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ORIGINAL RESEARCH

Genetic Knowledge Among Participants in the Coriell Personalized Medicine Collaborative

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Abstract Genetic literacy is essential for the effective integration of genomic information into healthcare; yet few recent studies have been conducted to assess the current state of this knowledge base. Participants in the Coriell Personalized Medicine Collaborative (CPMC), a prospective study assessing the impact of personalized genetic risk reports for complex diseases and drug response on behavior and health outcomes, completed genetic knowledge questionnaires and other surveys through an online portal. To assess the association between genetic knowledge and genetic education background, multivariate linear regression was performed. 4 062 participants completed a genetic knowledge and genetic education background questionnaire. Most were older (mean age: 50), Caucasian (90 %), female (59 %), highly educated (69 % bachelor's or higher), with annual household income over $100 000 (49 %). Mean percent correct was 76 %. Controlling for demographics revealed that health care providers, participants previously exposed to genetics, and participants with 'better than most' self-rated knowledge were significantly more likely to have a higher knowledge score ($p<0.001$). Overall, genetic knowledge was high with previous genetic education experience predictive of higher genetic knowledge score. Education is likely to improve genetic literacy, an important component to expanded use of genomics in personalized medicine.

Keywords Genetic knowledge · Genetic literacy · Health literacy · Education · Personalized medicine

Introduction

Health literacy is not solely dependent upon knowledge but rather the ability to understand healthcare information to make appropriate decisions (Ratzan and Parker 2000). The implications of low health literacy are not limited to a decreased understanding but are far more profound. Berkman and colleagues have shown that low health literacy results in an increased incidence of chronic illness, lower utilization of preventive health services and poorer self-reported health (Walter et al. 2004). Though there are many perspectives on how to improve patient outcomes, the implementation of personalized genomic medicine is one approach that is commonly cited as holding the greatest potential (Green et al. 2011; Hurle et al. 2013; Roberts et al. 2014).

As genome-based analyses become an ever larger component of clinical and preventive medical care, an understanding of basic genetics concepts as well as non-genetic risk factors for common complex (multifactorial) diseases will be increasingly necessary for individuals to make educated and informed choices regarding which tests to pursue and which actions to take to mitigate risks and improve health outcomes. Genetic education will also play a key role in keeping the general public informed of new developments in personalized

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medicine, and actively involved in any resulting public policy issues (Hurle et al. 2013; McInerney 2002; Roberts et al. 2014). Ultimately, the goal of personalizing medicine will only be fully realized with a well-educated populace, who can understand the implications of genetics for their own healthcare and become active participants in the process.

Genetic knowledge, also known as genetic literacy, refers to an individual's ability to understand and appreciate the basic principles of genetics for informed decision-making (Haga et al. 2013; Hurle et al. 2013; McInerney 2002). From a genomics perspective, this literacy should include an understanding that most common diseases (heart disease, diabetes, cancers) are complex diseases that are influenced by multiple genetic risk factors (that interact with one another as well as their environment), family history, and behavioral and lifestyle factors. To achieve genomic literacy, individuals should have the "capacity to obtain, process, understand, and use genomic information for health-related decision-making" (Hurle et al. 2013).

In the realm of basic genetic concepts the general public seems to have a grasp in some areas; however, overall, there remains a relatively low level of understanding among the majority of study participants (Bates 2005; Haga et al. 2013; Hurle et al. 2013; Lanie et al. 2004; Scheuner et al. 2008; Walter et al. 2004). For example, one of the larger studies reported to date, a Dutch survey by Henneman and colleagues, looked at the experiences, genetic knowledge, expectations for future medical genetic developments, and attitudes toward the use of genetic information among more than 800 members of the general public, aged 25 and older (Henneman et al. 2004). Overall, 57 % of respondents reported a perceived lack of genetic knowledge. Individuals with greater genetic knowledge in this study were more likely to have high self-rated genetic knowledge, younger age, high educational level, female gender, children living at home, employment as a health care provider, and familiarity with genetic testing. Moreover, while health professionals tended to score higher on genetic knowledge surveys compared to research participants that were not health professionals (Henneman et al. 2004), a systematic review of research literature related to the translation of genomic information to the management of complex disease found that health professionals report "feeling underprepared for assessing and managing genetic issues in their practice and lacking basic genetic knowledge" (Scheuner et al. 2008).

