PROJECT: HUMAN GENETIC DISORDERS

Background:

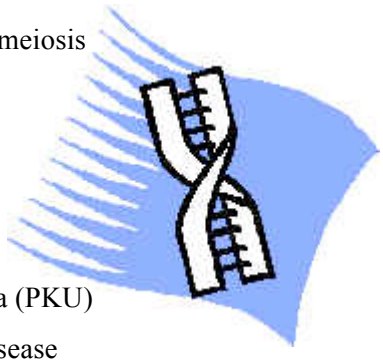
How do human genetic disorders or conditions occur? Does a single mutation, a mutation in one gene, or even a whole chromosome cause the disorder? Genetic disorders can affect many different aspects of human development: mental or intellectual development, height, the nervous system, and even gender. Sometimes during meiosis, there is an error called nondisjunction, in which the chromosomes fail to separate properly. Nondisjunction can result in other combinations of an individual's chromosomes, including the sex chromosomes. If the autosomes are affected (pairs #1-22) disorders such as Down Syndrome may result. When the sex chromosomes are involved, the new combinations can affect gender development in a variety of ways.

Purpose:

In this activity, you will be exploring the process of how human genetic disorders are inherited and how they develop. You will focus on a specific genetic condition and research how that particular condition shapes an individual.

Vocabulary:

Autosomal: refers to a trait whose gene is located on an autosome (non-sex chromosome)
Meiosis: cell division that produces egg cells and sperm cells
Mutation: a chemical change in a gene, resulting in a new allele; or, a change in the portion of a chromosome that regulates the gene (controls when a gene should make its protein)
Nondisjunction: failure of chromosomes to separate properly during one of the stages of meiosis
Sex-linked or X-linked: refers to a trait whose gene is located on the X-chromosome
Syndrome: set of symptoms that typically occur together



Procedure:

A. You and a partner will be assigned one of the following genetic disorders:

1) XO female (Turner's syndrome)
2) XXY male (Klinefelter's syndrome)
3) Xeroderma Pigmentosum
4) Down Syndrome
5) Achondroplasia (dwarfism)
6) Sickle-cell anemia
7) Cystic Fibrosis
8) Tay Sachs
9) Osteogenesis Imperfecta
10) Phenylketonuria (PKU)
11) Huntington's disease
12) Tourette Syndrome
13) Marfan's Syndrome
14) Hemophilia
15) Neurofibromatosis
16) Thalassemia
17) Gaucher Disease
18) Fragile X Syndrome

B. Research your particular genetic disorder

- The computer lab has been reserved for you to use the Internet to research your disorder. A list of websites to get you started is provided below. Be sure to expand your search to other sites. These will be included in your bibliography.

C. Design a Brochure explaining your disorder. Provide information to address the following questions:

- How would you recognize this condition in a family member? What physical & psychological **SYMPTOMS** are associated with this genetic condition?
- What is the **CAUSE** of this disorder? Is the inheritance pattern sex-linked, autosomal, recessive, dominant, or caused by mutation, chromosomal abnormality, or other mechanism?
- Is there any **TREATMENT** for this condition? Is there a way to prevent this condition?
- Is there a way to **SCREEN** individuals for this condition?
- What **PERCENTAGE** of the population is thought to have this condition? Is it more common in certain populations? Consider both ethnicity and geography.
- What are the **SOCEITAL** implications of these variant genetic conditions? How are individuals with this condition accepted by society?
- Your design must include a minimum of two (2) **GRAPHICS**. One should show an individual with this condition, the other should illustrate the inheritance pattern.
- Include a **BIBLIOGRAPHY** containing at least five (5) sources.

D. Complete a chart for all 18 genetic conditions. All brochures will be available in the classroom. You will read the brochures for each human genetic disorder record the information presented on a data table. You will be required to know about all of these conditions, so carefully read other group's reports and record accurate data.

Websites to get started:

- March of Dimes (Health Library; Fact Sheets) www.modimes.org
- http://www.healthweb.org/search.cfm
- http://www.genome.gov/10001204
- http://learn.genetics.utah.edu/content/disorders/whataregd/
- http://www.ornl.gov/sci/techresources/Human_Genome/medicine/assist.shtml

Genetic Disorder_____ **Names:** _____

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RUBRIC: HUMAN GENETIC DISORDERS BROCHURE PROJECT

Required Components	Points Possible	Points Received
Symptoms of Condition	2	
Genetic Cause of Condition	2	
Treatment for Condition	2	
Test or Screen Individuals for the Condition	2	
Percentage of Population Affected	2	
Societal Implications	2	
Layout and Design (including graphics)	2	
Bibliography	2	
TOTAL POINTS	16	