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BRIEF REPORT

REVISED Assessing knowledge of genetics in undergraduate students in Quito, Ecuador [version 2; peer review: 2 approved]

David Ortega-Paredes ^{1,2}, César Larrea-Álvarez², Michelle Herrera¹,

Esteban Fernandez-Moreira¹, Marco Larrea-Álvarez^{2,3}

¹Medicine School, Universidad de las Américas, Quito, Udlapark, Via a Nayón, Quito, 170124, Ecuador
²Education Unit, Life Science Initiative, Julian Estrella, Quito, 170607, Ecuador
³School of Biological Sciences and Engineering, Yachay-Tech University, Hacienda San José, Urcuquí, Imbabura, 100650, Ecuador

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Abstract

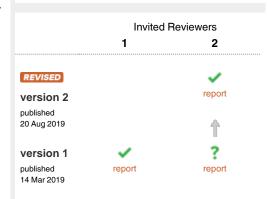
Knowledge of genetics is crucial for understanding genetic and genomic tests and for interpreting personal genomic information. Despite this relevance, no data are available about the level of knowledge of genetics in an Ecuadorian population. This investigation sought to survey such knowledge in undergraduate students affiliated with private and public institutions in Quito, the capital city of Ecuador. A total of 350 individuals responded to a validated questionnaire measuring knowledge of genetics. Scores ranged from 45% to 87% (mean: 66.8%), and students achieved slightly better results when asked about genetics and diseases (mean score: 68.3%) than when asked about genetic facts (mean score: 64.9%). Additionally, no significant differences in performance were found among students from private and public institutions. Surprisingly, the lower score obtained (45%) was from a guestion about how chromosomes are passed to the next generation. The highly educated status of the surveyed population could explain the overall results; nonetheless, the possibility that the correct responses were given by chance cannot be ignored. Therefore, the actual knowledge of genetics among the participants might be different than that revealed by the percentages of correct answers. Consequently, to achieve the goal of ensuring informed decision-making concerning genetic and genomic tests, it seems evident that the national education programs of Ecuador require improvement in the teaching of genetic concepts.

Keywords

Ecuador, knowledge of genetics, genetic literacy, undergraduate students, survey

Open Peer Review

Reviewer Status 🗸 🗸



- 1 Vasiliki Mollaki D, Hellenic National Bioethics Commission, Athens, Greece
- 2 Rebecca Carver (D), Norwegian Institute of Public Health, Oslo, Norway

Any reports and responses or comments on the article can be found at the end of the article.

Corresponding authors: David Ortega-Paredes (daortegap@gmail.com), Esteban Fernandez-Moreira (esteban.fernandez@udla.edu.ec), Marco Larrea-Álvarez (malarrea@yachaytech.edu.ec)

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Competing interests: No competing interests were disclosed.

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REVISED Amendments from Version 1

All observations were welcomed and much appreciated. Indeed, these permitted to improve the overall manuscript. Firstly, the misinterpretation of the terms "perspective", "attitude" and "actual knowledge" has been corrected. Moreover, sentences making use of the mentioned terms have been omitted or modified. Secondly, a justification for using such an instrument of measurement has been asked for. A detailed explanation is newly provided in the methodology section, explaining the reason why such a survey was chosen. Thirdly, the authors were asked to comment on the percentages of correct answers needed to qualify as "adequate" an amount of knowledge. The original and updated surveys do not provide a reference value for considering a percentage of correct answers as "adequate" (Fitzgerald-butt et al., 2016). Instead, they state that their study provides adequate references for knowledge of genetics. Therefore, we have avoided the use of "adequate" from the text. Additionally, the paper stated that the lower scores obtained on a question about how chromosomes are passed to the next generation were considered surprising. Nonetheless, relevant research has shown that the content of genetics in textbooks is lagging behind scientific developments. Indeed, the literature suggested by the reader (Gericke et al., 2014) has permitted to provide a better explanation of the results. Finally, the authors conclude that the possibility of correct responses was given by chance cannot be ignored - and that the actual knowledge might be less than that revealed by the answers. However, and as the reader indicates, if the responses were given by chance, it would be possible that the knowledge could be higher. We agree on this and have suggested, for further research, to add a section where the interviewed is asked to provide a degree of certainty of his or her answer, which could increase the efficacy of measuring.

