Research Article

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Mendelian or Multifactorial? Current Undergraduate Genetics Assessments Focus on Genes and Rarely Include the Environment

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Undergraduate genetics courses have historically focused on simple genetic models, rather than taking a more multifactorial approach where students explore how traits are influenced by a combination of genes, the environment, and gene-by-environment interactions. While a focus on simple genetic models can provide straightforward examples to promote student learning, they do not match the current scientific understanding and can result in deterministic thinking among students. In addition, undergraduates are often interested in complex human traits that are influenced by the environment, and national curriculum standards include learning objectives that focus on multifactorial concepts. This research aims to discover to what extent multifactorial genetics is currently being assessed in undergraduate genetics courses. To address this, we analyzed over 1,000 assessment questions from a commonly used undergraduate genetics textbook; published concept assessments; and open-source, peer-reviewed curriculum materials. Our findings show that current genetics assessment questions overwhelmingly emphasize the impact of genes on phenotypes and that the effect of the environment is rarely addressed. These results indicate a need for the inclusion of more multifactorial genetics concepts, and we suggest ways to introduce them into undergraduate courses.

KEYWORDS assessment, curriculum, environment, genes, genetics, undergraduate

INTRODUCTION

The inheritance and cause of most traits are best explained through multifactorial genetics, where many factors, including a combination of genes, the environment, and gene-by-environment interactions, play an important role (1–6). It is crucial that undergraduate students have an accurate understanding of multifactorial genetics for several reasons, including that advances in technology have led to the opportunity for them to become consumers of genetic information, data, and resources (5). For example, they can purchase DNA sequencing services to learn about ancestry, discover relatives, and learn about health risks (7–9). Genetics has also become a more prominent topic in the world news and social media, with these resources often acting as the primary source for public understanding of genetics (10–15). The COVID-19 pandemic is the most recent example of this, with these sources providing information about the intersection of genetics and personal susceptibility to contracting the virus (16, 17).

Undergraduate students are also naturally curious about complex traits at the intersection of genes and the environment, often driven by their desire to learn about concepts that they can connect with on a personal level (18, 19). As such, students are interested in human disease genetics, which requires an understanding of multifactorial genetics (19). Therefore, it is important that what is being taught in genetics courses aligns with modern genetics (20) and that the subsequent outcomes for student thinking, especially surrounding multifactorial genetics, are well understood (21, 22).

Genetics curricula can elicit deterministic thinking in students

Despite advances in the field of genetics to include a multifactorial understanding and student interest in complex human traits, historically, the focus in undergraduate genetics courses tends to be on simple Mendelian models, such as the effect of a single gene on a phenotype (e.g., yellow versus green pea color)

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(20, 23–25). While these examples provide an effective way to convey how traits are transmitted from one generation to another and how particular DNA mutations can have a significant impact on an organism, traits solely controlled by alleles of a single gene are rare in the natural world and, when overly emphasized, can increase deterministic thinking among students (26–29). Genetic determinism, in other words, that traits are completely controlled by the genetics of an individual, is problematic, as it is scientifically inaccurate and can mediate and exacerbate some forms of prejudice (22, 30–34). For example, common textbook examples about race and gender can further students' misunderstandings that differences have a genetic basis with no environmental influence (35).

Importantly, when multifactorial genetics concepts are incorporated into curricula, students are less likely to express views aligned with genetic determinism. For example, modifications to 7th to 12th grade genetics curricula that focus on multifactorial inheritance concepts, such as polygenic inheritance, the impact of the environment on phenotypes, gene by environment interactions, and trait malleability, resulted in a decrease in student belief that trait differences between racial groups are caused solely by genes (21, 22). Similarly, at the undergraduate level, modifications to a genetics curriculum to incorporate multifactorial concepts, specifically gene by environment interactions, decreased students' deterministic views on the role of genes on traits compared to those expressed after a course that emphasized the standard "Mendelian approach" (33). Given the implications that modifying the curricula in this way have on students' deterministic thinking, it is imperative that multifactorial genetics be incorporated into courses.

