INQUIRY & INVESTIGATION

Exploring Contemporary Issues in Genetics & Society: Karyotyping, Biological Sex, & Gender

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Abstract

In this two-part activity, high school biology students examine human karyotyping, sex-chromosome-linked disorders, and the relationship between biological sex and gender. Through interactive simulations and a structured discussion lab, students create a human karyotype and diagnose chromosomal disorders in hypothetical patients, as well as formulate a scientifically based argument distinguishing biological sex from gender. This activity reinforces biology concepts such as DNA structure and function, meiosis, and the inheritance of traits. In their arguments, students integrate prior knowledge with science content. This lesson connects the world of science with issues important in their own lives.

Key Words: *Genetics; karyotyping; sex-chromosome-linked disorders; gender; biological sex.*

Recently, science education has undergone another wave of reform intended to further strengthen the scientific literacy of all students. This reform movement, articulated in the National Research Council's (2012) report *A Framework for K–12 Science Education* and subsequently in the drafting of the *Next Generation Science Standards* (http://www.nextgenscience.org), expects students to meaningfully engage in the practices of science through authentic activities. To facilitate such a change, a set of performance expectations accompanies these new standards. Now more than ever, students will be held accountable for using models to explore concepts, constructing arguments from evidence, and communicating scientific information. This two-part activity develops high school biology students' science practices as they examine human karyotyping, sex-chromosome-linked disorders, and the relationship between biological sex and gender. Additionally, the activity has the potential to bolster the scientific literacy of all students by involving them in the study of science that directly connects with their interests and aligns with multiple national standards.

Karyograms are especially useful in determining instances of genetic abnormalities.

Background

Cytogeneticists use biotechnology tools such as a karyogram (a photomicrograph of the chromosomes in a cell) to study specific chromosome features like quantity and structure. This compilation of chromosomes is called a *karyotype* (Figure 1). In assembled karyotypes, chromosomes are ordered according to their size, banding pattern, and centromere position (http://www.ncbi.nlm.nih.gov). Karyograms are especially useful in determining instances of genetic abnormalities and, thus, can be used as an engaging way for students to explore multiple genetics concepts contained in national science education standards. For example, Turner syndrome (XO), the only known survivable case of monosomy (one copy of a chromosome), can be diagnosed through karyotyping by the absence of a second X or Y sex chromosome and serves as a real-life basis for studying nondisjunction (when chromatids fail to separate during meiosis I or II). Students can use sex-linked disorders like Turner syndrome to explore the ways in which nondisjunction mutations affect the functioning of an organism.

Sex-chromosome-linked disorders are those that occur on the sex chromosomes (X and Y chromosomes). Sex-linked disorders vary greatly in their symptoms and resulting complications. Disorders like Turner and Trisomy X syndromes can cause infertility and other complications (e.g., developmental delays), whereas other conditions have much subtler symptoms and often go unnoticed. For example, many individuals with Jacob's syndrome (47, XYY) exhibit phenotypes (physical appearance) that are considered normal and have normal sexual development. Thus, many affected individuals are unaware that they have this disorder, though the syndrome occurs in ~1 in every 1000 newborn males (Klug & Cummings, 2000).

The biological sex of an individual is often determined by the presence of specific sex chromosomes (XX for females, XY for males) and reproductive organs. However, classifying biological sex as a binary