Much of the exploration of genetic knowledge that has been completed has been performed in relatively small cohorts of either members of the general public or among individuals with some exposure to genetics through either their own diagnosis or the diagnosis of a child (Bates 2005; Haga et al. 2013; Henneman et al. 2004; Hurle et al. 2013; Lanie et al. 2004). Knowledge of genetics appears to be greater in the context of heredity than in the context of the structure or function of genes (Catz et al. 2005; Christensen et al. 2010; Haga et al. 2013; Kessler et al. 2007; Lanie et al. 2004; Molster et al. 2009). One such study (Lanie et al. 2004) surveyed 62 American adults ranging in age from 22 to 80 years using two open ended questions, "Can you tell me what you mean if you say that an ability or behavior is genetic?" and "Where do you think genes might be located in someone's body?". About one third of respondents indicated that they found the question about the meaning of "genetic" to be difficult to answer, and about half of respondents offered multiple answers in their response to this question. Only 34 % of respondents correctly identified the location of genes in the body as "everywhere/all over the body/in every cell". Twenty four percent of respondents indicated that the brain or mind was the primary location of genes, followed by 14 % who mentioned DNA or chromosomes in their response.

Despite this lack of functional knowledge, previously studied individuals endorse the idea that common complex diseases are caused by multiple variables including genes, behavior, and the environment but the amount of perceived influence of each of these variables changes with the disease or trait in question (Calsbeek et al. 2007; Fitzgerald-Butt et al. 2014; Human Genetics 2001; Molster et al. 2009). To further explore genetic knowledge among a large cohort (n=4 062) including healthy individuals, individuals with chronic disease (cancer, heart disease) and health care providers, we analyzed data gathered from participants of the Coriell Personalized Medicine Collaborative (CPMC) research study to quantify genetic knowledge and assess predictors of such knowledge.

Methods

The CPMC is an ongoing prospective study investigating the impact of personalized genetic risk reports for common complex diseases and drug metabolism on health behavior and outcomes (Keller et al. 2010a, b; Stack et al. 2011). The CPMC study has received human subjects approval from the Institutional Review Boards of the Coriell Institute for Medical Research and all collaborating institutions. The research activities described here are also covered under the CPMC study IRB approved protocol.

Participants

The CPMC study has been advertised through the Coriell and CPMC websites as well as through the websites of collaborating institutions, news articles and printed materials as described previously (Keller et al. 2010). Study participants have been recruited through one of four cohorts: the CPMC community cohort (apparently healthy members of the general public, n=2 839), the Fox Chase Cancer Center (individuals diagnosed with either prostate or breast cancer, n=82), the

Ohio State University Medical Center chronic disease cohort (individuals diagnosed with either hypertension or congestive heart failure, n=201), and the United States Air Force Medical Service cohort (which included both health care providers employed by the USAFMS as well as non-medically trained employees; e.g., administrative and office staff, IT support staff, etc., n=940). Participants must be at least 18 years of age, have a valid email address, view or attend an informed consent session, sign an informed consent document, and provide a saliva sample for genetic analysis.

The informed consent session was either conducted in person by a CPMC recruiter, or by video, in which case participants viewed a video of a CPMC recruiter presenting the study background information. There was no difference in content between the in-person and video presentations as the same script was used for both presentations. The informed consent session consisted of a 45 min presentation and included an introduction to the Coriell Institute for Medical Research, the Coriell Genotyping Facility, the CPMC study, an explanation of personalized medicine, an explanation of the CPMC study design, participant requirements, risks, benefits, and alternatives to participation, as well as an explanation of the collection of participant saliva for DNA analysis. In addition, for recruiting at Fox Chase Cancer Center (FCCC), an explanation of the collaboration between FCCC and CPMC was added for context.

Procedures

After consenting and providing a saliva sample, all CPMC participants complete required medical, family history, lifestyle, demographic and medication history questionnaires through a secure web-based portal (www.cpmc.coriell.org). Participants were also offered an optional baseline genetic knowledge survey comprised of 15 knowledge questions and 4 genetic education background questions. The CPMC web portal also offers text and multimedia format educational materials and a mechanism for participants to either request an in-person or telephone genetic counseling appointment or email specific questions to a genetic counselor. Genetic counseling is optional and is available to all study participants free of charge (Schmidlen et al. 2014).

