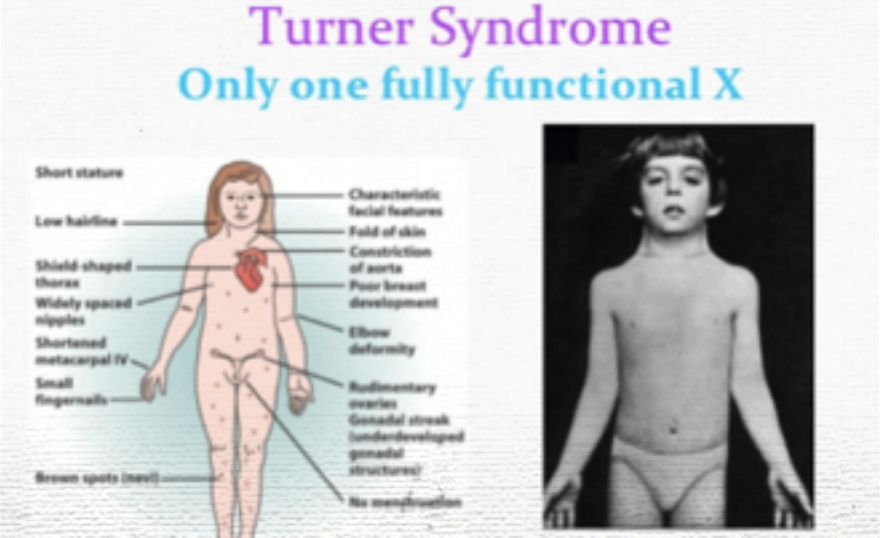
Turner's syndrome is an example of chromosomal aneuploidy since the female is lacking an X chromosome.

Genotype can be written as XO or X_



Symptoms include short stature, delayed puberty, infertility, heart defects, and certain learning disabilities.

Since Turner's syndrome is a chromosomal aneuploidy it does not impact one type of race over the other.