Any further responses from the reviewers can be found at the end of the article $% \left({{{\bf{r}}_{\rm{s}}}} \right)$

Introduction

Genetic and genomic testing have transformed our understanding of our health, personal well-being and recreational consumerism. Advances in powerful and cheap genetic analyses have allowed new opportunities to generate information about important conditions, such as cancer, diabetes, and cardiovascular diseases (Burton, 2015; Perkins et al., 2018; Rafiq et al., 2015; Roberts & Middleton, 2018; Wu et al., 2019). In recent years, access to pharmacogenomics, nutrigenomics, disease risk, ancestry and ethnicity tests, as well as access to sport genetic analyses, has become widespread in low- and middle-income countries. Such genetic and genomic practices are carried out by health care institutions and, moreover, direct-to-consumer (DTC) genetic tests are easily available on the internet (Covolo et al., 2015; Phillips, 2016). In Ecuador, a case study by the Red Cross found that rape, intimate partner violence and femicide rates are high. Ecuadorian laws offer mothers the right to ask for a free paternity test; a positive result automatically obliges fathers to provide support for their children. Additionally, genetic tests are routine in Ecuador for police investigating rape cases. For these reasons, increasing knowledge about how DNA can be a link between parents and children or between a sexual offender and a crime seems to be a powerful tool for women's empowerment. Several studies have demonstrated that the understanding and interpretation of personal genomic information is biased by one's own knowledge and appreciation of basic genetic facts, namely, their level of genetic literacy (Hooker et al., 2014; Lea et al.,

2011; Lontok et al., 2015; Rafiq et al., 2015). Evidently, a basic amount of genetic knowledge is essential to understand and interpret the results of genetic and medical analyses. Therefore, various studies have focused on assessing the impact of knowledge of genetics on perception of genetic facts and understanding of disease onset (Haga et al., 2013; Hollands et al., 2016; Lea et al., 2011). Unfortunately, despite the obvious necessity to determine knowledge of genetics, to our knowledge there is no available information regarding this matter in our country. Moreover, recent research has shown differences in quality between public and private higher education institutions in Colombia (Cayon et al., 2017). Therefore, it seems important to assess such differences in Ecuador. The data gathered from these kinds of studies could contribute to the development of programs to reinforce the teaching of genetics to a wider population, which will undoubtedly have a positive impact on national educational programs. Therefore, as a baseline report, we decided to determine the basic knowledge of genetics in undergraduate students in Quito, the capital city of Ecuador. This study provides on the student understanding of genetic concepts and the relation of genetics to disease in a relatively highly educated population based in a developing country. Furthermore, this investigation represents one of the first steps required for building the appropriate strategies to comprehensively assess knowledge of genetics and to ultimately increase the level of genetic literacy in the region.

Methods

Setting, recruitment and questionnaire

The main objective of this research was to assess the competence of undergraduate students, who do not follow programs involving biologically related courses (n=350 by convenience sampling method), to respond to a validated survey evaluating a minimum, amount of knowledge about genetics (Fitzgerald-Butt et al., 2016). This particular questionnaire is suitable at low knowledge levels and was developed for older teenage and young adult patients, along with parents in a pediatric setting. This survey was chosen because the targeted population is not involved in the life sciences/biology area, and, thus, are not prominently exposed to this type of information. Moreover, it appears useful to use an instrument measuring basic knowledge as a baseline report, especially in a region where no information about the competency of students in genetics is available. Surveys were carried out from August to October 2018. Individuals were recruited from 3 public and 4 private institutions located in Quito, the capital city of Ecuador. The identity of the institutions was handled in an anonymous form. The participants were approached at random inside the campuses and asked to fill out a questionnaire consisting of 18 statements, provided in Dataset 1 (Larrea, 2019), which measured both the actual knowledge of the associations of genetic conditions with diseases and the actual knowledge of genetic facts. For each question, the results are presented as the percentage of correct answers.

Statistical analysis

Pearson's chi-square test was used to determine the likelihood that the results (answers) supporting the null hypothesis are not due to chance. Additionally, Student's *t*-test was used to assess

whether the two groups, composed of publicly and privately educated students, presented any significant differences regarding their measure of knowledge about genetics (assuming equal variances). *P* values are reported using a Type I error level of 0.05, 0.01 and 0.001. All data analyses were carried out with MATLAB® version 9.9.9341360 (R2016a). A MATLAB script to repeat the analysis is available in Dataset 2 (Larrea, 2019).

Ethics approval

This survey was performed under the format of "common social topics". Because of the low-risk nature of the study, approval from a committee was not sought. The participants were informed about the objective of the questionnaire; the survey was voluntary and anonymous, and information that could put the person at risk was not collected. All surveyed students provided prior verbal consent. Written consent was not sought from the participants due to the low-risk nature of the study.