Once the required baseline questionnaires are completed, a CLIA certified in-house laboratory uses the Affymetrix Genome-Wide Human SNP 6.0 and DMET Plus genotyping arrays to generate genetic data that are used in customized risk reports to participants. For the current analyses, we excluded participants who completed the optional genetic knowledge questionnaire after viewing any personalized risk reports so as not to conflate previous genetic knowledge with genetic knowledge accumulated through participation in the CPMC study. However, we did not exclude participants that have read sample risk reports or educational materials that are publicly available on the CPMC website (www.cpmc.coriell.org).

As of June 30, 2014, 4 659 participants had completed the required medical, family, lifestyle and medication history questionnaires, and 4 062 (87 %) completed the genetic knowledge assessment before viewing any risk reports and were included in the following analyses.

Instrumentation

Participants completed a baseline genetic knowledge assessment (GKA) consisting of 15 unvalidated genetic knowledge questions which were either used in previously published studies (Christianson et al. 2010; Jallinoja and Aro 1999) or formulated for this study (see Table 1). Eleven of the 15 structured items were selected from a study that analyzed a large survey (n=1 216) with 16 items to evaluate general knowledge about genes and heredity in a population sample in Finland (Jallinoja and Aro 1999). Two questions (see Table 1) were selected from a telephone survey used in Guilford County, North Carolina (Christianson et al. 2010). Two additional items focused on complex disease and variants most often associated with complex disease, single nucleotide polymorphisms (SNPs), were developed by Coriell (see Table 1). The 15 structured questions were designed to capture knowledge of basic genetics, inheritance, Influence of gene/environment interactions on complex diseases, disease susceptibility and genetic variation.

Information relating to eight of the 15 genetic knowledge questions (3, 6, 8, 9, 10, 11, 14, and 15) was covered in the participant informed consent process either as part of the explanation of personalized medicine provided during the consent presentation or within the text of the informed consent document. Specifically, this included an explanation of the human genome (including the answer to question 11), genes (including the answers to questions 6 and 8 as well as information that is related to the answer to question 10), chromosomes (including the answer to question 9), SNPs (including information that is related to the answer to question 15), complex disease (including the answer to questions 3 and 14), and drug response. Participants would have had at least 1 month between the time they experienced the informed consent process and the time they would have received their electronic account information and been able to complete the genetic knowledge questionnaire.

Data Analysis

Following the recommendation of previous work (Jallinoja and Aro 1999), the final genetic knowledge score was calculated as the proportion of correct answers out of all 15 questions (# correct answers/15). In addition, 4 genetic education background questions included in the optional genetic

Table 1 Genetic knowledge questionnaire results: non-health care provider (Non-HCP) vs. health care provider [a] (HCP)

Genetic Knowledge Question	Answer	Non-HCP (n=3,388)	HCP (n=674)
		% Correct	
1. It is possible to see a gene with the naked eye. [b]	False	88.9	96.0
2. Healthy parents can have child with a hereditary disease. [b]	True	96.6	99.3
3. The onset of certain diseases is due to genes, environment and lifestyle. [b]	True	95.2	99.6
4. The carrier of a disease gene may be completely healthy. [b]	True	95.4	99.3
5. All serious diseases are hereditary. [b]	False	92.1	96.3
6. Genes are inside cells. [b]	True	75.7	89.6
7. The child of a disease gene carrier is always also a carrier of the same disease. [b]	False	71.0	84.3
8. A gene is a piece of DNA. [b]	True	77.7	90.4
9. A gene is part of a chromosome. [b]	True	71.1	87.2
10. All body parts have all of the same genes. [b]	True	46.7	58.8
11. It has been estimated that person has about 20,000 genes. [b]	True	37.4	42.7
12. A person's race and ethnicity can affect how likely they are to get a disease. [c]	True	95.5	98.4
13. Each of us has variations in our genes that make it more likely that we will get certain diseases. [c]	True	90.0	94.5
14. A "complex disease" is a health condition brought on by many genes and lifestyle and environment. [d]	True	66.5	84.9
15. A single nucleotide polymorphism or "SNiP" is a variation present in some individuals that stretches across a large section of DNA. [d]	False	15.0	22.4

