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Human Genetics: Pedigree Analysis Lab

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Introduction:

We can apply the principles of Mendelian genetics to humans in order to make predictions about the appearance of a trait in subsequent generations. There are some logistical difficulties in understanding human genetics. First, we must identify human traits which are controlled by a single gene (most are controlled by more than 1 gene and also environmental influences). It is often difficult to determine if environmental effects are more important than the genetics (for instance in the inheritance of tendencies toward heart disease or cancer). Second, humans are seldom willing to make the appropriate matings so that scientists can determine the mode of inheritance of a particular trait; plants and animals are much more cooperative. Instead, geneticists who study traits in humans must rely upon records of marriages, births, and deaths. These relationships are displayed in a chart on which we can trace the genetic relationships; these charts are called pedigrees.

A pedigree diagram expresses the relationships of individuals, their gender, and the presence / absence of a specific trait. To be of much use in a genetic analysis, the pedigree must include several generations, preferably from a family with many children.

Objectives:

To familiarize students with the construction of human pedigrees. Students will apply their knowledge of inheritance patterns to pedigrees. Students will identify the relationships between individuals in a pedigree and determine what inheritance pattern is present in a family based on which individuals in the pedigree are affected and unaffected. Then students will assign individuals who

are represented in a pedigree genotypes based on clues they interpret from the pedigree.

Pedigrees 1 - 5 can be found in the WORD or PDF files below this assignment in the weekly Moodle block

Consider the pedigree for cystic fibrosis. (See Pedigree 1.) An autosomal recessive gene (f) causes serious respiratory difficulties that usually lead to death of afflicted people by their late 30's.

The generations are indicated by the Roman numerals on the left side of the pedigree. The members of each generation (related or not) are indicated by Arabic numbers. Females are represented by circles and males are represented with squares. Individuals with cystic fibrosis are shaded gray (or black). Thus, the 2 affected individuals are those in the 3rd generation: III-2 and III-4.

You have already learned about the inheritance pattern of CF. However, you could have inferred it from the pedigree. Because the disease only affects siblings in one generation, the disease is most likely to have autosomal recessive inheritance.

Because we know that cystic fibrosis is autosomal recessive, we can write the genotypes (entirely or in part) of all of the individuals in this family tree. Recessive homozygous individuals (ff) express the disease. Each normal individual has at least 1 dominant allele (F). We know that the individuals II – 2 and II – 3 are heterozygous (because this couple has a child with CF). The genotypes of the other 2 individuals in generation II can only be written as F₋ because we do not know if they are FF or Ff.

QUESTIONS:

1. Using pedigree 1, list all of the possible genotypes of these individuals:
HINT: the easiest way to do this is to write letters (alleles) in, on the pedigree, for the people you know for sure. In this case, write the genotype that is homozygous recessive under the affected people. Then write out the genotypes of other

individuals that you can infer. After you have filled in the genotypes on the pedigree, then fill out this chart.

Individual Genotype

I-1

I-2

I-3

I-4

II-1

II-2

II-3

III-1

III-2

III-3

III-4

III-5

2. Look at PEDIGREE #2. Do you think the trait shown is autosomal dominant or autosomal recessive? List all of the characteristics of the pedigree that justify your answer.

Use the letters "B" and "b" for the alleles.

3. What is the most likely genotype of individual I-2?

4. What does the notation like an upside-down T between II-3 and II-4 mean?

5. What is the chance that III-3 will have an affected child?

6. What is the chance that III-4 will have an affected child?

7. What is the genotype of individual III-2?

The family depicted in PEDIGREE #3 has some individuals with achondroplasia, a form of dwarfism. Note: (1) The homozygous dominant genotype is lethal; unfortunately infants with this genotype have such small ribs they cannot breathe at birth. (2) All of the people in the pedigree who have the dominant allele have the express the achondroplasia phenotype.

8. For each of the Genotypes listed below write every individual in pedigree 3 that has that genotype (Write then as "I-1, I-2, etc):

Individuals with genotype "AA"

Individuals with genotype "Aa"

Individuals with genotype "aa"

Individuals with genotype "AA" or "Aa"

Individuals with genotype "Aa" or "aa"

9. Look at Pedigree 4. Do you think the trait shown is autosomal dominant or autosomal recessive or something else? List all of the characteristics of the pedigree that justify your answer.

10. Look at Pedigree #5. What does the double line between individuals III-2 and III-3 indicate?

11. What inheritance pattern is present in pedigree #5?

12. Why are III-2 and III-3 in pedigree #5 more likely to have an affected child?