

RESEARCH ARTICLE

Exploring Teaching of Genetic Inheritance in High School

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Abstract

Improving primary and secondary (K12) science education is of great interest and sustained qualified K-12 education has been described as key in preparing the next generation of scientists. Genetics is one of the major disciplines to be taught in K-12 and recent evidence suggests current educational approaches may be promoting student misconceptions and misunderstandings. Here we describe the experience of developing a specific lecture/activity to provide information on patterns of genetics inheritance to 12th grade students in an elective Advanced Biology course from a senior high school in western Pennsylvania. Observations allowed the identification of specific misconceptions and topics of the curricula that may need to be re-evaluated.

Introduction

According to the Academic Standard for Science and Technology of the Pennsylvania Department of Education (PA Department of Education, 2002), by grade 10, students should be able to describe how genetic information is inherited and expressed (Table 1). Pennsylvania obviously follows the National Science Education Standards that come from the National Academies of Science and Project 2061 which suggest that as a result of their activities in grades 9–12, all students should develop understanding of the molecular basis of heredity. The explicit requirement in these national standards of teaching basic Mendelian genetics was proposed to be a factor contributing to confusion regarding the deterministic nature of a single gene in phenotype control (Shaw et al., 2008). It is evident that the state standards in Pennsylvania emphasize Mendelian forms of inheritance (Table 1). Although most of the traits are consequence of the influence of many genes with or without environmental factors (multifactorial or complex inheritance), one can assume teachers are not necessarily going to explore this topic if it is not explicitly included in their state content assessments.

In a systematic review of 500 essays from U.S. students grades 9–12 about genetics and its importance, patterns of inheritance were a topic that revealed numerous misconceptions and misunderstandings for students (Shaw et al., 2008). In total, 80% of the essays that included a misconception regarding patterns of inheritance inaccurately described a basic tenet of Mendelian inheritance, despite the expected coverage of this material by ninth grade. In addition, students were frequently unable to accurately define DNA, genes, and chromosomes.

Although some studies have enumerated students' misconceptions in genetics (Lawson & Thompson, 1988; Lewis & Wood-Robinson, 2000; Lewis & Kattmann, 2004; Shaw et al., 2008), there is a lack of studies investigating how these misconceptions are created in the first place and how they could be avoided. This work is based on the results of a partnership created by the Geneticist – Educator Network of Alliances (GENA) project with the goal of developing a local leadership to locally support school districts in the area. Here we present preliminary findings obtaining during the introduction of material to an Advanced Biology



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Course tailored to emphasize multifactorial inheritance concepts.

Table 1. Genetics content related to patterns of inheritance in Biological Sciences that should be presented to Pennsylvania grade students

Grade 4	Grade 7	Grade 10	Grade 12
Pennsylvania's public schools shall teach, challenge and support every student to realize his or her maximum potential and to acquire the knowledge and skills needed to...			
Know that characteristics are inherited and, thus, offspring closely resemble their parents	Know that every organism has a set of genetic instructions that determines its inherited traits	Describe how genetic information is inherited and expressed	Explain gene inheritance and expression at the molecular level
Identify physical characteristics that appear in both parents and offspring and differ between families, strains or species	Identify and explain inheritable characteristics	Compare and contrast the function of mitosis and meiosis	Analyze gene expression at the molecular level
	Identify that the gene is the basic unit of inheritance	Describe mutations' effects on a trait's expression	Describe the roles of nucleic acids in cellular reproduction and protein synthesis
	Identify basic patterns of inheritance (e.g., dominance, recessive, co-dominance)	Explain the relationship among DNA, genes and chromosomes	Describe genetic engineering techniques, applications and impacts
	Describe how traits are inherited	Explain different types of inheritance (e.g., multiple allele, sex-influenced traits)	Explain birth defects from the standpoint of embryological development and/or changes in genetic makeup
	Recognize that mutations can alter a gene	Describe the role of DNA in protein synthesis as it relates to gene expression	
	Describe how selective breeding, natural selection and genetic technologies can change genetic makeup of organisms		

Material and Methods

R.F. and A.R.V. participated in the project called the Geneticist-Educator Network of Alliances (GENA; <http://www.ashg.org/education/gena.shtml>), which is funded by the National Science Foundation. This project aims to develop a network of 92 master Geneticist-Educator alliances to design teaching strategies related to standards and misconceptions in genetics that can improve general understanding of genetics by K-12. R.F. and A.R.V., after applying to be part in the GENA project, were matched by the GENA staff based on geographic location and participated in a 2-day workshop to initiate the plans to develop an activity to be applied in the classroom.