Genetics standards include multifactorial genetics concepts

Importantly, fostering student understanding of multifactorial genetics is a stated learning goal across published resources that define educational standards. The K–12 Next Generation Science Standards (NGSS) include the exploration of genetics and the environment; for example, the high school standard "Heredity: inheritance and variation of traits" includes the disciplinary core idea that "environmental factors also effect expression of traits" (36). At the undergraduate level, the Genetics Society of America developed a comprehensive learning framework that has multiple instances where the environment is mentioned, for example, under the "Transmission/patterns of inheritance" core category, it lists "Evaluate how genes and the environment can interact to produce a phenotype" as a genetics concept/learning goal (37). Finally, "Vision and Change," as outlined by the BioCore Guide includes the environment in many of its principle statements—for example, under "Information flow" the core concept category states "A genotype influences the range of possible phenotypes in an individual; the actual phenotype results from interactions between alleles and the environment" (38, 39). As standards continue to be updated to reflect the understanding of multifactorial genetics concepts, the expectation is that undergraduate genetics curricula will also be updated to include these concepts. However, whether or not multifactorial genetics is widely included in genetics curricula remains an open question.

Ways to investigate course curriculum

While published standards for undergraduate biology courses exist, there is no one standard undergraduate genetics curriculum across institutions in North America. As such, instructional tools such as syllabi and textbooks can be effective ways to investigate what is included in curricula (20, 40, 41). Previous work examined course syllabi and textbook topics to determine the extent to which undergraduate genetics courses included multifactorial concepts into their curriculum by investigating the order in which topics were presented to students (20). This work showed that genetics courses tend to focus on foundations of DNA, and multifactorial concepts were taught at the end of the course largely as "add on" topics. While these results provide valuable insight into the broader topics addressed in undergraduate genetics courses, an investigation of assessment questions across different tools used by undergraduate genetics instructors (textbook; published concept assessments; and open-source, peer-reviewed, curriculum materials) will identify which concepts are broadly covered and opportunities for further resource development.

Additionally, assessment questions can be used to investigate what is being taught in the undergraduate curriculum. Broadly, assessment questions are a measure of student learning and a defining feature of the course curriculum; therefore, they are an indicator of what concepts are emphasized and prioritized (41, 42). There are two perspectives to consider when identifying the role of assessment—the instructor and the student. Instructors design and give assessments that align with the learning goals of their course (42, 43). For students, assessment is a defining feature of the course curriculum and an indication of the concepts with which they should engage (42). Research into how students perceive assessment indicates that assessment content influences their effort distribution, approach to learning, and study behaviors (44, 45).

Study aims

Here, we analyzed over 1,000 genetics assessment questions from a commonly-used undergraduate genetics textbook; published concept assessments; and open-source, peer-reviewed, curriculum materials as a representative sample to determine how often questions about how traits are inherited and what causes them include multifactorial concepts. Using this pool of questions, we addressed the following research questions. (i) What proportion of assessment questions address the explanation of traits—how they are inherited and what causes them? (ii) What proportion of questions on the inheritance and causes of traits include multifactorial concepts? By taking inventory of what types of questions are currently being asked, undergraduate instructors can determine where current assessments provide coverage and where there are opportunities to develop questions that incorporate more multifactorial concepts into curricula.

TABLE 1
Peer-reviewed, freely available genetics concept assessments analyzed in this study

Concept assessment title	Total no. of questions	Reference
The genetic drift inventory (GeDI)	22	51
Genetics concept assessment (GCA)	25	64
Genetics literacy assessment instrument (GLAI)	17	65
Public understanding and attitudes towards genetics and genomics (PUGGS)	42	53
Total	106	

METHODS

Choosing assessments

We analyzed 821 end-of-chapter textbook questions from *Concepts of Genetics*, 12th edition (46). This textbook was chosen because it is widely circulated and available in 221 libraries worldwide, which surpasses the circulation of other genetics texts (N. Bishop, Mann Library Information and Public Services Assistant at Cornell University, personal communication, 20 April 2022). We also conducted a chapter comparison content analysis between this text and *Genetics: From Genes to Genomes*, 7th edition (47), which is circulated in 124 libraries, and found that the topics presented addressed similar concepts. Therefore, we used the pool of questions from Klug et al. (46) as a representative sample of textbook questions.

