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Assessing Genetic Literacy Awareness and Knowledge Gaps in the U.S. Population: Results from the Health Information National Trends Survey

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Abstract

Background/Aims—Public understanding of the role of genetics in disease risk is key to appropriate disease prevention and detection. This study assessed the current extent of awareness and use of genetic testing in the U.S. population. Additionally, the study identified characteristics of subgroups more likely to be at risk for low genetic literacy.

Methods—The study used data from the National Cancer Institute's 2017 Health Information National Trends Survey, including measures of *genetic testing awareness*, *genetic testing applications* and *genetic testing usage*. Multivariable logistic regression models estimated associations between sociodemographics, genetic testing awareness and genetic testing use.

Results—Fifty-seven percent of respondents were aware of genetic tests. Testing awareness differed by age, household income, and race/ethnicity. Most participants had heard of using tests to determine personal disease risk (82.58%) or inherited disease risk in children (81.41%), but less were familiar with determining treatment (38.29%) or drug efficacy (40.76%). Among those with genetic testing awareness, actual testing uptake was low.

Conclusions—A large portion of the general public lacks genetic testing awareness and may benefit from educational campaigns. As precision medicine expands, increasing public awareness about genetic testing applications for disease prevention and treatment will be important to support population health.

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Keywords

genetic literacy; genetic education; awareness; knowledge; knowledge gap; population survey; genetic testing; health information; health communication

As advances in genetic and genomic medicine are harnessed for the prevention, detection, and treatment of rare and common diseases, public genetic and genomic literacy will become increasingly important [1–3]. Indeed, widespread public participation is essential to achieving precision medicine-related public health aims [4–5]. In order to benefit from genetic testing, the public must first be aware of available genetic tests and their potential benefits across various types of genetic testing (e.g., risk assessment, personalized treatment, pharmacogenomics) [1,3]. Furthermore, genetic testing awareness will likely influence public participation in the largescale data collection efforts that drive continued discovery [6,7]. Promoting genetic and genomic literacy is therefore a significant public health goal.

A first step towards improving genetic literacy among the general public is understanding the public's current of awareness genetic testing. The National Cancer Institute's Health Information National Trends Survey (HINTS) is a leading source of health information about the general public. While past survey iterations have tracked several indicators of public knowledge and engagement with genetic and genomic medicine, including awareness and uptake of direct-to-consumer (DTC) genetic testing [8] and uptake of cancer-specific genetic tests [9–11], there is still a need to assess population-level estimates of awareness of precision medicine-related genetic testing [1].

The knowledge gap hypothesis suggests that awareness of advancements in genetic and genomic medicine is unlikely to be evenly distributed across the population [12]. For instance, race/ethnicity and sociodemographic variables, such as income and education levels, could play a role in genetic literacy. Prior population-based surveys found higher-income individuals more likely to be aware of DTC genetic testing [8] and both education and income predicted awareness of DTC nutrigenomic testing [13]. Racial/ethnic minorities are less aware of BRCA1/2 genetic testing [14] and even less likely to use it [15]. Adopters of genome sequencing have also been predominantly white and of higher SES [10,16]. Identification of subgroups with limited genetic awareness and low genetic literacy is a crucial step to inform targeted health communication efforts. To begin to identify disparities in precision medicine and genetic literacy among the general population, we analyzed data from HINTS 2017. Our primary objective was to determine prevalence of genetic testing awareness, including knowledge of specific applications for genetic tests; a secondary aim was to assess current usage of such tests among the general population.

Methods

This study used data from the National Cancer Institute's 2017 Health Information National Trends Survey (HINTS). HINTS is administered on an annual basis to a nationally representative sample of civilian, non-institutionalized adults to track health communication among the U.S. population, including awareness of novel health topics, information access, and health behaviors [17]. Data collection took place from January through April, 2017.

Administration of the 2017 HINTS survey was approved by the Institutional Review Board (IRB) at Westat and deemed exempt by the National Institutes of Health (NIH) Office of Human Subjects Research.

