Overview Of The Most Serious Genetic Disorders

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Sickle Cell Anemia, Pearson syndrome, Duchene Muscular Dystrophy, lactose intolerance, Familial Breast Cancer, polycystic disease, and Huntington's multiple repeats are some of the most serious genetic disorders. This paper gives an overview of the profile of each disease.

1. Sickle Cell Anemia
2. Name of Disease: Sickle Cell Anemia SNP based.
3. Nomenclature

- *Phenotype MIM number:* 603903
- *Gene locus MIM number:* 141900
- *Gene Symbol:* HBB
- The normal function of a gene: It encodes for the formation of beta-globin subunits. (National Library of Medicine, n.d.)

Brief Description:

It is a disease of the red blood cells resulting from gene mutation on the HBB gene. The HBB gene is found in chromosome 11 and encodes for beta-globin. The mutation results in the formation of HbS and incapacitates the oxygen-carrying capacity of the red blood cells. The disorder causes varied degrees of anaemia.

Chromosome location and gene coordinates: HBB gene on Chromosome 11

Figure 1: Chromosome location for Sickle cell anaemia

Inheritance Pattern:

The disease is a recessive trait. Phenotypic expression of the disease occurs when a person inherits two recessive genes from both parents so that they are homozygotes.

Genetic Basis:

Gene mutation causes the formation of sickle-shaped cells with reduced capacity to transport oxygen.

Interesting Fact:

The disease was discovered in 1910, but that does not imply that it had never existed before. It has been in existence for thousands of years. The nature of the disease became clear in the 1950s.

1. Pearson Syndrome
2. Name of Disease: Pearson Syndrome.
3. Nomenclature

- *Phenotype MIM number:* 612623
- *Gene locus MIM number:* 133170
- *Gene Symbol:* EPO
- *Normal function of gene:* hematopoiesis and normal functioning of mitochondria.

Brief Description:

The disorder affects multiple organs and systems in the body, especially the pancreas and bone marrow. This interferes with hematopoietic stem cells, hence the defective formation of blood cells. It also affects mitochondrial DNA, making respiration difficult.

Chromosome location and gene coordinates: 7q22.1

Figure 2: Chromosome affected by Pearson syndrome

Inheritance Pattern:

The mutations that cause Pearson syndrome occur spontaneously in the offspring.

Genetic Basis:

Multiple gene mutations cause defective erythropoiesis, faulty mitochondrial enzyme formation and defective pancreas function.

Interesting Fact:

Howard Pearson was the first person to describe the syndrome in 1979. Hence, the condition was named after him. Pearson was a pediatric oncologist and haematologist.

1. Duchenne Muscular Dystrophy
2. Name of Disease: Duchenne Muscular Dystrophy.
3. Nomenclature

- *Phenotype MIM number:* 310200
- *Gene locus MIM number:* 300377
- *Gene Symbol:* DMA
- The normal function of a gene encodes for the formation of dystrophin protein that helps in muscle contraction and nerve function. The protein works together with other proteins to strengthen muscle fibres.

Brief Description:

The disorder causes progressive weakness and degeneration of muscles due to a lack of dystrophin protein.

Chromosome location and gene coordinates: Xp21.1.

Figure 3: Chromosome location for Duchene Muscular Dystrophy.

Inheritance Pattern:

X-linked inheritance pattern

Genetic Basis:

The condition is linked to the X chromosome and is inherited alongside it. Since males have only one X chromosome, the condition is more prevalent in males than females.

Interesting Fact:

The disease was first reported in 1836 by Gioja and Conte, who identified it in brothers who had progressive muscle degeneration from age 10.

1. Lactose Intolerance.
2. Name of Disease: lactose Intolerance.
3. Nomenclature

- *Phenotype MIM number:* 223100
- *Gene locus MIM number:* 601806
- *Gene Symbol:* LCT
- The normal function of a gene: The LCT gene encodes for the formation of lactase enzyme, which is responsible for the digestion of lactose.

Brief Description:

Mutation of the LCT gene results in the formation of an abnormally short lactase enzyme that is incapable of carrying out the normal functions of the enzyme.

Chromosome location and gene coordinates: 2q21.3 located in MCM6.

Figure 4: Chromosome location for LCT gene

Inheritance Pattern:

Autosomal Dominant pattern.

Genetic Basis:

It is a recessive trait which is expressed only by homozygotes with cytosine residues on the alleles adjacent to the lactase gene.

Interesting Fact:

More than 75% of the global population suffers from lactose intolerance and cannot consume milk and milk products in adulthood.

1. Lactose Intolerance.
2. Name of Disease: Familial breast cancer.
3. Nomenclature

- *Phenotype MIM number:* 114480
- *Gene locus MIM number:* 600185 for BCRA2 and 603615 for RAD54L

- *Gene Symbol:* BRCA2
- The normal function of a gene: The BRCA2 gene regulates DNA repair of damaged DNA by producing the BRCA2 protein. (National Human Genome Research Institute, n.d.)

Brief Description:

BCRA2 is responsible for up to 10% of breast cancers. Defects in this gene are inherited along the familial line.

Chromosome location and gene coordinates: 17q21.31

Figure 5: Chromosome location for BRCA2 gene

Inheritance Pattern:

Autosomal Dominant pattern.

Genetic Basis:

Mutation on gene locus for BRCA1.

Interesting Fact:

A family history of breast cancer has been noticed in more than 21% of people with breast cancer.

1. Polycystic Kidney Disease.
2. Name of Disease: Polycystic Kidney Disease.
3. Nomenclature

- *Phenotype MIM number:* 173900
- *Gene locus MIM number:* 601313
- *Gene Symbol:* PKD1
- *The normal function of a gene* PKD1 encodes for polycystin-1, which is very active during prenatal life.

Brief Description:

In polycystic kidney disease, numerous fluid-filled cysts grow in the kidney. These cysts may eventually damage the kidneys and orchestrate kidney failure.

Chromosome location and gene coordinates:

16p13.3

Inheritance Pattern:

Autosomal Dominant pattern.

Genetic Basis:

Mutation in PKD1 gene.

Interesting Fact:

Most end-stage kidney diseases eventuate from PKD. The disease was first identified in 1586 by Jan Zigulist.

1. Huntington's Multiple Repeats.
2. Name of Disease: Huntington's multiple repeats.
3. Nomenclature

- *Phenotype MIM number:* 143100
- *Gene locus MIM number:* 613004
- *Gene Symbol:* HTT
- The normal function of a gene: The HTT gene encodes for the protein huntingtin. The protein ensures that brain neurons develop normally during prenatal life. (Clark et al., 2018)

Brief Description:

The disease makes brain neurons degenerate progressively, with symptoms beginning to develop at around 40 years of age.

Chromosome location and gene coordinates:

4p16.3

Inheritance Pattern:

Autosomal Dominant pattern.

Genetic Basis:

HTT gene causes a repeat of the CAG trinucleotide. In normal people, the segment repeats only about 35 times, while in people with Huntington's disease, the HTT gene mutates, causing the segment to repeat up to 120 times.

Interesting Fact:

The disease mainly affects the basal ganglia of the brain. Dr Huntington George was the first to describe the disease, and more than 80% of people with the condition have chorea as well. Hence, the name Huntington's chorea.

References

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