To identify published undergraduate genetics concept assessments, we searched the biology education research literature using databases such as PubMed and scanned the introduction section of manuscripts describing new genetics assessments. In total, there are four published, freely available concept assessments that focus on student understanding of genetics concepts, which include 106 questions (Table 1).

To find open-source, peer-reviewed undergraduate biology curriculum materials, we searched *CourseSource* genetics lessons published in the journal *CourseSource* (https://qubeshub.org/community/groups/coursesource/) between 1 January 2015 to 1 June 2021 (Table 2). *CourseSource* publishes lesson articles that employ evidence-based teaching strategies, are field-tested in undergraduate biology classrooms, and provide all the necessary details and supporting materials (e.g., slides,

TABLE 2
Peer-reviewed, freely available *CourseSource* lessons that were analyzed in this study

CourseSource lesson title	Total no. of questions	Reference
A clicker-based case study that untangles student thinking about the processes in the central dogma	17	50
A quick and simple natural selection role play	3	66
Fruit fly genetics in a day: a guided exploration to help many large sections of beginning students uncover the secrets of sex-linked inheritance	16	67
Furry with a chance of evolution: Exploring genetic drift with tuco-tucos	1	68
Homologous chromosomes? Exploring human sex chromosomes, sex determination and sex reversal using bioinformatics approaches	25	69
Interactive video vignettes (IVVs) to help students learn genetics concepts	10	70
Linking genotype to phenotype: the effect of a mutation in gibberellic acid production on plant germination	20	71
Meiosis: a play in three acts, starring DNA sequence	7	72
My dog is my homework: exploring canine genetics to understand genotype-phenotype relationships	42	73
Predicting and classifying effects of insertion and deletion mutations on protein coding regions	8	74
Sex-specific differences in meiosis: real-world applications	1	75
Teaching genetic linkage and recombination through mapping with molecular markers	18	76
The case of the missing strawberries: RFLP analysis	4	77
Why do some people inherit a predisposition to cancer? A small group activity on cancer genetics	28	57
Total	200	

TABLE 3
Complete list of codes used for each of the defined categories, as well as inter-rater reliability calculated for the jointly coded questions using Krippendorff's alpha

Category	Code	IRR (α)
Broad	Inheritance (i.e., inheritance of traits involving multiple generations)	0.94
	Causality (i.e., impact of allele(s) on phenotype)	0.93
	Excluded (i.e., genetic mechanisms or parameters for genetic drift)	0.95
Specific	Single gene	0.95
	Many genes	1
	E	n/a[a]
	G+E	0.84
	G×E	n/a[a]
Avg		0.95

[a]IRR, inter-rater reliability. An n/a indicates that no questions in the jointly coded subset were coded for these categories.

assessment questions) to replicate the lesson in additional classrooms. Lessons have learning goals and objectives that are aligned with professional society learning frameworks, including that from the Genetics Society of America (37). In total, 40 lessons were aligned with this framework; however, 26 lessons were not included in this study because they guide students through a protocol, such as working on lab procedures or bioinformatic/computational biology modeling activities. Of the lessons that were included, assessments that were specific to lab procedures were not included. The final question pool included 200 questions from 14 different lessons.

Analysis

Authors K.M.S., M.K.S., D.L., M.W., S.Y.A., and A.S. developed a codebook using an iterative coding process (48, 49). Through a process of coding four sets of questions, a complete codebook was created and can be found in the supplemental material (see Appendix S1 in the supplemental material). The codebook includes broad categories (e.g., inheritance) and specific categories (e.g., G+E) (Table 3).

Questions were first sorted into the broad categories of inheritance (i.e., inheritance of traits involving multiple generations), causality (i.e., impact of allele(s) on phenotype), or excluded (Table 3). Questions coded as excluded were questions that did not ask about the inheritance and causes of traits or mention a phenotype. For example, these questions asked about genetic mechanisms (e.g., When DNA polymerase reaches the nucleotides encoding the premature stop codon it will. . . [50]) and parameters of genetic drift (e.g., Agree/Disagree, Genetic drift is more pronounced in the island population than the mainland population in these first few generations [51]). If the coders applied the broad category codes of inheritance or causality codes, they subsequently coded a specific category for that assessment item (Table 3). The specific categories include single gene, many genes, environment (E), genes and the environment (G+E), or gene-by-environment interactions (G×E).