Triosomy 23

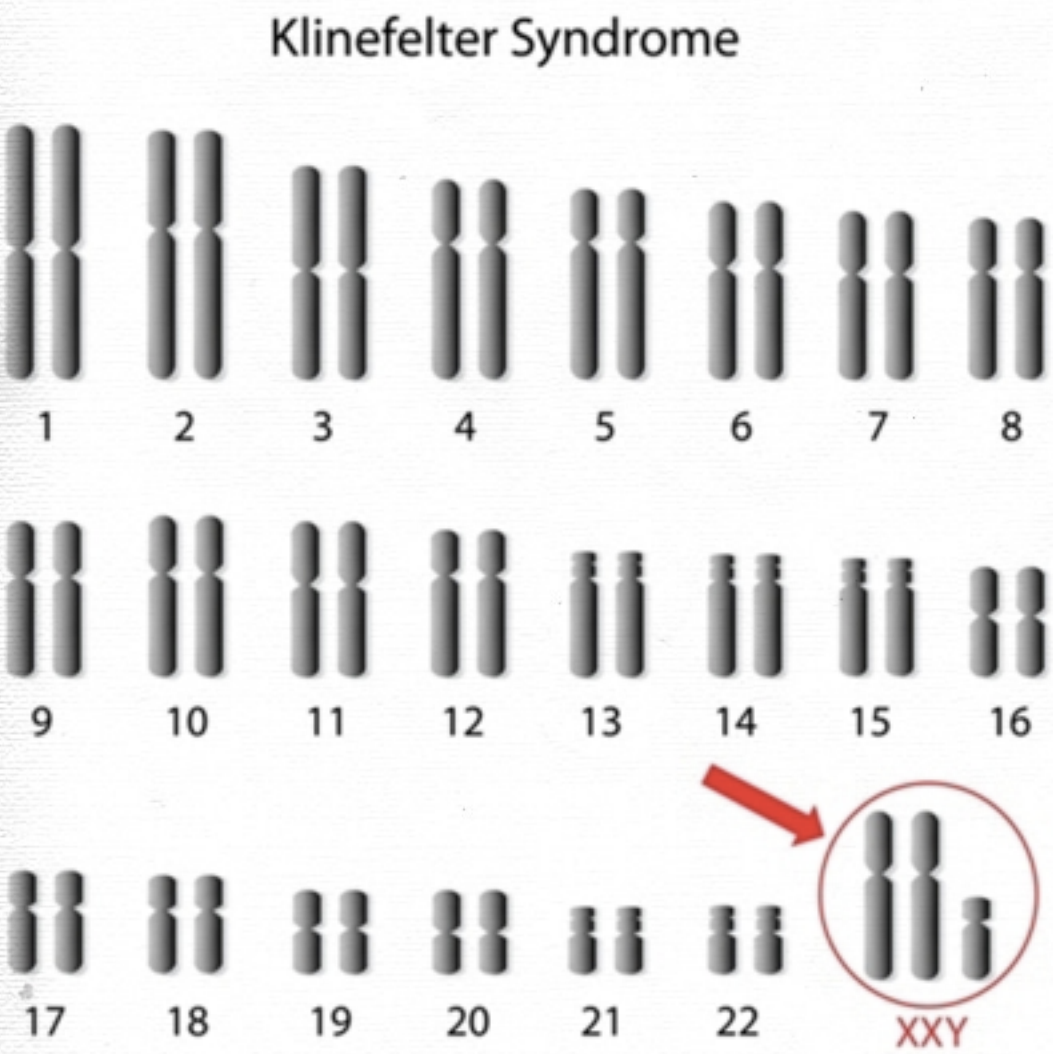
KLINEFELTER'S SYNDROME

KLINEFELTER'S SYNDROME IS AN EXAMPLE OF CHROMOSOMAL ANEUPLOIDY SINCE THE MALE HAS AN EXTRA X CHROMOSOME.

GENOTYPE CAN BE WRITTEN AS XXY

MALES BORN WITH KLINEFELTER SYNDROME MAY HAVE LOW TESTOSTERONE AND REDUCED MUSCLE MASS, FACIAL HAIR, AND BODY HAIR. MOST MALES WITH THIS CONDITION PRODUCE LITTLE OR NO SPERM.

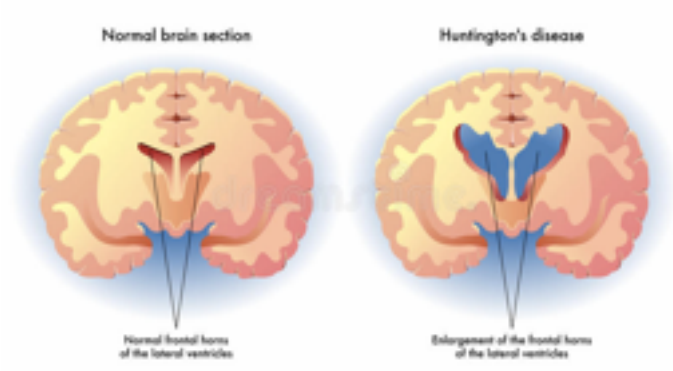
SINCE KLINEFELTER'S SYNDROME IS A CHROMOSOMAL ANEUPLOIDY IT DOES NOT IMPACT ONE GROUP OF PEOPLE MORE COMMON THAN OTHERS.

