**Are You A Mutant?**

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In Biology, a mutant is an organism or a gene arising or resulting from an instance of mutation. In this investigation, you will examine various mutations to determine whether you are a mutant. Are you ready for the truth? Here we go...

**Mutation 1**

Consider this. This outcome is produced as a result of 9 possible mutations in the coding regions of a gene. We will only focus on 2. For one mutation, on chromosome 2, 4 nucleotides are deleted leading to a premature stop codon after 55 altered amino acids. For the second mutation, on chromosome 2, a substitution of a purine for a pyrimidine at codon 268, which led to an amino acid substitution of histidine for glutamine.

1. Mutations arise from errors during which process?
2. Do these mutations arise at the transcription level or the translation level in protein synthesis? Explain.
3. For mutation one, is this mutation an example of a gene or chromosomal mutation? Explain your answer.
4. What is the specific type of mutation represented by mutation 1?
5. How did this specific mutation affect protein synthesis?
6. For mutation two, is this mutation an example of a gene or chromosomal mutation? Explain your answer.
7. What is the specific type of mutation represented by mutation 2?
8. How did this specific mutation affect protein synthesis?

Now that you have determined the possible types of mutations that can result in this condition, read the following below that explains the history of this mutation.

"Approximately 10,000 years ago, humans in Europe, parts of Africa, and a few other regions began to domesticate cows. Cattle herding provided people with an immediate benefit in the form of a reliable supply of meat. Cows also produce milk, which is a good source of nutrition, but the first cattle herders probably couldn't drink much of it. As infants, most mammals digest their mothers' milk with the help of a protein they produce, called lactase that breaks down lactose, the sugar in milk. But as mammals grow older, they stop producing lactase, with the result that they can no longer digest milk. Humans started out the same way, and many humans today remain lactose intolerant. They get indigestion when they try to drink milk or eat cheese.

But many people who descend from traditional cattle herders can still digest lactose. That's because they have inherited a mutation to a gene called LCT... It somehow disabled the off-switch for lactase production, and allowed people who carried it to drink milk into adulthood."- Carl Zimmer, "Under your skin", p. 14-15.

The mutation in the coding regions of the gene prevents humans from being able to digest milk. Therefore, are you a mutant? If your small intestine produces enough lactase to digest milk without experiencing symptoms such as bloating, diarrhea, gas, or pain or cramps in the lower belly, on the board, write a tally mark under the "mutant" category for mutation 1. If your small intestine cannot produce enough lactase to digest milk or cheese, on the board, write a tally mark under the "non-mutant" category for mutation 1.

**Mutation 2**

In this mutation of the OCA2 gene, the central six-base TAAATG sequence is replaced with the following mutated sequence- TAAGTC.

1. Mutations arise from errors during which process?
2. Does this mutation arise at the transcription level or the translation level in protein synthesis? Explain.
3. Is this mutation an example of a gene or chromosomal mutation? Explain your answer.
4. What is the specific type of mutation represented by this mutation?
5. How did this specific mutation affect protein synthesis?

Now that you have determined the type of mutation in this condition, read the following below that explains the history of this mutation.

Originally, humans all had brown eyes. New research has shown that people with blue eyes have a single, common ancestor. In fact, all humans with blue eyes are descendants of one individual with the mutation approximately 10,000 years ago.

A genetic mutation affecting the OCA2 gene in human chromosomes resulted in the creation of a "switch" which literally "turned off" the ability to produce brown eyes. The OCA2 gene codes for the production of the "P" protein, which is involved in the production of melanin, the pigment that gives color to our hair, eyes, and skin. The “switch”, which is located in the gene adjacent to OCA2 does not, however, turn off the gene entirely, but rather limits its action to reducing the production of melanin in the iris – effectively “diluting” brown eyes to blue. The switch’s effect on OCA2 is very specific therefore. If the OCA2 gene had been completely destroyed or turned off, human beings would be without melanin in their hair, eyes or skin color – a condition known as albinism.

Therefore, are you a mutant? If you have blue eyes, on the board, write a tally mark under the "mutant" category for mutation 2. If you have non-blue eyes, on the board, write a tally mark under the "non-mutant" category for mutation 2.

**Mutation 3**

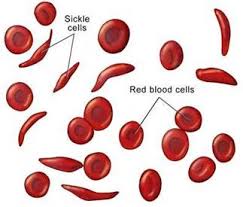
In this mutation, the sequence CCT GAG GAG AAG is replaced by CCT GTG GAG AAG.

1. Mutations arise from errors during which process?
2. Does this mutation arise at the transcription level or the translation level in protein synthesis? Explain.
3. Is this mutation an example of a gene or chromosomal mutation? Explain your answer.
4. What is the specific type of mutation represented by this mutation?
5. How did this specific mutation affect protein synthesis?

Now that you have determined the type of mutation in this condition, read the following below that explains the history of this mutation.

Sickle cell disease is a disorder that affects the red blood cells, which use a protein called hemoglobin to transport oxygen from the lungs to the rest of the body. Normally, red blood cells are round and flexible so they can travel freely through the narrow blood vessels.

