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Genetic Disorder

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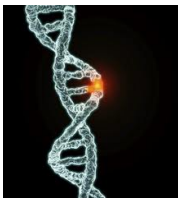
Gene Abnormalities

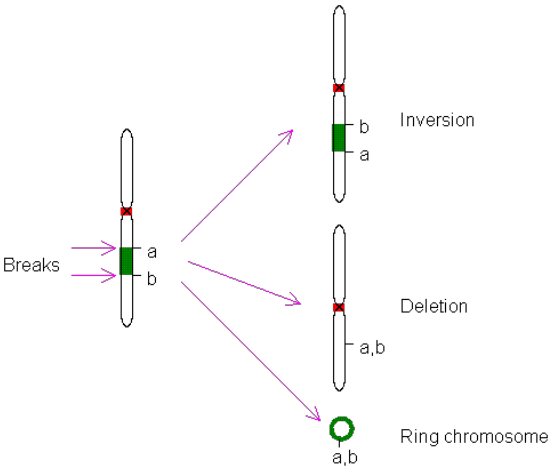


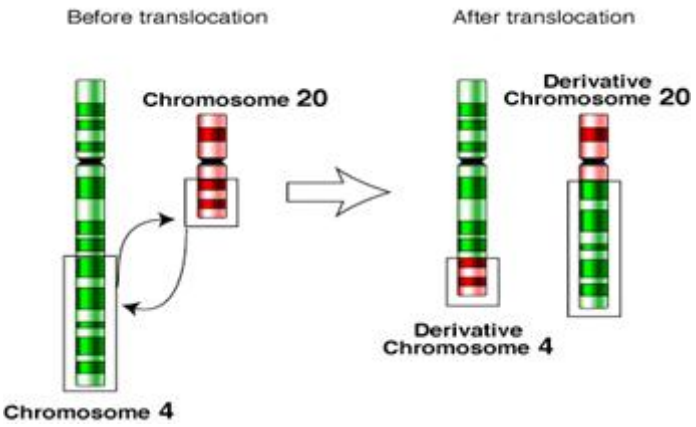
Abnormality of a gene leads to Genetic disorder. Even though cancer like disease is caused due to gene mutation, sometimes they can be influenced by environmental factors. Many disorders are uncommon and occurs in 1/1,00,000 persons.

Causes of Genetic Disorders in Humans

Genetic diseases in humans are caused due to abnormalities in genes or chromosomes. Such defects can be caused by the following mechanisms:

- **Mutations:** Sudden inheritable changes in the nucleotide sequence of a gene.
- **Aneuploidy:** Additional(Monosomy) or less Tetraploidy) number of chromosomes in an organism.
- **Deletions:** Loss of a part of chromosome.
- **Duplications:** Duplication of a portion of chromosome that results in extra amount of genetic material.
- **Inversions:** The nucleotide sequence got inverted and reattached at the original location of the chromosome.
- **Translocations:** Portion of chromosome has got transferred on to some other chromosome.





Types of Genetic Disorders in Humans

- **Autosomal Dominant Genetic Disorders:** Occurs by the inherited the defective gene from a single parent.
- **Autosomal Recessive Genetic Disorders:** Occurs only when an individual has got two defective alleles of the same gene, one from each parent.
- **Sex-Linked Disorders:** Occurs when sex chromosomes or genes get defected.
- **Multi-factorial Genetic Disorders:** Caused because of the genetic as well as environmental factors.

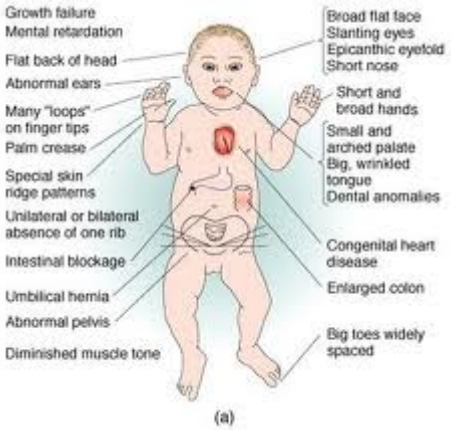
Common Genetic Disorders in Humans

Albinism

- A congenital disorder.
- Symptoms are little or completely no production of melanin in hair, skin and iris of the eyes.
- Caused due to inheritance of recessive alleles from parents.
- Can't be cured.

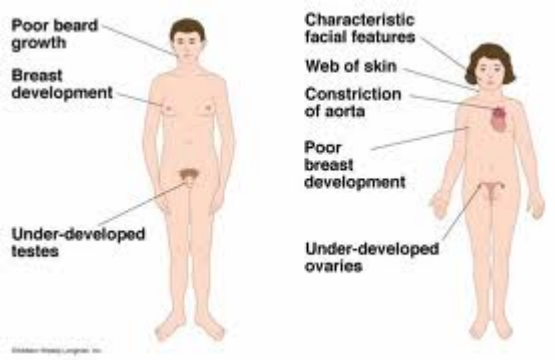
Alzheimer's Disease

- Alzheimer's disease is the most common form of dementia.
- Symptoms are memory loss, irritability, mood swings, confusion and language breakdown.
- Occurs because of the production of a small protein fragment called ABeta (A?).
- A degenerative disease.
- Can’t be cured
- Balanced diet, mental exercises and stimulation can prevent this disease.



Down Syndrome

- Trisomy 21 Caused because of extra copy of chromosome 21, that a child inherits from his/her parent.
- Symptoms are delays in mental as well as physical development of a child.



Klinefelter Syndrome

- XXY or XXY syndrome
- Sex linked genetic disorder
- Males have an extra X chromosome
- Symptoms are infertility,impaired physical, language and social developments.

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