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# Autosomal Recessive Genetic Disorder

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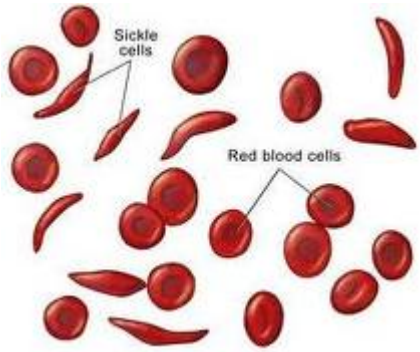
## What is autosomal recessive disorder?

An autosomal recessive disorder occurs when two copies of the gene must be mutated for a person. Even though an affected person has unaffected parents they may carry a single copy of the mutated gene. These types of parents are called as carriers. With this type of parents 25% chance with each pregnancy of affected child.

## Common Diseases

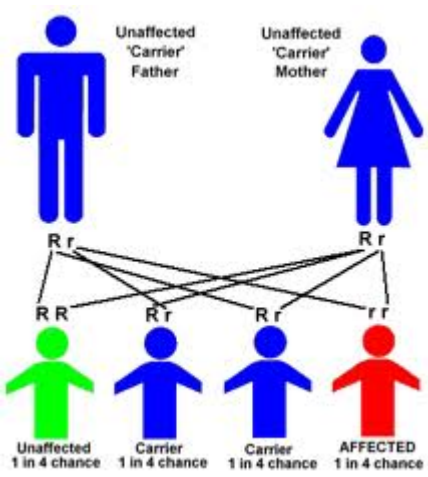


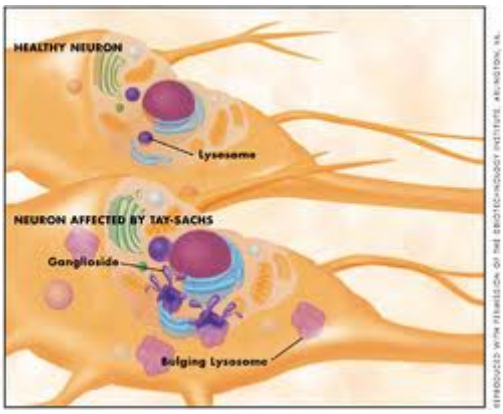

**Sickle Cell Disease**

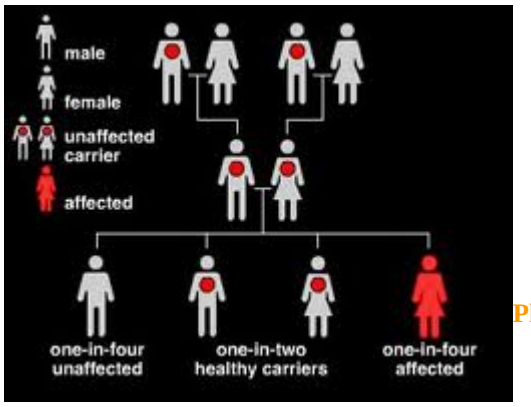
- Sickle cell disease is a blood disorder.
- Also known as sickle cell anemia.
- Inherited through autosomal recessive pattern.
- Cause damage to organs like lungs, kidney, spleen and brain.
- Reduced  life expectancy to 42 years for males and 48 years for females.

**Tay - Sachs disease**

- An autosomal recessive genetic disorder
- Destroys nerve cells in brain and the spinal cord.
- Neurological and physical disabilities are common.
- Present in infants of age of 3 to 6 months.
- Symptoms are losing motor skills mental disability, vision and hearing loss, occurrence of seizures and paralysis.
- No cure for the disease.

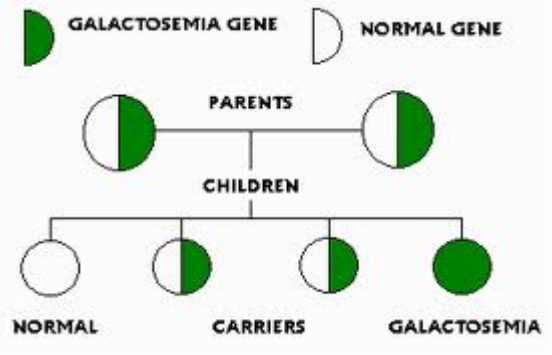



**Phenylketonuria(PKU)**

- PKU is an autosomal recessive disorder
- Causes deficiency in the enzyme phenylalanine hydroxylase
- Excess phenylketone can affect the development of the brain.
- Symptoms are mental retardation, seizures or brain damage.




**Galactosemia**

- Rare metabolic genetic disorder in human
- Galactosemia impairs body's ability to break down galactose.
- Symptoms are yellowing of skin, eyes, diarrhea, vomiting, and refusal to drink milk, malnourishment and also mental retardation.
- If milk or milk products are given to infants suffering from galactosemia, then accumulation of galactose in their system damages brain, eyes, liver and kidneys.


**Cystic Fibrosis**

- Cystic fibrosis is an inherited disease of the glands that secrete mucus and sweat.
- Lungs, liver, pancreas, sinuses, intestines and sex organs will be affected.
- Causes excess loss of salts through sweat
- Symptoms are dehydration, tiredness, weakness and elevated heart rate.
- No cure for cystic fibrosis.

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