Based on the data from the DNADay Essay Contest reported by Shaw et al. (2008), and from the evidence gathered by A.R.V. who served as judge for this same essay contest, an activity on "Patterns of Inheritance" was developed. The plan included a 45-minute lecture/activity that would cover the concepts of gene, chromosome, transcription, translation, mutation, types of mutation, monogenic disease, chromosomal disease, and multifactorial disease. The time allocated to the activity was decided based on the course schedule. The authors decided to use the muscle segment homeobox 1 gene (MSX1) and its involvement in the susceptibility to cleft lip and palate as the underlying theme of the lecture/activity. The current information on MSX1 and clefts is revised and summarized in Vieira (2008).

The lecture/activity was applied to one of the 12th grade Advanced Biology courses at Seneca Valley Senior High School, Pennsylvania, which is taught by R. F. The same course taught by another teacher did not have the activity. The Advanced Biology course is typically taken as a senior elective, although several juniors sign up for the class. Most students electing to take Advanced Biology have a strong interest in the sciences but may not

necessarily plan on majoring in Biology.

The lecture/activity was designed to have a 15-minute power point presentation followed by a hands-on manipulation of a segment of the MSX1 gene sequence (NCBI (<http://www.ncbi.nlm.nih.gov/>) entry AF426432). In this manipulation, students would predict the mRNA and protein sequences, as well as the consequence of mutations in the gene that were reported in the literature (reviewed in Vieira, 2008). One week prior to the day of the lecture, students anonymously responded to a series of questions prepared by their teacher (R.F.; Table 2). One week after the lecture, the students anonymously responded to the same series of questions. A subset of the students that took the same course with another teacher who did not apply the activity also anonymously responded to the same series of questions after they received the genetics material of the course. This project has University of Pittsburgh IRB approval (PRO08060233). Chi-square or Fisher's exact tests were used to compare the frequency of answers before and after the activity, as well as with the group that did not have the activity. A.R.V. attended the activity as an observer.

Table 2. Pre-post-assessment utilized

Pre and Post Assessment of the MSX1 Gene Mutation Lesson	
Directions: Please answer the following questions to your best ability using the choices provided	
1. A gene is	<ul style="list-style-type: none"> A. A region of DNA corresponding to a unit of inheritance B. A known mutation C. Another name for a chromosome D. Found only in animal cells
2. A Homeobox is a known sequence of DNA found within genes that are involved in the regulation of	<ul style="list-style-type: none"> A. Musical ability B. Development (morphogenesis) of animals, fungi and plants C. Behavioral development D. Sexual development
3. T/F The term mutation refers to a genetic condition with a negative outcome	
4. The definition of mutation is	<ul style="list-style-type: none"> A. A change to the nucleotide sequence of the genetic material of an organism B. The growth of an abnormal body part C. A change in the DNA code with results in absolute death D. The growth of a cancerous tumor
5. T/F Mutations are VERY common.	
6. The process of taking the information on a strand of messenger RNA and building an amino acid chain, which will become all or part of a protein molecule, is called	<ul style="list-style-type: none"> A. Polymerase B. Expression C. Transcription D. Translation
7. The process of obtaining a copy of the information in a gene as a strand of messenger RNA is called	<ul style="list-style-type: none"> A. Polymerase B. Expression C. Transcription D. Translation
8. T/F All genetic mutations are caused by the environment	
9. Mutations in DNA where a single base is added or deleted from the DNA sequence are called	<ul style="list-style-type: none"> A. a frameshift mutation B. a point mutation C. a deletion mutation D. an insertion mutation

