

## Genetic Disorders

A genetic disorder is an abnormality in the \_\_\_\_\_. They can range for a deletion of a gene to the deletion of an entire chromosome. List the types of genetic disorders.

**Williams Syndrome**-A rare genetic disorder affecting \_\_\_\_\_, \_\_\_\_\_ and \_\_\_\_\_

Genetic material is missing from chromosome \_\_\_\_\_ including the gene for \_\_\_\_\_ which gives blood vessels the \_\_\_\_\_ and \_\_\_\_\_.

How many genes are affected by this deletion? \_\_\_\_\_

Do the parents have the disorder? Why?

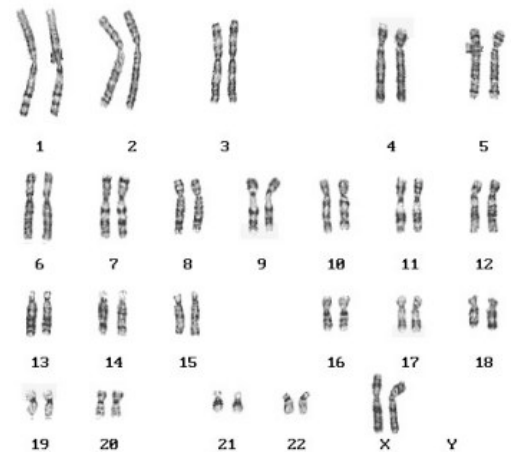
Symptoms include:

There is no cure but what should they avoid?

What can physical therapy do for them?

Non-disjunction-

What can a karyotype show us?

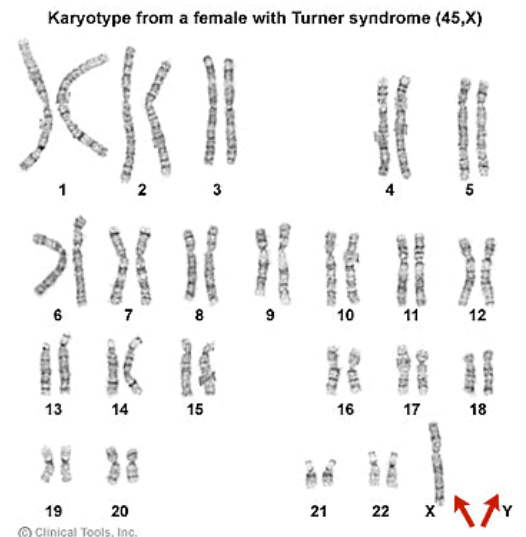


**Turner's Syndrome**- An incomplete or missing \_\_\_\_\_ chromosome affecting \_\_\_\_\_ and \_\_\_\_\_

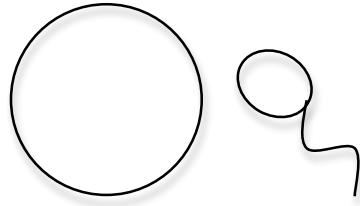
Girls with this disorder will be \_\_\_\_\_ than average and have abnormal \_\_\_\_\_

How is the karyotype different?

Why are they missing the X chromosome?



Either the sperm of the egg will be missing the X chromosome, leaving only 1 X



What are some physical symptoms of Turner's?

Why do the parents NOT have Turner's Syndrome?

What is the SHOX gene responsible for?

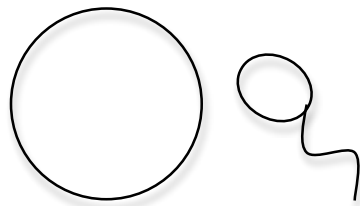
How can girls be treated for Turner's Syndrome?

What are the odds of being born with Turner's?

In most cases, what is responsible for the lack of the X-chromosome?

**Klinefelter's Syndrome**-Affects \_\_\_\_\_ giving them a total of \_\_\_\_\_ chromosomes ( XXY )

Either the sperm of the egg will have an extra X chromosome, leaving 2 x's and 1 Y



What is wrong with this karyotype?

