

which allows the creation of transgenic animals with targeted mutations. CRISPR-CAS9 is characterized from other methods by the capacity to detect many genes at once, it is an open door for studying complex human diseases that are caused not only by a single mutation but by many genes acting together.

Conclusion. The technique is already being engaged in numerous applications in different domains. Within the context of health it could pave the way to the development of new treatments for rare metabolic disorders like Gaucher and Hunter syndromes, mental diseases such as Autism and Alzheimer, and genetic maladies from Haemophilia through to Huntington's disease. It is also being utilized in the creation of transgenic animals to produce organs for transplants into human patients (in the case of pigs). This method was also investigated for gene therapy. Such therapy aims to insert normal genes into the cells of patients to cure various disorders. Several start-up companies have been founded to develop this technology commercially and large pharmaceutical companies are also exploring its use for drug discovery and development purposes.

GENE MUTATION

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Introduction. Firstly, a gene can be defined as the basic physical and functional unit of heredity. It can also be described as a particular succession of nucleotides in DNA or RNA that is typically found on a chromosome. Genes are tasked with conveying and relaying some or all traits by the identification of structure of a certain polypeptide, especially a protein or by being in charge of the function of other genetic materials.

Aim. Mutation can be defined as an alteration which takes place in our DNA sequence, which is typically caused by mistakes which occur when our DNA is copied. Mutations also take place due to environmental influences. Examples of such influences include ultraviolet lights, smoke from cigarettes and other basic examples. Mutations come in different sizes and so they are capable of affecting all the other parts of the body, from one single DNA building block to a huge segment of a chromosome which consists of several genes.

Materials and methods. We analyzed many sources of science literature to get enough information about this problem.

Results and discussion. A Gene Mutation is a change or an adjustment in the succession of the DNA which makes up a gene in such a way that the succession is different from what is found in regular people. Gene mutations are permanent. They can generally be classified in two major ways:

1. Hereditary Mutations:

These mutations are the ones that are passed down from parents that tend to stay throughout a person's life. These mutations are in basically every cell in the organism. Hereditary mutations can also be referred to as Germ-Line mutations. This is due to the fact that they are present in the parents' egg or sperm cells which are called germ cells.

2. Acquired Mutations:

These mutations are also called somatic mutations. They are mutations that happen at any point in the life time of an organism. They occur only in certain cells of the body unlike hereditary mutations that occur in every cell in the body. Acquired mutations happen due to environmental factors like the sun's ultraviolet rays. They can also occur due to errors made when the DNA copies itself during the process of cell division. Acquired or Somatic mutations can not be passed on from generation to generation.

There is a genetic mutation called "De Novo" mutation. De Novo mutations can either be Hereditary or Acquired. Sometimes, this mutation happens in a person's egg or sperm cell but it won't be

present in any other cell in the person's body. Sometimes it happens in the fertilized egg a little while after the egg and sperm cell come together. It is usually not possible to know when a De Novo mutation has occurred. As the fertilized egg divides, each resulting cell in the growing embryo will have the De Novo mutation.

When certain genetic disorders affect the child, De Novo mutations are usually the given explanation as to why this disorder has occurred, especially when the parents don't have this mutation in their body and cannot trace any family history of the disorder.

Some other ways in which we can classify genetic mutation include:

3. Spontaneous Mutation

Spontaneous Mutations are not premeditated. They usually occur without any known causes. These mutations could arise due to various reasons which may include development errors, metabolic errors, replication errors, growth errors, etc. They mostly take place at the birth of the child. Bigger genes have a higher chance for spontaneous mutations. This is due to the fact that there is a higher chance of replication error occurrence in them than in smaller genes. Spontaneous mutations occur very rarely but once they do they are stably inherited.

4. Induced Mutation:

Induced Mutations happen when an organism is exposed to mutagenic agents. Examples of common mutagenic agents may include ultraviolet lights, radiation, chemicals, etc

5. Somatic Mutation:

Somatic Mutations as earlier states are mutations which take place in the somatic cells of an organism. Somatic mutations can not be inherited because only germ cells undergo fertilization and it can only occur in some tissues and some parts of the body but it can be spread to other daughter cells if it occurs in progenitor cells. Progenitor cells are cells that possess the ability of further division. The most widely known type of somatic mutation is in cancer.