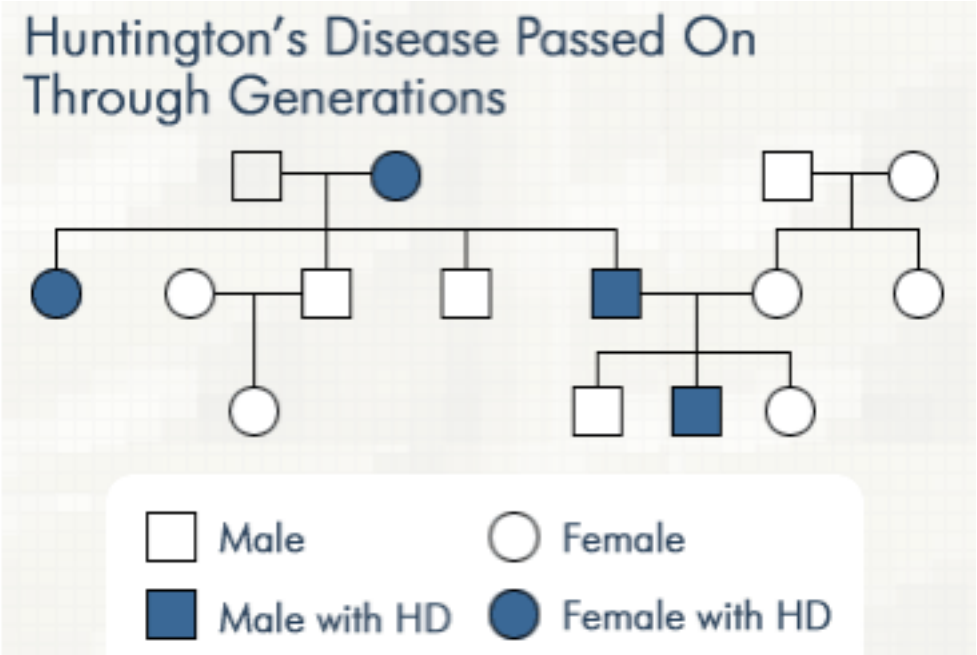


- A taller less muscular body than males there age.
- Broader hips and longer legs.
- Larger breast.
- Weaker bones.
- A lower energy level.
- Smaller penis and testicles
- Delay in puberty or go a parcel amount.
- Less facial and body hair following puberty.

HUNTINGTON'S DISEASE

HUNTINGTON'S DISEASES IS THE ONLY AUTOSOMAL DOMINANT GENETIC DISORDER DISCUSSED IN BIOLOGY ONE.





NOTE THAT THE PEDIGREE SHOWS HUNTINGTON'S DISEASE WITHIN EVERY GENERATION!

IMPACT ON BODY: AMNESIA, DELUSION, LACK OF CONCENTRATION, MEMORY LOSS, ABNORMALITY WALKING, INVOLUNTARY MUSCLE MOVEMENTS, IRRITABILITY, ANXIETY.

AREA OF PEOPLE: HUNTINGTON'S USUALLY IMPACTS PEOPLE IN THEIR 30'S OR 40'S

Cystic Fibrosis

CYSTIC FIBROSIS IS AN AUTOSOMAL RECESSIVE TRAIT.

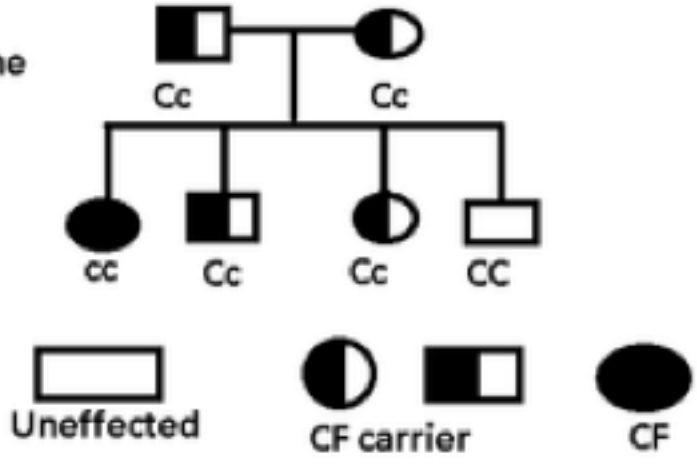
SYMPTOMS: COUGH, REPEATED LUNG INFECTIONS, INABILITY TO GAIN WEIGHT, FATTY STOOLS.