Results

In this research, we present the data gathered as a reference study outlining the knowledge of genetics in undergraduate students. Overall, 350 participants were enrolled in this research (average age: 21.8 years old, SD: \pm 2.8); individuals came from diverse backgrounds that did not involve life sciences or medicine. The results varied from 45% to 87% (mean: 66.8%, median: 65%) (Table 1). The responses to each question can be found in Dataset 3 (Larrea, 2019). The percentage scores were higher for the subsection regarding the relationship between genetics and the presence of illness (mean: 68.3%). The lower scores within this section were observed when

Table 1. Knowledge of disease related-concepts and genetic facts of undergraduate students with percentages of correct answers.

	Total popula (n=350)			Private institutions (n=170)		Public institutions (n=180)	
Disease-related concepts		% correct	p-Value ^a	% correct	p-Value ^a	% correct	p-Valueª
1.	Some diseases are caused by genes, environment and lifestyle. (T)	87	<0.001	89	<0.001	85	<0.001
2.	A gen is a disease. (F)	61	<0.001	63	<0.001	65	<0.001
3.	Healthy parents can have a child with an inherited disease. (T)	74	<0.001	76	<0.001	71	<0.001
4.	A person with altered (mutated) gene may be completely healthy. (T)	55	0.069	57	0.011	53	0.443
5.	All serious diseases are inherited. (F)	56	0.019	54	<0.001	59	0.014
6.	The child of a person with an inherited disease will always have the same disease. (F)	58	0.002	58	<0.001	58	0.032
7.	Altered (mutated) genes can cause disease. (T)	84	<0.001	88	<0.001	81	<0.001
8.	A genetic test can tell you if you have a higher chance to develop a specific disease (T)	80	<0.001	82	<0.001	78	<0.001
Av	erage percentage for this section	68.3		70		68.4	
Genetic facts							
1.	You can see a gene with the naked eye. (F)	59	<0.001	58	<0.001	61	0.004
2.	Genes are instructions for making proteins, which help the body grow and work properly. (T)	57	0.008	61	0.004	54	0.357
З.	A gene is a piece of DNA. (T)	77	<0.001	74	<0.001	80	<0.001
4.	Genes are inside cells. (T)	69	<0.001	71	<0.001	71	<0.001
5.	A chromosome contains many genes. (T)	78	<0.001	77	<0.001	79	<0.001
6.	Genes determine traits such as height, eye color and facial appearance. (T)	84	<0.001	82	<0.001	86	<0.001
7.	A person has thousands of genes. (T)	73	<0.001	74	<0.001	73	<0.001
8.	Identical twins have different sets of genes. (F)	49	0.915	47	0.307	53	0.443
9.	Humans have 20 pairs of chromosomes. (F)	58	0.004	53	0.027	61	0.004
10	. Parents pass both copies of each chromosome to their child. (F)	45	0.054	49	0.610	41	0.014
Average percentage for this section		64.9		63.8		68.6	
Overall average percentage		66.8		66.6		67	

^ap-values for determining answers provided by chance were calculated using Pearson's Chi squared test. T, true; F, false.

individuals were asked about the inheritance of diseases (mean: 56%, p=0.019) and when questioned about the health status of a person carrying an altered gene (mean: 55%, p=0.069). The percentage scores were lower for the subsection regarding genetic facts (mean: 64.9%). In particular, the students seemed to have difficulty answering correctly when asked about the quantity of chromosomes present in humans (mean: 58%, p=0.004) and about the number of copies of each chromosome passed down to the next generation (mean: 45%, p=0.054). In addition to the lower scores, the hypothesis that the questions were answered correctly without any previous knowledge (provided by chance) could not be significantly rejected. Generally, no differences in the overall knowledge of genetics could be found among students enrolled in private and public institutions (p=0.9405). Likewise, no differences between these two groups were observed regarding disease-related questions (p=0.7844) and genetic facts (*p*=0.7318).