6. Germ-Line Mutation:

Germ-Line Mutation is passed down from generation to generation. It is possible for germ line mutations to not occur in the parent organism but it will most definitely occur in the offspring. Germ-line mutations generally cannot be cured. It can be said that every inherited disorder is originated from the germ-line mutations. This mutation actually provides diversity in population (evolution of new alleles) but it is harmful to the organism sometimes.

7. Mitochondrial Mutation:

Mitochondria is the powerhouse organ of the cell. Mitochondrial DNA (mtDNA) is can be found in mitochondria and not in the nucleus. It has its own duplication and transcription machinery. An interesting fact about mitochondrial DNA is that it is inherited from only mothers, therefore it is called a maternally inherited DNA. If a mutation induced in the mitochondrial DNA is passed down to a male offspring, it cannot be passed down from him to his own offspring. Deletion, duplication, translocation, inversion, etc are some common types of mutation in the mtDNA.

8. Chromosomal Mutation:

Chromosomal mutations are passed down to generations depending on inheritance pattern. Some examples of mitochondrial DNA disorders are Kearns-Syre syndrome, Leigh syndrome, Non syndromic Hearing loss and others.

Other types of genetic mutations include:

9. Morphological Mutation:

Which influences the physical characteristics or phenotype of an organism. It alters physical properties like shape, size and color of the organism.

* Lethal Mutation:

Which leads to the death of the organism or has an effect on its survival.

* Conditional Mutation

* Biochemical Mutation

* Loss of Function Mutation

- * Gain of Function Mutation
- * Temperature - Sensitive; and many others.

Conclusions. A very small percentage of mutations give rise to genetic disorders. Most mutations do not have any effect on health or on development. Some mutations change the genes DNA sequence but do not change the function of the protein made by the gene, Some mutations do not have any noticeable effect on the phenotype of an organism.

Gene mutations that cause disease can rarely be seen in the general population. Frequently occurring genetic mutations (alterations that occur in more than 1% of the population) are called “Polymorphisms”. They happen frequently enough to be considered a normal occurrence in the DNA and are responsible for most of the normal differences between people. Polymorphism is also a natural phenomenon.

Scientifically, we can say that we are what we are today as a result of millions of mutations that have happened a long time ago.

AQUAGENIC URTICARIA

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Introduction. Water allergy is a rare form of physical urticaria, in which contact with water cause welts. It is a type of hives that cause a rash after you touch water, while taking a bath or a shower, in the rain, or in a swimming pool. In 1964 this disease was discovered by Shelley and Rawnsley.

Aim. It is an interesting disease with unknown pathogenesis and we want to investigate it properly.

Materials and methods. We have studied and analyzed some literature sources to describe aquagenic urticaria.

Results and discussion. The precise basic cause of aquagenic urticaria is poorly appreciated. Scientists suggest that allergy is not justified by water specifically, but more it could be an allergen in the water. There are some theories that chemical additives in water like chlorine cause a reaction in sensitive people by contacting with it.

In less than a few minutes being susceptible to water people with this disease can experience:

1. Abnormal redness and inflammation of the skin due to vasodilation.
2. Burning sensation
3. Inflammation
4. Abnormal growth compared to the skin around it

On the other side drinking water cause also other symptoms including:

1. Rash around the mouth
2. Difficulty swallowing
3. Wheezing
4. Difficulty breathing
5. Symptoms should be appeared within 30 to 60 minutes after drying the body.

To identify the nature of this illness the doctor should charge a bodily exam to detect the syndrome for the patient. He will check his psychiatric history and may also execute a water challenge test. Through the test, the doctor will apply a water press of 95°F (35°C) to upper body. This is done to call a reaction, symptoms should start in 15 minutes. The doctor will analyse the reaction of water challenge test and compare it to symptoms of aquagenic pruritus. This test normally can provoke itching and irritation, but doesn't cause hives or reddening.