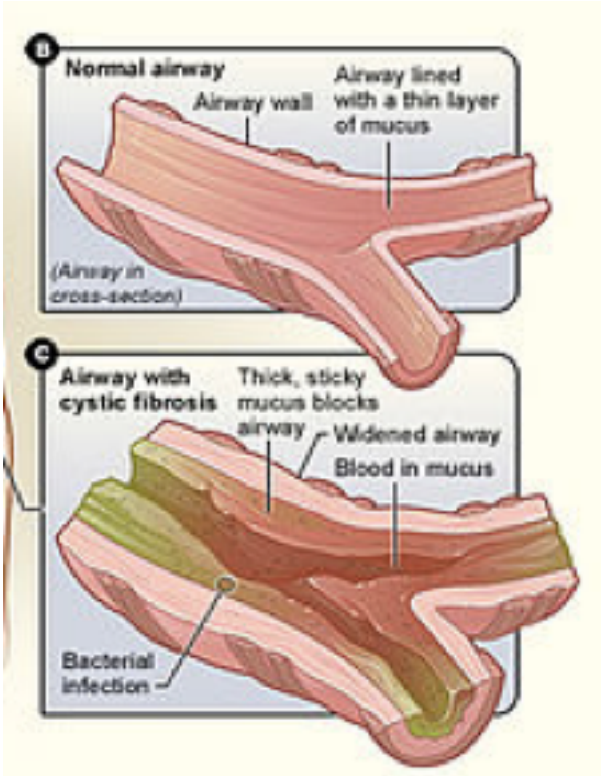
CYSTIC FIBROSIS AFFECTS THE CELLS THAT PRODUCE MUCUS, SWEAT, AND DIGESTIVE JUICES. IT CAUSES THESE FLUIDS TO BECOME STICK AND THICK WHICH PLUGS UP TUBES, DUCTS, AND PASSAGEWAYS.

CYSTIC FIBROSIS IS MOST COMMON WITHIN THE WHITE POPULATION IN THE UNITED STATES.