Authors M.K.S., K.M.S., and D.L. used the codebook to categorize the complete set of questions (see Appendix S1 in the supplemental material). The questions were split into two sets, jointly coded and individually coded. For the jointly coded set, the coders all coded a representative sample of 152 questions from all categories and across all assessments. We calculated inter-rater reliability for these questions using Krippendorff's alpha (52) and discussed discrepancies. The final average inter-rater reliability was calculated ($\alpha = 0.95$), and category inter-rater reliability is included in Table 3. The remaining questions were divided equally between K.M.S., D.L., and M.K.S. and independently coded. Once coding was complete, we calculated the percentage of questions that fell into each category.

RESULTS

To determine the range of question types across the entire question pool, we examined the frequency of inheritance, causality, and excluded questions (Fig. 1A). Inheritance questions, asking students about the inheritance of traits involving multiple generations, accounted for 16% of the total questions analyzed, and causality questions, asking students about the impact of allele(s) on phenotype, accounted for 11% of the questions (Fig. 1A). The majority of questions (73%) ask students about concepts that were excluded, such as questions about genetic mechanisms and questions about the parameters for genetic drift.

Because questions about inheritance and causality provide opportunities for students to explore multifactorial models, such as the interaction of genes with the environment, we analyzed the inheritance and causality questions by the specific categories of single gene, many genes, environment (E), genes and the environment (G+E), or gene by environment interactions (G×E). We found that when students are solving story

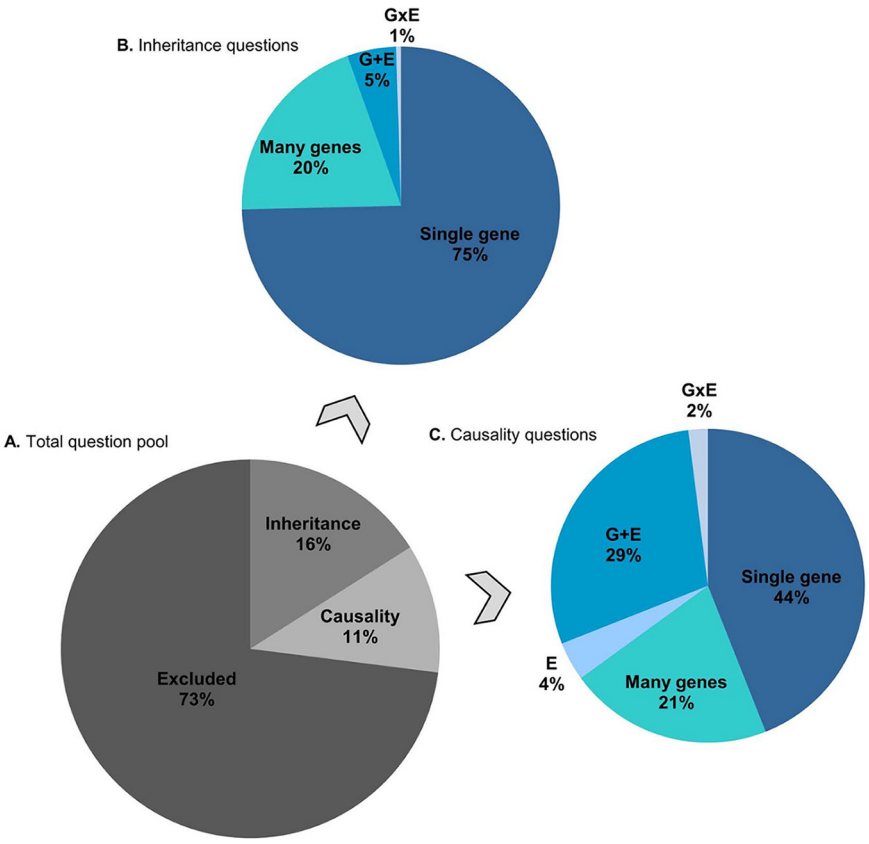


FIG 1. Breakdown of assessment questions that were coded into each of the categories. (A) Percentage of the total number of questions that were coded into each of the broad categories of inheritance, causality, and excluded. Percentage of inheritance (B) and causality (C) questions that were further characterized into single gene, many genes, environment (E), genes and the environment (G+E), and gene by environment (G×E) interactions.