[a] Health care providers included: physicians, nurses (NP, LPN, BSN, RN), and physician assistants

[b] Taken/adapted from Jallinoja and Aro 1999

[c] Taken from Christianson et al. 2010

[d] Written by Coriell staff

knowledge questionnaire were included in the analysis to capture self-rated genetic knowledge and previous exposure to genetic education through previous genetics coursework, books, websites or articles; through the CPMC website; or through genetic counseling (see Table 2). The required baseline demographic questionnaire included age, gender, income, education, and occupation.

A health care provider (HCP) variable was constructed in which any participant reporting that they were a physician, physician assistant, nurse practitioner, LPN, RN or BSN was considered a HCP and everyone else was considered a non-HCP. We used this variable as a proxy for individuals who we know have had medical training, although there are many other ways that participants could have acquired medical training that were not measured within our demographics questionnaire. We asked one additional question (When you have a health problem, where do you go to get information?) to which participants could choose "My doctor", "The internet", "Library", "Other", where choosing "Other" allowed participants to fill in an open text field.

Multivariate linear regression was used to explore the relationship between genetic education background and our calculated genetic knowledge score after controlling for the following covariates: recruitment cohort, gender, age, income, and education. For regression modeling recruitment cohort was coded as a factor; gender was coded as a binary variable; age was collected and coded as a continuous variable; income was re-coded as an ordered variable ranging from 1 to 5 and corresponding to the categories listed in Table 3; and education was recoded as an ordered variable ranging from 1 to 7 and corresponding to the categories listed in Table 3. Demographic covariates were tested independently for association with the genetic knowledge score, and covariates that were significantly associated ($p<0.05$) were retained in the multivariate linear modeling described below. However, race was not included as a covariate because of the problematic distribution of this variable: the majority of participants (90 %) were Caucasian, skewness=4.1, and kurtosis=20.5). Each of the following genetic education background variables was tested independently after controlling for demographic covariates: HCP, time spent reading the CPMC website, previous exposure to genetic education through college-level courses, genetic or personalized medicine websites, articles or books, previous genetic counseling, and self-rated knowledge of genetics. Missing or "don't know" responses to demographic covariates or genetic background questions were excluded, which in total impacted 50 participants. We also tested a combined model that included all of the genetic education background variables after controlling for the same set of demographic covariates. We executed the multivariate linear regression in R (Team 2013) with the lm function. We evaluated collinearity of the predictor variables in the

Table 2 Genetic education background questions

	n (%)	Genetic knowledge score: mean (SD)
How much time did you spend on the website reading about the CPMC study or about personalized medicine and genomics? (*n*=4055)		
Less than 5 min	1230 (30.3)	75.7 (18.3)
Between 5 and 30 min	1635 (40.3)	75.9 (15.8)
Between 30 min and 1 h	783 (19.3)	75.5 (15.4)
More than 1 h	407 (10.0)	75.7 (16.5)
Have you been exposed to genetics before enrolling in the CPMC (college-level courses, genetic or personalized medicine websites, articles or books)? (*n*=4043)		
Yes	2171 (53.7)	82.3 (12.3)
No	1826 (45.2)	68.7 (17.2)
Don't Know	46 (1.1)	58.6 (26.3)
Have you ever received genetic counseling? (*n*=4051)		
Yes	427 (10.5)	78.8 (14.6)
No	3605 (89.0)	75.6 (16.5)
Don't Know/Unsure	19 (0.5)	55.8 (29.5)
Compared to most people, how would you rate your knowledge of genetics? (*n*=4060)		
Better than most people	1329 (32.7)	85.5 (12.0)
About average	2289 (56.4)	72.9 (14.7)
Less than most people	442 (10.9)	61.1 (20.8)

combined model using the variance inflation factor (VIF) and found that all predictor variables had a VIF<2 (see Table S1); we therefore retained all predictor variables in the full model.

