

Genetic Disorder Research Project

DUE DATE: _____

You have been challenged with the task of incorporating your knowledge about cells, cell division, genetics, DNA, and human body systems to research a specific genetic disorder. Using the attached sheet of 50+ genetic disorders, choose one disorder you are most interested in researching. You will be the *EXPERT* on this particular disorder and you will present this information in a research paper. You have the opportunity to receive extra credit for creating a visual presentation of your choice. This will count for one informal test grade and up to 20 additional extra credit points.*If you would like to choose a disorder that is not on the list, please see me! There are so many more genetic disorders than the ones I have listed. I'll be glad to let you choose something else. I want this project to be interesting and relevant to YOU!

You will have 5+ days in class to work on this project, as well as no homework during the entire project so you can work at home as needed. We will have the ChromeBook cart in class the entire week, and you are welcome to use BYOT (laptops and tablets) every day to use as a tool. If you don't have a computer at home, be sure to take advantage of the computers in our classrooms and media center during class, as well as working lunch, PA, and REC if you need to!

PART I: RESEARCH PAPER

Guidelines for formatting your paper:

1. The paper must be IN YOUR OWN WORDS!!! You may NOT cut and paste information!
2. Create a title page including the name of the genetic disorder, your name, and class period
3. Must be typed using 12-point font, in Times New Roman ONLY
4. 1 inch margins, double spaced
5. Use correct grammar, punctuation, and spelling. Get a peer, parent, or teacher to edit!
6. You must include a bibliography page in APA format with at least 8 references. They may all be from the internet, but they MUST be reliable, credible sources. NO WIKIPEDIA AND REMEMBER, GOOGLE IS A SEARCH ENGINE AND NOT A SOURCE!! Please include the full website address.
7. Minimum of 4 pages, NOT INCLUDING title page and bibliography

You will need to include the following information in your paper:

1. What are the characteristics of the disease?
 - a. What is the name of the disorder?
 - b. Does it have a scientific name as well as a common name?
 - c. Who discovered the disorder and/or who contributed to the research?
 - d. What is the story/history of the disorder? Meaning, how was it discovered?
 - e. How common is the disorder in the general population? How many people currently are living with this disorder in the world? In the US?
 - f. Is the disorder more common in certain ethnic groups? Genders? If so, describe.
 - g. Is the disorder more common in certain parts of the world? If so, where and why?
2. How does the genetic problem affect the genetic code?
 - a. Do we know what chromosome it is on?
 - b. If so, do we know if it is dominant or recessive?
 - c. Is it autosomal or sex-linked?
 - d. Is it codominant or incomplete dominant? And what does this mean?
 - e. Who passes on the disorder: Mom or Dad, or both?
 - f. Is it passed on to the children?

3. What are the risk factors?
 - a. Does the disorder increase with mother's age, environmental factors, smoking, certain cancers, diet deficiencies, etc.?
4. How does the disorder manifest itself?
 - a. How does the disorder get diagnosed?
 - b. Are there prenatal tests available and what are they?
 - c. How does it make a person look or act?
 - d. Can you tell by looking at a person that they are affected?
 - e. How does it make the person different?
 - f. Does it start in infancy or is it adult onset?
 - g. Could this disorder have been prevented?
5. How does the disorder change the person's life?
 - a. How does it change the life of the affected individual?
 - b. Does the disorder affect the lifespan of the individual?
 - c. How does it change the life of the individual's family?
 - d. What does the individual have to do differently on a daily/weekly/monthly basis?
 - e. What preparations have to be made for later?
 - f. Is the disease progressive?
 - g. Can an individual with this disorder have children? What is the likelihood the children will be born with this or another disorder?
6. Is there any special medical care, assistance, or medicine required?
7. Are there any special services or support groups that can help?
8. What is the CURRENT status of research on this disorder?
 - a. If there is no cure, are we close to finding one?
 - b. Where is the research coming from?
 - c. Are there drug or surgical trials?
9. Include anything else you find about cures, research, or anything else you feel is important to add from your research.

EXTRA CREDIT: VISUAL PRESENTATION (up to 20 points)

Using the information you have gathered for your research paper, you will create a visual presentation that will educate your audience about your genetic disorder.

