http://www.bbc.co.uk/bitesize/standard/biology/inheritance/variation/revision/4/

TAQ 5.

A mutation occurs when an organism is born with a difference characteristic.

Such as six fingers, or three ears. Mutations occur naturally, it can be caused by the Sun, or simply when the DNA doesn't copy itself properly. The risk is increased if you are exposed to radiation, such as x-rays and UV light. Certain chemicals can also cause mutations, such as cancer. The mutation occurs when there are changes in the base sequence of the DNA. The mutations can produce new Alleles which will not function correctly.

De Novo Mutation An alteration in a gene that is present for the first time in one family member as a result of a mutation in a germ cell (egg or sperm) of one of the parents or in the fertilized ega itself.

CHD7. Chromo domain helicase DNA binding protein 7 CHARGE syndrome (birth defects, heart defects.

Breathing problems).

1.

DESCRIBE MOSAICISM AND PROVIDE ONE EXAMPLE

A genetic mosaic is a creature whose body is built of a mixture of cells of two or more Different genotypes. In mammals they arise by several different mechanisms

The fusion of two different zygotes, or early embryos, into one. (The reverse of the process that produces identical twins!) The resulting animal is called a chimera (after the monster in Greek mythology with a lion's head, goat 's body, and serpent's all The tetra parental mouse is a chimera formed this way. But on rare people and the same process can occur spontaneously in humans (especially those using in vitre feelingion).

his occurs with the occasional The sharing of blood supplies by separate energy. This occurs with the occasional fraternal cattle twins and also less often with human fraternal twins who have shared the same placenta. Flood utem cells of each twill see the bone marrow of the other. Only their bloot less are mosaic.

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During arly development, errors during mitosis can produce stem cells that go on to populate a tissue or organ with, for example, a chromosomal aberration (e.g., aneuploid). Example: Occasionally a baby is born with blood cells that have three copies of chromosome 21 (the same set responsible for Down syndrome). This can produce a leukemia-like illness that, fortunately, often disappears as that cell population declines.

All female mammals are mosaic for the genes on the X chromosome because of the random inactivation of one or the other X chromosome in all their somatic cells. [Discussion]

Anyone unlucky enough to have a cancer is a genetic mosaic because all cancers are made up of the descendants of cells carrying a suite of mutations not found in normal cells. [More]

Recent advances have enabled the coding portions of the genome of single cells to be sequenced. Early results indicate than even normal cells in an adult have Accumulated a suite of somatic mutations that differs from cell to cell. So all of us are genetic mosaics! However, the rate of somatic mutations in these normal cells is only a fourth of that in cancer cells.

http://www.nlm.nih.gov/medlineplus/ency/article/001317.htm