What are some symptoms of Klinefelter's?

When do some people find out they have Klinefelter's Syndrome?

How can Klinefelter's be treated?

What are the odds?

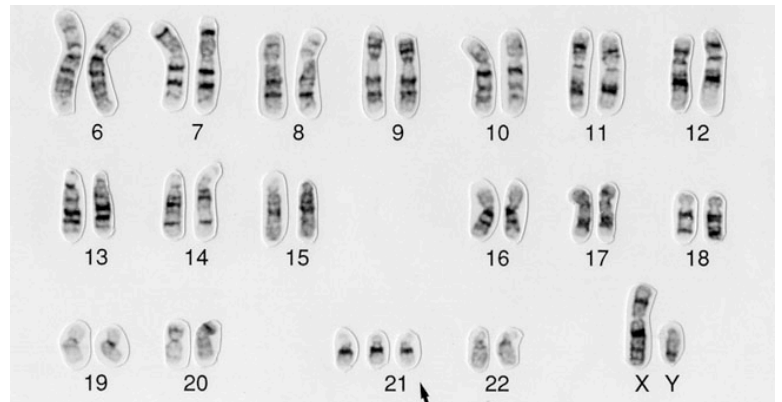
**Down's Syndrome**-Is a disorder caused by an extra copy of chromosome \_\_\_\_\_ sometimes called

\_\_\_\_\_

Why does this occur?

Will this be found in every cell? \_\_\_\_\_

What are some symptoms?



What are some other medical risks?

Can this be detected in the womb? YES or NO

Non-Invasive-

Invasive-

What are the odds of having a child with Down's Syndrome?

How can these odds increase?

Can this disorder be passed down? YES or NO

What does it mean to be an autosomal disorder?

What does it mean to be an autosomal recessive disorder?

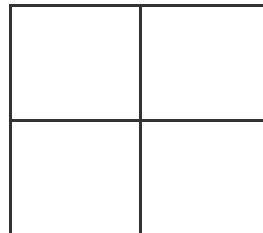
What does it mean to be a carrier?

What are the odds of passing down the disorder if one parent is a carrier and the other parent is normal?

\_\_\_\_\_%

G=Good Copy

g=Bad Copy

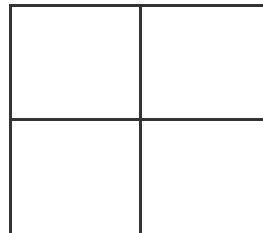


What are the odds of passing down the disorder if both parents are carriers?

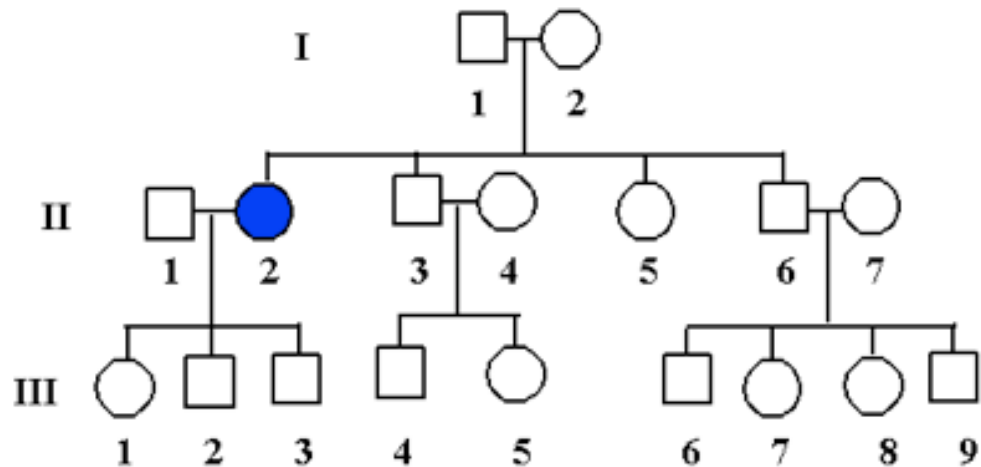
\_\_\_\_\_%

G=Good Copy

g=Bad Copy



Why are autosomal recessive disorders usually a total surprise?



