Parents’ Concern About Their Own and Their Children’s Genetic Disease Risk

Potential Effects of Family History vs Genetic Test Results

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Objective: To evaluate the effect of the genetic risk information source (family history vs genetic test results) on parents’ concern about their own and their children’s genetic disease risk.

Design: Randomized trial using a Web-enabled survey.

Setting: Internet survey.

Participants: National sample of 1342 parents.

Intervention: Parents first received a vignette about their hypothetical genetic risk, randomized as either a family history assessment or genetic test results. Next, parents received a vignette about their youngest child’s hypothetical genetic risk, similarly randomized.

Main Outcome Measure: Parents’ concern about their own and their child’s genetic disease risk.

Results: The response rate was 71.2%. Parents were more likely to be concerned about their own disease risk when the risk estimate came from a family history assessment vs a genetic test result (odds ratio, 1.96; 95% confidence interval, 1.44-2.68). In contrast, information source was not associated with parents’ concern about their children’s disease risk. Parents’ level of concern about disease risk was identical for themselves and their children 73% of the time in the same scenario. When concern differed, parents almost always reported greater concern about disease risk for their children.

Conclusions: Positive family history of disease generated greater concern about parents’ own risk of inherited disease than did genetic test results. This effect was not evident for parents’ concern about their children's risk. As genetic tests emerge and become increasingly available, physicians must not overlook the effect of family history on an individual’s concern about disease risk.

sult instead of a family history. We also sought to evaluate whether parents’ concern about their own genetic risk was similar to their concern about their children’s genetic risk.

Table 1. Randomized vignettes with genetic risk information based on different sources.

<table>
<thead>
<tr>
<th>Genetic Risk Based on Family History</th>
<th>Genetic Risk Based on Genetic Test Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>Imagine that you have family members with a disease that causes severe symptoms in adults. Having this “family history” means that you/your youngest child has a 30% chance of developing this disease. A 30% chance means that 3 out of 10 people will develop the disease. “How concerned are you that (you/your child) might develop this disease?”</td>
<td>Imagine that (you/your youngest) child gets a genetic test result that says he/she has a 30% chance of developing a disease with severe symptoms in adults. A 30% chance means that 3 out of 10 people will develop the disease. “How concerned are you that (you/your child) might develop this disease?”</td>
</tr>
</tbody>
</table>

Figure 1. Randomized vignettes with genetic risk information based on different sources.

OUTCOMES

After reading each vignette, participants responded to the question, “How concerned are you that [you/your child] might develop this disease?” Answers were recorded on a 4-point Likert scale (“very concerned,” “somewhat concerned,” “not too concerned,” and “not at all concerned”). For this analysis, we dichotomized this variable into those who reported concern (“very concerned” and “somewhat concerned”) vs those who reported less or no concern (“not too concerned” and “not at all concerned”).

INDEPENDENT VARIABLES

Demographic information on the participants included age, gender, race/ethnicity, educational status, income, and type of health insurance.

DATA ANALYSIS

We used US Census–based sampling weights provided by the survey vendor to obtain representative estimates for the US population. All the analyses were performed on the weighted sample. We used univariate analyses to determine the distribution of the demographics of the respondents.

We used χ² tests to assess parents’ level of concern about their own disease risk, comparing parents who received the family history results with those who received the genetic test results. We repeated this analysis, focusing on parents’ concern for their children’s disease risk. Of note, we do not report findings from adult respondents who did not have a minor child living in the household; their levels of concern about disease risk were not significantly different from concern expressed by parents (data not shown).

We then performed a simple logistic regression analysis to determine the odds of a parent reporting concern (either “very concerned” or “somewhat concerned”) about their own risk of disease after receiving genetic risk information from a family history vs a genetic test result. We repeated this analysis focusing on parents’ concern for their children’s disease risk. Based on our clinical experiences and expectations, we examined the interaction between parent age and gender for each of these analyses.

We used κ statistics and descriptive bivariate statistics to examine the consistency of parents’ reported concern for themselves compared with their concern for their children when parents received the same information source (family history or genetic test results) for themselves and their child.

We used χ² tests for categorical independent variables and logistic regression for continuous independent variables to explore the associations between demographic characteristics and the following outcomes: parents’ concern about their own risk of disease, parents’ concern about their child’s risk of disease, and parental report of greater concern for their children’s disease risk compared with their own. All analyses were performed using a software program (Stata 8; StataCorp, College Station, Texas).

RESULTS

The survey was presented to 1886 parents, of whom 1342 agreed to participate (response rate 71.2%). The demographic characteristics of the study population are given in the Table.

STUDY PARTICIPANTS

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EFFECT OF INFORMATION SOURCE ON PARENTS’ CONCERN ABOUT THEIR OWN AND THEIR CHILDREN’S GENETIC DISEASE RISK

When parents received disease risk information from family history vs a genetic test result they had an increased odds of reporting concern about their own disease risk (odds ratio, 1.96; 95% confidence interval, 1.44-2.68) (Figure 2). Women were more likely to be more concerned about their disease risk than were men, regardless of the scenario (odds ratio, 1.8; 95% confidence interval, 1.32-2.46). No other demographic characteristics listed in the Table were significantly associated with a greater parental level of concern about their own disease risk, regardless of scenario. We found no interaction between parent gender and parent age with respect to parental report of concern.

