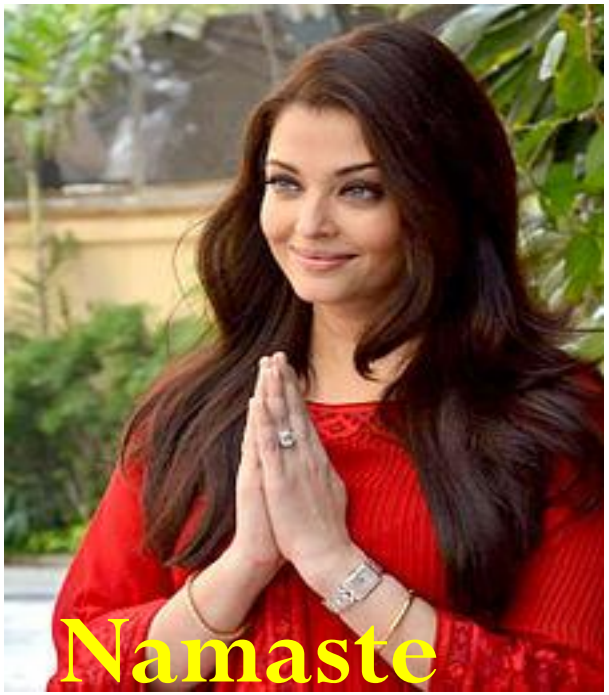
Genetic Disorder



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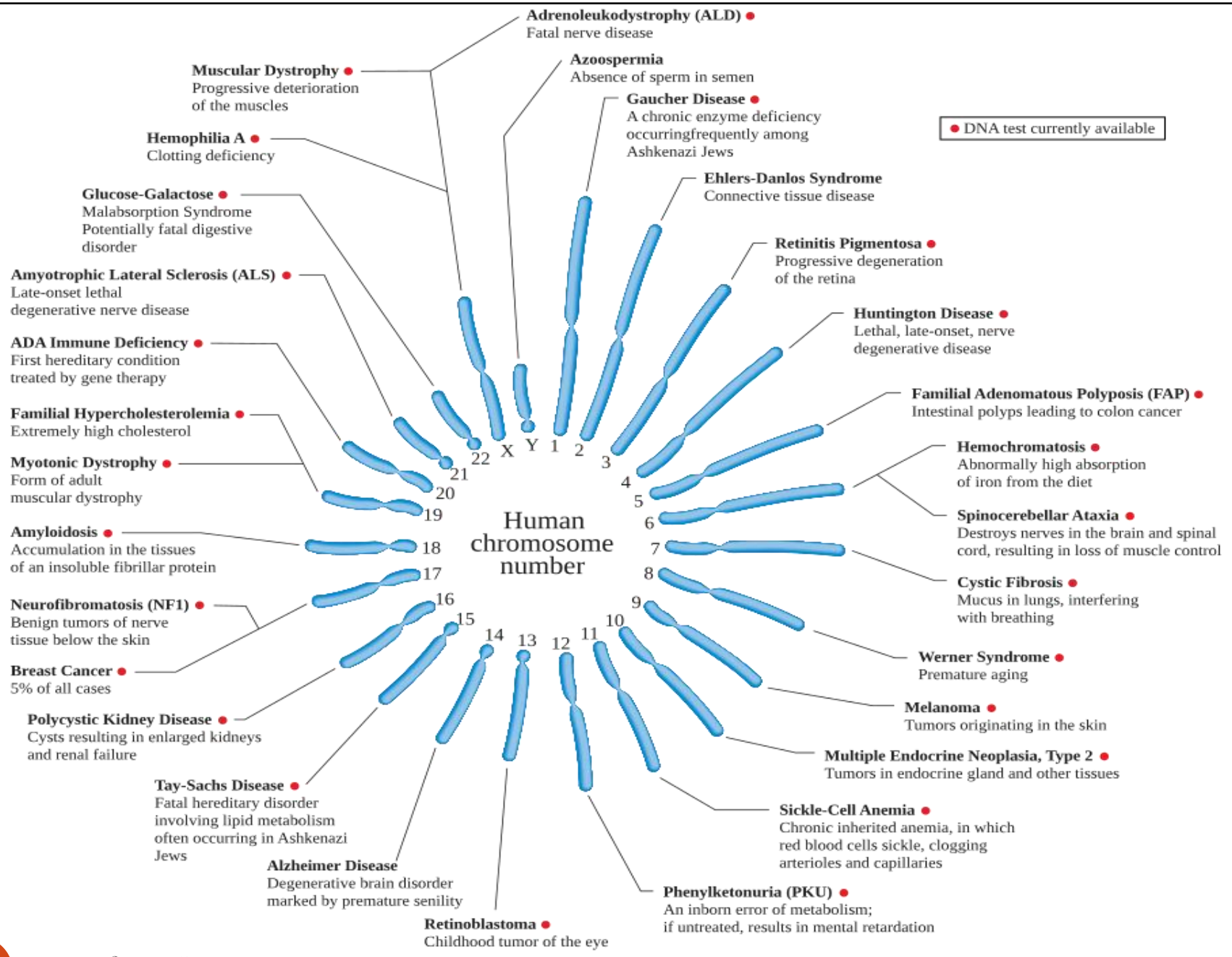
Genetic Disorder

