Names _____ Period _____

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

LabHandout

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 deter- mines 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:

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.

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.

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 the three 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:

Karyotypes for Jill and Andy Miller (parents)

Karyotype for Emily Miller (born with Down Syndrome)

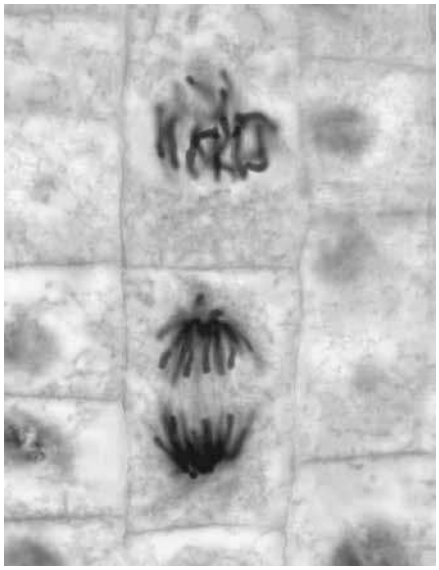
Karyotype for Andy Miller (born with Cri Du Chat Syndrome)

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 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. 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.

Chromosomes in a cell



Your teacher will provide a karyotype from Jill, Christopher, Emily and Andy so you can compare each person's karyotype.

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 (explanation 1).

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 opposite page.

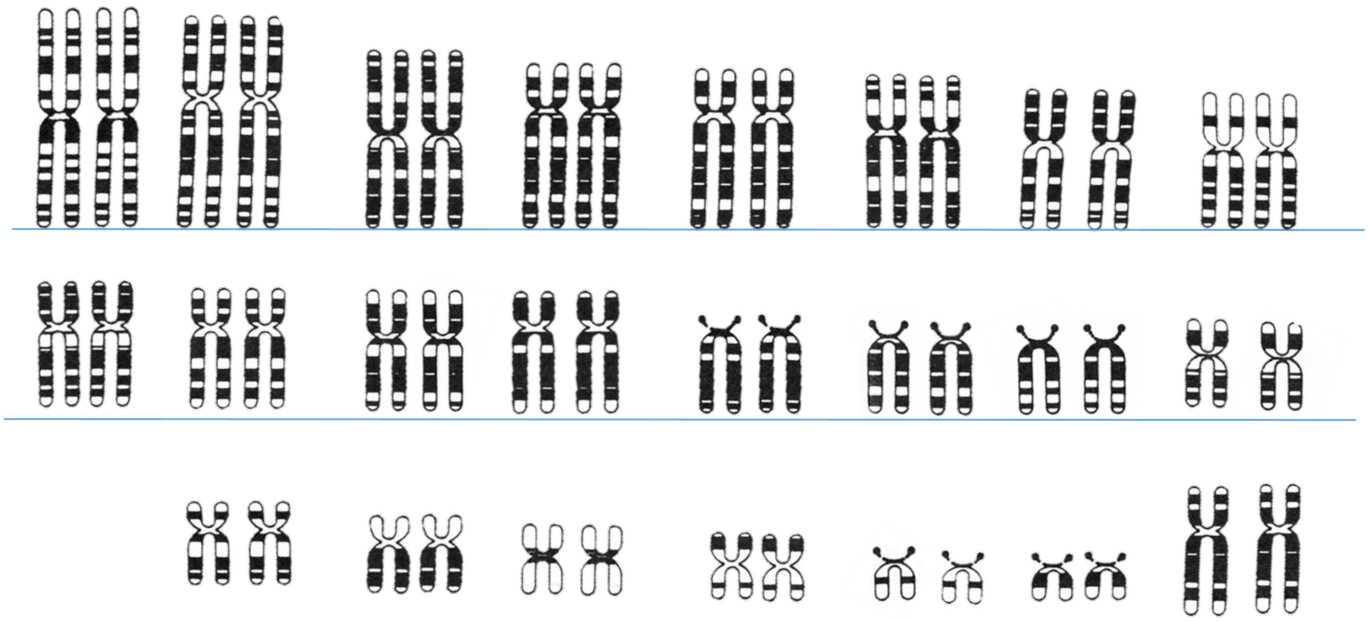
To share your argument with others, we will be using a round-robin format. This means

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.