problems about the inheritance of traits across multiple generations, the questions are overwhelmingly focused on single or many genes (Fig. 1B). Causality questions also overwhelmingly focused on single or many gene questions (Fig. 1C). Across all genetics assessment questions analyzed, only 9% of the questions ask about G+E and 0.7% ask about G×E (Fig. 1).

To investigate whether question content differed depending on if they came from textbooks; genetics concept assessments; or open-source, peer-reviewed undergraduate biology curriculum materials in *CourseSource*, we subdivided the questions according to source. The results from all three sources indicate that the most common question type is excluded questions (Fig. 2); although when comparing across the sources, concepts assessments included more questions about causality (Fig. 2B).

We then analyzed the inheritance and causality questions for each source by the specific categories of single gene, many genes, environment (E), genes and the environment (G+E), or gene by environment interactions (G×E). Within each of the question sources, questions about inheritance overwhelmingly focused on genes, including both single gene and many gene questions, and rarely included questions that included the environment (Fig. 2). For causality questions, there are different distributions depending on the source. Textbook (Fig. 2A) and concept assessment questions (Fig. 2B) are more likely to address the environment, largely through G+E questions. Although 33% of the causality concept assessment questions are G+E (Fig. 2B), the majority of these questions come from a single assessment, the PUGGS (public understanding and attitudes towards genetics and genomics), which explicitly measures public understanding of modern genetics and belief in genetic determinism (53). In contrast, *CourseSource* lessons contained the fewest number of questions that include the environment (Fig. 2C).

DISCUSSION

In a pool of over 1,000 genetics assessment questions from different sources, including textbooks, concept assessments, and open-source, peer-reviewed curriculum materials from *CourseSource*, very few questions ask about G+E and G×E concepts (Fig. 1 and 2). This outcome is notable considering that the majority of phenotypes result from the interaction of genes in the environment (1–3, 5, 6, 54), and there is widespread student interest in these more complex traits (18, 19). Furthermore, given that assessment is a key indicator of instruction and curricular content (42, 44, 55) and curriculum standards from elementary school through undergraduate include multifactorial concepts, such as the

