Lab 17. Chromosomes and Karyotypes: How Do Two Physically Healthy Parents Produce a Child With Down Syndrome and a Second Child With Cri Du Chat Syndrome?

Introduction

Mendel's model of inheritance is the basis for modern genetics. This important model can be broken down into four main ideas. First, and foremost, the fundamental unit of inheritance is the gene and alternative versions of a gene (alleles) account for the variation in inheritable characters. Second, an organism inherits two alleles for each character, one from each parent. Third, if the two alleles differ, then one is fully expressed and determines the nature of the specific trait (this version of the gene is called the dominant allele) while the other one has no noticeable effect (this version of the gene is called the recessive allele). Fourth, the two alleles for each character segregate (or separate) during gamete production. Therefore, an egg or a sperm cell only gets one of the two alleles that are present in the somatic cells of the organism. This idea is known as the law of segregation.

It was brilliant (or lucky) that Mendel chose plant traits that turned out to have a relatively simple genetic basis. Each trait that he studied is determined by only one gene, and each of these genes only consists of two alleles. These conditions, however, are not met by all inheritable traits. The relationship between traits and genes is not always a simple one. In this investigation, you will use what you know about the relationship between traits and genes to explain how two children from the same family inherited two different genetic disorders.

The first child is Emily. She was born with Down syndrome. Children with Down syndrome have developmental delays, a characteristic facial appearance, and weak muscle tone. In addition, these children have an increased risk of heart defects, digestive problems such as gastroesophageal reflux, and hearing loss. The second child is Andy, Emily's younger brother. He was born with cri du chat syndrome. Children with cri du chat syndrome have severe physical and mental developmental delays, distinctive facial features, a small head (microcephaly), a low birth weight, and weak muscle tone (hypotonia).

Christopher and Jill Miller are the parents of Emily and Andy and have been married for 15 years. Although the Millers were in their early forties when they had their first child, both of them were in excellent health. They both eat a well-balanced diet and exercise on a regular basis, and they do not smoke. The Millers therefore want to know why their daughter was born with Down syndrome and their son was born with cri du chat syndrome. Here are three potential explanations:

1. Down syndrome and cri du chat syndrome are both recessive genetic disorders. Christopher and Jill Miller each carried a recessive allele for these syndromes, and they each passed it down to their children.
2. Down syndrome and cri du chat syndrome are both caused by a chromosomal abnormality. Either the sperm cell from Christopher Miller or the egg from Jill Miller had a damaged, missing, or additional chromosome.
3. Down syndrome and cri du chat syndrome are both caused by toxins in the environment that alter genes. The children were exposed to these toxins before they were born.

Your Task

Determine which one of these explanations is most valid or acceptable.

The guiding question for this investigation is, **How do two physically healthy parents produce a child with Down syndrome and a second child with cri du chat syndrome?**

Materials

You may use any of the following materials during your investigation:

- Karyotype for Jill Miller
- Chromosome smear for Christopher Miller
- Chromosome smear for Emily Miller (born with Down syndrome)
- Chromosome smear for Andy Miller (born with cri du chat syndrome)
- 1–3 Karyotype placement grids
- Miller family pedigree

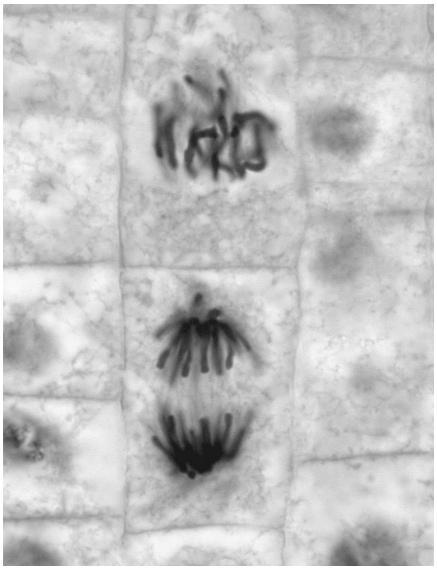
Safety Precautions

1. Wash hands with soap and water after completing this lab.
2. Follow all normal lab safety rules.

Getting Started

Unlike diseases that are transmitted from person to person, such as the flu or strep throat, people are born with cri du chat syndrome or Down syndrome. These syndromes therefore may have a genetic basis. One way to determine the underlying cause of a syndrome with a genetic basis is to produce a karyotype and then look for chromosomal abnormalities that may explain it.