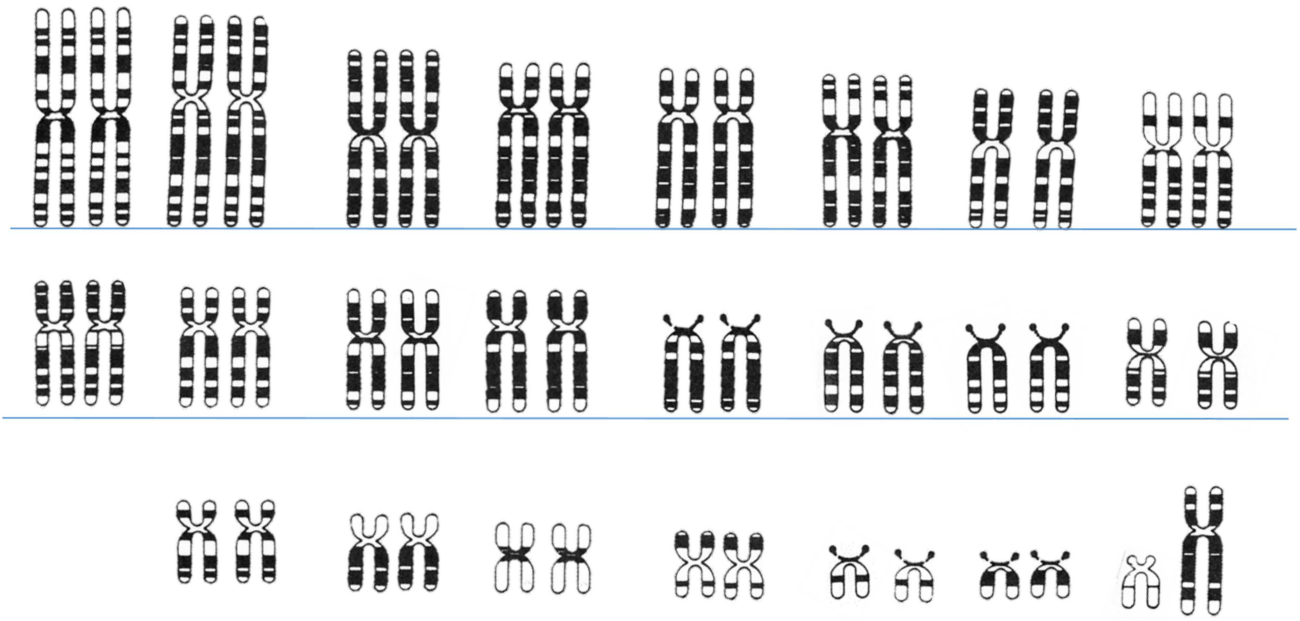
Once the argumentation session is complete, you will have a chance to meet with your group and revise your original argument. Your group might need to gather more data or design a way to test one or more alternative claims as part of this process. Remember, your goal at this stage of the investigation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