The survey included the following sociodemographic measures: sex, age, race/ethnicity, education, household income, metropolitan area (rural or urban, as defined by 2013 USDA rural-urban continuum codes), and personal and family cancer history. *Genetic testing awareness* was measured with a yes/no response to the item: "Doctors use DNA tests to analyze someone's DNA for health reasons. Have you heard or read about this type of genetic test?" Individuals who responded "yes" to this item were then asked to check all of "the following *genetic testing applications* [they] had heard of": "Determining the risk or likelihood of getting a particular disease," "determining the likelihood of passing an inherited disease to your children," "determining how a disease should be treated after diagnosis," and "determining which drug(s) may or may not work for an individual." To *assess genetic testing usage*, individuals who responded yes to genetic testing awareness were also asked if they had ever received any of the following types of genetic tests: ancestry, paternity, DNA fingerprinting, Cystic Fibrosis carrier, BRCA 1/2 and/or a Lynch Syndrome test. BRCA 1/2 and Lynch Syndrome were also combined to represent prevalence of cancer-related tests more generally.

Descriptive statistics were calculated to generate frequencies and weighted proportions for sociodemographic characteristics of the sample and for the four genetic testing application items. Multivariable logistic regression models estimated the associations between sociodemographic characteristics and genetic testing awareness and genetic cancer testing use.

Results

A total of 3285 responses were collected (Table 1). Over half (57.08%, n= 1878) of respondents were aware of genetic tests for health. Awareness differed across several sociodemographic groups. Among 1878 respondents aware of genetic tests, the majority had heard of using these tests to determine personal disease risk (82.58%) or inherited disease risk in children (81.41%) (Table 2). Respondents were less familiar with genetic testing for determining treatment (38.29%) or drug efficacy (40.76%). Over three quarters indicated that they had heard of multiple reasons for genetic testing (75.72%) and few (n = 106) had not heard of any of these applications (indicated by non-response to all four items).

Adults aged 75 or older were less likely to be aware of genetic tests, compared to the youngest age category (OR: 0.42, CI: 0.22, 0.77; Table 3). Individuals with household incomes over $75,000 were more likely to report awareness of genetic tests, compared to the lowest household income category (OR: 1.72, CI: 1.13, 2.60). Non-Hispanic Asian (OR: 0.31, CI: .18, .55) and black respondents (OR: 0.49, CI: .31, .78) were less likely be aware of genetic testing, compared to non-Hispanic white respondents. Genetic testing awareness did not differ across respondents' reported personal and family cancer histories.

Among those with genetic testing awareness, actual testing uptake was low. Of this subgroup, only 20.95% had undergone genetic testing of any kind, and 8.76% reported multiple tests. The most commonly reported types of tests were ancestry tests (11.11%), paternity tests (8.97%), DNA fingerprinting (8.51%), and Cystic Fibrosis carrier tests (6.87%). Only 5.36% had undergone at least one cancer-related test: 4.88% reported BRCA testing, and even fewer (2.52%) had undergone testing for Lynch Syndrome. Additionally, 23 individuals indicated that they had had both genetic cancer tests. Only personal cancer history predicted uptake of genetic cancer testing (OR: 2.69, CI: 1.01, 7.21; Table 3).

Discussion

Precision medicine has the potential to provide personalized care for patients with myriad conditions; however, broad clinical implementation of research findings (such as those from the All of Us Research Program [7]) will depend greatly on the patient's ability to understand the results of their genetic testing and subsequent preventive and therapeutic options. Analyses of data from the 2017 administration of HINTS revealed that many respondents had still not heard of genetic testing and, of those who had, most were unaware of its potential use for individualization of treatment. Those who were older, lower income, and members of racial/ethnic minority groups had lower awareness of genetic testing in general. Unfortunately, this is in keeping with previously published studies of genetic literacy and attitudes towards testing over the past several years [18–21].

Carefully designed educational interventions and public health messaging will be key to improving genetic testing awareness and literacy, especially for the aforementioned subpopulations. The creation and delivery of educational programming is often challenging; different modalities have been shown to work for different populations and repeated exposure may be necessary to move the needle on public genetic literacy [22]. For example, those with lower health literacy and numeracy may understand less of the genetic and genomic information in printed materials [23]. Educational outreach may be further complicated by media coverage of applications of genetically-based precision medicine, which can contain conflicting information and create a sense of false confidence among those who have lower genetic literacy skills [24]. Therefore, educational interventions centered around precision medicine and genetic testing need to be carefully designed with the target populations in mind, with the understanding that one educational intervention may not be effective for the general population as a whole.

