

Autosomal Recessive Genetic Disorder

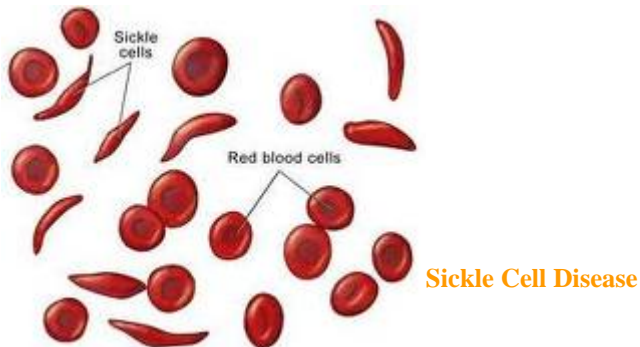
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What is autosomal recessive disorder?



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.

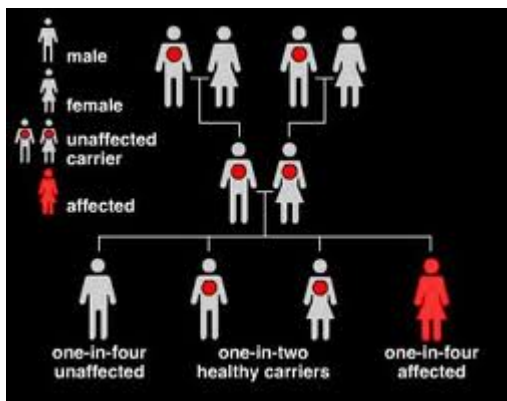
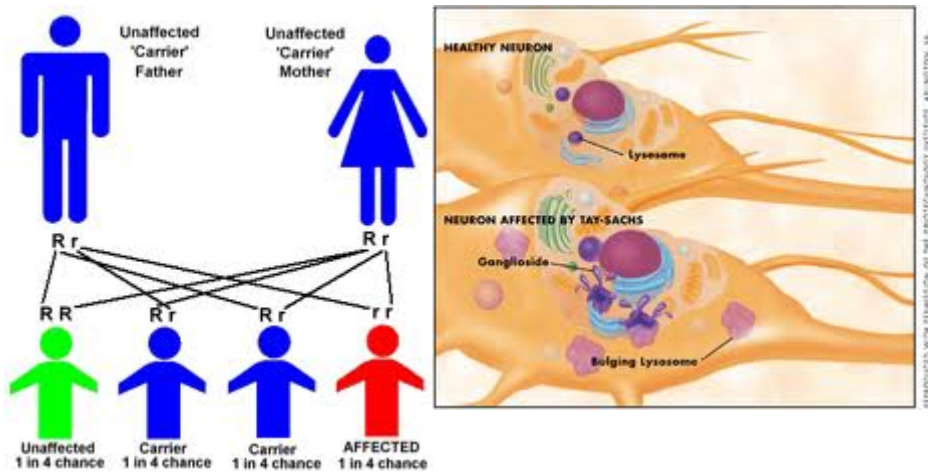
Common Diseases



- Sickle cell disease 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.

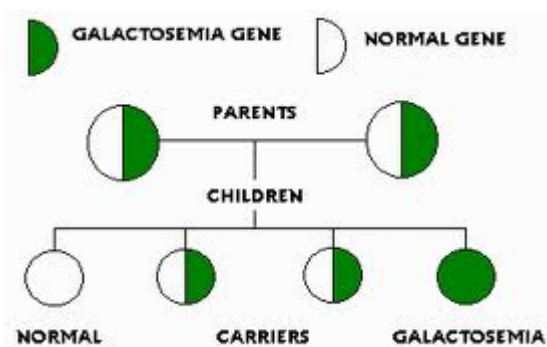
Tay - Sachs disease

- 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)

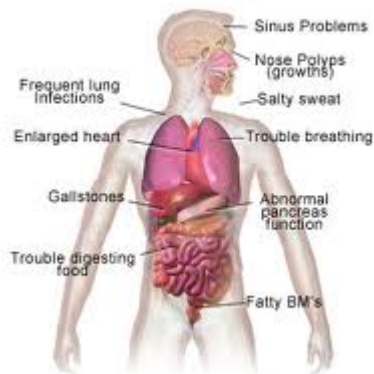
- PKU is an autosomal recessive disorder
- Causes deficiency in the enzyme phenylalanine hydroxylase
- Excess phenylketone can affect the development of the brain.
- Symptoms are mental retardation, seizures or brain damage.



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.

Health Problems with Cystic Fibrosis



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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