



## Short Report

# Knowledge of the Genetic Information Nondiscrimination act among individuals affected by Huntington disease

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The Genetic Information Nondiscrimination Act (GINA) of 2008 was the first US legislation to address genetic discrimination. We sought to assess understanding of GINA among individuals affected by the autosomal dominant condition, Huntington disease (HD). We conducted a cross-sectional survey of individuals with varying risk of HD to assess their familiarity with GINA. As a control, individuals were surveyed about their familiarity with the Health Insurance Portability and Accountability Act (HIPAA). Those who reported familiarity with GINA were asked about their knowledge of specific provisions of the legislation. The survey was offered to 776 participants and completed by 410 (response rate 53%). Respondents across all groups were less familiar with GINA (41% slightly, somewhat, or very familiar) than with HIPAA (65%;  $p < 0.0001$ ). Of individuals with or at risk for HD who reported some familiarity with GINA, less than half correctly identified GINA's protections, and less than 15% correctly identified its limitations. Thus, among individuals affected by HD, familiarity with and knowledge of GINA are low. The effectiveness of the legislation may be limited by this lack of knowledge.

### Conflict of interest

The authors have no conflicts of interest to report.

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The late Senator Ted Kennedy heralded the Genetic Information Nondiscrimination Act (GINA) as the 'first major new civil rights bill of the new century' (1, 2). President George W. Bush signed GINA on 21 May 2008 after more than 12 years of debate in the US Congress, making GINA the first federal legislation to address genetic discrimination.

The law has two parts. Title I regulations took effect on 21 May 2009 and prohibits group health plans from requiring an individual or family member to undergo genetic testing and from using genetic information to determine eligibility, benefits, or premiums. Title II

regulations took effect on 21 November 2009 and prohibits employers from requiring genetic testing and from using genetic information to make employment decisions (3, 4). GINA includes the provision that neither health insurers nor employers may use an individual's participation in clinical genetic research to make insurance or employment decisions. However, GINA does not prohibit insurers from using genetic information in life, disability, or long-term care insurance.

Huntington disease (HD) is an autosomal dominant, highly penetrant, neurodegenerative disorder in which the physical, cognitive, and behavioral symptoms begin

to emerge in adulthood (5). A predictive genetic test for HD was developed in 1993 (6). Therefore, GINA legislation is especially salient to those who are at risk for HD (7).

Little is known about the familiarity and knowledge of the legislation or the legislation's impact among individuals at risk for a genetic disorder. We sought to determine familiarity with GINA and knowledge of its specific provisions among individuals of families affected by HD. We hypothesized that this group of individuals would be particularly knowledgeable about GINA.

## Materials and methods

### Participants and survey

To better understand the genotype and phenotype of HD, the Cooperative Huntington's Observational Research Trial (COHORT) was undertaken in 2006. COHORT involved research participants at Huntington Study Group (8) research sites in the US, Canada, and Australia who were affected by or at risk for HD. A paper survey was distributed to US research participants from COHORT (9), aimed at evaluating the health and life choices of individuals affected by HD and included questions about GINA (see Appendix S1). The survey was given to COHORT participants at 20 participating sites in the US. A multidisciplinary team, consisting of clinical investigators and economists, created the survey (9). The University of Rochester's Institutional Review Board reviewed and approved this research study.

Members of COHORT were grouped into four categories based on their symptoms and genetic testing status: (1) individuals with clinically diagnosed HD; (2) individuals who pursued pre-symptomatic genetic testing prior to baseline, carried an expanded allele, but did not have clinically diagnosed HD; (3) first-degree or second-degree relatives of individuals in the first two groups; and (4) caregivers and spouses of individuals enrolled from group one or two.

### Outcome measures

We measured the familiarity of US participants with GINA. The survey asked participants to disclose their level of familiarity with HIPAA and GINA, which were assessed with the following questions: 'Are you familiar with the US Health Insurance Portability and Accountability Act (HIPAA)?' and 'Are you familiar with the US Genetic Information Nondiscrimination Act (GINA)?' Possible responses were 'No,' 'Slightly familiar,' 'Somewhat familiar,' and 'Very familiar.'

