Disorder Project

1. Name and describe the disorder. What part of the body does the disorder affect? (whole body, reproductive organs, lungs, etc.)

2. How is the disorder inherited?
   - chromosomal or gene disorder?
   - if gene disorder... sex-linked or autosomal?
   - if gene disorder... dominant, recessive, codominant, etc.?

3. Create a 4 generation pedigree, with at least 20 individuals that shows the transmission of the disorder. Include genotype, phenotype and a key/legend.

4. Symptoms, lifestyle, effects, outcome of the disorder, life expectancy

5. Is the disorder curable, treatable, lethal?

6. Fun fact/something interesting about the disorder
Section 15.2
Human Genetic Disorders
I Can...

• **LS 3.2** I can explain how changes in protein formation can result in changes in phenotype.

• **ETS 2.2** I can research ways karyotypes are used in diagnostic medicine.

• **ETS 2.2** I can identify patterns in common gene disorders (monosomy, trisomy, etc.) given a karyotype.
Key Questions

1. What are the effects of errors in meiosis?
2. How do small changes in DNA affect human traits?
Vocabulary

• Nondisjunction
Chromosomal Disorders

• The most common error in meiosis occurs when homologous chromosomes fail to separate, known as **nondisjunction**.
• Gametes with an abnormal number of chromosomes may result...
• ...leading to a disorder of chromosome numbers in the offspring.

• Examples
  • Down syndrome
  • Turner’s syndrome = (45,X)
  • Klinefelter’s syndrome = (47,XXY)
Down Syndrome

- Individual born with 3 copies of chromosome 21
- Also known as Trisomy 21
  - Trisomy = “three bodies”
- Down syndrome is the most common form of trisomy
- Range of cognitive disabilities and birth defects
From Molecule to Phenotype

• Changes in a DNA sequence can change proteins (by altering their amino acid sequences)
  • This may directly affect an individual’s phenotype
  • Sometimes, the effect is more subtle
    • Certain alleles are associated with tendencies to develop certain conditions (like heart disease, diabetes, and cancer)
    • Many other factors (like behavior, diet, and environment) can have a profound effect on whether these conditions actually develop
Disorders Caused by Individual Genes

- Sickle cell disease
- Cystic Fibrosis
- Huntington’s disease
Sickle Cell Disease

• One of the first recognized molecular diseases

• Two important discoveries...
  • Caused by a recessive allele
  • Hemoglobin in people with sickle cell disease is different from normal hemoglobin
    • (hemoglobin is the oxygen-carrying protein in red blood cells)

• Revealed the links between genes and abnormal proteins, and between proteins and human disease
Sickle Cell Disease

• Caused by a defective allele for beta-globin, one of the two polypeptides in hemoglobin
  • Makes hemoglobin less soluble... causing molecules to stick together when blood oxygen levels decrease
  • Hemoglobin clumps into long fibers and distorts the shape of red blood cells... “sickle-shaped”

• Sickle-shaped cells are more rigid than normal red blood cells... so they can get stuck in capillaries
Sickle Cell Disease

(a) Normal amino acid sequence

Thr 4  Pro 5  Glu 6  Glu 7

(b) Single change in amino acid sequence

Thr 4  Pro 5  Val 6  Glu 7

Normal red blood cells

Sickled red blood cells

Figure 3-13  Biological Science, 2/e

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Cystic Fibrosis

• Cystic fibrosis results from the deletion of 3 bases in the gene for a protein called CFTR.
• The loss of these bases removes a single amino acid from CFTR, causing the protein to fold improperly.
• Two copies of the defective gene are needed to produce the disorder... meaning the CF allele is recessive.
• Children with CF have serious digestive problems and produce a thick, heavy mucus that clogs their lungs and breathing passageways.
Huntington’s Disease

• Caused by a dominant allele for a protein found in brain cells
• The allele contains a long string of bases in which the codon CAG repeats over and over again.
• The reason why this causes the disease is still not clear.
• The greater the number of repeats, the earlier the disease appears and the more severe are its symptoms.
• Symptoms (mental deterioration and uncontrollable movements) usually do not appear until middle age.
Genetic Advantages

• CF Allele
  • Carried by roughly 1 in 25 people of European ancestry

• Sickle Cell Allele
  • Carried by approximately 1 in 12 people of African ancestry

• If these alleles produce diseases that can be fatal, why are they still around?
CF Allele and Typhoid

• More than 1000 years ago, cities in Europe had epidemics of typhoid fever, caused by a bacterium that enters the body through the digestive system.

• The proteins produced by the CF allele helped block the entry of this bacterium.

• Carriers of CF had an advantage, living in these cities with poor sanitation and polluted water.
Sickle Cell Allele and Malaria

• Malaria is common in west central Africa.
• Malaria is a mosquito-born infection caused by a parasite that lives in red blood cells.
• Carriers of sickle cell are highly resistant to the parasite.
Section 15.2 Exit Ticket

1. How can a karyotype illustrate a nondisjunction error during meiosis or large chromosomal deletions/duplications?
The End 😊