A **genetic disorder** is a health problem caused by one or more **abnormalities** in the genome. It can be caused by a mutation in a single **gene** (monogenic)or multiple **genes** (polygenic) or by a chromosomal abnormality


Adrenoleukodystrophy (ALD)
Fatal nerve disease
Muscular Dystrophy
Progressive deterioration of the muscles
Azoospermia
Absence of sperm in semen
Gaucher Disease
A chronic enzyme deficiency occurringfrequently among Ashkenazi Jews
Hemophilia A
Clotting deficiency
Ehlers-Danlos Syndrome
Connective tissue disease
Glucose-Galactose
Malabsorption Syndrome
Potentially fatal digestive disorder
Retinitis Pigmentosa
Progressive degeneration of the retina
Amyotrophic Lateral Sclerosis (ALS)
Late-onset lethal degenerative nerve disease
Huntington Disease
Lethal, late-onset, nerve degenerative disease
ADA Immune Deficiency
First hereditary condition treated by gene therapy
Familial Adenomatous Polyposis (FAP)
Intestinal polyps leading to colon cancer
Familial Hypercholesterolemia
Extremely high cholesterol
Hemochromatosis
Abnormally high absorption of iron from the diet
Myotonic Dystrophy
Form of adult muscular dystrophy
Spinocerebellar Ataxia
Destroys nerves in the brain and spinal cord, resulting in loss of muscle control
Amyloidosis
Accumulation in the tissues of an insoluble fibrillar protein
Cystic Fibrosis
Mucus in lungs, interfering with breathing
Neurofibromatosis (NF1)
Benign tumors of nerve tissue below the skin
Werner Syndrome
Premature aging
Breast Cancer
5% of all cases
Melanoma
Tumors originating in the skin
Polycystic Kidney Disease
Cysts resulting in enlarged kidneys and renal failure
Multiple Endocrine Neoplasia, Type 2
Tumors in endocrine gland and other tissues
Tay-Sachs Disease
Fatal hereditary disorder involving lipid metabolism often occurring in Ashkenazi Jews
Sickle-Cell Anemia
Chronic inherited anemia, in which red blood cells sickle, clogging arterioles and capillaries
Alzheimer Disease
Degenerative brain disorder marked by premature senility
Phenylketonuria (PKU)
An inborn error of metabolism; if untreated, results in mental retardation
Retinoblastoma
Childhood tumor of the eye
DNA test currently available
Human chromosome number
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y

There are three types of genetic disorders:

- Single-**gene disorders**, where a mutation affects one **gene**. Sickle cell anemia is an example.
- Chromosomal **disorders**, where chromosomes (or parts of chromosomes) are missing or changed. ...
- Complex **disorders**, where there are mutations in two or more **genes**.

Most common types

- Down syndrome

genetic chromosome 21 disorder causing developmental and intellectual delays.

- Cystic fibrosis

An inherited life-threatening disorder that damages the lungs and digestive system.

- Huntington's disease

An inherited condition in which nerve cells in the brain break down over time.

- Duchenne muscular dystrophy

An inherited disorder of progressive muscular weakness, typically in boys.

- Sickle cell anemia

A group of disorders that cause red blood cells to become misshapen and break down.

- Haemophilia

A disorder in which blood doesn't clot normally.

- Thalassemia

A blood disorder involving lower-than-normal amounts of an oxygen-carrying protein.

- Fragile X syndrome

A genetic condition causing intellectual disability

Treatment of genetic disorder

In general, quality of life is the perceived quality of an individual's daily life, that is, an assessment of their well-being or lack thereof. This includes all emotional, social and physical aspects of the individual's life. In health care, health-related quality of life (HRQoL) is an assessment of how the individual's well-being may be affected over time by a disease, disability or disorder.

Gene therapy is a medical field which focuses on the utilization of the therapeutic delivery of nucleic acid into a patient's cells as a drug to treat disease. The first attempt at modifying human DNA was performed in 1980 by Martin Cline,

Gene therapy may be classified into two types:

1.Somatic

2.germline

Somatic

In somatic cell gene therapy (SCGT), the therapeutic genes are transferred into any cell other than a gamete, germcell, gametocyte, or undifferentiated stem cell. Any such modifications affect the individual patient only, and are not inherited by offspring. Somatic gene therapy represents mainstream basic and clinical research, in which therapeutic DNA (either integrated in the genome or as an external episome or plasmid) is used to treat disease.Over 600 clinical trials utilizing SCGT are underway in the US. Most focus on severe genetic disorders,including immunodeficiencies, haemophilia, thalassaemia, and cystic fibrosis. Such single gene disorders are good candidates for somatic cell therapy. The complete correction of a genetic disorder or the replacement of multiple genes is not yet possible. Only a few of the trials are in the advanced stages.

Germline

In germline gene therapy (GGT), germ cells (sperm or egg cells) are modified by the introduction of functional genes into their genomes. Modifying a germ cell causes all the organism's cells to contain the modified gene. The change is therefore heritable and passed on to later generations. Australia, Canada, Germany,Israel, Switzerland, and the Netherlands prohibit GGT for application in human beings, for technical and ethical reasons, including insufficient knowledge about possible risks to future generations and higher risks versus SCGT. The US has no federal controls specifically addressing human genetic modification (beyond FDA regulations for therapies in general).

Ref.- From Wikipedia, the free encyclopedia

Down syndrome

A genetic chromosome 21 disorder causing developmental and intellectual delays

A boy with Down syndrome, one of the most common genetic disorders