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Genetics Beyond the Classroom: An analysis of undergraduate students' understanding of genetics and attitudes towards clinical genetics

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Honors Thesis

GENETICS BEYOND THE CLASSROOM: AN ANALYSIS OF UNDERGRADUATE STUDENTS' UNDERSTANDING OF GENETICS AND ATTITUDES TOWARDS CLINICAL GENETICS

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ABSTRACT

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Genetics is a field of study that has been difficult for high school students, college students, and the general public to fully grasp, comprehend, and apply to clinical settings. Other studies show that students who are given hands on experiences with difficult concepts, like personalized genomics, have a better attitude towards and retention of those concepts. The purpose of this study was to determine if an in-class lecture specifically discussing clinical genetics concepts and benefits in solving difficult polygenic diseases would increase student attitudes towards genetic testing and prenatal testing and their knowledge of genetics in general. Students were asked to take a pre- and post-survey evaluating their knowledge about genetics and attitude towards modern genetics. Students in the experimental group received an in-class lecture that used a real-life example of a complex disease to explain the benefits of genetic technology in receiving medical care. The control group took the pre- and post-surveys, but continued with normal academic lectures. Overall, having an in-class lecture devoted to clinical genetics and

applications of genetic technologies increases students' knowledge of modern genetics and leads to more positive attitudes towards genetic testing and prenatal diagnostic testing. Further research should be done to validate these findings and to build upon them. Specifically, future research should determine if the amount of time spent learning about real-life application correlates to student's attitudes and understanding.

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TABLE OF CONTENTS

LIST OF FIGURES

Introduction

Genetics is a field of study that has been difficult for both high school and college students to fully grasp and comprehend. Often, introductory biology students leave the course with an idea that genetics are based on a simple relationship – a gene causes a phenotype. This leads to the mistaken view that clinical genetics is a straightforward process with exact results [1]. However, as is the case when determining which gene mutations cause a specific disease, clinical diagnosis of a disease is a complex process in which we do not have all the answers. Having a skeletal understanding of genetics leads to skewed views about the importance of genetic research and new technologies [2, 3]. One such skewed view is the idea of genetic determinism. Individuals with a deterministic mindset believe that a individual's genes are fully responsible for a specific phenotype—often including personality, looks, and diseases—without acknowledging any role of environmental factors in these same phenotypes. This leads to a mistaken belief that having a specific gene variation, or mutation, means that they will have a certain phenotype. However, often times, a person's environment plays a large role in determining the final outcome of the trait. For example, an individual with a mutation in BRAC1 has a greater likelihood of developing breast cancer. This does not mean that they will, but that they are at a higher risk. Environmental factors, like smoking, or exposure to other carcinogens, will make the likelihood of breast cancer increase even more. However, there is still a chance that this individual may never develop breast cancer. By educating students about these complexities, their attitude towards use of modern genetics technology will become more favorable

and their genetic determinism views will decrease, thereby leading to increased awareness and better application of genetics to the clinical setting.

Having introductory biology students understand the importance and complexity of clinical genetics is vital to seeing growth in clinical genetics research. Clinical genetics is the application of human genetics to medical diagnosis and treatment of various diseases. As students understand the need for researchers and the many unanswered questions within human disease research, they may be drawn to this field of study [4]. As they are introduced to clinical genetics, they will be more willing to learn about careers in this sector [5] and be more willing to work with personnel in these settings regardless of their actual career choice. The earlier students are exposed to these options, the more able they are to pursue research as undergrads and to prepare for a career in one of these many fields.

Furthermore, the public at large has a poor understanding of genetic concepts [6]. This causes people to disapprove of genetic technologies and lack trust in genetic research and genetic professionals [7:11]. Technology that is currently available to the public is thus underused due to distrust and misinformation [12].