Participants who indicated any familiarity with GINA (i.e. 'Slightly,' 'Somewhat,' or 'Very') were asked to correctly identify the specific provisions of the legislation. Knowledge of GINA's provisions was tested with the following questions: 'Does GINA to your knowledge protect against genetic discrimination in the following areas: Health insurance? Life

insurance? Disability insurance? Long-term care insurance? Employment? Participation in research studies that include genetic testing, counseling, or education?' Possible responses were 'Yes,' 'No,' and 'Do not know.'

### Analysis

We compared age, sex, race, ethnicity, education, employment status, marital status, and genetic testing status between respondents and non-respondents using Wilcoxon–Mann–Whitney tests. We compared HD categories between respondents and non-respondents using a Pearson's  $\chi^2$  test. Among respondents, we compared familiarity with GINA and HIPAA using Pearson's  $\chi^2$  tests. Using Wilcoxon–Mann–Whitney tests, we conducted pairwise comparisons of HD mutation groups in their familiarity with GINA or HIPAA and in their knowledge of GINA provisions and limitations. We did not adjust results for multiple comparisons.

## Results

### Characteristics of respondents

The survey was offered to 776 participants of the COHORT study from 1 July 2009 through 30 June 2010, and completed by 410 participants for a response rate of 53%. One participant responded twice, so only the first questionnaire was included. The baseline demographics of respondents and non-respondents did not differ significantly (Table 1). Respondents were less likely to have clinically diagnosed HD (42% vs 55%;  $p=0.0001$ ). Among respondents, 4.6% reported discrimination in insurance (health, life, disability, and/or long-term care) and 3.4% reported discrimination in employment.

### Familiarity with and knowledge of GINA

Respondents across all groups were less familiar with GINA than with HIPAA (41.2% vs 64.9% 'Slightly familiar,' 'Somewhat familiar,' or 'Very familiar';  $p<0.0001$ ). Familiarity with HIPAA, but not GINA, differed among groups (Table 2). Among those who reported genetic discrimination in insurance, familiarity with GINA was higher than for those who did not report insurance discrimination (57.9% vs 41.1%;  $p=0.149$ ). Among those who reported genetic discrimination in employment, familiarity with GINA was higher than for those who did not report employment discrimination (64.3% vs 41.1%;  $p=0.085$ ).

### Knowledge of GINA provisions

Less than 50% of respondents who were familiar with GINA correctly identified GINA's provisions for protection against genetic discrimination in employment (46.2%), health insurance (43.2%), and participation in

## Knowledge of GINA among individuals affected by HD

Table 1. Demographic characteristics of respondents and non-respondents

	Respondents (N = 410)		Non-respondents <sup>a</sup> (N = 366)		p-Value <sup>b</sup>
Age (years) <sup>c</sup>	49.3 (12.2)		49.8 (12.9)		0.451
Female (% , n)	57.8	237	54.6	200	0.376
White (% , n)	92.0	377	92.3	337	0.846
Ethnicity (% , n reporting Spanish origin)	3.4	14	5.8	21	0.118
Education (% , n completing high school or more)	96.1	394	94.5	345	0.298
Employment status (% , n currently in labor force)	50.0	205	40.8	149	0.011
Marital status (% , n currently married)	70.5	289	69.9	255	0.850
HD category					< 0.001
Individuals with clinically diagnosed HD (% , n)	41.7	171	55.5	202	
Individuals with an expanded allele, but do not have clinically diagnosed HD (% , n)	14.4	59	7.7	28	
Individuals with first- or second-degree relatives with HD (% , n)	22.0	90	15.9	58	
Caregivers and spouses (% , n)	22.0	90	20.9	76	
Tested for expanded allele (% , n)	53.4	219	57.6	209	0.246

HD, Huntington disease.

<sup>a</sup>One individual has incomplete data on white, ethnicity, education, employment status, and marital status; two have incomplete data on HD category; and three have incomplete data on testing for expanded allele.

<sup>b</sup>p-Values refer to tests for differences between respondents and non-respondents;  $\chi^2$  test p-value for HD category; Wilcoxon–Mann–Whitney test p-values for other characteristics.