10. Which of the following types of mutation events causes the premature coding for a STOP codon?
 - A. Silent mutations
 - B. Missense mutations
 - C. Nonsense mutations
 - D. All mutations do this
11. Which of the following types of mutation events codes for a different amino acid?
 - A. Silent mutations
 - B. Missense mutations
 - C. Nonsense mutations
 - D. All mutations do this
12. An insertion or deletion of a nucleotide base which throws off the entire reading frame of a gene is known as a
 - A. Silent mutation
 - B. Frameshift mutation
 - C. Nonsense mutation
 - D. Missense mutation
13. The ultimate source/mechanism for the introduction of new alleles into a species is
 - A. Gene flow
 - B. Founder effects
 - C. Hybridization
 - D. Genetic drift
 - E. Mutation

Results

Eighty-six students (49 females and 37 males) were enrolled in the Advanced Biology course taught by R.F. The lecture activity was presented four times to groups of approximately 20 students. In general, students were captured by the images of individuals affected by clefts and the description of the genetic mechanisms underlying their condition. They manipulated a sequence of the *MSX1* gene and predicted mRNA and protein sequences, as well as consequences of different nucleotide changes at different locations of the sequence (Figure 1). They were informed about the National Center for Biotechnology Information (NCBI: <http://www.ncbi.nlm.nih.gov/>), the central repository of the sequence they were working on.

Figure 1. Student working on building mRNA and protein sequence from the coding DNA sequence of the *MSX1* gene



In the first course section of the day (taught to the first 25 students), teacher and students were confused at some point by the description of the DNA sequence, which was defined in the NCBI as mRNA. A number of students was "copying" the DNA to generate mRNA from the complementary strand of the DNA sequence and were obtaining an inverted mRNA sequence

when compared to the “answer.” In consequence, their predicted protein sequence was not corresponding to the expected. The confusion was clarified and the teacher modified her description of the activity in the subsequent sessions to avoid further confusion.

Seventy-six students anonymously responded the pre-assessment questions and 79 students responded the post-assessment. Thirteen students from the Advanced Biology course that did not have the lecture/activity anonymously responded to the questions after they received the genetics material from the other teacher. The frequency of the answers to the 13-question assessment is summarized in Table 3. The most interesting observations obtained from the evaluation of the assessments were:

(1) Question 3 tried to access the use of the term “mutation” as a “physical aberration” (such as what happened with the X-Man characters) versus a DNA sequence variation that can lead to mutation. On average, 35% to 40% of the students in any of the three assessments (before and after the lecture/activity and in the group, that did not have the lecture/activity) agreed that mutation referred to a genetic condition with negative outcome, even though mutation was defined in class as a “variation in the DNA sequence that can lead to disease.”

(2) In question 5, the proportion of the students in the group before the lecture/activity and in the group, that did not have the activity that agreed that mutations are very common was similar (66% and 77%). The proportion decreased to 44% for the group that was taught by R.F. after they received the lecture/activity. This was likely influenced by the introduction in the lecture/activity of the concepts “polymorphism” versus “mutation.” Whereas mutation was described as a variation in the DNA sequence that can lead to disease, “polymorphism” was defined as a “harmless” variation in the DNA sequence. The teacher explained that although these terms can be used interchangeably, many scientists tend to use mutation to refer to changes with harmful consequences and polymorphisms to variations with no known consequences.

(3) Questions 6 and 7 deals specifically with the concepts of transcription and translation. The proportion of correct answers in both the group before the lecture/activity and the group that did not receive the lecture activity was similar (35% to 41% and 38% to 46%). This proportion increased after the lecture/activity in the first group to 67% to 72% which suggested that the lecture/activity more effectively conveyed this information to students.

(4) Question 8 asks if the etiology of mutations is always environmental. Initially, 7% of the students agreed with this statement.

(5) Although after the lecture/activity at least 63% of the students defined correctly the specific mutations types (questions 9 through 12); just 38% were able to correlate mutation with the enrichment of the genetic variation in the population/species. For the class that did not have the lecture/activity, the proportion was much smaller (8%).