Results

Table 3 includes socio-demographic characteristics of the CPMC participants. Over 16 % of participants reported that they were employed as a HCP (*n*=674). More than half of the respondents (54 %) stated they "have been exposed to genetics before enrolling in the CPMC", 29 % of participants reported that they spent 30 min or more reading the CPMC website, and 11 % of participants reported that they had previously received genetic counseling. Furthermore, 33 % rated their knowledge of genetics compared to most people as "better than most people" (Table 2).

Across all respondents, the mean genetic knowledge score was 76 % correct (11.4/15), and psychometric analysis suggested adequate reliability (Cronbach's α=0.74) of the genetic knowledge score. The items most likely to be responded to correctly (>90 % correct; Table 1) included questions 1, 2, 3, 4, 5, 12, and 13. These questions tended to cover topics related to less technical and more general aspects of genetics (question 1), inheritance (questions 2, 5), and disease (questions 3, 4, 12, 13). The items least likely to be responded to correctly (<50 % correct) included questions 10, 11, and 15, all of which involve more specific, functional, and/or technical aspects of genetics.

Despite the time between the informed consent process and eligibility for the genetic knowledge questionnaire (at least 1 month), we explored the possibility that information covered in the informed consent document or informed consent session could have impacted (i.e., improved) the genetic knowledge score. We found that the average score (96 %) across questions in which the answers were covered in the informed consent process (questions 3, 6, 8, 9, 11, and 14) was the same as the average score (96 %) across questions in which the answers were not covered in the informed consent process (questions 1, 2, 4, 5, 7, 12, and 13). The average score across the questions that were related to but not answered in the informed consent process (questions 10 and 15) was lower, 49 %, and these two questions related to more specific information as discussed above.

To evaluate genetic knowledge in a variety of sub-populations, we included apparently healthy civilian participants, apparently healthy military participants, and participants with chronic disease. We used multivariate linear regression to compare the genetic knowledge score between all combinations of recruitment cohort after controlling for demographic covariates (gender, age, income, education). We found that the only significant difference (p-value<0.05) was between the CPMC community cohort and the Air Force cohort (p-value=1.28×10^{-15}). The average GK score for CPMC (76.44) was slightly higher than the average GK score for Air Force (75.33),

We also used multivariate linear regression to explore the relationship between our measured genetic knowledge score and aspects of genetic education background after controlling

Table 3 Socio-Demographic and Cohort Characteristics (n=4062)

	n (%)
Gender	
Male	1675 (41.2)
Female	2387 (58.8)
Age Mean (SD): 50.4 (14.9); Median: 51	
<25	102 (2.5)
25–35	643 (15.8)
35–45	690 (17.0)
45–55	911 (22.4)
55–65	988 (24.6)
65+	718 (17.7)
Race	
Caucasian	3611 (89.8)
African American	172 (4.3)
Native American	5 (0.1)
Asian	110 (2.7)
Hawaiian or Pacific Islander	5 (0.1)
Mixed Race	117 (2.9)
Education (n=4056)	
Some High School	10 (0.2)
High School Graduate	230 (5.7)
Vocational/Trade School	17 (0.4)
Some College	552 (13.6)
Associates degree	447 (11.0)
Bachelors degree	1124 (27.7)
Graduate degree	1676 (41.3)
Income (n=4019)	
<$25 K	194 (4.8)
$25 K - $49,999 K	478 (11.9)
$50 K - $74,999 K	654 (16.3)
$75 K - $99,999 K	721 (17.9)
>= $100 K	1972 (49.1)
Cohort	
CPMC Community	2839 (69.9)
Fox Chase Cancer Center	82 (2.0)
Ohio State University Medical Center	201 (5.0)
United States Air Force	940 (23.1)
Health care provider	
Yes	674 (16.6)
No	3388 (83.4)

for demographic factors (recruitment cohort, gender, age, income, education). The results from the model are presented in Table 4, and the R^2 was 0.29. We found that the amount of time a participant spent on the CPMC website reading about the study or about personalized medicine and genomics was only marginally significant when tested independently (p=0.04; Table S2) and not significant in the model that included all of the genetic education background variables (see Table 4). We found two genetic education background questions to be highly significant: participants reporting that they were exposed to genetics (genetic exposure) before enrolling in the CPMC (through college-level courses, genetic or personalized medicine websites, articles or books) and participants that rated their knowledge (self-reported genetic knowledge) higher than most people were significantly more likely to have higher genetic knowledge scores (p<0.001; Table 4; Table S3; Table S4).

