# Assignment Summary

For this assignment, you will research two disorders caused by a genetic mutation, and theorize how association between genetics and a health outcome is possible.

Background Information

Mutations alter the genetic information in a cell. They change the DNA sequence of a gene. The downstream effects from a mutation can be positive, negative or neutral. Neutral just implies that the mutation happens but there is neither a positive nor negative effect on an organism’s ability to survive. Of course, positive means there is a beneficial effect from a mutation. For example, sometimes random mutations in organisms create genetic variations that help them adapt and positively respond to a changing environment.

You are most likely familiar with the negative effects of mutations—the development of diseases and genetic disorders. Cells rely on proteins to function. Genes provide the information and blueprint to create these proteins. An alteration at the genetic level will disrupt protein synthesis, causing a protein to be made incorrectly or not at all. A cell that does not have the correct protein to function could result in abnormal development or a genetic disorder. Scientists have discovered various types of mutations:

* Missense (or point) mutations—change in a single DNA base pair that affects the amino acid used to build a protein.
* Nonsense mutations—change in one base pair that creates an amino acid signaling the cell to stop protein synthesis.
* Frameshift mutation—the reading frame of DNA (used during replication and transcription) is wrong due to the addition/removal of nucleotide bases.

These are just some examples of how mutations occur. It is important to realize that genes do not cause disorders. It is the disruption of the protein (from an alteration in DNA) that leads to a disease or disorder. Also, these mutations can have different effects on health, depending on where they are found and just how much they disrupt the process of protein synthesis.

Materials

* Internet
* Computer or word processor
* Graphic organizer

# Assignment Instructions

For this project, you are expected to submit 3 things:

1. Graphic organizer
2. Diagram
3. Responses to analysis questions

**Step 1: Prepare for the project.**

1. Read through the guide before you begin so you know the expectations for this project.
2. If there is anything that is not clear to you, be sure to ask your teacher.

**Step 2: Choose a disorder to research.**

1. Look at the following list of disorders and select **one** that you would like to research further.
   1. Crohn’s disease
   2. Sickle cell disease
   3. Turner syndrome
   4. Down syndrome
   5. Cystic fibrosis
   6. Klinefelter syndrome
   7. Parkinson’s disease
   8. Tay-Sachs disease
   9. Retinitis pigmentosa

**Step 3: Research the disorder.**

1. Find reliable sources, such as those listed below, and choose two sources to learn more about the disorder you selected in Step 2.
   1. Examples of reliable library of health condition sources:
      * <https://www.genome.gov/For-Patients-and-Families/Genetic-Disorders>,
      * <https://ghr.nlm.nih.gov/condition?initial=s>

**Step 4: Complete a graphic organizer.**

1. Collect information about the disorder and organize your findings, using the graphic organizer provided in the section below.
2. Make sure your notes are clear, concise, and truly offer enough detail about the disorder you have selected.

**Step 5: Draw a diagram showing how the disorder progresses.**

1. Look at your graphic organizer, and think about how the disorder is inherited or caused, along with its effects.
2. Write down the causes, including the type of mutations associated with the condition.
3. In the section below, draw a diagram that resembles a flow chart showing the progression of this disorder, beginning first with the gene and ending with the outcomes a person experiences.
4. Make sure your flow chart is specific as it relates to the disorder. For example: A point mutation occurred where A converted to G.

**Step 6: Complete the written analysis questions at the end of this project**

1. Analyze and explain the causes and effects of the disorder by answering the questions in the **Written Analysis** section below.

**Step 7**: **Include a list of the resources you used at the end of this document.**

**Step 8: Evaluate your project using this checklist.**

If you can check each box below, you are ready to submit your project.

* Did you select one disorder to research?
* Did you find two reliable resources to collect information?
* Did you fill out your graphic organizer completely?
* Did you complete your diagram being as specific as possible with the information provided?
* Did you complete your written analysis questions?
* Are your responses free of grammatical and spelling errors?
* Did you list the resources that you used at the end of this document?

**Step 9: Revise and submit your project.**

1. If you were unable to check off all of the requirements on the checklist, go back and make sure that your project is complete. Save your project before submitting it.
2. Turn in your graphic organizer, diagram, and analysis questions to your teacher. Be sure that your name is on it.
3. Submit your graphic organizer, diagram, and analysis questions through the virtual classroom.
4. Congratulations! You have completed your project.

**Graphic Organizer**

Use the following graphic organizer to fill in important details about the disorder.

|  |  |
| --- | --- |
| **Name of Disorder** |  |
| **Chromosome Affected** |  |
| **Mutation Involved** |  |
| **Other Causes** |  |
| **Mechanism of Action**  **on Cellular Level** |  |
| **Description of Symptoms** |  |
| **Effects** |  |

Mapping the Disorder

In the box below, draw a flow chart that shows how the disorder progresses from the alteration in DNA to the outcome in a person. Make sure to properly label each part/step of the diagram.

|  |
| --- |
|  |

Written Analysis

Answer the questions below.

1. Analyze how the cause of the disorder relates to a change in DNA at the molecular level.
2. Explain what happened in the cell as a result of the mutation and how this relates to the effects a person experiences with this disorder.
3. Write one scientific question that could be investigated further to gain more insight about treating and/or reversing the effects of this disorder in a person.

Resources Used