Chromosomes in a cell



A lab technician can create a karyotype by collecting a sample of cells from an individual. The sample of cells is then stained a dye that makes the chromosomes easier to see (see the figure to the right). Next, the chromosomes are photographed using a microscope camera. The pictures of the chromosomes are organized onto a grid by size, shape, and banding pattern. Medical professionals can then use the karyotype to look for chromosomal abnormalities such as a missing chromosome or the presence of too many chromosomes. A chromosomal abnormality can also be found on a single chromosome; for example, a chromosome might be shorter or longer than it should.

To create a karyotype for Christopher Miller and the two children, you will need to sort images of chromosomes taken from their cells according to length, pair any matching sets of chromosomes, and place them onto a grid. The final product is a karyotype (a picture of an individual's chromosomes). Your teacher will provide a karyotype from Jill Miller so you can see what a normal female karyotype looks like.

Your teacher will also provide you with a pedigree for the Miller family. This pedigree will provide you with important information about the extended Miller family. It will also show the members of the Miller family that were born with either Down syndrome or cri du chat syndrome. You can use the pedigree to determine if a recessive gene could have caused one or both of these syndromes.

Investigation Proposal Required? ☐ Yes ☐ No

Connections to Crosscutting Concepts and to the Nature of Science and the Nature of Scientific Inquiry

As you work through your investigation, be sure to think about

- the importance of identifying patterns,
- how scientists attempt to uncover causal mechanisms,
- how structure is related to function in living things,
- how the work of scientists is influenced by social and culural values, and
- the different methods that scientists can use to answer a research question.

Argumentation Session

Once your group has finished collecting and analyzing your data, prepare a whiteboard that you can use to share your initial argument. Your whiteboard should include all the information shown in the figure on the following page.

To share your argument with others, we will be using a round-robin format. This means that one member of your group will stay at your lab station to share your group's argument while the other members of your group go to the other lab stations one at a time to listen to and critique the arguments developed by your classmates.

The goal of the argumentation session is not to convince others that your argument is the best one; rather, the goal is to identify errors or instances of faulty reasoning in the arguments so these

mistakes can be fixed. You will therefore need to evaluate the content of the claim, the quality of the evidence used to support the claim, and the strength of the justification of the evidence included in each argument that you see. In order to critique an argument, you will need more information than what is included on the whiteboard. You might, therefore, need to ask the presenter one or more follow-up questions, such as:

- Is that the only way to interpret the results of your analysis? How do you know that your interpretation of your analysis is appropriate?
- Why did your group decide to present your evidence in that manner?
- Why did your group abandon the other alternative explanations?
- How confident are you that your claim is valid? What could you do to increase your confidence?

Argument presentation on a whiteboard

The Guiding Question:	
Our Claim:	
Our Evidence:	Our Justification of the Evidence:

Once the argumentation session is complete, you will have a chance to meet with your group and revise your original argument. Your group might need to gather more data or design a way to test one or more alternative claims as part of this process. Remember, your goal at this stage of the investigation is to develop the most valid or acceptable answer to the research question!