Previous research shows that students who are given hands on experiences with difficult concepts, like personalized genomics (sequencing, analyzing and interpreting your own DNA), in-lab genetics or plant pathology, will have a retention of these concepts and be more interested in learning more about these concepts and applying these concepts beyond classroom requirements [13:16]. It has also been shown that teaching genetic concepts by building upon foundational knowledge in hands-on meaningful lessons improves students' understanding of genetics [15,

Todd]. Increasing experience with and understanding of difficult genetics concepts in medical students led to increased support for research in those fields and for use of clinical genetics in the student's own practices [4].

The general public lacks an understanding of how genetic technologies are beneficial, lacks favorable attitudes towards using genetic technologies and has a high belief in genetic determinism [6]. This is specifically alarming in students who should use this knowledge later in their professional careers [4,17]. Currently, research has not been done to show any effect of having a specific real-world example lecture devoted to improving understanding and acceptance of these practices.

In this study, we gave students a pre-survey, allowed students to attend class for one week, and then distributed a post-survey. These surveys measured changes in students' tendency to view genetics as deterministic, their understanding of the application of specific genetic technologies, and their willingness to receive treatment or diagnosis using these technologies [18]. The survey specifically addressed technologies that include gene therapy, genetic testing, prenatal genetic testing and personalized genomics and pharmocogenetics (which is the study of identifying the most effective drugs for an individual based on their genes). During the week between surveys, one class was taught an intervention lecture that focused on clinical genetics and its application when diagnosing and treating tuberous sclerosis. The control class continued into the next unit with normal academic lectures.

The purpose of this study was to determine if an in-class lecture specifically discussing clinical genetic concepts and benefits when solving difficult polygenic diseases (diseases caused by an interaction of multiple genes), like tuberous sclerosis, would shift student's attitudes more favorably towards genetic testing and prenatal testing and increase their knowledge of genetics in general.

Methods

Students were asked to take a pre-survey evaluating their attitude towards genetic determinism, knowledge about genetics and attitude towards modern genetics. Students in the experimental group took part in an in-class learning experience that exposed them to the complexity of variant classification. The lecture included a brief introduction into variant classification, a real-life scenario of variant classification, and students searching online to solve the scenario. As part of the wrap up, students were told the options that an individual would have in the case study and how genetic technologies would enable the individual and health care professionals to make the best decision with knowledge. Students were also introduced to ways in which they can be part of clinical genetics now and in the future. Students in the other class did not receive this lecture and instead moved forward to the next unit in their academic curriculum. After the class, students in both treatment groups were given a post-survey identical to the pre-survey to see how their views and knowledge had shifted.

Ethics statement

The Institutional Review Board for Human Subjects at Brigham Young University approved this research and granted permission for use of human subjects in this study; written consent was obtained from all participants.

Sampling

Students were invited to participate from two different sections of an introduction course to Biology for Majors that were taught by two different professors. Each student was required to participate in two surveys and was given course credit for completing the survey. In the survey, consent was received from the students to use their survey data in this research. Each class had 130 participants, a total of 260 potential subjects.

After given the opportunity to complete both surveys, only students who fully completed both the pre- and the post-survey were used in the study, all others were discarded. A total of 117 students from the intervention class and 22 students from the normal class completed both the pre- and post-survey and were therefore used in this study. It seems that receiving class credit for this assignment was not emphasized to the control class and therefore reduced the sample size. Data is available upon request.

Data collection

The survey distributed to the students was designed by Carver et al. to measure Public Understanding and Attitudes towards Genetics and Genomics

(PUGGS) and emphasizes the respondent's understanding of genetic determinism and attitudes toward various genetic technologies [18]. Of specific interest in this study were the students' responses about understanding genetic determinism and their attitudes towards genetic testing.

All students in the two different classes were given a link via their learning platform to a Qualtrics survey that displayed all the questions from the PUGGS questionnaire.

Responses were coded based on the PUGGS key to compute scores for both the pre- and post-surveys. To find the difference in students' understanding and attitudes, the pre-survey score was subtracted from the post-survey score.