Discussion

In this report, we portray the percent of correct answers to an 18-item questionnaire measuring a minimum amount of knowledge about genetics. Overall, this Andes-located population of undergraduate students demonstrated some basic knowledge toward genetic concepts and their relation to diseases. Nonetheless, student knowledge on facts about genetic proved to be less strong. This tendency was observed in both privately and publicly educated individuals with no significant difference. These results are lower in comparison to the published reports on general populations that have made extensive use of similar survey instruments to determine knowledge about genetics. For instance, Haga & colleagues (2013) found higher scores in a general population based in the US. However, similar scores to those reported here were found by Jallinoja & Aro (1999) in a study performed on a general population in Finland. Furthermore, a group composed of adolescents and young adults suffering from congenital heart disease scored similar results (Fitzgerald-Butt et al., 2016). Notably, the present results are somewhat higher than those obtained from a Dutch population suffering from asthma, diabetes mellitus type II and cardiovascular disease (Calsbeek et al., 2007). It is evident that demographic differences may account for the variances in the results. Nevertheless, these results may also imply notable differences between Ecuadorian, US and European science and health education programs (Lontok et al., 2015). The lowest scores obtained were for the two questions involved in how chromosomes are passed down to the next generation. These outcomes might be related to the hitherto reported conceptual variation in biology textbooks, which have been shown to have detrimental repercussions regarding the students understanding of conceptual knowledge, models in particular, within the context of genetics (Gericke et al., 2014; Gericke et al., 2013). This means that students may not understand the power of genetics to address important issues for the Ecuadorian population, such as determining paternity, solving crimes or understanding our ethnic genetic background. To the best of our knowledge, this study is the first to report a measurement of knowledge of genetics in an Ecuadorian population.

Additionally, the presented results indicate that the probability of participants providing correct responses by chance could not be significantly discarded (Table 1). This fact implies that the actual knowledge might be different from the one asserted by the percentages of correct answers. Therefore, to have a better understanding on the actual knowledge, we suggest to implement, for each question, a section in which the interviewed is asked to provide a degree of certainty for his or her answer. Indeed, such an analysis has been applied to measure diabetic patients' knowledge about the illness, and was shown to be useful in determining more efficaciously their degree of mastery about the subject (Leclercq, 2010). Individuals affiliated with private and public universities responded with similar accuracy. The observed average scores might reflect the high level of education of the respondents. It is worth mentioning that the interviewed people did not follow any biologically/medically related courses. Furthermore, this study provides adequate estimates of the knowledge of genetic and its relation to disease in a non-specialized population. It is important to note that the participants' knowledge may not be as strong as it appears. As mentioned earlier, the scores do not differ substantially from the earlier studies making use of similar surveys. Nonetheless, the scores were lower than those obtained from a study performed in the US (Haga et al., 2013) where genetic education is constantly improving (Lontok et al., 2015). Based on these observations, a revision of the genetic content covered in educational programs and the implementation of science popularization initiatives seem imperative.

Some limitations of this study should be mentioned. This investigation did not attempt to address the differences in knowledge of genetics among groups classified by characteristics such as sex, ethnic group, age, family history of inherited diseases or level of education. Instead, this study was intended to be focused solely on a general undergraduate population not studying biology or medicine. Furthermore, more universities in different cities should be sampled to have a national perspective on students' insights about genetics. Overall, these results provide a glimpse of the students' standpoint toward genetics and its involvement in disease. Nevertheless, more effort is decisively needed to design and execute plans that will ensure an optimized method to measure knowledge of genetics in a larger and more diverse population. The data generated using these approaches will be proven essential when designing educational programs involving genetics and health. The application of such programs will be fundamental for the general population to avoid misunderstandings about genetics and to avoid the incorrect utilization of scientific terms.

Follow-up studies will try to explore the knowledge about genetics and the attitudes toward related subjects, including genetic testing, stem cells, regenerative medicine and genetically modified organisms (GMOs). The expected results will provide improved insight into the population's knowledge and will serve as a foundation for developing better strategies to increase the level of genetic literacy in our community.

Data availability

Extended data

Open Science Framework: Assessing genetic knowledge in Ecuador. https://dx.doi.org/10.17605/OSF.IO/ZUVMN (Larrea, 2019)

This project contains the following extended data:

- Dataset 1.pdf (the questionnaire in the original Spanish language and its translation into English)
- Dataset 2.pdf (the MATLAB script to reproduce the analysis)

Underlying data

Open Science Framework: Assessing genetic knowledge in Ecuador. https://dx.doi.org/10.17605/OSF.IO/ZUVMN (Larrea, 2019)

This project contains the following underlying data:

• Dataset 3.csv (a spreadsheet containing all responses to the evaluation)

Grant information

The author(s) declared that no grants were involved in supporting this work.

