

## LEARNING PROGRESSION IN GENETICS

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Abstract. This learning progression describes progressive levels of understanding for core concepts in modern genetics. The progression extends from 5<sup>th</sup> to 10<sup>th</sup> grade. We have organized the core ideas in this learning progression around two questions in the discipline: (a) how do genes influence how we, and other organisms, look and function? And (b) Why do we vary in how we, and other organisms, look and function? We identified eight big ideas that are needed to successfully reason about these questions. The target performances of this progression thus involve generating several types of mechanistic explanations: explanations that link our genotype to our phenotype; explanations of the processes by which our genes are passed on from generation to generation and how they contribute to genetic variation; and explanations of the sources of variation in phenotype (including environmental interactions with our genes). The learning progression describes three levels of understandings that allow the progressive construction of the explanations described above. Associated with the big ideas are learning performances that range in their complexity and can help determine a student's level of understanding.

## The Domain

Modern genetics extends the explanatory models of classical genetics into the realm of the cellular and molecular mechanisms that underly the patterns of inheritance described by the classical models. Over the past several decades, there have been numerous scientific and technological advances in this domain. The current state of affairs increases the demand for scientific literacy placed on the public because many of the novel technologies and discoveries are entering the public realm. Citizens are expected to be able to make decisions about genetic screening, stem cell research, genetically manipulated foods, etc. Without a sound understanding of core ideas in genetics such decisions are, at best, uninformed.

While important to know, concepts in modern genetics are particularly challenging to understand (e.g. Stewart & Van Kirk, 1990; Venville & Treagust, 1998; Lewis & Kattmann, 2004). Given that we cannot teach, or expect 9<sup>th</sup> or even 12<sup>th</sup> grade students to master, all the intricacies of this domain. It is therefore imperative to identify a limited set of core ideas in the domain that will provide students with a coherent framework for understanding phenomena and issues in modern genetics. The big ideas we lay out for our progression are organized around two pivotal questions in the discipline: (a) how do genes influence how we, and other organisms, look and function? And (b) Why do we vary in how we, and other organisms, look and function? The first question deals primarily with the genetic mechanisms involved in linking genotype to our phenotype within an individual. For example, if an individual inherits the gene for sickle cell anemia how does that gene bring about the various symptoms associated with this disorder? The big idea here involves the understanding that genes contain information about proteins, proteins carry out a multitude of functions and ultimately result in our phenotype, that the expression of

genes can be turned on or off (gene expression is regulated), and the genetic information is replicate during cell division (such that all cells have the same genetic information).

The second question, on the other hand, deals primarily with the transmission of genetic information across generations and how environmental factors interact with our genetic makeup. For example, individuals of varying skin color tend to tan to a different degree; what are the causal mechanisms that underlie interactions between an environmental factor, like UV rays, and our genes for skin color. The big idea here involves an understanding of the patterns and mechanisms of gene transfer across generations (meiosis, Mendel's model of inheritance), where variation in the genetic makeup arises in these processes, and how the environment can influence such variation. For each question we have identified several conceptual components (big idea components) for which we describe a progression in understanding.

We chose to focus our progression on deepening students' understanding of conceptual content rather than inquiry practices in the domain (foreground big ideas and background practices). Research in genetics education has suggested several important inquiry practices that need to be part of genetics learning such as constructing, revising and arguing about models of genetic phenomena (e.g. Buckley, et al, 2004; Stewart, Hafner, Johnson & Finkel, 1992; Stewart, Cartier, & Passmore, 2005; Cartier & Stewart, 2000). We acknowledge the importance of these practices for genetics learning and include them as part of performances that can be used to assess students' understanding. However, because these inquiry practices are not specific to genetics (Driver, Leach, Miller, & Scott, 1996; Duschl & Osborne; 2002; Kuhn, 1993) we elected not to explicitly include them in our genetics learning progression.

## The Learners

The learning progression describes the deepening of the big ideas across grades 5-10. We chose this range for two key reasons. First, we wanted to begin the discussion of core ideas in genetics earlier than is currently done according to the standards (NSES and AAAS). However, it was important that we while still had support in the research literature that children at that age can reason about these ideas productively (we discuss our assumptions about children's prior knowledge in a later section). Second, we wanted to ensure that the core ideas are treated during the course of compulsory education. While our progression can easily be extended into advanced placement courses and college level, we did not want to leave key ideas out of the compulsory curriculum. We do not assume that the progression is aimed at high achieving students and believe that all students can develop deeper understandings of modern genetics through carefully designed instruction and with the support of competent teachers.