Classroom comparison

Each section of the BIO 130 class was taught by a different professor with a different teaching style, but both sections had similar learning outcomes and basic curriculum for the course as a whole and specifically for the unit on genetics. Each section had one week remaining in their unit on Genetics when the PUGGS pre-survey was available to take, and it remained open until the night of the intervention lecture. When the experimental class received the intervention lecture, the other class moved on to their next academic unit. Both classes then had the opportunity to take the post-survey for a week after the intervention (treatment) or normal (control) class period.

Intervention lecture

Two guest presenters gave the intervention lecture instead of the class's typical professor. The lecture focused on the importance of reliability in clinical genetic testing by teaching what genetic testing is, how genetic testing can be applied, and how gene variants are classified through a real-life example.

Students were taught that a clinical genetic test informs clinicians about why a certain disease may be caused and allows the clinician to better treat the disease. Limitations of genetic testing were discussed after explaining what a genetic test is. Four limitations were addressed: we only know a fraction about the entire human genome, what we do know tends to be probabilistic and not concrete, polygenic diseases are more common than monogenic diseases and harder to know which gene(s) cause the disease, and epigenetics and environmental factors are always at play, but it is difficult to know the exact effect of these factors on the disease phenotype.

Students were then taught about six different types of genetic testing and why they would each be used. These included pharmacogenetic testing, newborn screenings, diagnostic testing, predictive and pre-symptomatic testing, carrier testing and prenatal testing. Examples of scenarios in which each of these tests would be used were presented to the students. Cancer treatments were an example of using pharmacogenetics, Cystic Fibrosis *CFTR* testing was used as an example for both newborn screenings and carrier testing, and *BRAC1* and Angelina Jolie illustrated the use of predictive and pre-symptomatic testing.

When prenatal testing was discussed, abortion and abortion laws were touched on as well as the benefits of having prenatal testing done even if the couple has no plans of carrying out an abortion.

The lecture then switched to focusing on how genetic variants are classified and why this is important in receiving accurate genetic test results. Tuberous sclerosis was introduced as our disease of interest and phenotypic malformations like benign brain tumors were considered. The variants in *TSC1* and *TSC2* that are associated with tuberous sclerosis were introduced, including statistical information about how common the disease is and how it is inherited.

In particular, the class was told that *TSC2* c.2476 C>A (p.Leu826Met), a mutation in the *TSC2* gene, is a genetic variant that was associated with tuberous sclerosis. Without being told how this variant was classified, the class was divided into two random groups and asked to read one of two papers about this variant. Students were asked to decide based on the information in the paper if the variant caused tuberous sclerosis or not. They were guided to read the abstract and some of the results that had been highlighted previous to the lecture. Highlighted portions typically illustrated the main conclusions that the authors were drawing from their experiment. Both papers concluded that *TSC2* c.2476 C>A (p.Leu826Met) caused tuberous sclerosis [19,20].

Upon coming to this conclusion, students were told that this was a misclassification and that in fact more people carry that mutation than have the disease. They were taught about the classification phrase, “too common to cause disease.” When a variant is too common to cause disease, it is seen in the general

population at a higher rate than the disease it supposedly causes. This implies that the variant cannot actually cause a rare disease; otherwise, we would expect more people with the variant to also have the disease.

After introduction of this idea, students were exposed to the idea of crowdsourcing primary literature to better understand which variants have statistically significant research results indicating that they are pathogenic and how this in combination with clinical genetics can improve clinical diagnosis of complex genetic diseases. They were invited to be a part of this initiative and were then given an opportunity to ask questions.

Instrumentation: PUGGS survey

Students took the Public Understanding and Attitudes towards Genetics and Genomics Survey (PUGGS) created by Carver et al. [18]. It consists of five different sections: background information, belief in genetic determinism, knowledge about gene-environment interaction, knowledge about modern genetics and genomics, and attitudes towards applications of modern genetics and genomics. Students took the survey as written by Carver et al. except they did not complete the background information.