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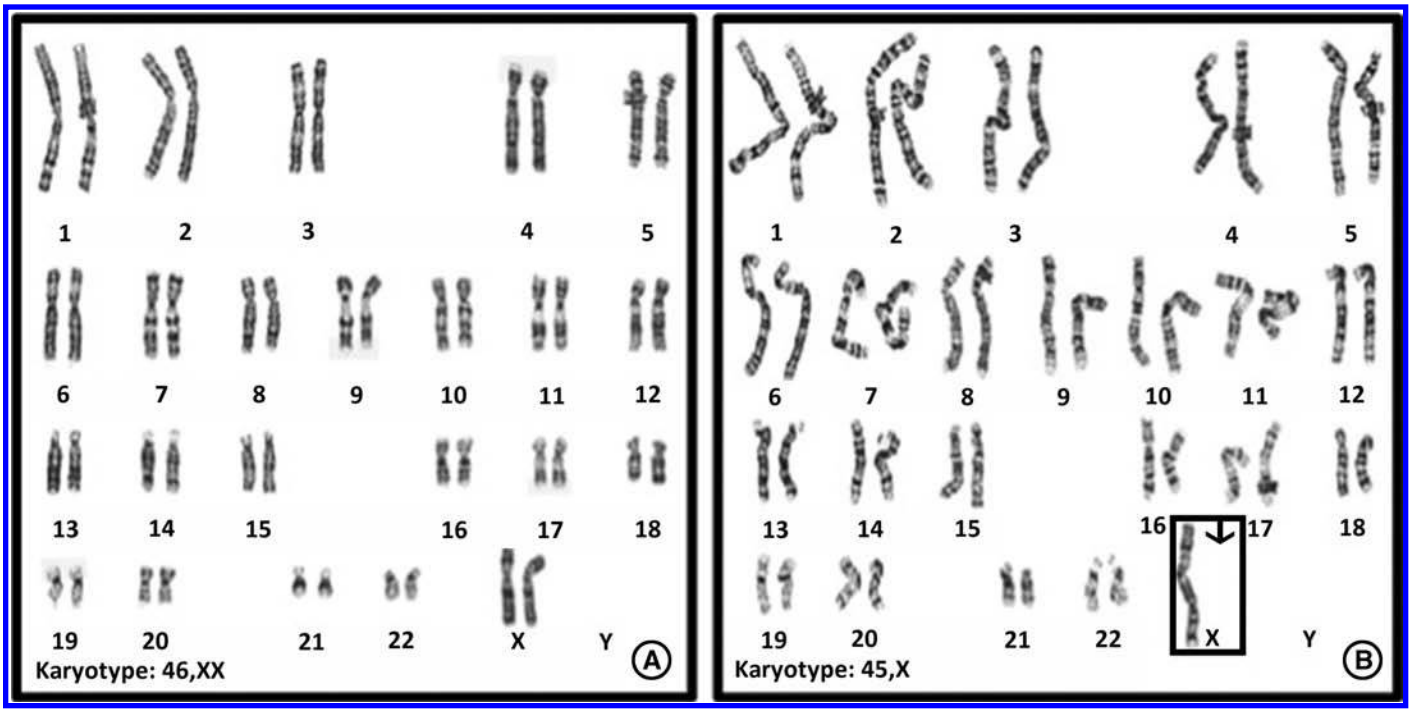

Figure 1. Comparison of (A) a typical human female karyotype with (B) the karyotype of an individual with Turner syndrome. Note the absence of a second sex chromosome in B. (Images from http://www.biology.iupui.edu/biocourses/N100/2k2humancsomaldisorders.html.)

(i.e., male–female) is often problematic because not all individuals adhere to these norms. For example, the SRY gene (sex-determining region of the Y chromosome) is characteristic in males and leads to testis formation. Yet it is plausible for a male to carry the female genotype (genetic composition) XX but still be considered male, because one X chromosome contains a copy of this SRY gene (http://www.ncbi.nlm.nih.gov/books/NBK22246/). Moreover, Klinefelter syndrome (47, XXY), the result of nondisjunction of the X chromosome during meiosis I, produces simultaneous male and female sexual development in individuals. Yet, outwardly, people with Klinefelter often exhibit many phenotypic characteristics associated with being male – for example, tall height and broad shoulders. Hence, the terms *sex* and *gender* are often used interchangeably, despite socially recognized differences between the two.

Social scientists contend that gender includes the ways individuals act and look. Moreover, gender is taught through the process of socialization. Thus, gender is driven by socially determined norms, such as females wearing makeup or males playing contact sports. These actions produce different feelings, relationships, and skills in individuals. Although it may be socially acceptable to view gender as a binary (male–female, boy–girl), this is problematic and marginalizes those who do not fit neatly into one gender category (Lorber, 2010).