A primary strength of this study is its use of data from a nationally representative survey (HINTS) administered in 2017, which allows for the determination of current population-level estimates of genetic testing awareness and use. One limitation is in the cross-sectional nature of the survey, which limits the potential for evaluation of causal relationships. Additionally, the item regarding genetic testing usage only provided six categories of testing, including two cancer-related tests, and thus do not reflect the broad array of genetic testing clinically available to patients; therefore, our estimation of uptake may be lower than actual usage. As specific information on types of personal and family cancer history was limited, the study did not examine uptake of BRCA or Lynch Syndrome testing among the subset of individuals meeting clinical guidelines for these genetic tests. Finally, the survey did not

explore lifecourse factors that may influence testing uptake. For example, it is possible that cystic fibrosis carrier screening is more prevalent among young adults as part of family planning and/or among new parents, alongside neonatal screening programs for this condition.

For successful public engagement with genetic health interventions, it is important to ensure that all stakeholders, including members of the general population, have an understanding of the capabilities and limitations of genetic testing [3]. The results presented here indicate that a substantial proportion of the general public currently lacks awareness of genetic testing, knowledge gaps still exist, and a broad swathe of the public is unfamiliar with certain applications of testing relevant to precision medicine (e.g., using genetic tests to inform treatment after diagnosis and use of pharmaceuticals). This suggests the public may benefit from targeted public health communication to disseminate this knowledge. Additionally, among the subset of the population who was aware of genetic tests, knowledge of testing for risk assessment was more common than for determining optimal disease treatment. As applications for precision medicine expand, increasing public awareness about genetic testing applications through carefully selected and designed educational interventions will support informed decision-making and enhance population health.

References

1. Hurle B, Citrin T, Jenkins JF, Kaphingst KA, Lamb N, Roseman JE, Bonham VL. What does it mean to be genomically literate?: National Human Genome Research Institute meeting report. Genet Med. 2013; 15:658–663. [PubMed: 23448722]
2. Khoury MJ, Bowen MS, Burke W, Coates RJ, Dowling NF, Evans JP, Reyes M, St Pierre J. Current priorities for public health practice in addressing the role of human genomics in improving population health. Am J Prev Med. 2011; 40:486–493. [PubMed: 21406285]
3. McBride CM, Bowen D, Brody LC, Condit CM, Croyle RT, Gwinn M, Khoury MJ, Koehly LM, Korf BR, Marteau TM, McLeroy K, Patrick K, Valente TW. Future health applications of genomics: Priorities for communication, behavioral, and social sciences. Am J Prev Med. 2010; 38:556–565. [PubMed: 20409503]
4. Hawgood S, Hook-Barnard IG, O'Brien TC, Yamamoto KR. Precision medicine: Beyond the inflection point. Sci Transl Med. 2015; 7:1–3.
5. Khoury MJ, Iademarco MF, Riley WT. Precision public health for the era of precision medicine. Am J Prev Med. 2016; 50:398–401. [PubMed: 26547538]
6. Collins FS, Varmus H. A new initiative on precision medicine. N Engl J Med. 2015; 372:793–795. [PubMed: 25635347]
7. National Institutes of Health: All of Us Research Program. 2017. Retrieved from https://allofus.nih.gov
8. Agurs-Collins T, Ferrer R, Ottenbacher A, Waters EA, O'Connell ME, Hamilton JG. Public awareness of direct-to-consumer genetic tests: Findings from the 2013 US Health Information National Trends Survey. J Cancer Educ. 2015; 30:799–807. [PubMed: 25600375]
9. Mai PL, Vadaparampil ST, Breen N, McNeel TS, Wideroff L, Graubard BI. Awareness of cancer susceptibility genetic testing: The 2000, 2005, and 2010 National Health Interview Surveys. Am J Prev Med. 2014; 46:440–448. [PubMed: 24745633]
10. Roberts MC, Taber JM, Klein WM. Engagement with genetic information and uptake of genetic testing: The role of trust and personal cancer history. J Cancer Educ. 2017; 20:1–8.
11. Taber JM, Chang CQ, Lam TK, Gillanders EM, Hamilton JG, Schully SD. Prevalence and correlates of receiving and sharing high-penetrance cancer genetic test results: Findings from the Health Information National Trends Survey. Public Health Genomics. 2015; 18:67–77. [PubMed: 25427996]