The survey was distributed to students through a Qualtrics survey link that was available to students online via an assignment in their learning management system. Each section included either Likert-scale or true/false questions in order to gauge attitudes towards and correct knowledge of the different topics. For the most part, the questions focused on examples within human genetics.

Belief in genetic determinism

The first section was about the student's belief in genetic determinism. Students were given a chart listing various traits, including physical traits, personality traits and diseases, and then asked to indicate if the trait is influenced by environmental differences or genetic differences. This was done with a Likert-scale response format with the options, "Only environmental difference contribute to the trait", "Mainly environmental differences contribute to the trait", "Both genetic and environmental differences contribute to the same extent to the trait", "Mainly genetic differences contribute to the trait" or "Only genetic differences contribute to the trait." Answers to this set of question reflect an individual's belief that genes influence phenotypic traits more than environmental factors. A low score means students do not understand which traits are caused by genes or the environment and that they think genes play a larger role in the development of these traits.

For categories that are genetic, like color-blindness and blood type, students received a score of 4 for saying "Only genetic differences contribute to the trait," and a score of 0 for anything else. They received points for knowing that genes are in fact fully responsible for these traits, but lost points for any other response.

For categories that are environmental, like interest in fashion, political beliefs and religious beliefs, students received a score of 4 for indicating that "Only environmental difference contribute to the trait." All other answers received a score of 0. Again, they received points for correctly understanding which traits are caused by genes and which are not, but lost points if they lacked this understanding.

For the remaining categories that have both a genetic and environmental component such as height, developing cancer, or diabetes, students received a 4 for indicating, “Both genetic and environmental differences contribute to the same extent to the trait.” A high score indicates correct understanding of gene and environment interaction and thus weighing these traits equally shows that the students understands both factors are at play without overemphasizing the role of genes.

Since these traits are influenced by both genetics and environment and since a lower score means that students are more likely to believe that genes have a stronger influence than environmental factors, the remaining categories were scored as follows: 3 for “Mainly environmental differences contribute to the trait,” 1 for “Mainly genetic differences contribute to the trait,” and 0 for both “Only environmental difference contribute to the trait” and “Only genetic differences contribute to the trait.” Higher points were awarded for acknowledging that both genes and environment influence these traits, but more points were awarded for stating that environmental differences play a larger role. An example of this would be height. An individual can have genes that are favorable for tall height, but if they are raised in poor environmental conditions where they do not receive enough food, tall height will not occur. Therefore, only 1 point was awarded for listing genetic factors as the main contributor to these types of traits. 1 point was awarded instead of 2 because this statement shows a lack of correct knowledge and it also shows a tendency to believe more deterministically and a low score indicates belief in genetic determinism. Finally, scores of 0 were awarded to both extremes because

these students do not have a correct understanding of what influences a phenotypic trait.

The score for each individual question was then added up to create a total pre- and post-score for this section of questions. A high score indicates that students have an accurate understanding of which traits are caused by the environmental, genetic or a combination of factors. Meanwhile, a low score indicates a belief in genetic determinism due to the fact that the student overestimates or misunderstands the role that genes plays in these traits.

Gene-Environment and modern genetics and genomics knowledge

The next two sections, "Gene-Environment Knowledge" and "Modern Genetics and Genomics Knowledge," consisted of true and false questions about whether and how environment and genes interact, characteristics of the genome, gene function and expression, and epigenetics (molecular changes to DNA that affects protein expression and thereby phenotype) [18]. One point was given to a student for correctly answering the question, and no points for any incorrect answer. Again, the score for each section was added together for a total score to be used to compare the pre- and post-survey scores.

Genetic technology

The final section focused on student attitudes towards genetic technology. This section was composed of four subsections: gene therapy, genetic testing, prenatal genetic diagnosis and personalized medicine and pharmacogenomics. Each

category had a list of statements that students were asked to share their feelings about. Using a Likert-scale, they could respond “Strongly Disagree”, “Disagree”, “Agree”, or “Strongly Agree.” These statements included ideas like being willing to use genetic technologies, feeling apprehension about new genetic technologies and when it may or may not be appropriate to use these technologies. Attitudes that were strongly positive received a score of 3, mostly positive a 2, mostly negative a 1, and strongly negative a 0. A high score indicates that a student has favorable attitudes towards applications and development of genetic technology, while a low score reflects negative feelings and apprehension towards these same applications.