Sex-chromosome-linked disorders like Klinefelter syndrome provide a valuable forum for the examination and subsequent discussion of biological sex and gender in the science classroom, allowing students the space and voice to examine connections between science and their lives. Individuals with sex-chromosome-linked disorders often have symptoms and complications that place them outside socially accepted gender norms (e.g., infertility, breast development in males). This ultimately leads to questions like "What does it mean to be a girl or boy?" – about which many students are simply unsure. Such questions may seem trivial to individuals who fit neatly into either gender label, but for students who do not, these are questions of paramount importance, with real consequences for not fitting neatly into those categories.

Karyotyping functions as a simple diagnostic tool in the medical fields and can also be used to reinforce multiple abstract genetics concepts such as the molecular basis of heredity and inheritance patterns. Yet, because the process is labor intensive, traditional karyotyping cannot easily be conducted in the science classroom (Baker & Jones, 2006). The following activity provides an engaging way to bridge the scientific and social worlds of students while maintaining rigorous study of introductory genetics concepts. It is appropriate for a range of students and can easily be adapted to serve the academic demands of multiple high school biology classes. I have used this particular activity with 9th- and 10th-grade introductory and honors biology classes and taught it midway through a genetics unit. To most effectively teach this lesson, students should already be familiar with biology concepts such as meiosis and the relationship between chromosomes and DNA function. For instance, prior to this activity, students spend several class periods learning about the fundamentals of meiosis through lecture, readings, and creating models. Additionally, students have investigated relationships among DNA, chromosomes, protein coding, and trait expression.

○ Procedure

The objectives for this two-part activity are to engage students in the practices of science, particularly (1) modeling a hypothetical human karyotype by using shape, centromere position, and banding patterns to arrange chromosomes; (2) using simulations to explore

ways in which DNA sequence mutations can alter gene function and phenotype; (3) using information contained within karyotypes to diagnose sex-chromosome-linked disorders; (4) constructing a scientifically based argument distinguishing biological sex and gender; and (5) making inferences about the ways in which science and society uphold these constructs. To facilitate student-centered active learning and complex thinking, this activity has been designed from the 5-E lesson-planning format (Bybee et al., 2006). 5-E lesson plans are divided into distinct phases intended to support science as inquiry: Engage, Explore, Explain, Elaborate, and Evaluate. Activity components will be described according to these phases in the following section. This entire activity occurs over four 50-minute class periods.

○ Materials & Resources Needed

One per student. **Note:** All worksheets listed below are available at http://plaza.ufl.edu/brownjc/.

- Computers with Internet access
- *Human Karyotyping* worksheet
- *Sex-Chromosome-Linked Disorders* homework sheet
- *Images of Gender* worksheet

○ Part I: Human Karyotyping and Sex-Linked Disorders

Over the course of two 50-minute class periods, this first lesson introduces students to human karyotyping and sex-chromosome-linked disorders. Students use interactive simulations to model hypothetical human karyotypes in addition to identifying and diagnosing sex-linked disorders. This lesson also provides rich opportunities for students to explore the various ways in which information stored in DNA mediates the function and expression of genes.

Engage

During a whole-class Q&A session, students first review concepts such as DNA structure and meiosis. I also elicit students' relevant prior knowledge at this time, through questions such as "What are some ways to determine whether or not a person has a chromosomal disorder?" To assess any changes in students' ability to make a scientifically based argument over time, I also have them argue for or against the statement "There is no difference between your biological sex and gender," providing evidence to support their assertion. Responses reveal much about students' initial conceptions and the connections they make between science and themselves. Take, for example, a 9th-grade male student's response, reproduced as Figure 2.