12. Viswanath K, Finnegan JR. The knowledge gap hypothesis: Twenty-five years later. In: Burleson B, editorCommunication Yearb. Vol. 19. Thousand Oaks, CA: Sage Publications; 1996. 187–227. 1996
13. Goddard KA, Duquette D, Zlot A, Johnson J, Annis-Emeott A, Lee PW, Bland MP, Edwards KL, Oehlke K, Giles RT, Rafferty A, Cook ML, Khoury MJ. Public awareness and use of direct-to-consumer genetic tests: Results from 3 state population-based surveys, 2006. Am J Public Health. 2009; 99:442–445. [PubMed: 19106425]
14. Gammon AD, Rothwell E, Simmons R, Lowery JT, Ballinger L, Hill DA, Boucher KM, Kinney A. Awareness and preferences regarding BRCA1/2 genetic counseling and testing among Latinas and non-Latina white women at increased risk for hereditary breast and ovarian cancer. J Genet Couns. 2011; 20:625–638. [PubMed: 21691939]
15. Butrick M, Kelly S, Peshkin BN, Luta G, Nusbaum R, Hooker GW, Graves K, Feeley L, Isaacs C, Valdimarsdottir HB, Jandorf L, DeMarco T, Wood M, McKinnon W, Garber J, McCormick SR, Schwartz MD. Disparities in uptake of BRCA1/2 genetic testing in a randomized trial of telephone counseling. Genet Med. 2014; 17:467–475. [PubMed: 25232856]
16. Lewis KL, Han PK, Hooker GW, Klein WM, Biesecker LG, Biesecker BB. Characterizing participants in the ClinSeq genome sequencing cohort as early adopters of a new health technology. PLoS One. 2015; 10:e0132690. [PubMed: 26186621]
17. Finney Rutten LJ, David T, Beckjord EB, Blake K, Moser RP, Hesse BW. Picking up the pace: Changes in method and frame for the Health Information National Trends Survey (2011-2014). J Health Comm. 2012; 17:979–989.
18. Christensen FS, Jayaratne TE, Robets JS, Kardia SL, Petty EM. Understandings of basic genetics in the United States: Results from a national survey of black and white men and women. Public Health Genomics. 2010; 13:467–76. [PubMed: 20203477]
19. Haga SB, Barry WT, Mills R, Ginsburg GS, Svetkey L, Sullivan J, Willard HF. Public knowledge of and attitudes toward genetics and genetic testing. Genet Test Mol Biomarkers. 2013; 17:327–335. [PubMed: 23406207]
20. Kaphingst KA, Blanchard M, Milam L, Pokharel M, Elrick A, Goodman M. Relationships between health literacy and genomics-related knowledge, self-efficacy, perceived importance, and communication in a medically underserved population. J Health Comm. 2016; 21:58–68.
21. Ashida S, Goodman M, Pandya C, Koehly LM, Lachance C, Stafford J, Kaphingst KA. Age differences in genetic knowledge, health literacy, and causal beliefs for health conditions. Public Health Genomics. 2011; 14:307–316. [PubMed: 20829577]
22. Kusnoor SV, Koonce TY, Levy MA, Lovly CM, Naylor HM, Anderson IA. My cancer genome: evaluating an educational model to introduce patients and caregivers to precision medicine information. AMIA Jt Summits Transl Sci Proc. 2016:112–121. [PubMed: 27570660]
23. Lea DH, Kaphingst KA, Bowen D, Lipkus I, Hadley DW. Communicating genetic and genomic information: Health literacy and numeracy considerations. Public Health Genomics. 2011; 14:279–289. [PubMed: 20407217]
24. Abrams L, Koehly LM, Hooker GW, Paquin RS, Capella JN, McBride CM. Media exposure and genetic literacy skills to evaluate Angelina Jolie's decision for prophylactic mastectomy. Public Health Genomics. 2016; 19:282–289. [PubMed: 27427958]

Table 1

Respondent Characteristics, Health Information National Trends (HINTS) Survey V Cycle 1, 2017 (n= 3285)