C = normal gene

c = CF gene





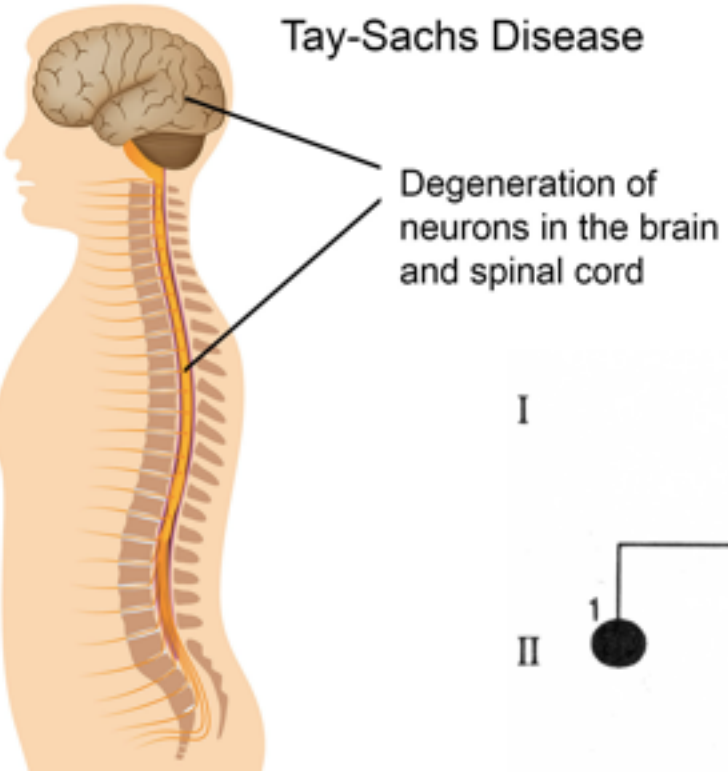
Tay Sachs

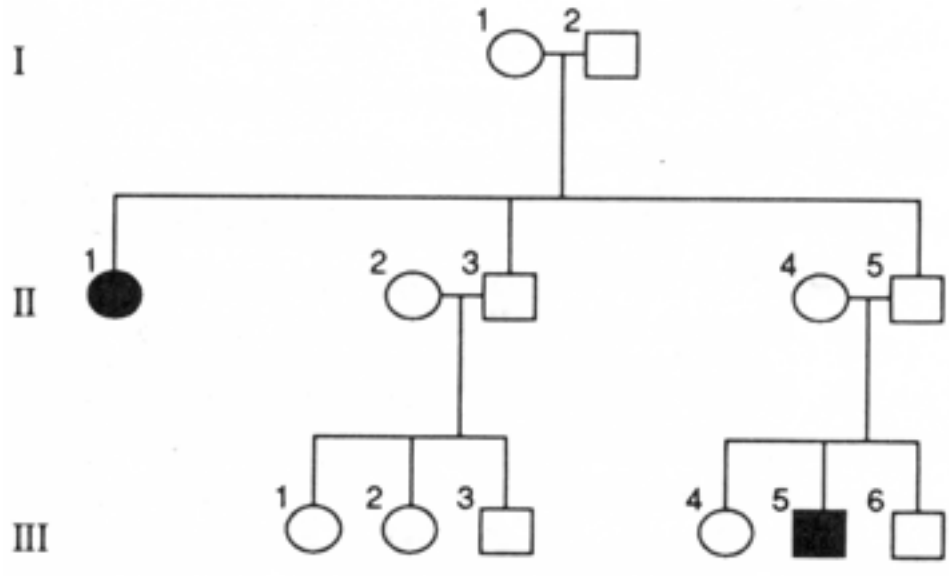
TAY SACHS IS AN AUTOSOMAL RECESSIVE TRAIT.

SYMPTOMS: SLOWED DEVELOPMENT AROUND SIX MONTHS OF AGE, USUALLY DEATH BY AGE FOUR. MUSCLE WEAKNESS, PROBLEMS WITH COORDINATION, LOSS OF BODY FUNCTIONS.

IN TAY-SACHS A FATTY SUBSTANCE IN THE BRAIN DESTROYS THE NERVE CELLS.

CYSTIC FIBROSIS IS MOST COMMON IN EASTERN EUROPEAN JEWS.





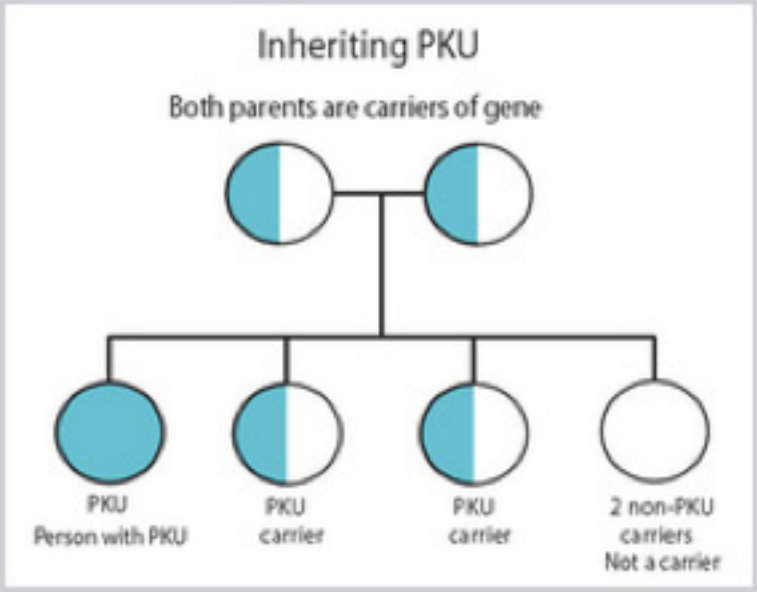
Phenylketonuria (PKU)