Explore

In the Explore phase, teams of two or three students complete the Human Karyotyping worksheet as they interact with simulations from the Learn.Genetics and Explore Learning websites (http://learn.genetics.utah.edu and http://www.explorelearning.com) and visit the National Center for Biotechnology Information (NCBI) website (http://www.ncbi.nlm.nih.gov/) to garner information about the molecular basis of heredity, including mechanisms through which genetic information is transferred from parent to offspring (Figure 3). The Learn.Genetics site, constructed and maintained by the University of Utah, provides many high-quality teacher and student resources to facilitate the learning of genetic concepts, all free of charge. At this site, students arrange chromosomes according to their distinct size, centromere position, and banding patterns, ultimately constructing a hypothetical human karyotype. However, because this site only lets students interact with a normal male karyotype, I also have them visit Explore Learning to identify karyotypes of individuals with various chromosomal conditions, including sex-chromosome-linked disorders. Although the Explore Learning website is commercially available, teachers who choose not to purchase a license can still access the Human Karyotyping simulation as a guest for 5-minute trial sessions. Because this brief access time can be problematic, I also recommend using the karyotyping exercise available through The Biology Project created by the University of Arizona as an alternative (http://www.biology.arizona.edu/human_bio/activities/karyotyping/karyotyping.html). This exercise allows students to arrange and analyze the karyotypes of three hypothetical patients, ultimately diagnosing chromosomal disorders. Although not all of the patients' disorders are sex-linked, this activity engages students in additional practice arranging homologous chromosomes according to their distinct characteristics, interpreting completed karyotypes, and diagnosing conditions.

After becoming familiar with karyotyping basics, students then visit the Chromosome Map at the NCBI website (http://www.ncbi.nlm.nih.gov/books/NBK22266/) to record pertinent information about three chromosomes of their choice, including approximate number of genes on the chromosome, any diseases associated with each chromosome, and specific disease characteristics. Students have told me that visiting the NCBI site helped them think about science topics in concrete ways, because they often know or have heard of someone with a chromosomal disease. The NCBI site offers vast, credible stores of information and can be used to increase the rigor of this activity for advanced biology classes. For example, students can continue exploring the Chromosome Map to identify genes that are involved in regulatory or structural functions versus protein coding, as well as speculate on environmental and heritable factors that influence genetic mutations such as those associated with cancer.

Explain

There are two ways in which I typically facilitate the Explain phase: the first is through a team check-in and the second is through a small-group debriefing. Over the course of each class period, we

> "Sex is what you are on the inside. Gender is what you look like on the outside. This is because you are either a male or a female. Sex has to do with your chromosomes and gender is like what you see."

Figure 2. A ninth-grade male student's original argument response to the statement, "There is no difference between your biological sex and gender."

* Log onto www.explorelearning.com then select the ***Human Karyotyping*** Gizmo.

* A chromosomal disorder occurs when a person's cells do not have the correct number of chromosomes. The table below lists three common chromosomal disorders.

Disorder Name	Description of Disorder	Subject (Circle C, D, or E)	Symptoms (List 1-2 from DIAGNOSIS)
Down syndrome	Extra Chromosome 21	C D E	
Klinefelter syndrome	Extra X in male (XXY)	C D E	
Turner syndrome	Single X in female (XO)	C D E	

NCBI

* Visit the National Center for Biotechnology Information (NCBI) ***Chromosome Map*** at www.ncbi.nlm.nih.gov/books/NBK22266/.

*Compare the ***Karyogram of a human male*** map with Subject A's karyotype from the Gizmo.

Chromosome #	Approx. # of Genes	Disease associated with chromosome	One characteristic of the disease*

Figure 3. Snapshots of the Human Karyotyping worksheet (http://plaza.ufl.edu/brownjc).

have a midpoint team check-in where I assess group progress. During this time, student teams report to me (1) their current progress on the Human Karyotyping worksheet; (2) one thing they have learned toward the lesson objectives (see Procedure); and (3) one thing they are struggling with. Because students are accustomed to this practice, it typically takes no more than 1 minute per group, or a maximum of 6–8 minutes. Yet it provides me with invaluable assessment information, and I can then visit each group with a personalized "trouble-shooting" plan. During the small-group debriefing sessions, students answer objectives- and standards-aligned questions such as "What information is contained within chromosomes?" and "How are viable mutations inherited?" Students then share their answers with the whole class, which provides great fodder for an in-depth discussion of genetics-based content and often leads to questions that can be answered through the next activity.