<sup>c</sup>Age is listed as mean (SD).

genetic research studies (32.0%; Table 3a). Less than 15% of respondents who were familiar with GINA correctly identified GINA's lack of protection in life insurance (14.8%), long-term care insurance (11.8%), and disability insurance (10.1%; Table 3b). Individuals who carried an expanded allele but did not have clinically diagnosed HD were more knowledgeable about protection in employment and health insurance than individuals with clinically diagnosed HD, first- or second-degree relatives of individuals with HD, and caregivers and spouses. At the 5% level, familiarity of GINA's provisions and limitations did not differ between those who reported discrimination in insurance or employment and those who did not report genetic discrimination (data not shown), with one exception. Among those who reported employment discrimination, knowledge of GINA's protection in employment was higher than for those who did not report genetic discrimination (88.9% vs 47.0%;  $p = 0.015$ ).

### Discussion

Contrary to our hypothesis, familiarity of GINA among a cohort of individuals at risk for genetic discrimination was low. Actual knowledge of key provisions and limitations of this civil rights legislation was even lower, with less than half of all individuals familiar with the legislation aware of its protection against genetic discrimination in health insurance, employment, and research, and less than 15% aware of its absence of protection in life insurance, long-term care insurance, and disability insurance.

The lack of knowledge of GINA was present among individuals affected by HD, who have historically either experienced or been at risk for genetic discrimination

(10–14), and are a population that the legislation seeks to protect. Those who are pre-symptomatic for HD may be more familiar with genetic discrimination and GINA by virtue of having had genetic testing, and likely, genetic counseling. Issues of genetic discrimination may be discussed in genetic counseling. Genetic discrimination has been reported worldwide among populations at risk for genetic disease (15, 16). Concern about genetic discrimination is frequent among individuals at risk for HD, and this concern may influence behaviors and result in high levels of distress (17). Knowledge of one's genetic testing results has the potential to inform an individual's health care choices and guide personal decisions (18, 19). As researchers continue to develop predictive genetic tests for additional diseases, more individuals will have the opportunity to identify their disease susceptibilities before symptomatic presentation of disease.

This study was limited by its timing, response rate, and its focus on a single condition. The survey was conducted approximately 1 year after GINA was signed into law, after many high-profile news sources and scientific journals published stories on the passage of GINA (4, 20–22), but prior to complete implementation of the law. Current familiarity and knowledge of GINA may be higher or lower. While the response rate to this survey was modest at 53%, the characteristics of the respondents and non-respondents were similar. In addition, the population sample was relatively large (over 400 respondents) and sampled across 20 sites in the US. Respondents may have more knowledge about GINA than the general population, given their education level, participation in the study, and their responsiveness to the survey. Finally, this study was

Table 2. Familiarity with GINA and HIPAA among respondents

HD category	Familiarity with GINA					Familiarity with HIPAA					N	χ <sup>2</sup> p-value <sup>b</sup>
	Not familiar	Slightly familiar	Somewhat familiar	Very familiar	Blank	Not familiar	Slightly familiar	Somewhat familiar	Very familiar	Blank <sup>a</sup>		
Individuals with clinically diagnosed HD (% , n)	59.1	20.5	12.9	4.7	2.9	43.9	19.9	15.2	19.9	1.2	171	< 0.001
Individuals with an expanded allele, but do not have clinically diagnosed HD (% , n)	101	35	22	8	5	75	34	26	34	2	59	0.001
Individuals with first- or second-degree relatives with HD (% , n)	57.8	17.8	16.7	7.8	0.0	18.9	18.9	25.6	35.6	1.1	90	0.001
Caregivers and spouses (% , n)	52	16	15	7	0	17	17	23	32	1	90	< 0.001
χ <sup>2</sup> square	55.6	16.7	22.2	4.4	1.1	33.3	13.3	31.1	21.1	1.1	90	< 0.001
	50	15	20	4	1	30	12	28	19	1		

p-Value<sup>c</sup> = 0.482

p-Value<sup>c</sup> = 0.001

GINA, Genetic Information Non-discrimination Act; HIPAA, Health Insurance Portability and Accountability Act.

<sup>a</sup>'Blank' refers to respondents that did not answer the question.

<sup>b</sup>p-Values refer to tests for difference in 'Familiarity with GINA' and 'Familiarity with HIPAA' responses across that row.

<sup>c</sup>p-Values refer to tests of heterogeneity across all four groups.