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Rebecca Carver 🔟

Department of Communication, Norwegian Institute of Public Health, Oslo, Norway

I have now read the revised version and the authors have made appropriate changes.

Competing Interests: No competing interests were disclosed.

I confirm that I have read this submission and believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard.

Version 1

Reviewer Report 21 May 2019

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Department of Communication, Norwegian Institute of Public Health, Oslo, Norway

This study provides valuable insight into the level of knowledge of genetics in an Ecuadorian population.

Introduction

Be careful with terminology regarding "perspectives" towards genetics, as this is not the same as knowledge about genetics. The same goes for "attitudes".

Study design

The first sentence in the methods section does not make sense. How is this a "baseline report on the attitude among undergraduate students toward genetic concepts?" What do the authors mean? Knowledge is not the same as attitudes.

The authors of the chosen instrument (Fitzgerald-Butt et al 2016¹) have stated in their paper that the instrument can be used for older teenage and young adult patients and parents in the pediatric setting, and that is most informative for individuals with below average genetic knowledge. How is this instrument applicable to the current study's Ecuadorian student population, which is a very different sample population than the one for which the instrument was developed? What were the reasons for choosing this particular instrument? Can the authors justify their choice of instrument?

Results

What percentage of correct answers is "adequate" in the chosen instrument? Is there a reference value in the original instrument that can used to compare the results of this survey? (E.g. what minimum percentage of answers should the respondents answer correctly in order to have an adequate amount of knowledge?).

Discussion

The results show that students achieved slightly better results when asked about genetics and diseases than when asked about genetic facts. One possible explanation for this could be that the questions about disease relate more to people's lives than genetic facts. The authors state that they were surprised by the lower score obtained on a question about how chromosomes are passed to the next generation, but students may regard this type of "textbook knowledge" as less relevant to their everyday lives, and thus be less inclined to remember it. There is a lot of literature on how the genetics content in textbooks is lagging behind modern scientific developments – e.g. see previous literature by Gericke et al. and Dougherty et al. – and this may be an explanation for the results in this study. The authors might like to comment on this.

Conclusions

The authors conclude by saying that despite the relatively adequate score overall (66.8% correct answers), the possibility that the correct responses were given by chance cannot be ignored – and that the actual knowledge of genetics among the participants might be less that that revealed by their answers. Consequently, the authors assert; "it seems evident that the national education programs of Ecuador require improvement in teaching of genetic concepts". However, if the responses were given by chance, is it not also possible for the actual knowledge of genetics among the participants to be *higher* than that revealed here? Could the authors comment on this?

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Is the work clearly and accurately presented and does it cite the current literature? $\ensuremath{\mathsf{Yes}}$

Is the study design appropriate and is the work technically sound? Partly

Are sufficient details of methods and analysis provided to allow replication by others? Yes

If applicable, is the statistical analysis and its interpretation appropriate? $\ensuremath{\mathsf{Yes}}$

Are all the source data underlying the results available to ensure full reproducibility? Yes

Are the conclusions drawn adequately supported by the results? Partly

Competing Interests: No competing interests were disclosed.

Reviewer Expertise: Science communication, Public understanding of Genetics, Science Education, Public Health and Survey design

I confirm that I have read this submission and believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard, however I have significant reservations, as outlined above.

Reviewer Report 07 May 2019

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Vasiliki Mollaki 问

Hellenic National Bioethics Commission, Athens, Greece

The study aimed to assess the knowledge of genetics in a selected population of students from both public and private institutions.

The work is described in sufficient detail (regarding the samples size, study design and statistical analysis). The authors acknowledge the study limitations.

Although similar studies have been performed in other countries, this is the first study assessing genetic knowledge in Ecuador. The result of the study could be proved useful for policy makers and educational programs.

Is the work clearly and accurately presented and does it cite the current literature? $\ensuremath{\mathsf{Yes}}$

Is the study design appropriate and is the work technically sound? $\ensuremath{\mathsf{Yes}}$

Are sufficient details of methods and analysis provided to allow replication by others? $\gamma_{\mbox{es}}$

If applicable, is the statistical analysis and its interpretation appropriate? $\ensuremath{\mathsf{Yes}}$

Are all the source data underlying the results available to ensure full reproducibility? $\gamma_{\mbox{es}}$

Are the conclusions drawn adequately supported by the results? $\ensuremath{\mathsf{Yes}}$

Competing Interests: No competing interests were disclosed.

Reviewer Expertise: Human genetics, molecular genetics, bioethics

I confirm that I have read this submission and believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard.

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