To determine students’ overall understanding and feelings towards genetic technology and genetic concepts, the scores of each subsection were added together. For consistency, all sections scored with a Likert-score were divided by the highest possible score value (either a 4 or a 3) so that all answers would be standardized to a correct or more favorable answer receiving a score of 1 and an incorrect or unfavorable answer receiving a score of 0.

Statistics

The samples were first analyzed for normality using the Shapiro-Wilk normality test. Each category (“Gene-Environment Knowledge”, “Modern Genetics and Genomics Knowledge”, “Gene Therapy Attitudes”, “Genetic Testing Attitudes”, “Prenatal Genetic Testing Attitudes”, “Personalized Medicine and Pharmacogenetics Attitudes”, and “Total Score”) has a $p < 0.001$, indicating data that are not normally distributed. Therefore, we assumed all categories violated assumptions of normality.

Simple summary statistics were run for each category for each treatment using R. Outliers were identified in the dataset using the rule that an outlier is any point that falls one and a half times the inter-quartile range below quartile one or any point that falls one and a half times the inter-quartile range above quartile three. Rather than reducing an already small data set, students were considered to be outliers only if he or she was an outlier in his or her "Total Score" on the PUGGS survey. These students were excluded from any further analysis. This reduced our sample size to 105 subjects who received the intervention lecture and 21 subjects in the normal classroom.

To test whether scores between the pre- and post-surveys differed between the intervention and normal classroom groups, we compared the classes using a Mann-Whitney U non-parametric two-sample test on each questionnaire category. To test whether scores within the individual treatments significantly increased or decreased, Wilcoxon signed rank tests were performed. All tests, including the Shapiro-Wilk, Mann-Whitney U, and Wilcoxon signed rank tests, were run in R.

Results

Having an intervention lecture focusing on genetic variation and testing improves understanding and attitudes towards genetics concepts and genetic testing. Overall, students who received the intervention lecture were both more knowledgeable and felt more favorable about the ideas presented in the survey (Fig 1A, B, C).

Genetic testing attitudes

The most significant result seen is that of the interventions group's attitude towards genetic testing (Fig 1A). The experimental group showed a more positive attitude of genetic testing when compared to the control group ($p = 0.02$, $U = 797$, $Z = 2.05$). The control group had a more negative view of genetic testing after their unit on genetics ($p = 0.01$, $Z = -2.33$), whereas the experimental group had an almost significant result of being more favorable after the intervention ($p = 0.078$, $Z = 1.42$).

Prenatal genetic diagnostic attitudes

Students who received the intervention lecture had a more favorable view of prenatal genetic testing than their counterparts ($p = 0.05$, $U = 857$, $Z = 1.63$) (Fig 1B). The experimental group moved towards more favorable attitudes ($p = 0.001$, $Z = 3.09$) in comparison to the control group ($p= 0.82$, $Z = -0.23$).

Modern genetics and genomics knowledge

The intervention group showed no change in understanding of Modern Genetics and Genomics while the control group seemed to decrease in understanding ($p = 0.05$, $U = 852$, $Z = 1.65$) (Fig 1C). The median in the experimental group is 0, showing no change in knowledge but the control group decreased by a point, showing less understanding ($p = 0.016$, $Z = -2.14$). This shows that students who received intervention had a better retention of modern genetics knowledge.

Gene-Environment interaction knowledge

In both the experiment and the control groups, no change was seen in their knowledge of which traits are influenced more by genes or environment ($p = 0.15$, $U = 951$, $Z = 1.02$) (Fig 1D).

Gene therapy attitudes

In both the experiment and the control groups, no change was seen in their attitudes towards gene therapy ($p = 0.18$, $U = 966$, $Z = 0.92$) (Fig 1E).