Table 3. Frequency of answers from students before and after the activity and from the students that did not have the activity

	Pre-Assessment	Post-Assessment	Pre-Assessment	Outgroup	Post-Assessment	Outgroup
Question 1						
Answer Correct	72	78	72	13	78	13
Answer Incorrect	4	1	4	0	1	0
P-Value	0.14		0.52		0.86	
Question 2						
Answer Correct	24	48	24	12	48	12
Answer Incorrect	52	31	52	1	31	1
P-Value	0.00020		0.0005		0.02	
Question 3						
Answer True	27	31	27	5	31	5
Answer False	49	48	49	8	48	8
P-Value	0.63		0.23		0.24	
Question 4						
Answer Correct	65	77	65	13	77	13
Answer Incorrect	11	2	11	0	2	0
P-Value	0.006		0.16		0.74	
Question 5						
Answer True	50	35	50	10	35	10
Answer False	26	44	26	3	44	3
P-Value	0.007		0.2		0.02	
Question 6						
Answer Correct	27	53	27	5	53	5
Answer Incorrect	49	26	49	8	26	8
P-Value	0.00001		0.24		0.04	
Question 7						
Answer Correct	31	57	31	6	57	6
Answer Incorrect	45	22	45	7	22	7
P-Value	0.00001		0.72		0.06	
Question 8						
Answer True	5	0	5	0	0	0
Answer False	71	79	71	13	79	13
P-Value	0.03		0.44		1.0	
Question 9 (both C and D were considered correct)						
Answer Correct	40	50	40	3	50	3
Answer Incorrect	36	29	36	10	29	10
P-Value	0.17		0.03		0.006	
Question 10						
Answer Correct	4	63	4	10	63	10
Answer Incorrect	72	16	72	3	16	3
P-Value	0.0000001		0.0000001		0.27	
Question 11						
Answer Correct	17	53	17	7	53	7
Answer Incorrect	59	26	59	6	26	6
P-Value	0.00001		0.02		0.16	
Question 12						
Answer Correct	52	59	52	12	59	12
Answer Incorrect	24	20	24	1	20	1
P-Value	0.39		0.06		0.12	
Question 13						
Answer Correct	24	30	24	1	30	1
Answer Incorrect	52	59	52	12	59	12
P-Value	0.77		0.06		0.04	

Discussion

This paper reports observations made in a 12-grade classroom of a high school in western Pennsylvania of students particularly interested in biology. Although the scope of this work does not allow for any specific conclusions, it serves the basis to several inquiries that could eventually be directly tested.

The state standards described in Table 1 suggest that the role of DNA in protein formation should be understood by the 10th grade. However, less than 50% of the students assessed correctly defined transcription and translation. After the classroom activity, this proportion improved to 70%. The Seneca Valley Senior High School offers a required biology course in 9th grade which includes a genetics component. The 9th grade Honors Biology course outline includes Mendelian genetics, evolutionary genetics, and genetic technologies. These topics are specifically linked to Pennsylvania anchors. Additional genetic material will be only available to students if they choose to take Advanced Biology as an elective course in the senior high school. We can speculate that half of the students, after taking genetics, still do not have a full understanding of the transcription and translation processes. One possibility is that, although the material was presented, it is offered in such a way that students do not assimilate these concepts. Evidence from more senior students already in medical school (Lujan & DiCarlo, 2006) demonstrates that students tend to prefer multiple modes of information presentation (visual, auditory, reading/writing, or kinesthetic) over a single mode of information presentation. Most students are able to learn effectively as long as the teacher provides a blend of visual, auditory, reading/writing, and kinesthetic activities. However, some students prefer one of the modalities over the other three so strongly that they struggle to understand the subject matter unless special care is taken to present it in their preference (Miller, 2001). The traditional lecture format assumes that all students are auditory learners and acquire the same information presented orally at the same pace without dialogue with the presenter. The lecture/activity we design incorporated a kinesthetic activity with an auditory and visual activity. The students also received a review paper on clefts (Vieira, 2008) after the activity. The challenge here is requesting already overwhelmed high school teachers to incorporate different approaches to present an already long list of topics lined up in the curricula. Lectures are a good way to present large amounts of information in a short period of time and traditionally have been the preferred choice of teaching strategy. The balance that it is needed is the amount of information presented versus how much of that information is assimilated.

