

# Genetic Disorders Presentation

Genes are the individual units of DNA, the building blocks of life. When mistakes or mutations occur, they can cause genetic disorders. Hereditary, or germline mutation, can be passed on from generation to generation. Acquired, or somatic mutation, may be caused by environmental stress.

Use reference and circulating library books for background information. Journal articles and some information on the web will help you round out your report.

## TOPICS

Choose one of these topics for your research. If you have another genetic disease you would prefer to use, please discuss it with your instructor.

Achondroplasia	Fragile X	Phenylketonuria (PKU)
Color Blindness (color Vision Deficiency)	Galactosemia	Prader-Willi Syndrome
Cri du Chat	Hemophilia	Progeria
Cystic Fibrosis	Huntington's Disease	Sickle Cell Disease
Diabetes Mellitus	Klinefelter Syndrome	Tay Sachs
Down's Syndrome	Marfan Syndrome	Triple X Syndrome
Fanconi Anemia	Muscular Dystrophy (Duchenne's)	Turner's Syndrome

## ASSIGNMENT

Your assignment is to design a PowerPoint presentation to describe and discuss a specific genetic disorder. The information will be presented orally in class and the PowerPoint slides will be added to the e-classroom. You are encouraged to this web site and Chapter 15 in the text.

The following criteria will be used to evaluate your presentation:

1. The information must be current and correct.
2. The information must be presented in a concise manner. Do the research first, and then design the slides. Use bullets appropriately and avoid wordiness.
3. Do NOT copy information from your references.
4. Organize the information in a logical manner.
5. The information should be understandable. Do not use terms you cannot pronounce or do not understand. Please consult me for help.
6. The presentation should have visual appeal but stay away from overly dramatic displays. (flying words, the "typed" effect, weird noises etc.)

The following points must be included in your slides and oral presentation:

1. Briefly describe the disorder. Include characteristics, symptoms, age of onset and any relevant ethnic information. Do NOT list every symptom or characteristic. Include only the most relevant and important points.
2. Discuss any unusual or unique features of the disorder.
3. Discuss the genetic basis for the disorder.
  - a. Is the disorder inherited?
  - b. Is the disorder a result of a mutation?
  - c. Is the disorder a result of too many or too few chromosomes?
4. Explain the inheritance by doing one of the following: construct a punnett square to illustrate path of inheritance, describe and illustrate with a diagram
5. Describe the prognosis for individuals with this disorder. Are there any medical treatments available? What is the average life expectancy? Are there genetic tests available?
7. Finally, provide a references slide in proper MLA format.

## REFERENCE BOOKS

Begin your research by looking at Reference Books for an overview of the disease. A few of these diseases have circulating books available. [Check the library catalog](#) for other books.

REF RB155.5 .G35 *Gale Encyclopedia of Genetic Disorders*

Diagnosis, treatment, prognosis. The bibliographies include organizations and websites.

REF RB155.5 .G455 Massimini, Kathy, ed. *Genetic Disorders Sourcebook*.

Several pages on each covered disease, including a bibliography and other resources to contact.

REF RB155 .G3593 King, Richard A., ed. *Genetic Basis of Common Diseases*.

Scientific overview of diseases, with graphs and extensive bibliography.

REF RB155.5 .W96 Wynbrandt, James. *Encyclopedia of Genetic Disorders and Birth Defects*.

Brief overview of diseases in alphabetical order. Good starting point.

## FINDING JOURNAL ARTICLES

Research Journal Articles for the most recent information. Use *peer reviewed* or *scholarly* (articles) for authoritative information.

### [Health Reference Center: Academic](#)

Includes articles from medical journals, reference books, and dictionary definitions.

### [PubMed](#)

Index to articles in medical journals. More thorough than the other indices but usually only abstracts, not full text.

### [Proquest: Research Library](#)

Articles from less scientific journals. Easier to understand.

## **WEB SITES**

Search these web sites for information. You may also want to try a web search on your own, but be sure to determine the authority of your source.

### [Medline Plus Genetic Disorders](#)

Links to the latest research on genetic disorders. Includes overviews.

### [Genetic Science Learning Center](#)

Basic information on Genetic Disorders.

### [Yahoo! Directory of Genetic Disorders](#)

Links to information on many specific disorders

### [Merck Manual of Diagnosis and Therapy](#)

Disease diagnosis, prevention, and treatment.

### [University of Washington Health Links](#)

Searchable index to health information at both professional and student level.

### [National Center for Biotechnical Information](#)

Database of genetics and genetic information, intended for professionals.