Name(s):	Period:	Date:

Mutations and Genetic Disease

HASPI Medical Biology Lab 18a

Scenario

Genetic diseases are caused by abnormalities, or mutations, in an individual's DNA. These mutations can be small, involving a single nucleotide, more involved affecting the nucleotide sequence, or extensive resulting in the loss or addition of entire chromosomes. Since genes are the instructions for proteins, a mutation in the genes results in the protein being formed incorrectly. The protein is then not able to perform the function it was originally meant to perform. Once a mutation occurs in an individual, that mutation can be passed down and inherited in offspring of an affected individual. These mutations are present in the DNA of every cell in an affected individual, and therefore are extremely difficult to cure. At this time, there are no cures available for genetic disorders, only treatments, but researchers are still working to discover a cure.

Guidelines

You will be creating a brochure, poster, newsletter, PowerPoint presentation, or written report for an assigned genetic disease.

A minimum of a paragraph explanation of each of the following must be included with a title for each section(question):

1. A **title** page with your **name, class,** and **date** with an **image** related to your disease.
2. **WHAT IS IT?** Identify the disease.
3. **CAUSES:** On what chromosome is the defective gene located, or which chromosome abnormality is responsible for the disease?
4. **SYMPTOMS:** Explain how the disease affects a person's body.
5. **INCIDENCE:** How many people have it, or birthrate frequency, and what age/sex?
6. **INHERITANCE:** Determine whether your disorder is autosomal dominant, autosomal recessive, X-linked recessive, chromosomal error, or multi-factorial (polygenic disorder). Chromosomal errors may take several types: a particular missing chromosome (e.g. a missing X chromosome), an extra chromosome (e.g. an extra chromosome 21), or a damaged chromosome (e.g. part of a chromosome deleted). A multi-factorial disorder is caused by several genes, or by a combination of genetic and environmental factors.
7. **DIAGNOSIS:** Describe the tests that are performed in order to diagnose the disease (blood test, x-ray, MRI…). Can a person be directly tested to see if she/he has the gene? Can amniotic fluid be tested to see if a baby is going to have this genetic disease?
8. **TREATMENT:** What types of therapies are currently <u>being done</u> for the disease? What therapies are currently <u>being researched</u> for the disease?
9. **DISCOVERY:** How long have we known about the disease? Has the source been commonly known for years, or did we recently figure out the cause of this disease?
10. **SUPPORT:** What is the name of a support organization for people who have this disease, or for their family members and friends? Where is the chapter closest to San where you live located?
11. **PROMOTION:** Are there any celebrities who have the disease or are a spokesperson to promote awareness of the disease? Are there any special events centered on this disease?

You MUST show references for the researched information!!!

Name(s): **Period:** **Date:**

List of possible diseases to research: (this is only a small list of diseases -- don't limit yourself to just the diseases on this list. Feel free to choose a disease of someone you know who has been diagnosed)

- **Alzheimer's disease** (AD; degenerative dementia)
- **Amyotrophic lateral sclerosis** (ALS; Lou Gehrig's Disease; progressive motor function loss leading to paralysis and death)
- **Ataxia telangiectasia** (AT; progressive brain disorder resulting in loss of muscle control and cancers)
- **Breast cancer** (make sure you focus on information involving the BRCA 1 and BRCA 2 genes)
- **Charcot-Marie-Tooth** (CMT; loss of feeling in ends of limbs)
- **Congenital adrenal hyperplasia** (CAH; hormone deficiency; ambiguous genitalia and male pseudohermaphroditism)
- **Cystic fibrosis** (CF; disease of lung and pancreas resulting in thick mucous accumulations and chronic infections)
- **Duane syndrome** (DS; eye movement disorder)
- **Duchenne muscular dystrophy/Becker muscular dystrophy** (DMD; severe to mild muscle wasting, deterioration, weakness)
- **Dystonia** (DYT; muscle rigidity, repetitive twisting movements)
- **Fanconi anemia, group C** (FA; anemia, leukemia, skeletal deformities)
- **Factor V-Leiden** (FVL; blood-clotting disorder)
- **Fragile X syndrome** (FRAX; leading cause of inherited mental retardation)
- **Gaucher disease** (GD; enlarged liver and spleen, bone degeneration)
- **Hemochromatosis** (iron overload in blood)
- **Hemophilia A and B** (HEMA and HEMB; bleeding disorders)
- **Hereditary hemochromatosis** (HFE; excess iron storage disorder)
- **Huntington's disease** (HD; usually midlife onset; progressive, lethal, degenerative neurological disease)
- **Klinefelter's syndrome** (affects male reproductive system)
- **Myotonic dystrophy** (MD; progressive muscle weakness; most common form of adult muscular dystrophy)
- **Neurofibromatosis type 1** (NF1; multiple benign nervous system tumors that can be disfiguring; cancers)
- **Parkinson's disease** (PD; degeneration of the central nervous system)
- **Phenylketonuria** (PKU; progressive mental retardation due to missing enzyme; correctable by diet)
- **Adult polycystic kidney disease** (APKD; kidney failure and liver disease)
- **Prader Willi/Angelman syndromes** (PW/A; decreased motor skills, cognitive impairment, early death)
- **Sickle cell disease** (SS; blood cell disorder; chronic pain and infections)
- **Spinocerebellar ataxia, type 1** (SCA1; involuntary muscle movements, reflex disorders, explosive speech)
- **Spinal muscular atrophy** (SMA; severe, usually lethal progressive muscle-wasting disorder in children)
- **Thalassemias** (THAL; anemias - reduced red blood cell levels)
- **Tay-Sachs disease** (TS; fatal neurological disease of early childhood; seizures, paralysis)
- **Turner's syndrome** (absence of female sex chromosome)