○ Part II: Sex-Chromosome-Linked Disorders, Biological Sex, & Gender

In this structured discussion lab, students connect biology content learned in the previous activity with the concepts of biological sex and gender. Student teams construct a scientifically based argument in which they distinguish biological sex from gender, drawing from related biology concepts as evidence to support their assertions.

Elaborate

In preparation for this portion of the activity, which also typically occurs over two class periods, students first complete a Sex-Chromosome-Linked Disorders homework assignment. Through this assignment students learn about the causes, risk factors, symptoms, and treatment of four sex-linked disorders. In addition, fictional accounts of an individual living with each condition are provided to make concepts more concrete. Although this homework assignment is not required to participate in the next day's activity, students become so engaged that I often experience 100% homework completion rates with my classes.

I begin by posing select reflection questions from the homework assignment to gauge student understanding. I also take this time to review content from Part I. Next, I project several images of individuals and ask students to use the Images of Gender worksheet to record their observations and decide on each individual's gender. Students have listed attributes such as breasts, long hair, and long fingernails under the *female* category, whereas the *male* category typically contains attributes such as abdominal muscles, broad shoulders, and facial hair. At this point, students individually complete an "identity circle" in the Images of Gender worksheet where they list words or phrases that describe elements of their identity (http://www.nsrfharmony.org/protocol/doc/paseo.pdf) (Figure 4). This particular activity feature allows students space to share their backgrounds and also sets the stage for connecting sensitive topics, such as gender, to science content while constructing an argument. As an extension, students can further support their arguments through age-appropriate content on sociological topics like gender. Teaching Tolerance (http://www.tolerance.org) is one example of a website that offers a wealth of credible classroom resources connecting science instruction to such topics. Activities and readings are organized by grade level and subject area, making resource selection easier.

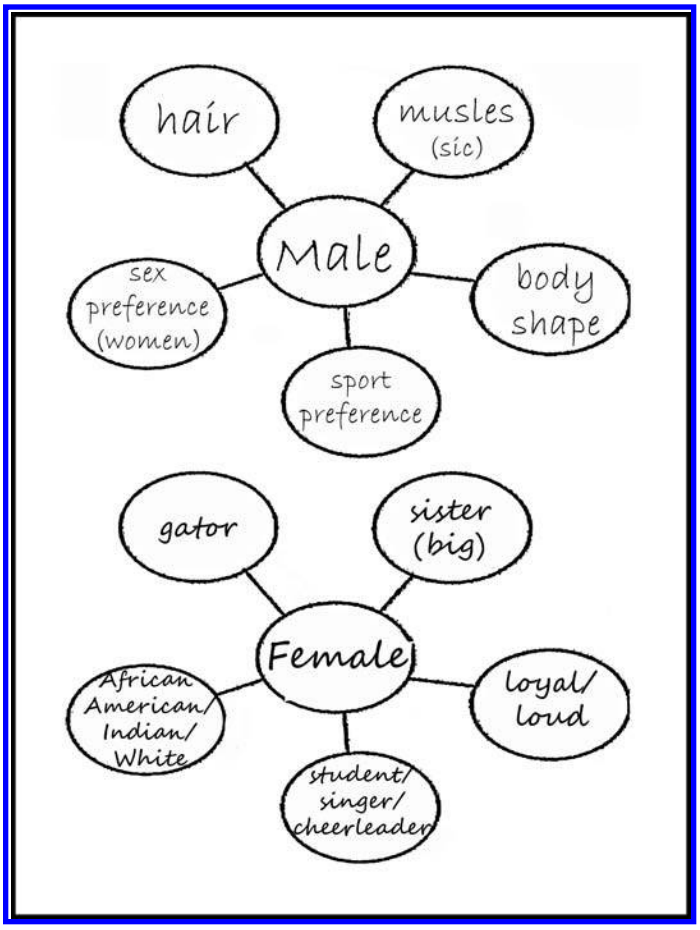

Figure 4. Complete male (above) and female (below) student "identity circles."