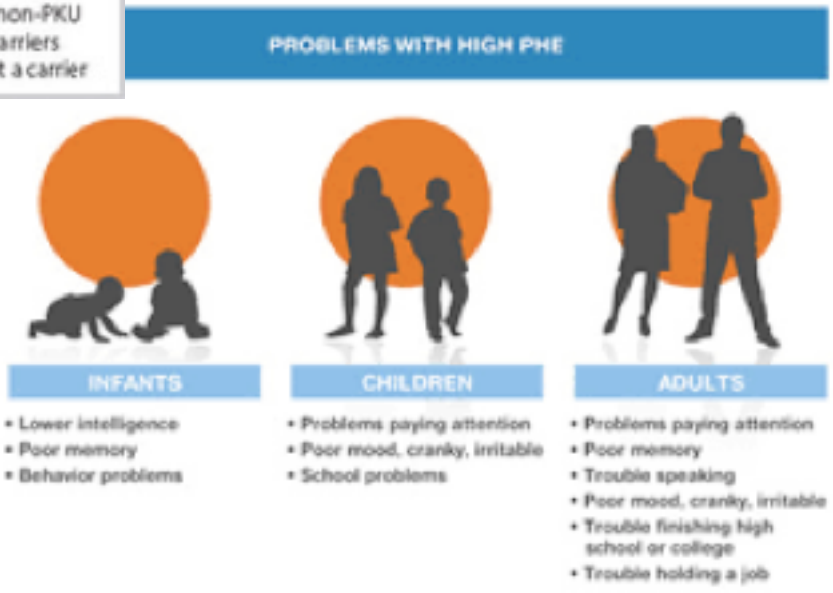
PHENYLKETONURIA IS AN AUTOSOMAL RECESSIVE TRAIT.

SYMPTOMS: BRAIN DAMAGE, INTELLECTUAL DISABILITIES, BEHAVIORAL SYMPTOMS, OR SEIZURES.

A BIRTH DEFECT THAT CAUSES AN AMINO ACID CALLED PHENYLALANINE TO BUILD UP IN THE BODY.

PKU IS MOST COMMON NATIVE AMERICAN AND NORTHERN EUROPEAN BACKGROUNDS.





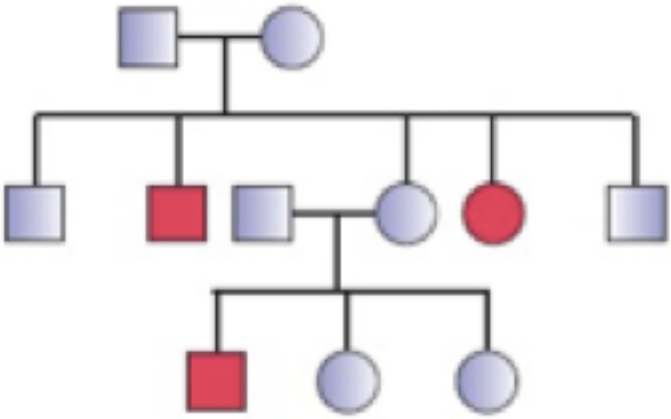
Sickle Cell Anemia

SICKLE CELL ANEMIA IS AN AUTOSOMAL RECESSIVE TRAIT.

SYMPTOMS: INFECTIONS, PAIN, FATIGUE.

RED BLOOD CELLS CONTORT INTO A SICKLE SHAPE (LIKE A CRESENT MOON). THIS CAUSES CELLS TO DIE EARLY LEAVING A SHORTAGE OF RED BLOOD CELLS. THESE CELLS CAN ALSO BLOCK BLOOD FLOW CAUSING PAIN.

SICKLE CELL ANEMIA IS MOST COMMON IN PEOPLE OF AFRICAN DESCENT.



Normal Red Blood Cell

Sickle Cell