**Cystic Fibrosis**-Mainly affects northern \_\_\_\_\_ and \_\_\_\_\_ cultures. The odds are about \_\_\_\_\_ births.

It is caused by an amino acid deletion at \_\_\_\_\_ on chromosome \_\_\_\_\_. This prevents the enzyme that breaks down \_\_\_\_\_ around the lungs from forming.

Explain how a person is affected by cystic fibrosis.

There is no cure but treatments include:

Life expectancy is usually in the mid \_\_\_\_\_ up to late \_\_\_\_\_.

**Tay Sach's**-Is a \_\_\_\_\_ disorder that causes a progressive degeneration of the \_\_\_\_\_. It is caused by a defect of the \_\_\_\_\_ gene on chromosome \_\_\_\_\_ resulting in a toxic accumulation of \_\_\_\_\_ in the \_\_\_\_\_ and \_\_\_\_\_. Life expectancy is about \_\_\_\_\_ years.

There is no cure but how can you help children with the disorder?

**Sickle Cell Anemia**-Is a disorder involved in the \_\_\_\_\_ and their ability to carry \_\_\_\_\_. The amino acid \_\_\_\_\_ replaces \_\_\_\_\_ and the hemoglobin protein does not fold properly.

What happens to the shape of the blood cell and why is that a problem?

How long do the sickle cells live compared to a normal blood cell?

How is the spleen affected?



What are the treatments?

There is not really a cure, but what is the best option?

What are the odds and who is mainly affected?

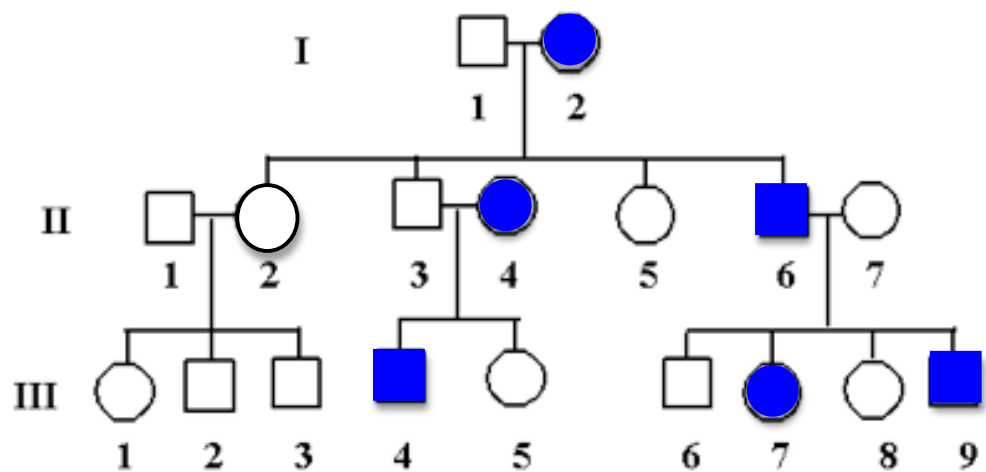
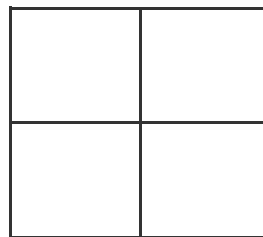
What is the life expectancy for both males and females?

What does it mean to be an autosomal dominant disorder?

If one parent is affected, what are the odds the offspring will have the disorder? \_\_\_\_\_%

G=Good Copy

g=Bad Copy



Are males or females more likely to get an autosomal dominant disorder?

**Huntington's Disease**-A genetic defect on chromosome \_\_\_\_ causing the \_\_\_\_\_ to repeat \_\_\_\_\_ - \_\_\_\_\_ instead of the normal \_\_\_\_\_-\_\_\_\_\_ times. Symptoms usually develop in the \_\_\_\_\_.

