Manuscript Title

Exploring Teaching of Genetic Inheritance in High School

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Summary

The manuscript was received on January 11, 2018 and was peer reviewed by one reviewer and an editor.
The initial recommendation of Major Revision was made on April 03, 2018.
The first revision was submitted on April 04, 2018 and was re-evaluated by the editor.
The manuscript was accepted for publication on April 10, 2017.

Peer Review Comments

Review - Exploring teaching genetics inheritance in high school. (n.b. should be genetic)

This is a complex manuscript for a number of reasons. First it seems to be centered around a 45 minute lecture activity that covers “concepts of gene, chromosome, transcription, translation, mutation, types of mutation, monogenic disease, chromosomal disease, and multifactorial disease.”

This description left me more confused than enlightened, since each of these topics is complex in an of itself, topics that could easily absorb multiple class periods each. In particular, it appears that the concept of a gene presented to students is missing ideas such as the importance of various transcriptional and other regulatory elements. Similarly, the authors are surprised by students’ thinking about the origin of mutations, but it is completely unclear as to whether students were introduced to, or understand the relative significance of replication associated
versus environmental origins of mutation. Without such an understanding, there is no reason to expect students to embrace an accurate view of the mutational process.

If the basic message of the paper is that genetics, as currently taught, is promoting misconceptions, this is not surprising given that the instruction the students are subjected to appears to seriously trivializes the concepts involved (this might well be a meta-criticism of the standards driving the curriculum). For example, there appears to be no 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

(see Exac Browser page: http://exac.broadinstitute.org/gene/ENSG00000163132) seems seriously inexact, since it omits a description of introns (and intron-exon junctions) as well as 5' and 3' untranslated regions, and regulatory (promoter/enhancer) sequences, where terms such as non-sense, mis-sense, and frameshift mutations do not apply.

There are also a number of issues associated with the assessment instrument. As an example, in question 2, it is incorrect to say that a homeobox is a sequence of DNA; a homeobox is a protein sequence domain involved in (generally) binding to and regulating gene activity. Question 3 seems to force students into an inappropriately dichotomous choice. In question 4 it is not clear that student appreciate that some mutations are insertions or deletions (or movements) of DNA and in question 5, what the term "very" implies, in what context (the whole genome versus a single genomic position). In question 9, the presumption appears to be that the mutation occurs within the coding region of a gene, but these do not apply to non-coding regions (where insertion and deletion are more appropriate answers). Finally, in question 13, can students articulate how an allele is the same or different from a mutation?
In sum, I would think that the manuscript needs to be seriously reconsidered to determine exactly which aspects of instruction lead to which misconceptions, an analysis that I would suspect would dramatically impact how these topics are presented to students, and what types of assessments can be used to reveal the accuracy of students' working knowledge. If all that is desired is to recognize multiple choice responses, then instruction needs to be tuned to these questions (rather than an actually understanding of genetic processes).

Finally, the example of the Msx1 gene is complex, as its phenotype in mouse also involves tooth formation, and it is not clear the depth to which instruction goes into what is (cleft palate) a complex developmental process (can, for example, students use OMIM (http://omim.org/entry/142983) to better understand the genotype-phenotype relationships associated with Msx1 mutations?