The hemoglobin molecule has two parts: an alpha and a beta. Patients with sickle cell disease have a mutation in a gene on chromosome 11 that [codes](http://learn.genetics.utah.edu/content/disorders/singlegene/sicklecell/) for the beta subunit of the hemoglobin protein. As a result, hemoglobin molecules don't form properly, causing red blood cells to be rigid and have a concave shape (like a sickle used to cut wheat). These irregularly shaped cells get stuck in the blood vessels and are unable to transport oxygen effectively, causing pain and damage to the organs.



Therefore, are you a mutant? If you have sickle cell disease, on the board, write a tally mark under the "mutant" category for mutation 3. If you do not have sickle cell disease, on the board, write a tally mark under the "non-mutant" category for mutation 3.

**Mutation 4**

In this mutation, during cell division, chromosomes fail to separate correctly, resulting in one copy of chromosome 12 (instead of the required 2).

1. Is this mutation an example of a gene or chromosomal mutation? Explain your answer.
2. What is the specific type of mutation represented by this mutation?

Now that you have determined the type of mutation in this condition, read the following below that explains this mutation.

The majority of human chromosomal abnormalities occur in the [autosomes](http://anthro.palomar.edu/abnormal/glossary.htm#autosome) (chromosomes that do not determine the gender of an organism).  Most of these abnormalities are monosomies (1 copy of a chromosome) or trisomies (3 copies of a chromosome).  The majority of fetuses with autosomal monosomies spontaneously abort early in pregnancy. The only exception is fetuses with monosomy 21 (one copy of chromosome 21).

Therefore, are you a mutant? If your mother experienced spontaneous abortion with you, on the board, write a tally mark under the "mutant" category for mutation 4. If you are currently alive, on the board, write a tally mark under the "non-mutant" category for mutation 4.

**Mutation 5**

In this mutation, during cell division, chromosomes fail to separate correctly, resulting in three copies of chromosome 21 (instead of the required 2).

1. Is this mutation an example of a gene or chromosomal mutation? Explain your answer.
2. What is the specific type of mutation represented by this mutation?

Now that you have determined the type of mutation in this condition, read the following below that explains this mutation.

Down syndrome is a chromosomal condition that is associated with intellectual disability, a characteristic facial appearance, and weak muscle tone (hypotonia) in infancy. All affected individuals experience cognitive delays, but the intellectual disability is usually mild to moderate.

People with Down syndrome may have a variety of birth defects. About half of all affected children are born with a heart defect. Digestive abnormalities, such as a blockage of the intestine, are less common.

Therefore, are you a mutant? If you have Down syndrome, on the board, write a tally mark under the "mutant" category for mutation 5. If you do not have Down syndrome, on the board, write a tally mark under the "non-mutant" category for mutation 5.

**Mutation 6**

More than 25 mutations in the CNGB3 gene have been identified in people with this condition in which a nucleotide is deleted.

1. Does this mutation arise at the transcription level or the translation level in protein synthesis? Explain.
2. Is this mutation an example of a gene or chromosomal mutation? Explain your answer.
3. What is the specific type of mutation represented by this mutation?
4. How did this specific mutation affect protein synthesis?

Color vision deficiency is the inability to distinguish certain shades of color or in more severe cases, see colors at all. The term "color blindness" is also used to describe this visual condition, but very few people are completely color blind. Most people with color vision deficiency can see colors, but they have difficulty differentiating between

* particular shades of reds and greens (most common) or
* blues and yellows (less common).

The most common form of color deficiency is red-green. This does not mean that people with this deficiency cannot see these colors at all; they simply have a harder time differentiating between them. The difficulty they have in correctly identifying them depends on how dark or light the colors are, therefore red and green are perceived as identical. It is inherited in an X-linked recessive manner and affects 6% of males.

Another form of color deficiency is blue-yellow. This is a rarer and more severe form of color vision loss than red-green since persons with blue-yellow deficiency frequently have red-green blindness too. In both cases, it is common for people with color vision deficiency to see neutral or gray areas where a particular color should appear.

Therefore, are you a mutant? If you have red-green colorblindness, on the board, write a tally mark under the "mutant" category for mutation 6. If you do not have red-green colorblindness, on the board, write a tally mark under the "non-mutant" category for mutation 6.

**Discussion Questions**

Based on this information, what can you conclude about the following?

1. Using the information in this activity and your own background knowledge, are mutations beneficial, harmful, or neutral? Explain.
2. Do mutations result in immediate death?
3. If a mutation occurs in a skin cell, can this mutation be passed on to the offspring? Explain.
4. Examine the completed chart of your peers. Which type of mutation (gene or chromosomal) is more common? Explain why.
5. Examine the completed chart of your peers. What can you infer about the frequency of mutations? Explain.
6. Examine the completed chart of your peers. Make a conclusion on the frequency of harmful mutations in the future. Do you predict an increase or decrease?