* 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 justificated by water specifically, but more it could be an alergen 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 vosadilation.
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 syndome for the patient. He will check his psychiatric history and may also execute a water challenge test. Throught 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.

There is no treatement for aquagenic urticaria. However, there are some treatment options available to reduce symptoms. Antihistamines are medications used to treat allergy-like symptoms. Your doctor may recommend you take a prescription antihistamine to calm your hives after coming into contact with water. If you have a severe case of aquagenic urticaria and can't breathe, you may need to use an epipen. Epipens contain epinephrine, also known as adrenaline. They are only used as an emergency alternative for severe allergic reactions. Epipens increase blood pressure to reduce swelling and hives. They help the lungs function when they're constricted.

Conclusion. People with aquagenic urticaria can drink water, but they shouldn't let it get on the skin. They should limit rituals cleaning to 5 minutes having shower once or twice a week. Moreover, ration is also important, people with such disease should become vegetarian to make their body produce less oil.

PROGERIA

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Introduction. Progeria is a rare syndrome in children characterized by physical symptoms suggestive of premature old age. It is a very common autosomal dominant genetic disorder, in which people have symptoms similar to those of aging, but at a very early age. Those who born with this disorder usually live in their mid-teens to early twenties. It is a genetic condition that occurs because of a transformation of genes and is rarely inherited, that those who suffer from it usually do not live for a period of time when they can have children. Although progeria is a term that applies in its exact meaning to all diseases that include symptoms of premature aging, it is often found in people with Hutchinson-Guitdford-progeria syndrome (HGIPS). Scientists are interested in progeria disease itself because it may reveal important information about the natural aging process. It develops during a cellular division in a new fertilized egg or in the parents gametes.

Progeria is a Greek word what is divided into two parts: pro, which means premature, and gerya, which means aging, causes premature aging, so those who suffer from it live at an average of fifteen years only. People develop with great acceleration, so that the average age of children increases from five to ten times, compared to normal. The first case of this disease was clearly described in 1886 by Jonathan Hutchinson and was also described separately in 1897 by Hastings Guipford. The disease was later called by their names and now it is called Hutchinson-Guilford progeria syndrome.

Aim. The aim of the study was to inquire into progeria disease analyzing different data.

Materials and methods. We have researched information about progeria to study more about causes and symptoms of it.

Results and discussion. It cannot be said that progeria is a hereditary disease because it is not transmitted from parent to patient, but rather is caused by a genetic defect. A genetic mutation occurs in fetus during the months of pregnancy. The gene responsible for this disease have been remained unknown for a long period of time, due to many difficulties, including small number of patients and distributed between long distances, which made it more difficult to conduct studies. However, scientists were able to find the reason in 2003. They studied the genetic system of twenty patients and their families. There were similarities among 18 patients carrying the same change, located in the LMNA (lamine A/C) gene on the first chromosome at position 22, and the cause of the defect was substitution of one nitrogenous base, which led to the change of the amino acid guanine to amino acid adenine. It caused the disease, because change in one gene is responsible for abnormal protein progerin, which cannot function properly, holding