Personalized medicine and pharmacogenetics attitudes

In both the experiment and the control groups, no change was seen in their attitudes towards personalized medicine and pharmacogenetics ($p= 0.41$, $U = 1067$, $Z = 0.24$) (Fig 1F).

Belief in genetic determinism

The understanding that genes alone do not determine your phenotype, nor are genes always the strongest predictor of phenotype, did not change significantly between the experimental group and the control ($p = 0.88$, $U = 1283$, $Z = 1.19$), as seen in Fig 1G.

Total score

Overall when all scores are summed together, there was no significant change in students' understanding of genetics and in their attitudes towards genetics and genetic technologies (p = .08, U = 891, Z = 1.38) (Fig 1H).

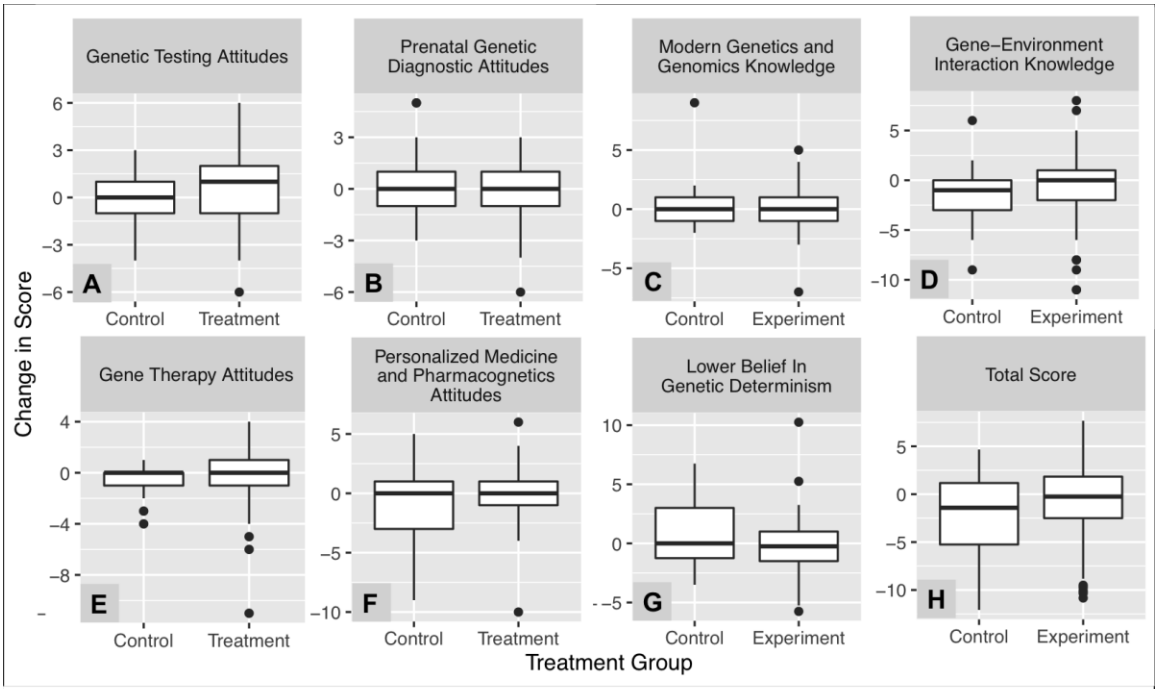

Figure 1: Changes in Student Survey Scores. Students took the PUGGS assessment before and after either an intervention lecture in the experiment group or a normal class period in the control group. Statistically significant differences are evident in Genetic Testing Attitudes (A: p = 0.02), Prenatal Diagnostic Attitudes (B: p = 0.05), and Modern Genetics and Genomics Knowledge (C: p = 0.05). Statistical differences are calculated based on Mann Whitney U test. (Panel D: p = 0.15; E: p = 0.18; F: p = 0.88; H: p =0.08)