## Process for Characterizing Target Performances

In the process of defining our target performances we relied on multiple sources of input. The NSES and AAAS Benchmarks provided information about current expectations for genetic literacy by 12<sup>th</sup> grade. A recent study by Venville and Donovan (2005) reported on what experts in genetics think are important ideas/themes for the public to understand in this discipline. We incorporated the four themes into our progression at the relevant levels. We also relied on our own experiences developing instructional materials and studying their implementation (Duncan & Tseng, submitted; Gelbart & Yarden, 2006; Rogat & Krajcik, 2006) and on similar work by others (Berenfeld et al., 2004; Buckley et al., 2006; Stewart, Cartier & Passmore, 2005). In addition, a recent discussion of a learning progression in genetics based on project 2061 strands

maps (Roseman et al., 2006) informed our sequencing of ideas over the course of the progression. Broadly construed the target goal of the progression is to deepen students' understandings of the relationships between the genetic, meiotic, and molecular models of inheritance (Stewart, Cartier & Passmore, 2005). Below we discuss in more detail a few of our expectations of what student at the upper level of the progression should know and be able to do to give the reader a sense of what these expectations look like.

By the end of 10<sup>th</sup> grade students should have an understanding of how the genetic information is translated into proteins, and how proteins affect our physiology. Students should be able to explain genetic phenomena at the macro level using mechanisms that occur at the cellular and molecular levels. Thus, students should be able to model the physiological consequences of removing a certain protein, or of mutating the gene for that protein. These ideas are reflected in the NSES standards for grades 9-12 on the molecular basis of heredity.

We acknowledge that 9<sup>th</sup> grade students may only have rudimentary understandings of chemistry, however, there are certain biologically relevant chemical properties and structures that they can reason about (Duncan & Tseng, submitted; Berenfeld et al., 2004; Rogat & Krajcik, 2006). This existing research supports the notion that students at this level, with carefully structured instruction, can reason about the role of proteins in genetic phenomena and the consequences of genetic mutations to proteins and ultimately the phenotype.

By the end of 10<sup>th</sup> grade students should also understand that while all cells have the same genetic content they express different proteins. The goal here is not for students to know the specific mechanisms by which gene expression is regulated; but rather to understand that gene expression is regulated. For example, students should be able to explain which proteins might be expressed in different cell types and how a genetic disorder might affect some tissues

but not others. Such understandings should also allow students to explain various genetic phenomena that are influenced by environmental factors such as tanning, and the effect of global climate change on the male/female ratios in reptiles. Both the understanding that genes code for proteins (discussed above) and the understanding that gene expression is regulated are critically important to reasoning in modern genetics and have been identified as such by the NSES and by experts in the field of genetics (Venville & Donovan, 2005).

There are a few ideas we did not think were valuable for students to know but are currently part of the curriculum- such as memorization of all the steps in the process of meiosis, or memorization of the structure of DNA. We do not believe that learning these steps and structure promotes a useful understanding of genetics nor do they provide explanatory power. There were also some ideas we did not include in our progression because their value did not seem to outweigh the cost in time and effort of learning them. For example, we felt that understanding some of the techniques used in genetics research and applications, such as sequencing, fingerprinting, etc, were valuable to know because they would help students understand their value and limitations given different contexts (such as a court case). However, given limited time and resources we do not believe that learning these techniques, often at the expense of learning other ideas, was worth while enough to include in the progression

#### Process for Characterizing the Starting Place

We have some assumptions about where the learners might be at the start of the progression in terms of prior knowledge and experience. Research on young children's understanding of inheritance suggests that even nine year olds have a theory of kinship that allows them to differentiate biological inheritance and cultural transmission (Solomon & Johnson, 2000; Springer & Kiel, 1989; Venville, Gribble & Donovan, 2005). This theory of

kinship does allow for inheritance of acquired traits but favors the inheritance of biologically functional features that are inborn (Springer & Kiel, 1989). Work by Venville, Gribble, and Donovan (2005) indicates that 9-15 year olds have heard of genetic terms like genes, DNA, and many explained resemblance between parents and offspring as cause by genes/DNA. Their understanding of how genes resulted in such features was lacking, thus a theory of genetics per se (reference to genetic mechanisms) was not evident in their reasoning.