Jill Miller Karyotype



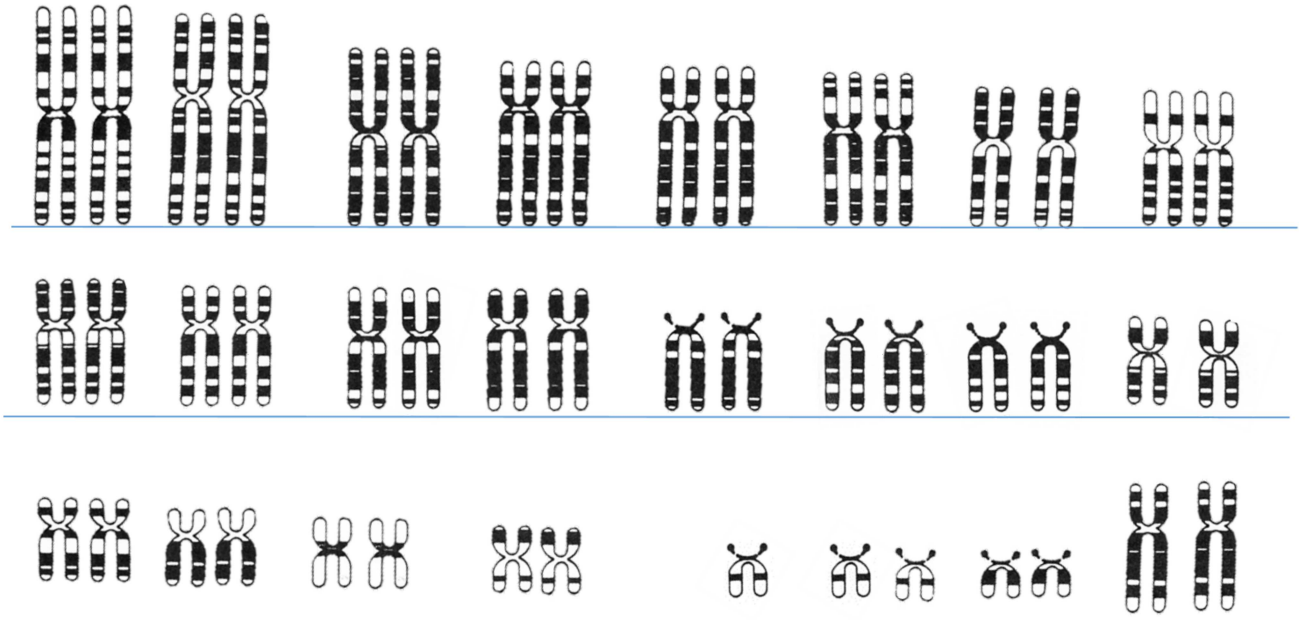
Normal
Female

Christopher Miller Karyotype



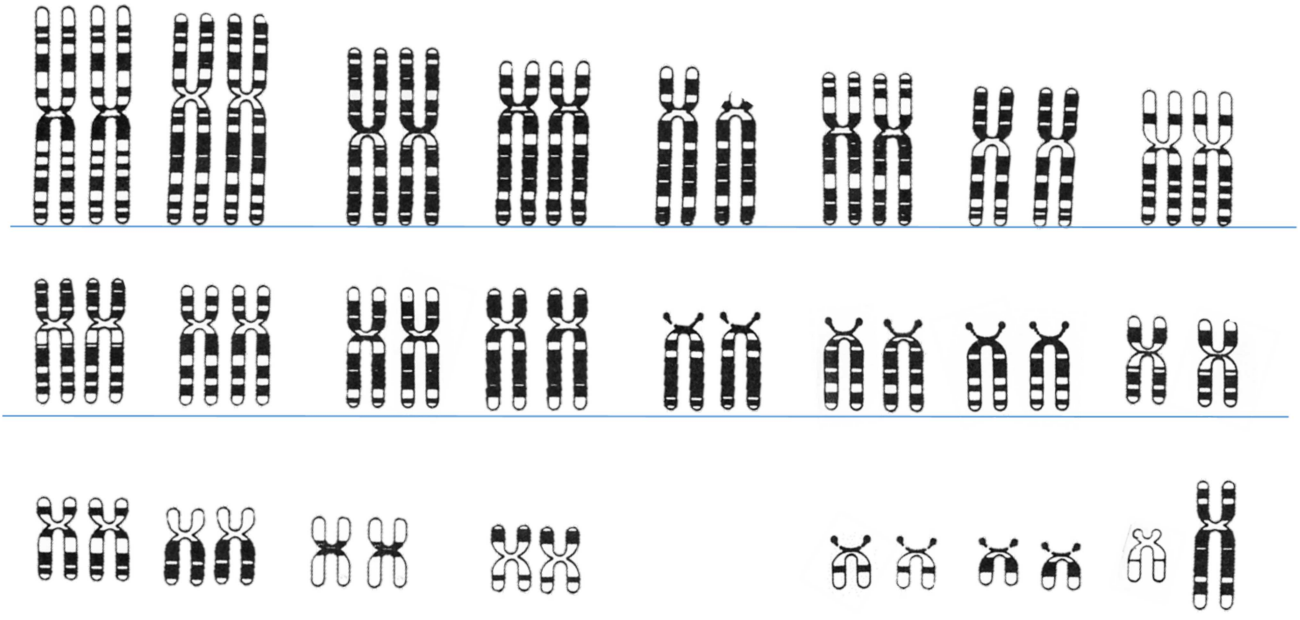
Normal
Male

Emily Miller



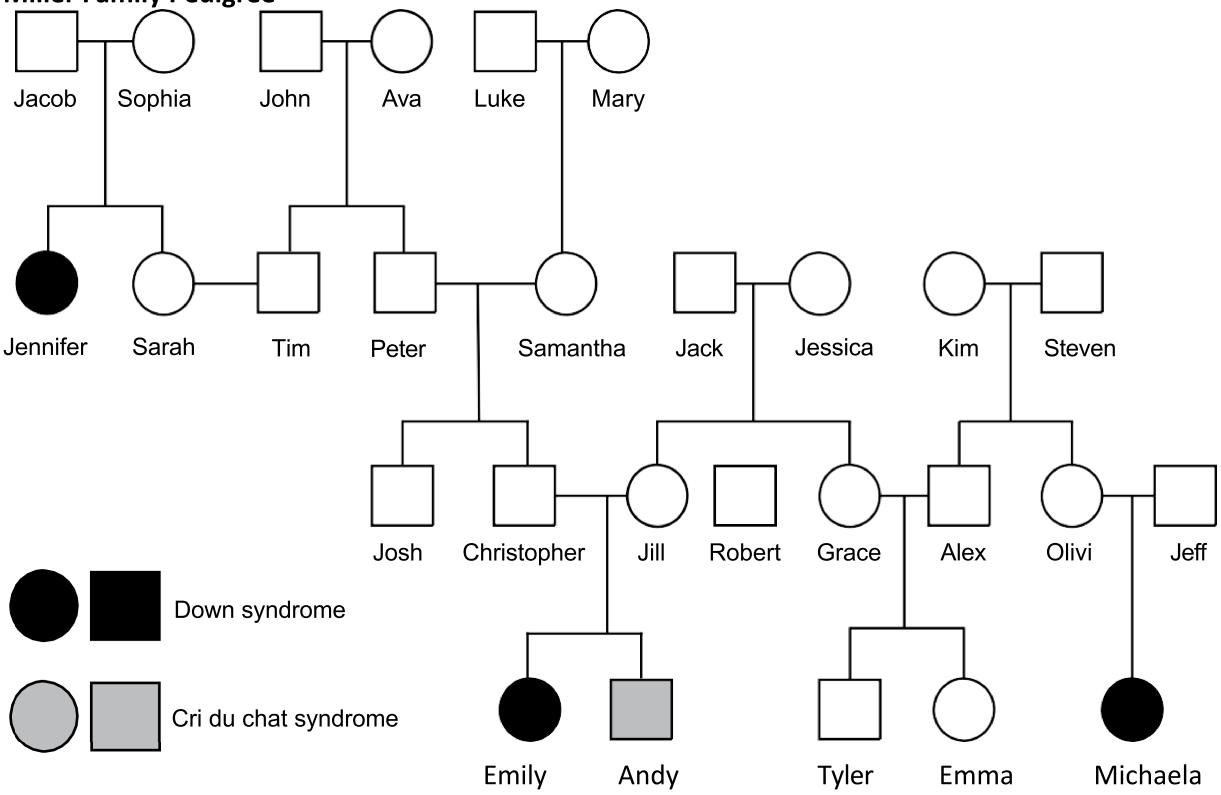
Down's
Female

Andy Miller



Cri du
Chat

Miller Family Pedigree



Lab Questions:

1. Looking at Jill's Karyotype:

Is this karyotype from a male or a female?

Male
Female

How do you know?

2. Looking at Christopher's Karyotype:

Is this karyotype from a male or a female?

Male
Female

How do you know?

3. Looking at Emily's Karyotype:

What is different between her karyotype and Jill and Christopher(normal):

Does Emily person have Down syndrome?
(Circle answer)

Yes
No

How do you know? What caused her Down Syndrome?

4. Looking at Andy's Karyotype:
What is different between his Karyotype and Jill and Christopher (normal):

Does Andy have Cri Du Chat?
(circle answer)

Yes
No

How do you know? What caused his Cri du chat syndrome?