Table 3. Knowledge of GINA's provisions and limitations by respondents<sup>a</sup>

HD category	Life insurance				Long-term care ins.				Disability insurance				
	No	Yes	Do not know	Blank	No	Yes	Do not know	Blank	No	Yes	Do not know	Blank	N
	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	
Individuals with clinically diagnosed HD (% <i>, n</i> )	9.2	24.6	56.9	9.2	9.2	23.1	55.4	12.3	9.2	27.7	50.8	12.3	65
Individuals with an expanded allele, but do not have clinically diagnosed HD (% <i>, n</i> )	6	16	37	6	6	15	36	8	6	18	33	8	27
Individuals with first- or second-degree relatives with HD (% <i>, n</i> )	3	9	15	0	2	9	16	0	2	8	17	0	38
Caregivers and Spouses (% <i>, n</i> )	25.6	23.1	43.6	7.7	17.9	7.7	64.1	10.3	17.9	15.4	59.0	7.7	39
Overall (% <i>, n</i> )	14.8	25.4	54.4	5.3	11.8	20.7	60.4	7.1	10.1	24.3	59.2	6.5	169
	25	43	92	9	20	35	102	12	17	41	100	11	
(b) GINA limitations													
HD category	Employment				Health insurance				Research studies				
	Yes	No	Do not know	Blank	Yes	No	Do not know	Blank	Yes	No	Do not know	Blank	N
	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	(%)	
Individuals with clinically diagnosed HD (% <i>, n</i> )	38.5	7.7	43.1	10.8	35.4	6.2	49.2	9.2	35.4	6.2	50.8	7.7	65
Individuals with an expanded allele, but do not have clinically diagnosed HD (% <i>, n</i> )	25	5	28	7	23	4	32	6	23	4	33	5	27
Individuals with first- or second-degree relatives with HD (% <i>, n</i> )	16	1	10	0	18	1	8	0	9	2	16	0	38
Caregivers and spouses (% <i>, n</i> )	48.7	7.7	33.3	10.3	33.3	15.4	43.6	7.7	25.6	12.8	48.7	12.8	39
Overall (% <i>, n</i> )	46.2	6.5	40.8	6.5	43.2	7.7	43.8	5.3	32.0	8.3	53.8	5.9	169
	78	11	69	11	73	13	74	9	54	14	91	10	

GINA, Genetic Information Non-discrimination Act.

<sup>a</sup>Response to whether GINA protects against genetic discrimination in each area, where 'Yes' is correct. 'Blank' refers to respondents who did not answer the question.

restricted to families affected by a single genetic disorder, HD. Whether the results are generalizable to other inheritable conditions (e.g. breast cancer, Alzheimer disease) or to the general population remains to be established.

Despite these limitations, our study highlights the lack of familiarity with important and immediately relevant legislation. GINA provides individuals with genetic civil rights protections, similar to civil rights already established based on race, sex, religion, and disability. After enactment of the Americans with Disabilities Act of 1990 (ADA), public knowledge was limited (23). Among individuals who indicated that they were knowledgeable about ADA legislation, they overestimated their familiarity, similar to what we found among individuals in COHORT who indicated familiarity with GINA. Discrimination based on disability is still a major concern of the US government and since the signing of the ADA, various government positions and offices have been established to promote awareness of the policies of the legislation (24–27). The history of the ADA may mirror the current status of GINA and genetic rights. Knowledge of genetic rights may also allay fears of genetic discrimination within the general population and remove some stigmatization surrounding genetic testing and participation in research. Lack of knowledge about GINA, the first federal legislation aimed at preventing genetic discrimination, may limit its impact. Although several groups (28–30) focus on increasing public knowledge of genetic rights, greater education is needed. As has been done for the ADA, the Department of Justice and other federal agencies could work to promote GINA by developing a Web site, toll-free number, and other educational resources for GINA. Multiple federal agencies work to promote the ADA; similar resources allocated to GINA could help ensure that GINA achieves its intended impact. Genetic counselor practice guidelines for Alzheimer disease include suggested discussion of the benefits and limitations of state and federal genetic discrimination legislation (31). Practice guidelines for HD could be updated to include a similar suggestion. The protections enabled by the legislation – more than a decade in its making – likely depend on greater awareness of the law.

### Supporting Information

The following Supporting information is available for this article:

Appendix S1: Section C: Privacy and Insurance

Additional Supporting information may be found in the online version of this article.

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