Young students are also aware of genetic variation, however, they are more likely to ascribe such variation to humans and animals rather than plants (Clough & Wood-Robinson, 1985). While able to explain that both parents contribute to the offspring's traits, young students do not assume equal contribution. Rather, they favor more maternal contribution to female offspring and larger paternal contribution to male offspring (Clough & wood-Robinson, 1985). Taken together existing research suggests that by 5-6<sup>th</sup> grade students have a rudimentary theory of genetics that can be elaborated on with careful instruction.

In terms of experiences with phenomena young children are familiar with the idea of kinship and the outcomes of reproduction. It is likely that they have seen puppies and kittens and noticed similarities and differences between the offspring and parents. They may also know twins and may have noticed the similarities and differences between them (some due to environmental influences). They may also know of some genetic diseases or that some conditions are inherited. Thus, they may have interacted with individuals that have diabetes or a heart condition and are were told that these are "genetic". It is unlikely, however, that they have any experiences with the mechanisms involved in genetics at the cellular or molecular level. While they may have seen pictures of DNA and chromosomes, their ability to make sense of these or relate them to phenomena they are familiar with is limited.

## Framework for Describing Change in Knowledge and Practice

We represent our learning progression in a format of a table with the columns denoting the different levels of understanding (there are three in our progression) and the rows denoting the components of the big ideas (see Table 1). Thus each cell in the table represents a version of a particular big idea component appropriate for a particular level in the progression. We can also incorporate learning performances into the table as a second row below each big idea component (for an example see Table 2). Cells associated with learning performances describe a particular performance students should be able to do if they have a particular level of understanding for a specific component of the big idea.

Movement along our progression entails developing understandings of the big idea components that grow in sophistication; i.e. developing more sophisticated versions of the big ideas over time. As any form of representation, our learning progression table has affordances and constraints. It affords seeing the different versions of the big ideas and mapping the types of changes we expect over time. However, it fails to represent some important aspects of the progression, such as connections between big idea components and how those connections may change over time. Thus each component appears to develop independently of the other components. This is clearly not the case and there are many important connections between big idea components that likely influence the ways in which the different versions of understandings develop. The incorporation of learning performances into the learning progression is somewhat clunky as these are superimposed on the progression table. Here too there is no way to represent

connections between performances or how performances for one components impact performances in another.

In this section we present the components of the big ideas discussed above. We already noted that these big ideas are organized around two key questions and here we describe three of the eight components (four per question) in more detail to give the reader a sense of the nature and grain size of what we call big ideas in genetics. These components represent the rightmost column of the progression table and thus constrain the content of the cells in the table. In the next section we will describe the development of one of these components over time (essentially a row in our table).

### *Components of the Big Ideas*

#### *A. All Organisms have Genetic Information that is Hierarchically Organized and That is Replicated During Cell Division*

A key aspect of reasoning in genetics is the understanding that genes contain information and that all organisms have DNA as their genetic material (Venville & Treagust, 1998). In order to understand some of the technologies used by geneticists and genetic counselors it is important to also understand the organizational structure of the genetic material. Specifically, that each DNA molecule comprises a chromosome and that different organisms have a different set of chromosome but in all organisms the chromosomal set is replicate during cell division. This replication of the genetic material is highly regulated to ensure the continuity of the complete genetic content.

#### *B. The Genetic Information Specifies Protein Structure*

Understanding genes as information units is not enough to be able to explain how the genetic information influences our traits and features. It is critical to understand what that

information specifies and how (Duncan, in press; Venville & Donovan, 2005). Genes specify the type and order of amino acids that make up a protein. Each set of three nucleotides in the DNA specifies one amino acid in the protein chain. Without an understanding of what the genetic information specifies it is impossible to understand the intermediate steps that link genotype to phenotype.

*C. Proteins Have a Central role in the Functioning of All Living Organisms and They are the Connection Between Genotype and Phenotype*

Along with understanding that the genetic information specifies the amino acid sequence of proteins, which then determines the protein's structure and function, students need to understand the different types of functions proteins have. As well as how these functions, or their absence, affect the subsequent structure and functions of cells, tissues and organs (phenotype). The big idea of proteins as central biological entities that link genes to their phenotypes is necessary to understanding why some genetic disorders, such as diabetes and dwarfism, can be treated by giving patients a protein (insulin or growth hormone), or what precisely occurs when you insert a gene from one organism into another.