List at least three symptoms:

List at least three unusual movements:

List at least three things that happen when dementia sets in.

There is no cure but the goal is to make the person \_\_\_\_\_ as long as possible.

People with Huntington's usually die within \_\_\_\_\_ years after diagnosis. They often die from \_\_\_\_\_

X-linked disorders are associated with the \_\_\_\_ chromosome. If it is recessive and you are a female you are a carrier, which means you are \_\_\_\_ affected. If you are a male, you only have one copy of the \_\_\_\_-chromosome, so you will have the disorder. If the disorder is X-linked dominant it does not matter if you are a male or a female.

$X_g$ =No Problem or Disorder

$X_b$ =Disorder or Problem

Y = Male

DOMINANT


RECESSIVE


What are the odds if the mother is a carrier and the father is normal?

$X_g$ =No Problem or Disorder

$X_b$ =Disorder or Problem

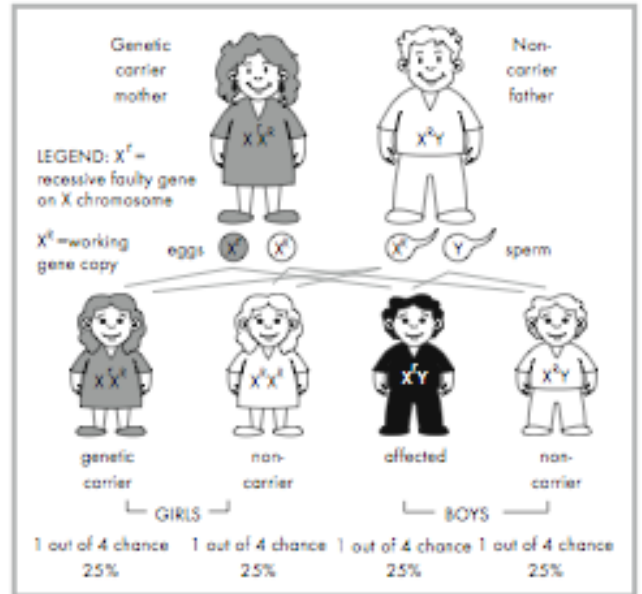
Y = Male

\_\_\_\_\_ % \_\_\_\_\_

\_\_\_\_\_ % \_\_\_\_\_

\_\_\_\_\_ % \_\_\_\_\_

\_\_\_\_\_ % \_\_\_\_\_

What are the odds if the mother is a carrier and the father is normal?

$X_g$ =No Problem or Disorder

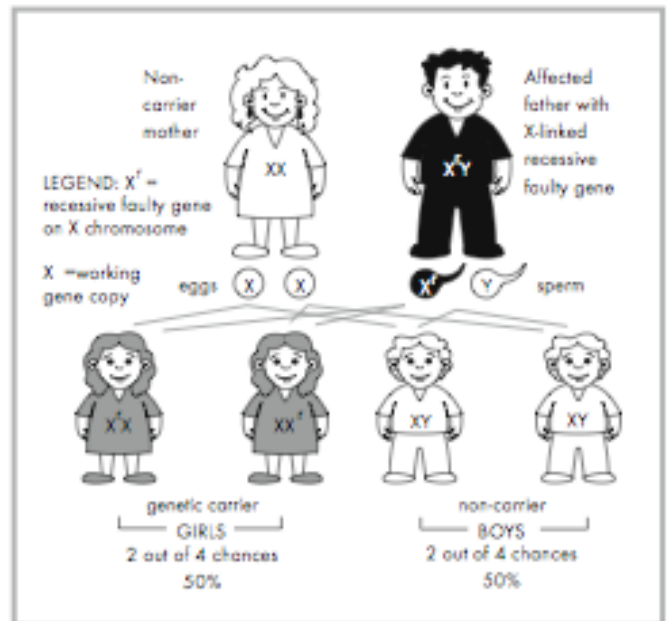
$X_b$ =Disorder or Problem

Y = Male

\_\_\_\_\_ % \_\_\_\_\_

\_\_\_\_\_ % \_\_\_\_\_

\*\*\*\*The girls are all \_\_\_\_\_



**Hemophilia**-One or more of the bloods \_\_\_\_\_factors is missing. What are some symptoms?

What is a possible treatment?