The quality of the information assimilated by the students can also vary. In the case of our lecture/activity, definitions of types of mutations of the DNA sequence were presented. After the lecture/activity, the students that attended it were more often able to correctly define the types of mutations defined in class, compared to students from the other class that did not attend the lecture/activity, in which these concepts were not presented. However, a much lower proportion of students were able to elaborate on the consequences in the population/species of mutation events, although the proportion was higher in the group that attended the lecture/activity. Naming types of mutations is much less relevant for an average future college student than understanding the consequence of these mutations at the population level, in particular for individuals interested in a career path in science.

Although our written assessment was relatively short, we were able to identify that the required 9th grade biology course did not overcome the typical misconception of believing that all genetic mutations are caused by the environment. Since the Advanced Biology course is an elective course in the Seneca Valley Senior High School, we can suggest that this is the proportion of high school graduates that do not identify genetics as a major cause for mutations in the population.

Ideally, hands on lessons like the one developed and utilized in the senior high Advanced Biology course need to be brought down to the 9th grade biology classes. Although a very successful lesson, it is believed that it would have had more of an impact on younger aged students. Unfortunately, the curriculum alignment in many schools requires teachers at the senior high level to re-teach many fundamental concepts which should be concrete. Hands on lessons like the one developed should increase a student's ability to incorporate these fundamentals so that they are able to build on them once they reach the higher-level science classes.

It is important to reflect about these initial findings in light of a number of limitations related to our assessment. We designed one single 45-minute session lecture activity that covered concepts of gene, chromosome, transcription, translation, mutation, types of mutation, monogenic disease, chromosomal disease, and multifactorial disease. These topics are complex in and of themselves, topics that could easily absorb multiple class periods each. In addition, certain concepts, such as "gene," presented to students did not include further detail

on the importance of various transcriptional and other regulatory elements. We also could not assess whether students were introduced to or understand the relative significance of replication associated with the origin of mutation versus environmental origins of mutation, and we cannot further conclude if the students have an accurate view of the mutational process.

To perform the activity, we are describing in this report, there was a need to select how much detail to include in a 45-minute session. We made the attempt to link various mutations (the original events leading to changes in genomic DNA sequence) to alleles, which are found at various frequencies in a population. Similarly, the description for the *MSX1* gene omitted a description of its intron and intron-exon junctions, as well as 5' and 3' untranslated regions, and regulatory (promoter/enhancer) sequences, where the mutation terminology we emphasized (nonsense, missense, and frame shift mutations) do not apply.

Hence, the limitation of having just a 45-minute session to present a complex material is reflected in the instrument we used to assess the students. In question 2, we used the term homeobox to refer to the specific sequence of DNA, although homeobox is a protein sequence domain involved in (generally) binding to and regulating gene activity. In question 3, we forced students into a dichotomous choice. In question 4, we cannot appreciate if students know that some mutations are insertions or deletions. In question 5, we used the term "very" to imply events in the whole genome and not a single genomic position. In question 9, the presumption appears to be that the mutation occurs within the coding region of a gene, although we know these events also occur to non-coding regions but their consequences are not well understood at this time. Finally, in question 13, we assume students can articulate how an allele is the same or different from a mutation.

It is our general feeling that, although the current Pennsylvania state standards for K-12 science education are well written, it is not practical to cover in the curricula all the information suggested to be taught concerning genetics. The attempt by teachers to present all the information may be one source of student misconceptions. Sustaining qualified K-12 science teachers is another challenge. Emphasis on Mendelian patterns of inheritance is likely another issue, since those concepts cannot fully explain the majority of human traits and diseases. Innovative approaches to allow for the preparation of the next generation of scientists have been proposed (Bush et al., 2008), such as the seeding of university science departments with science faculty with specialized science education roles within their disciplines that could help keeping K-12 science teachers better informed about the field of genetics.

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