Defining the Intermediate Steps and Transitions

In this section we describe in more detail movement along the progression for the second big idea component: *B. The Genetic Information Specifies Protein Structure*. The progression for this idea begins in grades 5-6, at this stage students are expected to understand that genes contain information. Research has shown that young students often conceive of genes as particles that are correlated with traits (Venville & Treagust, 1998). The goal at this level is to help students

develop the more sophisticated understanding that genes are instructions for traits. The notion of genetic information should set the stage for the next level of the progression.

At the second level of the progressions, grades 7-8, students deepen their understanding of genes as information by learning that genes contain instructions for proteins. At this level students do not learn the mechanism by which the genetic information is translated into proteins or exactly what the genetic code specifies about proteins. For this level it is sufficient for students to understand that the genetic content specifies one type of biological entity- the protein. Thus, changes to the instructions will affect the protein's ability to function. Understanding the role of proteins in our bodies is part of the third component (see above component C). At this level students thus move from understanding the genes are information to understanding what that information specifies.

The third, and final, level of understanding (grades 9-10) entails understanding the processes by which the genetic instructions are interpreted by the cell- i.e. the central dogma of molecular biology. At this level students learn about the genetic code and that each triplets of nucleotides codes for one amino acid in protein chain. They learn that changes to the nucleotide sequence can therefore alter the composition, and hence the properties, of the protein. They also further their understanding of the genetic information by learning that not all DNA sequences code for proteins; much of the sequence is non-coding.

Overall we can characterize the movement along this row of the table as gaining a deeper understanding of what the genetic information specifies and how. It is also characterized as a movement from the macro to the molecular; students begin by linking the genetic information to traits, then to protein in cells, then to the molecular units in proteins. In terms of performances students should be able to give more precise and mechanistic explanations of what the genetic

instructions determine and how. They should also be able to construct more sophisticated models that describe how a change to the genetic instructions might affect the phenotype.

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Table 1.

*Learning Progression in Modern Genetics*

Question: How do genes influences how we, and other organisms, look and function?

Big idea: All organisms have genetic information to specify the structure of proteins that carry out a multitude of functions and ultimately result in our phenotype. The genetic information is replicated in cell division and its expression is tightly regulated.

Components of Big Idea	Level 1- Grades 5-6	Level 2- Grades 7-8	Level 3- Grades 9-10
A. All organisms have genetic information that is hierarchically organized and that is replicated during cell division.	Humans, animals, plants, and bacteria have genes (genetic information) in their cells.	The genetic information is found in the chromosomes of cells (often in the nucleus). Organisms have a set of chromosomes (karyotype). All cells of an organism have the same chromosomal set (except gametes). The chromosome set is duplicated and then passed on to new cells through cell division (mitosis).	Genes are nucleotide sequences that make up segments of the DNA molecule. DNA molecules make up chromosomes that make up our genome. The process of cell division involves DNA replication and is tightly regulated.
B. The genetic information specifies protein structure.	Genes contain information about our physical structures and functions.	Genes have information for making proteins. Proteins carry out cellular functions and build cellular structures.	The genetic code is translated into a sequence of amino acids that makes up the protein. Some sections DNA do not code for proteins (non-coding).
C. Proteins have a central role in the functioning of all in living organisms and they are the connection between genotype and phenotype	Cells have to carry out many essential functions to live. Within cells organelles do specific functions. The structure of cells, tissues, and organs determines their function. Our body is composed of multiple levels of organization and changes at one level may affect another (cascade of effects in hierarchical system).	Proteins are like little machines that do the work of the cell. Proteins have shapes and properties that afford their functions. There are different types of proteins (enzymes, receptors, etc) Changes to genes result in changes to proteins, which affect the structure and function of cells. Cells in turn affect the structure and function of tissues/organs, etc.	Proteins have particular three-dimensional shape determined by their amino acid sequence. Proteins have many different kinds of functions that depend on their specific properties. There are different types of genetic mutations that can affect the structure and thus function of protein and ultimately the phenotype.
D. All cells have the same genetic information but	Different cells have some common and some different	Different cells have different collections of proteins. Proteins	All cells have the same genetic content but gene expression is