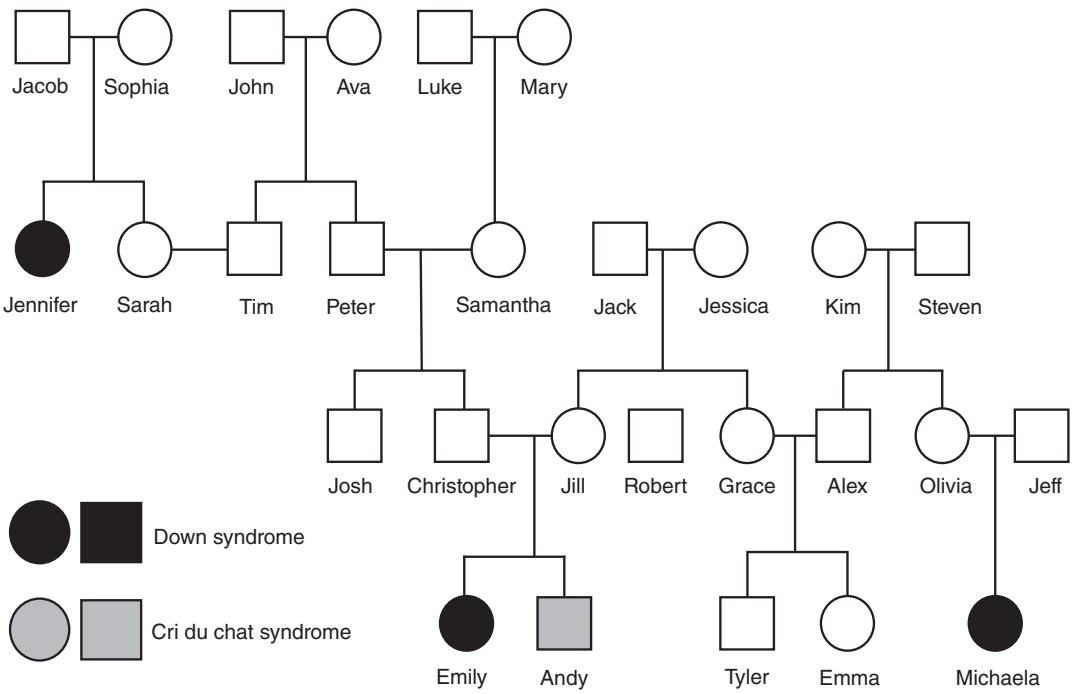
Report

Once you have completed your research, you will need to prepare an investigation report that consists of three sections that provide answers to the following questions:

1. What question were you trying to answer and why?
2. What did you do during your investigation and why did you conduct your investigation in this way?
3. What is your argument?

Your report should answer these questions in two pages or less. This report must be typed, and any diagrams, figures, or tables should be embedded into the document. Be sure to write in a persuasive style; you are trying to convince others that your claim is acceptable or valid!

Miller family pedigree



Lab 18. DNA Structure: What Is the Structure of DNA?

Introduction

We know that genes are made of DNA because scientists were able to demonstrate that DNA and proteins are found in the nucleus of cells, and, more importantly, that DNA (and not protein) is able to transform the traits of organisms. Oswald Avery, Colin MacLeod, and Maclyn McCarty made this discovery in 1944. Their research showed that it is possible to transform harmless bacteria into infectious ones with pure DNA. They also provided further support for their claim by demonstrating that it is possible to prevent this "'transformation" with a DNA-digesting enzyme called DNase.

However, knowing that genes are made of DNA and that DNA is able to store the genetic information of an individual is a little like having a parts list to a 747 jumbo jet. It tells what is important, but it tells you little about how it works. To figure out how DNA works—that is, how it is able to store genetic information—scientists had to figure out its structure. In this investigation, you will duplicate the work of the two scientists who first figured out the structure of DNA—James Watson and Francis Crick.

Your Task

Use the available data to develop a model that explains the structure of DNA.
The guiding question of this investigation is, **What is the structure of DNA?**

Materials

You may use any of the following materials during your investigation:

- Pop beads (DNA kit)
- Fact sheet about DNA

Safety Precautions

1. Safety goggles or glasses are required for this lab.
2. Wash hands with soap and water after completing this lab.
3. Follow all normal lab safety rules.

Getting Started

To answer the guiding question, you will need to develop a model for the structure of DNA. In science, models are explanations for how things work or how they are structured. Scientists often need to develop models to explain a complex phenomenon or to understand the structure of things that are too small to see (such as the structure of an atom or the structure of a molecule of DNA). Scientists use drawings, graphs, equations, three-dimensional representations, or words to communicate their models to others, but scientists only use these physical objects as a way to illustrate the major components of the model.

You will need to create a three-dimensional representation of your model for the structure of DNA using pop beads. Remember that more than one model may be an acceptable explanation for the same phenomenon. It is not always possible to exclude all but one model—and also not always desirable. For example, physicists think about light as a wave and as a particle, and each model of light's behavior is used to think about and account for phenomena differently.

Investigation Proposal Required? ☐ Yes ☐ No

Connections to Crosscutting Concepts and to the Nature of Science and Scientific Inquiry

As you work through your investigation, be sure to think about

- the importance of identifying patterns;
- the importance of examining proportional relationships;
- how the way an object is shaped or structured determines many of its properties or functions;
- how science, as a body of knowledge, changes over time; and
- the different methods that scientists can use to answer a research question.