Teacher Notes for Genetics and Probability – Sex Ratios of Births[1]

In this minds-on analysis and discussion activity, students analyze the inheritance of sex chromosomes. Students use a Punnett square to predict the sex ratio of births and compare their prediction to data for individual families and for the entire US. As students analyze the reasons why many real families deviate from Punnett square predictions, they learn about the probabilistic nature of inheritance and the limitations of Punnett square predictions.

Before beginning this activity, your students should have a basic understanding of meiosis, fertilization, and Punnett squares. For this purpose, I recommend:
- "Understanding How Genes Are Inherited via Meiosis and Fertilization" (https://serendipstudio.org/exchange/bioactivities/meiosisRR%20)
- "Introduction to Genetics – Similarities and Differences between Family Members" (https://serendipstudio.org/exchange/bioactivities/geneticsFR)

Learning Goals
In accord with the Next Generation Science Standards:[2]
- This activity helps to prepare students for the Performance Expectation, HS-LS3-3, "Apply concepts of statistics and probability to explain the variation and distribution of expressed traits in a population."
- Students engage in two Scientific Practices:
  - Developing and Using Models: "Develop and/or use a model… to predict phenomena, analyze systems,…"
  - Constructing Explanations: "Apply scientific ideas, principles, and/or evidence to provide an explanation of phenomena…"
- This activity provides the opportunity to discuss two Crosscutting Concepts:
  - Systems and System Models: Models can be used "to predict the behavior of a system, [but] these predictions have limited precision and reliability due to the assumptions and approximations inherent in the models".
  - Cause and Effect: Students "suggest cause and effect relationships to explain and predict behaviors in complex natural and designed systems. They also propose causal relationships by examining what is known about smaller scale mechanisms within the system".

Additional Content Learning Goals
- The behavior of chromosomes during meiosis and fertilization provides the basis for understanding the inheritance of genes.
- The processes of meiosis and fertilization can be summarized in Punnett squares which can be used to predict the genotypes and phenotypes of offspring.
- Each fertilization event is independent of other fertilization events, so the genetic makeup of each child is independent of the genetic makeup of any siblings.
- Quantitative predictions from Punnett squares are accurate for large samples, but random variation in the genetic makeup of the sperm and egg that unite to form each zygote often results in substantial discrepancies between the Punnett square predictions and the outcomes observed in small samples such as individual families.

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[1] By Dr. Ingrid Waldron, Dept Biology, Univ Pennsylvania, © 2024. These Teacher Preparation Notes and the related Student Handout are available at https://serendipstudio.org/exchange/bioactivities/geneticsSRB
[2] http://www.nextgenscience.org/sites/default/files/HS%20LS%20topics%20combined%206.13.13.pdf and http://www.nextgenscience.org/sites/default/files/Appendix%20G%20-%20Crosscutting%20Concepts%20FINAL%20edited%204.10.13.pdf

This activity will help to counteract this common misconception:
- Students often fail to recognize the probabilistic nature of Punnett square predictions and inheritance.

Instructional Suggestions and Background Biology

To maximize student learning, I recommend that you have your students work in pairs to complete groups of related questions. Student learning is increased when students discuss scientific concepts to develop answers to challenging questions. After students have worked together to answer each group of related questions, I recommend having a class discussion that probes student thinking and helps students to develop a sound understanding of the concepts and information covered.

If your students are learning online, I recommend that they use the Google Doc version of the Student Handout available at https://serendipstudio.org/exchange/bioactivities/geneticsSRB. To answer question 1, students can either print the relevant page, draw on it and send pictures to you, or they will need to know how to modify a drawing online. To answer online, they can double-click on the relevant drawing in the Google Doc to open a drawing window. Then, they can use the editing tools to answer the question.[3]

You may want to revise the Word document or Google Doc to prepare a version of the Student Handout that will be more suitable for your students. If you use the Word document, please check the format by viewing the PDF.

If you would like to have a key with the answers to the questions in the Student Handout, please send a message to iwaldron@upenn.edu. The following paragraphs provide additional instructional suggestions and background information.

The Student Handout omits several complexities. For example, actual sex ratios at birth deviate slightly from the Punnett square prediction.[4] Slightly more males than females are born (51.2% males in the US in 2000, slightly lower for African-Americans and slightly higher for Asian-Americans). This slight deviation from the Punnett square model may be the result of higher mortality for female embryos and fetuses.

The Y chromosome contains the SRY gene, which stands for Sex-determining Region of the **Y** chromosome. If a zygote has a **Y** chromosome with the SRY gene, the embryo will develop testes and male anatomy; if a zygote does not have a **Y** chromosome with the SRY gene, the embryo will develop ovaries and female anatomy. The SRY gene codes for a protein that binds to regulatory DNA and activates multiple genes that stimulate the gonads to develop into testes instead of ovaries. The testes secrete testosterone and other chemical messengers that stimulate the genitalia to develop into penis, scrotum, vas deferens, etc. In the absence of the SRY gene,

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[3]To insert text
1. At the top of the page, click Insert.
   - To place text inside a box or confined area, click Text Box and drag it to where you want it.
2. Type your text.
3. You can select, resize and format the word art or text box, or apply styles like bold or italics to the text. When you are done, click Save and Close.