Participants who reported that they previously received genetic counseling were not significantly more likely to have a higher genetic knowledge score in the model containing all of the genetic education background variables (p=0.08; Table 4); however, when tested separately after controlling for demographic factors only, this variable was significant (p=0.006; Table S5), suggesting that previous genetic counseling experience had an impact on participant's genetic knowledge score, but that this effect was not as strong as other genetic education factors.

In addition, we found that participants reporting that they are employed as a HCP (physician, nurse practitioner, physician assistant, LPN, RN, BSN) are more likely to have a higher genetic knowledge score (HCP mean score 83 % versus Non-HCP mean score 74 %; Table S6), a result that remains significant after controlling for demographic factors and other genetic education background variables (p<0.001; Table 4). Given the significant difference between the CPMC community and AF cohorts described above, we additionally ran the combined models separately for HCPs and non-HCPs in each cohort (Tables S7, S8, S9, and S10). Within the AF cohort, education was significantly associated with genetic knowledge score only for the HCPs (Table S7). Self-reported genetic knowledge was significant in AF HCPs and non-HCPs (Tables S7, S8). Non-HCP AF participants that have been exposed to genetics before enrolling in the CPMC were significantly more likely to have a higher genetic knowledge score (p-value=2.45×10^{-10}; Table S8); however, the significance of previous exposure was much less extreme for the AF HCPs (p-value=0.02; Table S7). Within the CPMC community cohort, we found a similar distinction in the significance level of previous exposure to genetics, which was less extreme in the HCPs (p-value=0.01; Table S10) compared to the non-HCPs (p-value=5.57×10^{-25}; Table S9). In addition, education and self-reported genetic knowledge were significant in HCPs and non-HCPs in the CPMC community cohort (Tables S9 and S10).

Discussion

Mean genetic knowledge score among participants in the CPMC research study was 76 % with previous genetic

Table 4 Linear regression model results

	B coefficient	β coefficient	SE	*t*-value	*p*-value
Intercept	0.78	–	0.02	43.34	<0.001
Cohort (CPMC vs. AF)	0.02	0.06	0.01	2.61	0.009
Cohort (FCCC vs. AF)	<0.01	0.01	0.02	0.21	0.832
Cohort (OSU vs. AF)	0.01	0.46	0.01	0.44	0.661
Gender	0.02	0.12	<0.01	3.46	<0.001
Age	<0.01	−0.01	<0.01	−5.79	<0.001
Income	<0.01	<0.01	<0.01	1.03	0.304
Education	0.01	0.08	<0.01	7.96	<0.001
Health care provider	0.03	0.1	0.01	5.43	<0.001
Time spent	<0.01	<0.01	<0.01	−0.24	0.807
Genetic exposure	0.07	0.26	0.01	13.25	<0.001
Genetic counseling	−0.01	−0.05	0.01	−1.74	0.083
Self-reported genetic knowledge	−0.08	−0.24	<0.01	−18.70	<0.001

education experience predictive of higher levels of genetic literacy. When corrected for demographics, health care providers, participants previously exposed to genetics (through previous genetics coursework, books, websites or articles), and participants with 'better than most' self-rated knowledge were significantly more likely to have a higher genetic knowledge score ($p<0.001$).

CPMC participants demonstrated similarly high levels of genetic knowledge to that reported by Haga et al., who also studied genetic knowledge in the context of common, complex diseases, in a study of 300 individuals recruited from the general public in Durham, North Carolina (Haga et al. 2013). While the Haga et al. study had more young individuals and more African American individuals than the CPMC cohort, they also had a mostly highly educated (65 % with a college degree), Caucasian (60 %), female (70 %) study population. Though the survey questions were not identical, both our cohort and that of Haga et al.(Haga et al. 2013) had greater genetic knowledge than previously described cohorts using a similar questionnaire in a European general public population (64 %) (Jallinoja and Aro 1999) and a European patient population (46 %) (Calsbeek et al. 2007). The European cohorts were both comprised of mostly female Caucasian individuals however their education levels were more reflective of the general European population.