different cells express different genes.	structure and functions.	carry out the housekeeping and unique functions of the cell.	regulated.
<p>Question: Why do we, and other organisms, vary in how we look and function?          Big Idea: There are patterns of gene transfer across generations (Mendel’s model of inheritance). Cellular mechanisms drive these patterns (meiosis) and result in genetic variation. The environment interacts with our genetic makeup leading to variation.</p>			
Components of Big Idea	Level1- Grades 5-6	Level 2- Grades 7-8	Level 3- Grades 9-10
E. Transmission of the genetic information through the process of meiosis	The genetic information is passed on from parent to offspring. Each parent contributes half the information.	Chromosome sets are randomly assorted into gametes through the process of meiosis. From a set of 2N (full set) you get cells with N (half a set).	Meiosis has stages. The first stages are similar to mitosis. Then 4N cell is split into four N cells. During this process chromosomes can swap parts (recombination).
F. Patterns of correlation between genotype and phenotype and the molecular mechanisms that cause them.	For a given trait there are variations. Different organisms have different versions of the trait.	Each gene has two versions of the information (alleles), one on each chromosome in the set (per individual). There are patterned correlations between the variants of the genes and the resulting phenotype (Mendel’s model of inheritance).	The gene variants differ in their nucleotide sequence resulting in different or missing proteins that affect our phenotype. Dominance and recessiveness can be explained at the molecular level. Recessive and dominant alleles are essentially categorical definitions.
G. Changes to the genetic information can cause changes in phenotype, and such variation in the DNA can serve as a measure of phylogenetic relationships	The genetic information can sometimes be changed. Some changes can be beneficial, others harmful, and some neutral to the organism in its environment. Different organisms vary in how they look and function. Even within a group of organisms there is variation in traits.	Changes in the genetic information can result in changes to the structure and function of proteins. Genetic changes create the variation needed for evolution. There are different types of chromosomes some, like X and Y, vary in boys versus girls. The karyotypes of different species vary. Differences are in terms of the structure and number of chromosomes	Mutations can result in changes to the amino acid sequence. Mutations are a source of variation in a population and can be used as an evolutionary clock. Genomes vary in their genes composition. The Nucleotide sequence of genes varies between organisms in a species. Evolutionary proximity of species is directly related to the extent of similarity of their genomes.
H. Environmental factors	The environment can affect our	The environment can influence	Environmental factors can cause

can interact with our genetic information	traits. Even organisms that are related may end up looking or behaving differently.	cell function through changes at the protein level (type and amount).	mutations in genes or alter gene expression.
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Table 2.

*Learning Performances for the Progression*

C. Proteins have a central role in the functioning of all in living organisms and they are the connection between genotype and phenotype	<p>Cells have to carry out many essential functions to live. Within cells organelles do specific functions. The structure of cells, tissues, and organs determines their function.</p> <p>Our body is composed of multiple levels of organization and changes at one level may affect another (cascade of effects in hierarchical system).</p>	<p>Proteins are like little machines that do the work of the cell. Proteins have shapes and properties that afford their functions. There are different types of proteins (enzymes, receptors, etc)</p> <p>Changes to genes result in changes to proteins, which affect the structure and function of cells.</p> <p>Cells in turn affect the structure and function of tissues/organs, etc.</p>	<p>Proteins have particular three-dimensional shape determined by their amino acid sequence. Proteins have many different kinds of functions that depend on their specific properties.</p> <p>There are different types of genetic mutations that can affect the structure and thus function of protein and ultimately the phenotype.</p>
Associated performances	<p>Students can describe the functions of a living cell and identify the organelle responsible for carrying out the function.</p> <p>Students explain how the structure of a particular cell (nerve, skin, etc) affords and constrains its function.</p> <p>Students construct a model the different tissues of an organ, e.g. heart, and their role in the organ's function. Students predict how the organ's function will change if a constituent tissue was altered.</p>	<p>Students list a few proteins and their functions.</p> <p>Students explain how a genetic mutation might affect a cellular function.</p> <p>Given a description of genetic disorder at the tissue/cell level (e.g. deformed blood cells) students create a mechanistic model of what might be happening at the cellular and molecular level.</p> <p>Students predict the effects at the cellular and tissue level of a change to the function of a particular protein.</p>	<p>Students explain how a genetic mutation might affect the structure and function of a protein.</p> <p>Students predict how specific mutations will change the amino acid sequence of a protein and what the resulting change in the protein's properties might be.</p> <p>Students create a model linking a genetic mutation to the symptoms of a disorder at different levels of biological organization.</p>