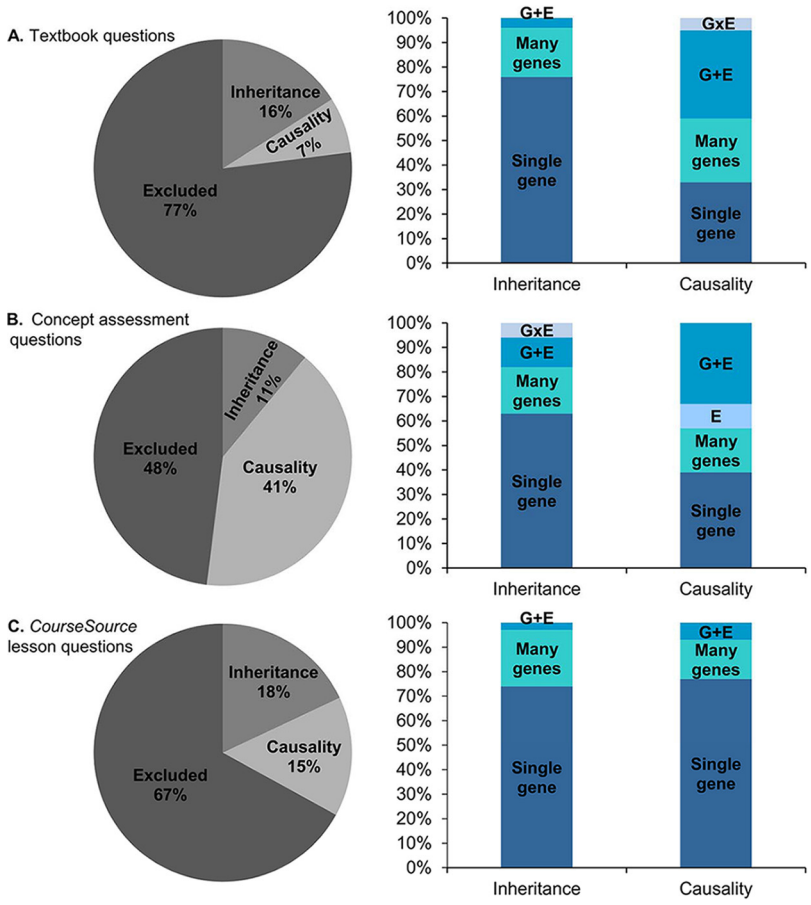


FIG 2. Breakdown of assessment questions by each of the three question sources as follows: end-of-chapter genetics textbook questions (A), concept assessments (B), and *CourseSource* lesson questions (C). For each source, the pie chart depicts the percentage of the total number of questions that were coded into each of the broad categories of inheritance, causality, and excluded. The bar chart depicts the inheritance and causality questions further characterized into single gene, many genes, environment (E), genes and the environment (G+E), and gene by environment (G×E) interactions.

intersection of genetics with the environment (36, 37, 39), there are several opportunities to expand assessment question content.

While limited, there are questions that ask about environmental factors (Fig. 1 and 2). These questions tend to focus on causality rather than inheritance patterns and largely come from end-of-chapter textbook questions or a single assessment instrument—the PUGGS (53) (Fig. 2A and B). While these types of questions provide students entry into thinking about the intersection of genes and the environment, they do not integrate the environment into genetic inheritance problem solving, which is a hallmark of undergraduate genetics instruction (56). Therefore, we suggest that there is a need for the development of undergraduate genetics learning materials and assessments that are designed to meet multifactorial genetics learning goals and motivate student learning. A shift toward a more multifactorial approach has additional benefits in that it has been shown to reduce deterministic thinking in students at both the K–12 and undergraduate level (21, 22, 33) and, therefore, has important outcomes for student thinking.

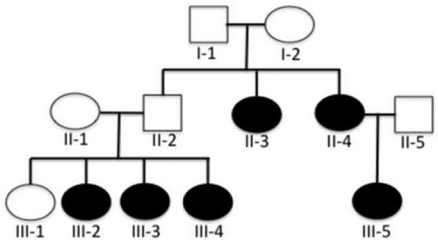
Examples of ways to incorporate multifactorial concepts into curricula

While there is evidence that there is a need to include more questions on multifactorial genetics concepts, making these changes is difficult, as it is time-consuming and it can be challenging to find easy-to-understand multifactorial examples (25). One way, suggested by McElhinny et al. (20), is to make wording changes to existing questions. For example, Fig. 3 shows an assessment question from a *CourseSource* genetics lesson (57). This example already has created a story about the inheritance of alleles of a single gene (the *BRCA1* gene). Additional questions could be added to broaden the scenario to include the environment as follows. For $BRCA1^{+}$/$BRCA1^{-}$, females who have this particular allele of $BRCA1^{-}$ have a high chance of developing early onset breast cancer. Why is this chance not 100%? What environmental factors might affect someone's chance of developing breast cancer?

Based on our experience leading professional development for undergraduate genetics instructors (SABER West workshop) (58), we also suggest introducing E, G+E, and G×E concepts using interaction graphs that model the relationship

The *breast cancer 1* (*BRACA1*) gene has been implicated in breast and ovarian cancer. Below is a pedigree of a family showing the incidence of breast cancer with a particular BRACA1$^-$ allele.

- BRACA1$^+$/BRACA1$^-$ females who have this particular allele of BRACA1$^-$ have a high chance of developing early onset breast cancer.
- There are no BRACA1$^-$/BRACA1$^-$ individuals in this family.
- BRACA1$^-$ mutations usually cause breast cancer in females but not in males; one member in generation I is heterozygous for the mutant allele; individuals II-1 and II-5 are BRACA1$^+$/BRACA1$^+$.



Which mode of inheritance is most consistent with the information in the pedigree?