[4] In general, we cannot extrapolate from Punnett squares to the percent of all babies with specific genotypes unless we know the prevalence of each allele in the reproducing population. For inheritance of sex chromosomes, we can extrapolate from the Punnett square to the percent of male babies in large samples, because we know that every mother has two X chromosomes and every father has an X and a Y chromosome.

the gonads develop into ovaries, and in the absence of testosterone the genitalia develop into clitoris, labia, uterus, etc.; this results in the female anatomy of an **XX** baby. Rarely, the **Y** chromosome does not have an SRY gene, which results in an **XY** baby with female anatomy. Also rarely, an **X** chromosome has an SRY gene, which results in an **XX** baby with male anatomy.[5]

It should be noted that the **X** chromosome has many crucial genes that are not found on the Y chromosome. Therefore, a zygote must have at least one **X** chromosome to survive and develop into an embryo.

The data in the table on the top of page 2 of the Student Handout are for the 36 people in the 12 nuclear families in three generations of descendants of a woman who was born in the early twentieth century. These data illustrate that a Punnett square does not reliably predict the outcome for any individual family.

Discussion of random variation will help your students to reconcile their experience of variation in outcomes in real world families with the predictions of Punnett squares in the classroom. Random variation almost always averages out in large samples, so the predictions of the Punnett square model are more accurate for large samples.

These analyses illustrate both:
- the usefulness of the Punnett square model of inheritance (predicting the percent male in large samples of children and the probability that a child will be male) and
- the limitations of the Punnett square model (not accurately predicting the makeup of individual families or the sex of a specific child, both of which vary due to random variation in which sperm fertilizes an egg).[6]

At the end of this activity, I suggest that you discuss the Crosscutting Concept that models can be useful "to predict the behavior of the system, [but] these predictions have limited precision and reliability due to the assumptions and approximations inherent in models". A model is a simplified representation of reality that highlights certain key aspects of a phenomenon and thus helps us to better understand and visualize the phenomenon. Many students tend to think of a model as a physical object and may not understand how a Punnett square, chemical equation or

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[5] Additional genes on multiple chromosomes contribute to the normal development of male and female reproductive organs. Defects in these genes can lead to anomalies in the development of male or female reproductive organs, e.g. due to defective hormone receptors or defective enzymes to produce hormones. Examples are:
- Androgen Insensitivity Syndrome results from lack of functional molecular receptors for testosterone and dihydrotestosterone. Due to the lack of these molecular receptors, testosterone and dihydrotestosterone do not affect the cells in the fetal genitalia of an XY fetus with Androgen Insensitivity Syndrome, so female external genitalia develop. These individuals are raised and live as females, but they have testes instead of ovaries. They are infertile. This syndrome is typically detected when a teenage female fails to menstruate.
- Congenital Adrenal Hyperplasia (also called Adrenogenital Syndrome) develops when an enzyme needed to produce cortisol is defective or missing, resulting in abnormal hormonal feedback which leads to excessive production of androgens by the adrenal cortex. The elevated androgen levels in a XX fetus result in varying degrees of masculinization of the external genitalia. As a result, the baby's sex may appear ambiguous or even be mistaken for male.

For a more complete account of differences in sexual development in individuals who are 46XY, see https://www.ncbi.nlm.nih.gov/books/NBK279170/. Other anomalies in sexual development are due to too many or too few copies of the sex chromosomes in each cell (e.g. Kleinfelter and Turner Syndromes).

[6] These points are also illustrated by the Coin Flip Genetics section of "Genetics" (https://serendipstudio.org/sci_edu/waldron/#genetics).

diagram can be a model. It may be helpful to introduce the idea of a <u>conceptual model</u>. As noted in *A Framework for K-12 Science Education*, "Conceptual models allow scientists… to better visualize and understand a phenomenon under investigation… Although they do not correspond exactly to the more complicated entity being modeled, they do bring certain features into focus while minimizing or obscuring others." [7] If your students are not familiar with conceptual models, you may want to <u>give examples</u> of conceptual models that students may have used, e.g., a map, a diagram of a football play, sheet music, or an outline of a paper the student is writing.

Additional Genetics Learning Activities
<u>Genetics – Major Concepts and Learning Activities</u>
https://serendipstudio.org/exchange/bioactivities/GeneticsConcepts
Part I summarizes key concepts in genetics. Part II presents common misconceptions. Part III recommends an integrated sequence of learning activities on the biological basis of genetics, plus seven human genetics learning activities. These learning activities develop student understanding of key concepts and counteract common misconceptions. Each of these recommended learning activities supports the Next Generation Science Standards (NGSS; https://www.nextgenscience.org/).

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[7] Quotation from <u>A Framework for K-12 Science Education: Practices, Crosscutting Concepts, and Core Ideas</u> (available at http://www.nap.edu/catalog.php?record_id=13165).