Students then return to their Part I teams and have a structured discussion around questions such as these:

- Imagine your cousin decided to undergo a surgical procedure to change his sex. Does his gender change after the surgery? Does his sex change? Use evidence from the Sex-Chromosome-Linked Disorders homework handout to support you answer.
- Are individuals with sex-linked disorders less female or less male because of their disorder? Use evidence from the Human Karyotyping worksheet to support your answer.

To best facilitate this discussion, I structure it by assigning students roles (e.g., record keeper, clarifier), providing explicit directions for participation (e.g., "Treat each other with respect"), and requiring a format for questions and responses. Specifically, once a question has been posed, group members are allowed 1 minute to think about their response. When students respond, they are allowed to do so without interruption. This format is intended to ensure that each person is fully listening to teammates during dialogue. Because this content can be sensitive, students do not have to share if they do not feel comfortable. They are responsible, however, for recording group members' responses on the Images of Gender worksheet. It has been my experience that students talk openly in their groups, and some have expressed to me that this activity connects science to their lives. After students complete the discussion, they use evidence from their Human Karyotyping and Images of Gender worksheets to construct a group argument distinguishing biological sex from gender. This activity concludes with a combination of small-group and whole-class discussions in which the groups first have 2 minutes to present their arguments, and then we open the floor to questions and comments. Although groups are held responsible for citing evidence from this particular activity only, they often refer to biology concepts previously learned in their argument, including cell cycle and meiosis. Figure 5 shows the same 9th-grade male student's revised argument to the statement "There is no difference between your biological sex and gender."

> "We disagree that biological sex is the same as gender because someone's gender comes from how other people see them, but their biological sex results from the sex chromosomes they inherited from their parents. We see gender as something that other people also decide and that you can change. Someone can have one biological sex and a completely different gender. Like someone with Turner syndrome. They have only one sex chromosome because of an error during meiosis when the chromosomes did not separate and this leads to aneuploidy or a lack of chromosomes. So, the person's biological sex would be most likely female because the X is the only chromosome she has. But she might dress and think of herself like a man, so her gender would be male but her sex is female."

Figure 5. A ninth-grade male student's revised argument response to the statement, "There is no difference between your biological sex and gender" after completing Parts I and II of the Human Karyotyping and Sex-Chromosome-Linked Disorders activity.

Evaluate

Both individual and group understanding can be assessed during this activity. I have used a rubric to grade individually completed assignments such as the Sex-Chromosome-Linked Disorders homework, in which I evaluate responses on the basis of how thoroughly they answered each question. When assessing individual and group understanding in the Human Karyotyping activity, as well as when the groups present their final arguments, I have found it helpful to also use the Knowledge Integration framework (Linn & Elyon, 2006). This scoring rubric gauges the degree to which a student response connects scientifically accepted ideas as they justify a claim or explain a concept. These evaluation methods allow me to remain consistent in my grading among the different classes.

○ Conclusion

In this two-part activity, high school students examine human karyotyping, sex-chromosome-linked

disorders, and the relationship between biological sex and gender. Considering that high school is a time when students often desire to conform to socially imposed norms, I have found that giving students opportunities to connect sensitive topics like gender to science concepts is of utmost importance. This activity occurs midway through a genetics unit, after students have learned concepts such as DNA function and meiosis. The activity content reinforces multiple science ideas and sets the stage for an in-depth study of the processes of Mendelian inheritance and gene expression. Moreover, it contextualizes abstract genetics concepts within students' lives, thereby creating meaningful learning experiences (Basu & Calabrese Barton, 2007; National Research Council, 2012). Additionally, students take the lead as they engage in several science practices, including using models, constructing an argument from evidence, and communicating scientifically and socially based information, thereby bolstering their science literacy. Though strengthening science proficiency is always a goal of high-quality instruction, students also benefit when bridges are built between their lives and the world of science, propelling their curiosity beyond classroom walls.

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