Discussion

Having an in-class intervention lecture that focuses specifically on clinical genetics and making this process real to students through an authentic case study increased students' knowledge of modern genetics and genomics and led to more favorable attitudes towards genetic testing and prenatal genetic diagnosis. In our study, we showed that student opinions of genetic testing and pre-natal diagnostic

testing became more positive when a real-life scenario about a genetic disease was used to illustrate how different genetic technologies (including genetic testing and prenatal diagnostics) enable a more informed opinion on how to proceed with medical treatment. This implies that these students will be better prepared to receive genetic testing or other clinical genetic technologies in the future [21]. Additionally, they will be more likely to understand how genetics can benefit their field of interest and be open to applying genetics within their field [4].

Knowledge of Genetics

The survey given to the students assessed their knowledge of basic genetic principles, including true and false questions assessing whether and how genes and the environment interact and a section of true and false questions about genetics concepts typically taught in a high school biology classroom [18].

Having a correct understanding of genetics is vital to understanding why research should be done in this field and how and why genetic technologies are helpful in a research setting. We did not expect to see a difference between the two groups in this area since the intervention lecture did not focus on teaching basic genetic truths, but expounded upon what the students should already know. In fact, we expected both classes to increase in this area due to the similar class curriculum that was followed during the time frame of the survey, but not due to the intervention.

However, we observed a significant decrease in the control group's understanding of basic genetic and genomic concepts in comparison to the

intervention group (Fig 1C). This may be due to a decrease in retention of new information in a setting that did not stress hands-on interaction with the material, differences between the professors' teaching style previous to the intervention, or due to the small sample size of the control group.

Attitudes towards genetic testing and pre-natal diagnostics

For the most part, a person's view of genetic technology is positive as long as it is seen to maintain a natural order, meaning that it enables an individual to live a more normal life [18]. Unfortunately, clinical genetics is often not viewed positively [22]. There are mixed opinions from different studies indicating that knowledge in genetics may lead to either more positive or more negative attitudes towards genetic technologies [8:10,23,24].

In our study, we showed that student opinions of genetic testing (Fig 1A) and pre-natal diagnostic testing (Fig 1B) became more positive when students were exposed to a real-life scenario about a genetic disease and when they were taught how different genetic technologies (including genetic testing and prenatal diagnostics) enable a more informed opinion on how to proceed with medical treatment.

Therefore, in addition to teaching about basic genetic technologies, the applications of these technologies need to be explained in the context of real-life scenarios. Teaching in the context of real life, gives the students a more informed view of the benefits of these technologies and how they enhance medical decisions.

Students have more favorable views when medical and other applications are relevant to their own experience (Frymier, Roszkowski).

Limitations

One of the largest limitations of this study is the small sample size, particularly in the control population. While the study was designed with the intent to have two samples of about 100 students each, this expectation was not met due to low participation by the partnering teacher's class. Further research may better clarify these conclusions when a larger sample is used.

An additional limitation was the experimental setup in the study design. It was not possible within the scope of this experiment to randomly assign students to either a normal class lecture or to the intervention lecture. Therefore, there may be other confounding variables influencing these results. These include having two different professors teaching each class, two different teaching of the professors, and having the experimental group spend an extra day on the topic of genetics compared to the control class. Further studies in which students are randomly assigned to one of two treatment groups each of which is instructed by the same professor may better clarify these conclusions.

Conclusion

Having an in-class lecture devoted to clinical genetics and applications of genetic technologies in a real-life scenario increases students' knowledge of modern genetics and leads to more positive attitudes towards genetic testing and pre-natal

diagnostic testing. Courses that teach genetics should approach genetic technologies and modern application, especially medical applications, by walking students through a real-life scenario.

Further research should be done to validate these findings and to build upon them. Specifically, future research should determine if the number of exposures to real-life applications correlate to student's attitudes and understanding. Additionally, determining if student engagement with a professional in the field correlates to better attitudes and understandings should also be studied.

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