A. Autosomal dominant
B. Autosomal recessive
C. X-linked dominant
D. X-linked recessive
E. More than one of the above is possible

If a man has a BRACA1 mutation (remember BRACA1$^-$/BRACA1$^-$ individuals do not exist), what is the chance he will pass the mutation on to his son?

A. 100%
B. 75%
C. 50%
D. 25%
E. 0%

FIG 3. Example of gene-focused questions from "Why do some people inherit a predisposition to cancer? A small group activity on cancer genetics" lesson published in *CourseSource* (57).

between genotypes, the environment, and their effect on a phenotype (Fig. 4). These models can be adapted for populations with two genotypes (genotype A represents alleles of one or more genes and genotype B represents alternative alleles of the same gene or genes) and two different environments (environment 1 and environment 2).

If instructors want to explore a phenotype that is largely controlled by genes (Fig. 4A), they could use single gene human mutations that have phenotypic outcomes and explore those phenotypes in two different environments. For example, instructors could compare males who have a wild-type version of the X-linked *opsin1 long wave sensitive* gene (*OPN1LW*) and see color to males who have a mutation in this gene and are red-green colorblind (59). Regardless of the environment these males are placed in, one group will detect color and one will not.

For exploring a phenotype that is largely controlled by the environment (E) (Fig. 4B), flamingos and their plumage color provides a good example scenario. In this example, flamingos' plumage color, rather than variation in genotype, is largely determined by a diet that is rich in carotenoids (60). In other words, if the flamingos have little variation in their genotype, the differences in phenotypic expression are determined by the environment. Instructors could present students with the scenario where they measure the plumage color of two flamingo populations across two environments as follows: a diet rich in carotenoids and a diet low in carotenoids.

For exploring a phenotype that is controlled by both genes and the environment (G+E) (Fig. 4C), skin cancer risk as determined by pigmentation and UV exposure provides an example scenario. In this example, variants of the *melanocortin-receptor 1* gene (*MC1R*) as well as UV exposure impact skin cancer risk (61). Instructors could present students with the scenario where groups of people with two different *MC1R* genotypes, one with the wild-type *MC1R* genotype and another with a red hair color (RHC) *MC1R* variant, are in two different environments, high and low UV. Both genotypes experience an increase in skin cancer risk in the high-UV environment, but individuals with the RHC *MC1R* variants have an overall greater risk of skin cancer regardless of the UV level (61).

For exploring a phenotype that is controlled by a gene by environment interaction (G×E) (Fig. 4D and E), *Mimulus guttatus* flowering time during different seasons provides one example (62). Instructors could present students with the scenario where they observe the flowering time of two *Mimulus guttatus* plant populations that are genetically distinct (i.e., different families) and grown in chambers simulating spring or fall germination conditions (Fig. 4D). In the simulated spring environment, genotype A has a longer flowering time when compared to genotype B. In the simulated fall environment, the flowering time stays fairly consistent for genotype A and decreases for genotype B.

Exploring scenarios that span the range of multifactorial concepts across multiple organisms will provide students with context and ways to visualize these complex ideas. Following this exploration, we suggest creating a summative activity where