Characteristic	n	Weighted %
Sex		
Male	1254	48.97
Female	1784	51.03
Age		
18-34	367	21.90
35-49	655	28.67
50-64	1063	30.10
65-74	676	11.10
75+	385	8.23
Income		
Less than $20,000	559	17.44
$20,000 to < 35,000	423	12.23
$35,000 to < 50,000	386	14.88
$50,000 to < 75,000	530	19.12
$75,000 or more	1064	36.34
Education		
Less than high school	217	8.67
High school graduate	616	22.94
Vocational or technical	228	9.45
Some college	714	23.36
College graduate or more	1406	35.57
Race/Ethnicity		
Non-Hispanic White	1868	65.69
Non-Hispanic Black	409	10.28
Hispanic	427	15.75
Non-Hispanic Asian	138	5.54
Non-Hispanic Other	111	2.74
Metropolitan area		
Rural	437	14.17
Urban	2848	85.83
Personal cancer history		
Yes	504	8.64
No	2756	91.36
Family cancer history		
Yes	2252	72.79
No	754	27.21

Table 2

Frequencies and weighted proportions of genetic testing applications indicated by respondents who were aware of genetic testing (n= 1878)

Genetic testing application	No n (Weighted %)	Yes n (Weighted %)
Determine risk or likelihood of getting a particular disease.	345 (17.42)	1533 (82.58)
Determining how a disease should be treated after diagnosis.	1152 (61.71)	726 (38.29)
Determining which drug(s) may or may not work for an individual.	1133 (59.24)	745 (40.76)
Determining the likelihood of passing an inherited disease to your children.	375 (18.59)	1503 (81.41)

Table 3

Weighted, fully adjusted multivariable logistic regression models predicting genetic testing awareness and usage by sociodemographic variables, OR (95% CI)

Variable	Heard of genetic tests (n = 3285)	Had a genetic cancer test (n = 1878)
Sex		
Male	ref	ref
Female	1.19 (0.88, 1.61)	1.42 (.51, 3.95)
Age		
18-34	ref	ref
35-49	0.60 (0.37, 0.97)	.42 (.14, 1.23)
50-64	0.69 (0.42, 1.13)	.39 (.12, 1.29)
65-74	0.64 (0.40, 1.03)	.30 (.07, 1.38)
75+	**0.42 (0.22, 0.77)** **	.37 (.10, 1.43)
Income		
Less than $20,000	ref	ref
$20,000 to < 35,000	1.12 (0.74, 1.69)	.40 (.05, 2.96)
$35,000 to < 50,000	0.98 (0.55, 1.75)	.60 (.10, 3.49)
$50,000 to < 75,000	1.24 (0.79, 1.97)	.60 (.10, 3.49)
$75,000 or more	**1.72 (1.13, 2.60)** *	.48 (.09, 2.49)
Education		
Less than high school	ref	ref
High school graduate	0.99 (0.52, 1.89)	.76 (.02, 39.34)
Vocational or technical	0.95 (0.41, 2.21)	1.15 (.02, 54.08)
Some college	1.37 (0.66, 2.87)	.83 (.03, 25.57)
College graduate or more	1.93 (0.97, 3.84)	1.25 (.04, 38.67)
Race/Ethnicity		
Non-Hispanic White	ref	ref
Non-Hispanic Black	**0.49 (0.31, 0.78)** **	1.42 (.15, 13.60)
Hispanic	0.71 (0.44, 1.15)	.40 (.04, 3.73)
Non-Hispanic Asian	**0.31 (0.18, 0.55)** ***	.80 (.12, 5.17)
Non-Hispanic Other	1.06 (0.44, 2.53)	1.92 (.47, 7.81)
Metropolitan area		
Rural	0.81 (0.52, 1.26)	1.59 (.37, 6.79)
Urban	ref	ref
Personal cancer history		
Yes	1.10 (0.80, 1.52)	**2.69 (1.01, 7.21)** *
No	Ref	ref
Family cancer history		
Yes	1.32 (0.90, 1.95)	1.54 (.58, 4.04)
No	Ref	ref

Note: Boldface indicates statistical significance

*
p < .05

**
p < .01

***
p < .001).