We have also extended previous work evaluating genetic knowledge among health care providers to include a larger and more diverse sample of health care providers. Our study includes 674 health care providers practicing in the United States, military ($n=260$) and civilian ($n=414$). A 2004 study included a smaller sample of European health professionals ($n=57$), and consistent with their work (Henneman et al. 2004), we have found that participants who are employed as HCPs are significantly more likely to have a higher genetic knowledge score.

Topically, we found higher knowledge of questions related to heredity and the relationships between genes, environment and disease (90 % or greater responding correctly) as compared to specific information about genes, chromosomes, cells and body (80 % or lower responding correctly). In particular, CPMC participants had more trouble answering (<50 % correct; >22 % answered don't know; see Table 1) three true/false questions that contained more specific information related to genes and genetic variation: Q10: 'All body parts have all the same genes' [true], Q11: 'It has been estimated that a person has about 20,000 genes' [true], and Q15:'A single nucleotide polymorphism is a variation present in some individuals that stretches across a large section of DNA' [false]. The answer to question 11 and information related to the answers of questions 10 and 15 was included in the informed consent presentations. This finding is consistent with previously reported smaller studies that have identified a trend toward greater understanding of genetics within the context of heredity rather than in terms of the structure or function of genes (Calsbeek et al. 2007; Christensen et al. 2010; Condit 2010; Haga et al. 2013; Jallinoja and Aro 1999; Kessler et al. 2007; Lanie et al. 2004; Molster et al. 2009).

While the content of questions 10, 11, and 15 is relatively specific and technical and therefore not important for patients that are interested in adopting personalized medicine as part of their health care, this type of information is relevant to health care providers who may be responsible for interpreting and communicating genetic test results within the context of personalized medicine. It is notable that only 22 % of HCPs correctly answered question 15, only 43 % of HCPs correctly answered question 11, and only 59 % of HCPs correctly answered question 10. These questions pertain to specific information that is commonly known among individuals with specialized training in genetics, but may not be as accessible to individuals with clinical education in other fields.

Nevertheless, clinicians that plan to interpret genetic test results and communicate these results to patients should have a clear understanding of the types of variants that are included in a given test.

Study Limitations

This study has several limitations. Based on the demographics of the study population, results are not generalizable to the United States population at large, to any other disease population, or to any other healthcare provider population. The study relies on self-reported data and, therefore, is subject to reporting bias. The study population consists of individuals who selected themselves into a study on complex disease genetics, which could mean that they have greater interest in genetics and therefore perhaps greater genetic knowledge than the general population. In addition, participants were exposed to the answers to six questions (3, 6, 8, 9, and 11) and exposed to information related to two questions (10 and 15) during the informed consent session. The genetic knowledge score was based on a 15-item unvalidated true-false scale which means we cannot be certain of the accuracy and adequacy of the survey questions in the assessment of genetic knowledge. However, we found significant known correlates of genetic knowledge in agreement with most published findings. Strengths of the study are the high completion rate (87 %) of the genetic knowledge questionnaire and the large sample size (n=4 062).

Practice Implications

Taken together, our results suggest that CPMC participants have a good overall understanding of general concepts in genetics and disease, particularly when they report having previous exposure to genetic education or previous medical training. This indicates that genetic education can result in a measurable increase in knowledge and that individuals do successfully self-identify as more or less knowledgeable in genetics. The deficits in genetic knowledge that were observed, such as understanding what the term "SNP" means, may not translate directly to a deficit in patient genetic literacy. This level of genetic knowledge may not be necessary for individuals to integrate information and make appropriate healthcare decisions. It does however suggest the need for laboratories reporting genetic information and healthcare providers communicating genetic information to ensure that the information provided is made available in terms that can be understood by patients.

We concur with the position of other authors that steps should be taken to facilitate the dissemination and understanding of genetic knowledge among the general public so that genetic knowledge can be readily applied when patients are faced with a need for it (M.J. Dougherty et al. 2014; Jallinoja and Aro 2000). Previously suggested avenues for these efforts have included: improving the high school biology school curriculum (M. J. Dougherty et al. 2011), improving college curriculum for non-science majors (Hott et al. 2002), making key genetic concepts part of core competencies for health care providers (Genetics 2007; McInerney et al. 2012), improving access to and availability of genetic counseling (Jallinoja and Aro 2000), improved accuracy of genetics media coverage (Brechman et al. 2011), as well as the development of new and the vetting of existing online genetic education tools (Alliance 2010). All of these efforts should be made with the goal of increasing genetic knowledge as a step towards improvement of overall genetic literacy.

