Types of gene mutations

Gene– A gene is a special strand of DNA that contains information about everything in our body. The genes make proteins to construct things in our body. Humans have about 25, 000 genes.

Homeotic Genes

Description: A homeotic gene is any collection of genes that manage the development of early embryonic stage of organisms, these genes produce proteins that tell cells to create different parts of the body

Mutation: If a mutation occurs in the homeotic gene an organism will not develop properly and may have displaced body parts, for example, a fly may grow a leg on its head instead of an antenna

Leptin Receptor

Description: The leptin receptor protein is produced by the LEPR gene, it manages energy balance and body weight. The leptin receptor protein is found on the exterior of cells in many organs and tissues of the body. The leptin receptor is activated by a hormone called leptin that connects to the receptor. Usually, the body’s fat cells release leptin in accordance to their size. As fat cells enlarge, they produce more leptin. This increase in leptin indicates that fat stores are increasing. The binding of leptin to its receptor send a series of chemical signals to the hypothalamus and give a sense of fullness.

Mutation: If a mutation in the leptin receptor occurs it may lead to excessive hunger, obesity, and reduced production of sexual development hormones

Retinoblastoma

Description: The RB1 gene provides instructions for creating a protein called pRB. This protein regulates cell growth and keeps cells from irregular division. Under certain conditions, pRB stops other proteins from DNA replication. Since DNA replication must happen before a cell can divide, regulation of this process helps prevent tumor growth. pRB also interacts with other proteins to control cell survival, cell self-destruction, and the process by which cells grow to carry out special tasks.

Mutation: If a mutation occurs in the RB1 gene a person may develop bladder cancer, retinoblastoma (an eye cancer that develops in the retina), lung cancer, breast cancer, osteosarcoma (a type of bone cancer), and melanoma (a type of skin cancer).

Insulin

Description: The INS gene provides commands for the production of the hormone insulin, insulin regulates glucose levels in the blood. Insulin is produced by the pancreas. Glucose is the primary source of energy for most cells in the body. Insulin is produced in a precursor form called proinsulin, which contains a single sequence of amino acids. The proinsulin sequence is cut to make individual pieces called the A and B chains, which are connected together by connections called disulfide bonds to form insulin.

Mutation: Mutations in the INS gene have been linked to permanent neonatal diabetes mellitus. Babies with this disorder have a low birth weight and develop increased blood sugar levels within the first 6 months of life. An INS gene mutation can also cause type 1 diabetes and other disorders that involve insulin production and blood sugar control.

Red Hair Colour

Description: The MC1R gene provides instructions for making a protein called the melanocortin 1 receptor. The receptor is found on the surface of melanocytes (specialized cells that create melanin). Melanin provides colour to skin, hair, eyes and the retina. Melanocytes produce two different types of melanin, eumelanin and pheomelanin. The amounts of these two pigments help decide the color of a person's hair and skin. The more eumelanin a person has the darker their hair and skin is. People with more eumelanin have increased protection from damage caused by UV radiation. People with more pheomelanin usually have red or blond hair, freckles, and lighter skin that is prone to skin damage caused by UV radiation.

Mutation: If a mutation occurs in this gene people develop oculocutaneous albinism type 2. People who have this mutation have light-colored hair and eyes, pale white skin, and vision conditions. A mutation in this gene can also increase the risk of developing skin cancer.

Boy in the Bubble Disease

Description: The Boy in the Bubble Disease also known as SCID, is Severe Combined Immunodeficiency. Children that develop this disorder do not have a working immune system. Children affected by SCID can also become sick from viruses present in some vaccines. These vaccines (such as Measles, Polio etc.) don't harm children with a healthy immune system. However, children with SCID may develop severe, life-threatening infections from the vaccines. There are various forms of SCID. The most common type is linked to the X-chromosome, making this disorder exclusive to males.

Symptoms: Babies with this disorder generally have infections occurring within the first few months of life, these infections are dangerous and may even be life-threatening, they may include pneumonia, meningitis etc. Other symptoms include failure to gain weight or grow normally, continuous mouth or throat infections, and a family history of immunodeficiency or infant deaths due to infections.

Diagnosis: Early diagnosis of this disorder is usually uncommon since the disease is extremely rare. The average age babies are diagnosed with SCID is about six months, generally because of reoccurring infections and improper development. If the mutation leading to SCID in a family is known, a test can happen through sequencing DNA from the fetus. Since SCID is so rare prenatal testing of a baby with no family history of the disorder is not usually done since the test is so expensive.

Prognosis: Without treatment most babies die within the first year of life.

Treatment Options: The most effective treatment for SCID is a bone marrow stem cell transplant. A bone marrow transplant from a tissue-matched sibling offers the greatest chance for curing SCID. However, most patients do not have a matched sibling donor, so transplants from a relative or unrelated matched donor are often carried out. These types of transplants have a less chance of succeeding compared to transplants from a matched, related donor. Transplants that are done in the first three months of life have the highest success rate.

SRY Gene

The SRY gene gives instructions for producing the sex-determining region Y protein. This protein is involved in male sexual development. The X and Y chromosomes determine whether a fetus will become a male or female. Females usually have two X chromosomes (XX), while males usually have one X chromosome and one Y chromosome (XY). The SRY gene is located on the Y chromosome. The sex-determining region Y protein produced from this gene attaches to specific regions of DNA and begins processes that cause a fetus to develop male gonads and stop the development of female reproductive organs.