

# Genetic Diseases

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You are now familiar with the role of DNA in our bodies; how it is replicated and expressed within a cell. You also know how DNA can accumulate mutations and what this can do to the protein that may be coded by that sequence. Also keep in mind how DNA is organized into multiple chromosomes. During mitosis and meiosis, it is crucial that chromosomes be separated equally between the daughter cells.

In our current unit, we will discuss how traits (carried on our chromosomes) are inherited from one generation to the next. You will learn how to predict the probability of a trait being passed on and how this information is practical for potential parents and genetic counselors.

Through this assignment, you will choose a genetic disease which you will research. You will create a wiki page about your disease and post it on the Pingry server. In addition you will give a brief (5 minute) oral presentation to the class. And provide a brochure of your disease and syndrome.

During your research, look for the following:

- What is the genetic cause of the disease?
- What gene is mutated or chromosome altered?
- What is the pattern of inheritance of the disease (dominant or recessive)?
- What are the symptoms and characteristics of the disease?
- What are the current methods of treatment/cure for the disease?

Guidelines for the wiki page that you will be creating:

- Treat your wiki as if you are presenting the information live to a group of people.
- Create your own work. If you use material from another source, be certain that you give proper credit to that source.
- Follow appropriate formatting.
- You will use at least three (3) citations. One of which has to be print sources.
- Include the following:
  - Introduction of the disease
  - Genetic information
  - Symptoms
  - Treatments and cures
  - Appropriate images and pictures
  - Contact information for support groups, etc.

Your brochure:

- will provide a summary of your wiki page.
- a color copy will be provided to your teacher.
- make enough copies to distribute to your classmates. These copies should be black and white.

In the ever increasing volume of information available on the World Wide Web, it is important to consider the reliability of your sources. For this project, you may want to avoid personal websites (although you may use them to obtain personal accounts of those living with a particular disease).

Here are some links that may be helpful:

<http://www.bbc.co.uk/health/genes/disorders/types.shtml>

<http://rarediseases.info.nih.gov/index.html>

### Suggested genetic diseases:

Achondroplasia	Marfan's Syndrome
Achromatopsia	Moebius Syndrome
Acid maltase deficiency	Mucopolysaccharidosis (MPS)
Adrenoleukodystrophy	Nail Patella Syndrome
Aicardi Syndrome	Nephrogenic Diabetes Insipidus
Alpha-1 Antitrypsin deficiency	Neurofibromatosis
Androgen Insensitivity Syndrome	Niemann-Pick Disease
Apert Syndrome	Osteogenesis Imperfecta
Arrhythmogenic Right Ventricular Dysplasia	Porphyria
Ataxia Telangiectasia	Prader-Willi Syndrome
Barth Syndrome	Progeria
Blue Rubber Bieb Nevus Syndrome	Proteus Syndrome
Canavan Disease	Retinoblastoma
Cri Du Chat Syndrome	Rett Syndrome
Crigler-Najjar Syndrome	Rubinstein-Taybi Syndrome
Cystic Fibrosis	Sanfilippo Syndrome
Dercum's Disease	Shwachman Syndrome
Ectodermal Dysplasia	Sickle Cell Disease
Fanconi Anemia	Smith-Magenis Syndrome
Fibrodysplasia Ossificans Progressiva	Stickler Syndrome
Fragile X Syndrome	Tay-Sachs Disease
Galactosemia	Thrombocytopenia Absent Radius (TAR) Syndrome
Gaucher Disease	Treacher Collins Syndrome
Glutaric Aciduria	Tuberous Sclerosis
Hemochromatosis	Turner's Syndrome
Hemophilia	Urea Cycle Disorder
Huntington's Disease	von Hippel-Lindau Disease
Hurler Syndrome	Waardenburg Syndrome
Hypophosphatasia	Williams Syndrome
Klinefelter Syndrome	Wilson's Disease
Krabbes Disease	
Langer-Giedion Syndrome	
Leokodystrophy	
Long QT Syndrome	

**Projects are to be completed by: October 26, 2011**

**Oral Presentations will be given on: October 27, 2011**