Research Recommendations

To aid in targeting genetics educational programs, further work should be done to tease apart any potential differences in genetic knowledge based on the specific type of genetics exposure (college-level course, genetic or personalized medicine websites, articles or books) that were considered only collectively in this study. In addition to type, duration of exposure required to make a meaningful impact on genetic knowledge would also be worthy of further exploration. We had very few participants (n=53) that reported spending more than an hour on the CPMC website reading about genomics, personalized medicine or the CPMC study. Given that we restricted the current study to participants who had not yet received a CPMC risk report and that our study cohort is overall highly educated this is not surprising. However, future research that evaluates online educational materials such as the ones included on the CPMC website would be useful in assessing the impact of internet resources on public genetic literacy over time.

Improvement of public genetic literacy is an educational challenge worthy of a variety of approaches; however some approaches may be more feasible than others. Development of books and print articles may not be preferable given printing costs and the inability to rapidly update print information to keep pace with new developments in the field of genetics. Similarly, school curricula change is challenged by lengthy turnaround time for implementation, localized control over course content, and uneven teacher quality (M.J. Dougherty et al. 2014). Therefore we put forth that it may be particularly productive to continue to develop and improve online genetics education tools and resources. The internet is an almost ubiquitous tool for public health education. Indeed, 34 % of the study participants told us that they get information from the internet when they have a health problem.

Similarly, a recent Pew Health Online survey (Fox and Duggan 2013) found that 72 % of internet users, which encompasses 59 % of the general US adult population, reported accessing health information online, primarily via search

engines like Google rather than targeted health education sites like WebMD. Development and maintenance of genetic education websites and online tools like the CPMC web portal may prove to be a complementary and relatively short term and cost effective approach to advancing public genetic education in addition to modernizing public science education curricula.

While the internet is a widely utilized and increasingly accessible source of health information, it does require users to have more advanced information-filtering skills to achieve the maximum benefit. Studies by Pew Research Center (Center 2002) and Wathen et al. (Wathen and Burkell 2002) have shown that the general public uses both relevant indicators of quality, like finding the same information on multiple websites, as well as irrelevant indicators, like website layout, in their determination of the credibility of information encountered online. We suggest that an additional goal of public health research and education should be to evaluate and provide guidance on how to determine if health and genetic information encountered online is of high quality or not.

In addition to the 34 % of participants that told us that they get information on the internet when they have a health problem, 54 % told us that they get information directly from their doctor when they have a health problem. Our study suggests that HCPs generally have higher genetic literacy than non-HCPs (as measured by our genetic knowledge score); however, questions related to specific and technical aspects of genetic literacy were difficult for both non-HCPs and HCPs. Given that an estimated 41 % of the general US adult population are not accessing health information online (Fox and Duggan 2013), it is also critical for public health research and education to provide support and direction to HCPs for accessing high quality and up to date genetic and genomic information. As genome-based analyses are being increasingly utilized in clinical and preventive medical care, future work that also evaluates both health care provider and patient comprehension of genetic and genomic test reports should be used to identify critical gaps in genetic literacy and to construct best practices for test providers to help address the informational needs of both patients and providers. Additionally, further research to study the influence of genetic and health literacy on patient health behaviors and patient outcomes is also warranted in order to fully leverage the potential of personalized medicine to improve patient health behaviors and ultimately reduce disease risk.

Conclusions

This study found that genetic knowledge was high among a large, cohort including healthy individuals, individuals with chronic disease, and health care providers. Previous exposure to genetic education was correlated with higher levels of genetic knowledge suggesting that education may improve genetic literacy, a significant factor influencing the utility of genomics in personalized medicine.

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Conflict of Interest Authors Schmidlen, Scheinfeldt, Zhaoyang, Kasper, Sweet, Gordon, Keller, Stack, Gharani, Daly, Jarvis, and Christman declare no conflict of interest.

Human Studies and Informed Consent All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all participants for being included in the study.

Animal Studies No animal studies were carried out by the authors for this article.

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