

Lab 15: Chromosomes & 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 (mother)
- Karyotype for Christopher Miller (father)
- Karyotype for Emily Miller (born with Down syndrome)
- Karyotype for Andy Miller (born with cri du chat syndrome)
- Miller family pedigree

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.

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 figure to the right). Next, the chromosomes are photographed using a microscope camera. The pictures of the chromosomes are organized on to 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 analyze the karyotypes for Christopher Miller and the two children, you will need to look closely at images of chromosomes taken from their cells according to length, then paired with matching sets of chromosomes, and placed onto a grid. The final product is a karyotype (a picture of an individual's chromosomes). Your teacher will provide karyotypes from the Miller family.

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.

Chromosomes in a cell



Report

Once you have completed your research, you will need to prepare an investigation report that consists of four sections (be sure to have section headings):

1. Introduction: Give some background information on the topic. Explain what question you were trying to answer and include a hypothesis. (Background info, research question and hypothesis)
2. Procedure: What did you do during your investigation and why did you conduct your investigation in this way? (How you collected and analyzed data)
3. Data: Include a data table and/or graph to show your results. Be sure to include a title for your table or graph with labels for the variables.
4. Conclusion: What is your argument? (Claim - Evidence - Reasoning)

Your report should answer these questions in two pages or less. The report must be typed, and any diagrams, figures, or tables should be embedded into the document. Type your report on Google Docs (12 point font, double-spaced) and share it with your teacher. Your report will be graded based on the rubric in the class syllabus.