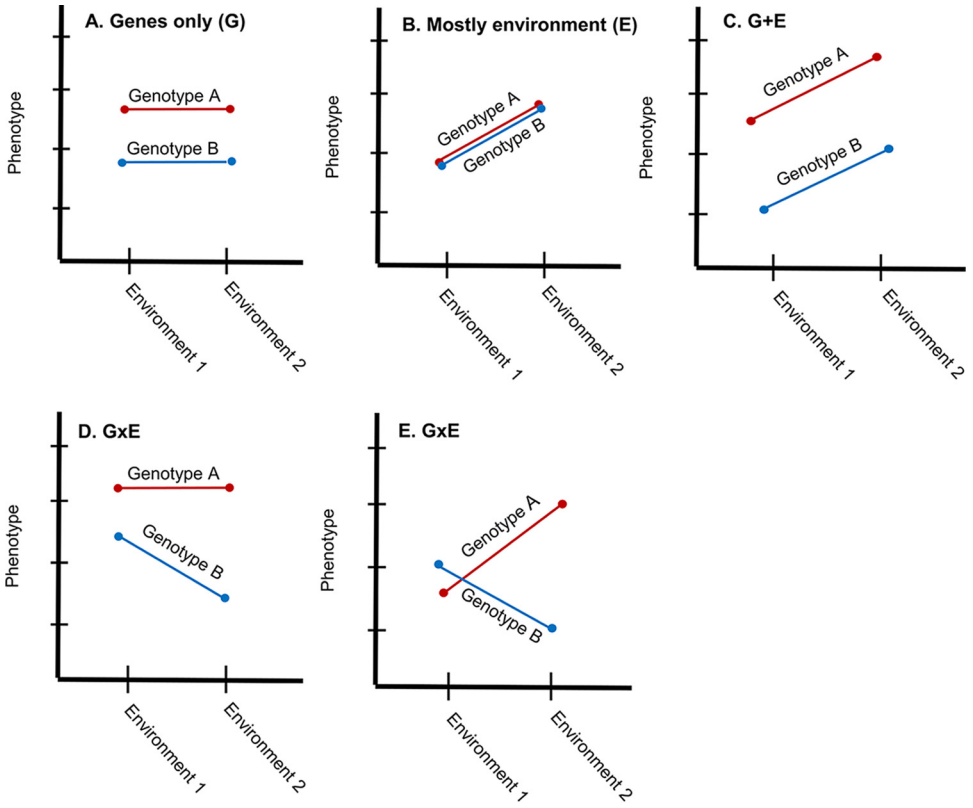


FIG 4. Graphical representations of multifactorial genetics concepts, including the effect of genes only on a phenotype (A), the effect of mostly the environment only on a phenotype (B), and the effect of genes and the environment on a phenotype (C). Panels D and E show examples of two different gene by environment interactions.

students are presented with a novel experimental design and predict the results if the outcome is influenced by genes only, the environment, G+E, or G×E. This summative activity using contrasting cases (63) provides an opportunity for students to think more critically about these concepts and apply what they have learned to novel scenarios.

Limitations and future directions

This study is based on a representative sample of over 1,000 genetics assessment questions from three broad sources. Because there is no single widely used undergraduate genetics curriculum, this work provides a general overview of what is likely covered—the emphasis on genes only may not be the case at a specific course level. Notably, open-source, peer-reviewed undergraduate biology curriculum materials have the fewest questions that include the environment (Fig. 2C). These platforms offer the greatest potential for undergraduate biology instructors from multiple institution types to share their innovative multifactorial genetics lessons and make an impact on the field. Instructors have several venues to publish these materials including *CourseSource* and the JMBE Curriculum or Tips and Tools section. Additionally, providing opportunities for instructor professional development will be an important part of increasing the prevalence of multifactorial genetics into undergraduate courses. In these spaces, instructors can build their expertise and become more comfortable with these complex topics while also engaging with other instructors to form support networks.

Once more materials are available, it will be important to conduct future studies that assess how multifactorial genetics questions affect student learning and views on genetics determinism, and to determine the appropriate target number of multifactorial assessment questions that should be included in undergraduate courses. Furthermore, while it is still an open debate, studies have advocated for a restructuring of course content to move multifactorial genetics to the beginning of courses instead of at the end (18, 20, 23). The development of more resources and an increase in the number of courses that include multifactorial concepts will provide opportunities to investigate the efficacy of restructuring versus not.

Conclusions

Results from this study indicate that there are few published undergraduate genetics assessment questions that include the effect of the environment on a trait, and questions that include the environment tend to focus on causality rather than inheritance. These results suggest a need for reform to undergraduate genetics curricula to include assessment questions with more multifactorial genetics concepts. For instructors who want to incorporate these concepts into their curriculum, we provide examples of ways to begin this process. These changes provide the opportunity to

bring the curriculum up to date with the current understanding of the field, engage student interest, and align with curriculum standards. They may also have important outcomes for student thinking that lead to downstream effects for students as consumers of genetic information and materials outside of the classroom.

SUPPLEMENTAL MATERIAL

Supplemental material is available online only.

SUPPLEMENTAL FILE 1, PDF file, 0.1 MB.

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