Is life pretty normal if you are careful?

What is the life expectancy?

**Duchenne Muscular Dystrophy**-Involves rapidly worsening muscle \_\_\_\_\_. Symptoms usually appear before age \_\_\_\_\_ and include:

The ability to walk may be lost by age \_\_\_\_\_ and are usually bound to a \_\_\_\_\_.

There is no cure but treatment aims to maximize \_\_\_\_\_. Death usually occurs by age \_\_\_\_\_, usually from \_\_\_\_\_disorders.

**X-Linked Dominant**-When the abnormal gene from one parent is capable of causing \_\_\_\_\_, even though the other parent is normal.

What are the odds if the mother is normal and the father is affected?

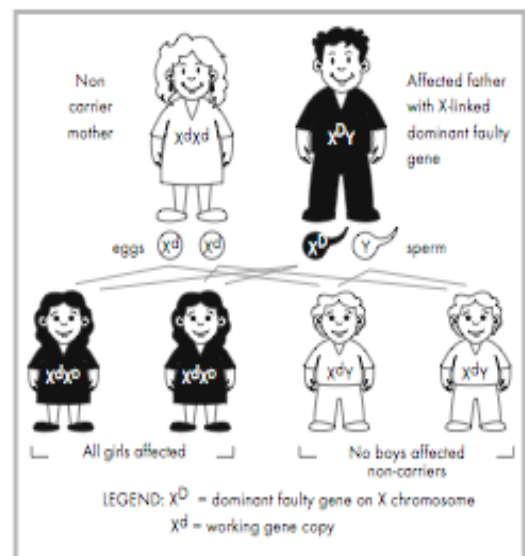
$X_g$ =No Problem or Disorder

$X_b$ =Disorder or Problem

$Y$  = Male

\_\_\_\_\_ % \_\_\_\_\_

\_\_\_\_\_ % \_\_\_\_\_

What are the odds if the mother is affected and the father is normal?

$X_g$  = No Problem or Disorder

$X_b$  = Disorder or Problem

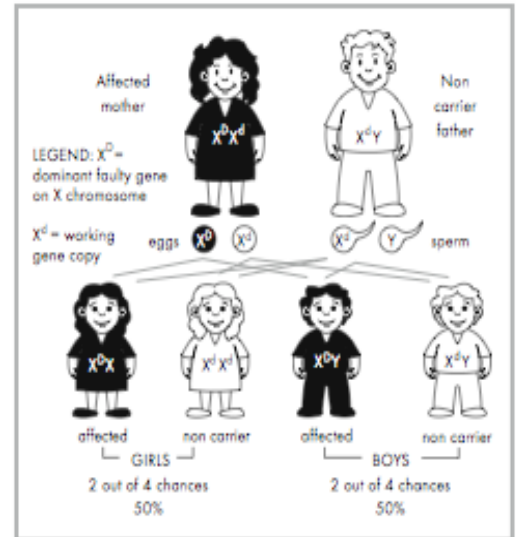
Y = Male

\_\_\_\_\_ % \_\_\_\_\_

\_\_\_\_\_ % \_\_\_\_\_

\_\_\_\_\_ % \_\_\_\_\_

\_\_\_\_\_ % \_\_\_\_\_




\*\*\*\*Half of the children will be affected not based on sex.

Rett Syndrome-It is a \_\_\_\_\_ disorder seen almost exclusively in \_\_\_\_\_.

Onset is between \_\_\_\_\_ months and life expectancy is about \_\_\_\_\_ years

List some symptoms: