Genetics – Major Concepts, Common Misconceptions, and Learning Activities¹

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; <u>https://www.nextgenscience.org/</u>).

Part I – Key Concepts

Key Concepts from the Next Generation Science Standards (NGSS)²

• Students will gain understanding of several <u>Disciplinary Core Ideas</u>:

- LS1.A: Structure and Function "All cells contain genetic information in the form of DNA molecules. Genes are regions in the DNA that contain the instructions that code for the formation of proteins."
- LS1.B: Growth and Development of Organisms "In multicellular organisms individual cells grow and then divide by a process called mitosis, thereby allowing the organism to grow. The organism begins as a single cell (fertilized egg) that divides successively to produce many cells, with each parent cell passing identical genetic material (two variants of each chromosome pair) to both daughter cells. Cellular division and differentiation produce and maintain a complex organism, composed of systems of tissues and organs that work together to meet the needs of the whole organism."
- LS3.A: Inheritance of Traits "Each chromosome consists of a single very long DNA molecule, and each gene on the chromosome is a particular segment of that DNA. The instructions for forming species' characteristics are carried in DNA."
- LS3.B: Variation of Traits "In sexual reproduction, meiosis can create new genetic combinations and thus more genetic variation. Although DNA replication is highly regulated and remarkably accurate, errors do occur and result in mutations, which are also a source of genetic variation."

For additional information about NGSS Performance Expectations, Crosscutting Concepts, and Science and Engineering Practices and how the recommended learning activities support these learning goals, see the Teacher Preparation Notes for each recommended learning activity.

More Specific Key Concepts

<u>Genetics</u> is primarily concerned with <u>two questions</u>:

- How do our genes influence our characteristics?
- How are genes transmitted from parents to offspring?

This overview and the suggested learning activities will show that:

- understanding the functions of DNA and proteins provides the basis for understanding how genes influence an organism's characteristics (Key Concept 1 below)
- understanding meiosis and fertilization provides the basis for understanding how genes are transmitted from parents to offspring (Key Concepts 2 and 3 below).

¹ By Dr. Ingrid Waldron, Department of Biology, University of Pennsylvania, 2022. These Teacher Notes are available at <u>http://serendipstudio.org/exchange/bioactivities/GeneticsConcepts</u>.

² <u>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</u>

1. Genes in DNA \rightarrow Proteins \rightarrow Characteristics

- Genes in <u>DNA</u> provide the information necessary to make proteins, and <u>proteins</u> carry out many biological functions and thus influence our <u>characteristics</u>.
- More specifically:

nucleotide sequence in the DNA of a gene

 \rightarrow nucleotide sequence in messenger RNA (mRNA) *transcription*

 \rightarrow amino acid sequence in a protein *translation*

→ structure and function of the protein (e.g. normal hemoglobin vs. sickle cell hemoglobin)

 \rightarrow person's **characteristics** or **traits**

(e.g. normal health vs. sickle cell anemia)

- <u>Different alleles</u> (different versions of the same gene) code for different versions of a protein which can result in <u>differences in phenotype</u> (an organism's appearance or other observable characteristics). Differences in phenotype can also result from differences in <u>gene expression</u> and effects of the <u>environment</u>.
- A person is <u>homozygous</u> for a gene if both alleles for that gene are the same. A person is <u>heterozygous</u> if they have two different alleles for the gene.
- For some pairs of alleles, the phenotype of a heterozygous individual is the same as the phenotype of one of the two types of homozygous individual. The allele that results in the same phenotype for both a heterozygous individual and a homozygous individual is <u>dominant</u>. The other allele is <u>recessive</u>.
- In other cases, neither allele is completely dominant or completely recessive. In <u>incomplete</u> <u>dominance</u>, the phenotype of a heterozygous individual is halfway between the phenotypes of the two homozygous individuals. In <u>codominance</u>, both alleles affect the phenotype of the heterozygous individual.
- A single gene often influences more than one phenotypic characteristic (<u>pleiotropy</u>). Many phenotypic characteristics are influenced by more than one gene (<u>polygenic inheritance</u>).

2. Meiosis and Fertilization → Inheritance

- The <u>behavior of chromosomes during meiosis and fertilization provides the basis for understanding</u> <u>the inheritance of genes</u>.
- As a result of <u>meiosis</u>, each egg receives one copy of each gene from the mother and each sperm receives one copy of each gene from the father. When the gametes unite in <u>fertilization</u>, the zygote that develops into the child receives one copy of each gene from the mother and another copy of each gene from the father. Repeated mitosis ensures that each cell in a child's body has the same genes as the zygote.
- Because children get their genes from their parents, they tend to resemble their parents and their siblings. However, meiosis results in genetically diverse sperm and eggs which, together with random fertilization, results in genetic diversity of the zygotes/children produced by the same mother and father; this can result in phenotypic differences within families.

3. Punnett Squares → Probabilistic Predictions of Inheritance

- The processes of meiosis and fertilization can be summarized in <u>Punnett squares</u> which can be used to predict the genotypes of offspring.
- Each fertilization event is <u>independent</u> 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 <u>random variation</u> 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.

Part II – Common Misconceptions³

Sometimes our teaching may inadvertently reinforce some of these misconceptions (especially b-e). The learning activities proposed in Part III help to counteract these misconceptions.

- a. Students often do not understand the distinctions between a chromosome, a gene and an allele. Similarly, students often do not understand the difference between a chromatid and a chromosome.
- b. Each trait is influenced by a single gene, and each gene influences only one trait (not recognizing how common polygenic traits and pleiotropy are).
- c. There are only two alleles for each gene.
- d. Genes are the sole determinants of traits (not recognizing environmental influences).
- e. Dominant traits are the most common traits (which is true for some genes, but not all).
- f. A person who doesn't have a characteristic lacks the gene for this characteristic (not recognizing that the person has other alleles for this gene).
- g. All genetic conditions are inherited (not recognizing the important role of new mutations or mistakes in meiosis in causing some genetic conditions).
- h. All mutations are harmful (which is often true, but not always).
- i. Students often fail to recognize the probabilistic nature of Punnett square predictions and inheritance.

Part III – Recommended Learning Activities

<u>Tables 1 and 2</u> present recommended learning activities that will help students develop a solid understanding of the key concepts and will also counteract common misconceptions. Each title of a learning activity links to:

- a brief description of the activity
- a Student Handout (available as a Word file, a PDF file and, for the analysis and discussion activities, a GoogleDoc)
- Teacher Notes which include prerequisites, instructional suggestions (including a list of supplies for the hands-on activities), background biology, and an explanation of how the activity supports the Next Generation Science Standards (NGSS).

<u>Table 1</u> presents an integrated sequence of learning activities that engage students in science practices as they learn basic genetics concepts. Each of these learning activities supports the NGSS. Except for the first, each learning activity is available both as an analysis and discussion activity and as a handson activity. This sequence begins with three activities to help students understand DNA structure and replication and the basic molecular biology of how genes influence characteristics. Some teachers may prefer to use only the first of these activities before beginning the discussion of mitosis, meiosis and

³ These misconceptions are taken primarily from <u>http://knowgenetics.org/common_misconceptions/</u> and <u>https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2278104/</u> (especially tables 5 and 6).

fertilization and inheritance and then return to other aspects of basic molecular biology after completing the discussion of transmission genetics.⁴

| Analysis and Discussion Activities | Hands-On Activities |
|--|---|
| Understanding the Functions of Proteins and DNA | |
| DNA Structure, Function and Replication | DNA |
| <u>How Genes Can Cause Disease –</u> | How Genes Can Cause Disease – Introduction |
| <u>Understanding Transcription and Translation</u> | to Transcription and Translation |
| <u>Mitosis and the Cell Cycle – How the</u> | <u>Mitosis and the Cell Cycle – How a Single</u> |
| <u>Trillions of Cells in a Human Body</u> | <u>Cell Develops into the Trillions of Cells in a</u> |
| <u>Developed from a Single Cell</u> | <u>Human Body</u> |
| Understanding How Genes Are Inherited via | Meiosis and Fertilization – Understanding |
| Meiosis and Fertilization | How Genes Are Inherited |
| Introduction to Genetics – Similarities and | <u>Genetics</u> |
| Differences between Family Members | + |
| + Genetics and Probability – Sex Ratios of | <u>Were the babies switched? – The Genetics of</u> |
| Births | <u>Blood Types</u> |

Table 1. Recommended Sequence of Basic Genetics Learning Activities

<u>Table 2</u> presents human genetics learning activities that introduce students to important genetics concepts and counteract common misconceptions. Each of these learning activities engages students in science practices and supports the NGSS.

Table 2. Recommended Human Genetics Analysis and Discussion Learning Activities

| Introduction to Genetics – Similarities and Differences between Family Members | |
|---|--|
| The Genetics of Sickle Cell Anemia and Sickle Cell Trait | |
| Genetics and Probability – Sex Ratios of Births | |
| A mistake in copying DNA can result in dwarfism. | |
| How Mistakes in Cell Division Can Result in Down Syndrome or Death of an Embryo | |
| Mutations and Muscular Dystrophy | |
| Soap Opera Genetics – Genetics to Resolve Family Arguments | |

<u>Table 3</u> presents supplementary genetics learning activities, which may be of interest for specific teaching purposes. Most of these are not explicitly aligned with the NGSS (as designated by parentheses).

⁴ For molecular biology key concepts and additional learning activities, see <u>Molecular Biology: Major Concepts and</u> <u>Learning Activities</u>.

Table 3. Supplementary Genetics Learning Activities

Cell Differentiation and Epigenetics

Learn Genetics; this website is a rich source of videos and explanations of multiple aspects of genetics

DNA from the Beginning

Dragon Genetics – Understanding Inheritance

Cootie Genetics

Genetics Jeopardy Review Game