Choose one of the following to display your disorder:

- ❖ Brochure or pamphlet
- ❖ Google Presentation
- ❖ Poster or tri-fold presentation board
- ❖ Video, including but not limited to a skit, music video, or infomercial

Your visual presentation should include the following:

- ❖ Color and graphics, including meaningful/necessary photos, charts, and/or diagrams
- ❖ Important facts organized in a neat way that readers can easily comprehend
- ❖ Correct spelling, grammar, and punctuation
- ❖ Evidence that you UNDERSTAND what you are presenting
- ❖ An attractive, neat layout that shows your best effort!
- ❖ Ten minute minimum for video presentation
- ❖ 12 slide minimum for Google Presentation

List of Possible Genetic Diseases and Disorders to Research

Achondroplasia (Dwarfism)	Edwards Syndrome	Patau Syndrome (Trisomy 13)
Adrenal Hyperplasia	Fabry Disease	Pheylketonuria
Adrenoleukodystrophy	Familial Dysautonomia	Polycystic Kidney Disease
Albinism	Gardner Syndrome	Prader-Willi Syndrome
	Gaucher's Disease	Proteus Syndrome
Angelman Syndrome	Hemochromatosis	Progeria
Autism	Hemophilia	
Breast Cancer	Huntington Disease	
Burkitt's Lymphoma	Ichthyosis	Rett's Syndrome
	Jacobsen Syndrome	SCID
Celiac Disease	Klinefelter Syndrome	Sickle Cell Anemia
Colorblindness	Klippel-Feil Syndrome	
Crohn's Disease	Leukodystrophy	Smith-Magenis Syndrome
Cystinosis	Marfan Syndrome	Spinal Muscular Atrophy
Cystic Fibrosis	Moebius Syndrome	Spinocerebellar Ataxia
Dercum Disease	Monosomy 9p (Alfi's Syndrome)	Tay-Sachs Disease
DiGeorge Syndrome	Myotonic Dystrophy	Tourette Syndrome
Down Syndrome	Neurofibromatosis	Turner Syndrome
Duane Syndrome	Noonan Syndrome	Von Hippel-Lindau Syndrome
Duchenne Muscular Dystrophy		Wilson Disease

Below are some examples of reliable and credible sources to use to get you started. These sources may not necessarily be related to your specific disorder, but they can help you with terms you may not understand or background information about your project. You may start with these, but since you are required to research for this project, you may use NO MORE THAN FOUR of these sources.

Genetic and Rare Conditions Site: <http://kumc.edu/gec/support>

Genetic Disorder Library: <http://learn.genetics.utah.edu/units/disorders/whataregd>

The National Human Genome Research Institute: <http://www.nhgri.nih.gov/>

A Gene Map of the Human Genome: <http://www.ncbi.nlm.nih.gov/science96/>

What is Genetic Testing?: <http://lbl.gov/Education/ELSI/Frames/genetic-testing-f.html>

Genetics Education Center: <http://kumc.edu/gec/>

List of Specific Genetic Disorders and Links: <http://www.genome.gov/10001204>

Research Paper Grading Rubric

Research Paper Requirements	Possible Points
Formatting	
Information in your own words, no cutting/pasting	5
Layout	
✓ 12 point font, Times New Roman	4
✓ 1 inch margins, double spaced	4
Title Page	
✓ Name and class period	1
✓ Name of genetic disorder	4
Grammar, Punctuation, and Spelling	5
Bibliography	
✓ Minimum 8 sources	4
✓ APA format, cited correctly	2
✓ Sources are reliable and credible	4
Minimum 4 pages	4
Information (See instruction sheet for details)	
(#1) Characteristics of disease	7
(#2) Genetic information	7
(#3) Risk factors	7
(#4) Symptoms	7
(#5) Affects	7
(#6) Medical care	7
(#7) Services and support	7
(#8) Current Research	7
(#9) Other important information	7
Total Possible Points	100

Visual Presentation Rubric

Visual Presentation Requirements	Possible Points
Use of appropriate graphics, photos, charts, or diagrams	5
Correct spelling, grammar, and punctuation	2
Evidence that you UNDERSTAND what you're presenting	3
Attractiveness, neatness, and organization	7
Minimum requirements met	3
✓ 10 minute minimum for video	
✓ 12 slide minimum for Google Presentation	
Total Possible Points	20