Parents’ level of concern about their children’s genetic risk of disease showed a pattern that was distinct from parents’ concern about their own risk. We found no significant difference between parents’ concern about their children’s disease risk and the source of the risk information (Figure 3) (P = .29).

PARENTS’ CONCERN FOR THEMSELVES COMPARED WITH THEIR CONCERN FOR THEIR CHILDREN

When parents were randomized to the same information source for themselves and their child, 73.3% of the time they reported identical levels of concern. Parents were somewhat more consistent in their reported levels of concern when the risk information came from family history vs a genetic test (kappa = 0.67 vs kappa = 0.54). When parents rated concern differently, they almost always reported a higher level of concern for their children than for themselves (Figure 4). Parents were more likely to report a higher level of concern for their child than for themselves when the source of the risk information for

### Table. Characteristics of Parent Respondents

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Weighted Estimate, % (Unweighted No.)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age, mean (range), y</td>
<td>37.9 (37.3-38.4)</td>
</tr>
<tr>
<td>Female sex</td>
<td>58.3 (852)</td>
</tr>
<tr>
<td>Race/ethnicity</td>
<td></td>
</tr>
<tr>
<td>White, non-Hispanic</td>
<td>63.6 (912)</td>
</tr>
<tr>
<td>Black, non-Hispanic</td>
<td>11.9 (166)</td>
</tr>
<tr>
<td>Hispanic</td>
<td>17.0 (200)</td>
</tr>
<tr>
<td>Other, non-Hispanic</td>
<td>7.5 (84)</td>
</tr>
<tr>
<td>Educational level</td>
<td></td>
</tr>
<tr>
<td>&lt;High school</td>
<td>13.8 (156)</td>
</tr>
<tr>
<td>High school</td>
<td>30.8 (390)</td>
</tr>
<tr>
<td>Some college</td>
<td>28.2 (448)</td>
</tr>
<tr>
<td>≥Bachelor’s degree</td>
<td>27.2 (348)</td>
</tr>
<tr>
<td>Annual income, $</td>
<td></td>
</tr>
<tr>
<td>&lt;15 000</td>
<td>13.2 (156)</td>
</tr>
<tr>
<td>15 000 to &lt;30 000</td>
<td>18.2 (214)</td>
</tr>
<tr>
<td>30 000 to &lt;60 000</td>
<td>35.6 (483)</td>
</tr>
<tr>
<td>60 000 to 100 000</td>
<td>23.6 (349)</td>
</tr>
<tr>
<td>&gt;100 000</td>
<td>9.4 (140)</td>
</tr>
<tr>
<td>Insurance status of respondent a</td>
<td></td>
</tr>
<tr>
<td>Private</td>
<td>67.4 (940)</td>
</tr>
<tr>
<td>Public</td>
<td>21.8 (246)</td>
</tr>
<tr>
<td>None</td>
<td>10.8 (126)</td>
</tr>
</tbody>
</table>

aThirty participants did not respond.
Individuals' concern about their genetic risk of disease can significantly influence the decisions they make about their health.\textsuperscript{11-15} It is possible that the source from which individuals receive their genetic risk information can affect their level of concern. In hypothetical scenarios posed to a nationally representative sample, we found that parents were more likely to be concerned about their own genetic risk of disease when they received this information from a family history assessment rather than from a genetic test result. On the other hand, when contemplating their children’s genetic risk of disease, parents' concern was not influenced by the information source. Parents were most consistent in their reported level of concern about their own and their children's disease risk when the disease risk information came from a family history. To our knowledge, this is the first study to examine the independent effect of the source of genetic risk information while focusing on parents and their children.

These findings regarding the influence of family history on parents' concern about their own disease risk indicate that the public may not view genetic tests as deterministically as some may have feared. Rather, it seems for many adults that family history—in effect, one’s “observed” genetic destiny—trumps inherited disease risk as measured by genetic tests. Having family members who develop a disease may lead individuals to interpret their risk as more salient compared with test results that may seem more abstract.

In previous studies of hypercholesterolemia and Alzheimer disease, individuals with family histories of these diseases were influenced more strongly by a combination of genetic testing and family history results than by family history information alone.\textsuperscript{6,14} Following on this work, in the present study, we sought to contrast the separate, independent effects of information provided in the context of family history vs in the context of a genetic test result. We did so using hypothetical scenarios to provide the participants the opportunity to imagine a disease that might be most relevant to their lives. These findings are relevant in the context of the increasing availability of direct-to-consumer genetic testing, through which individuals without a known family history of disease may seek genetic testing information. We did not find evidence to support the fact that genetic testing results would increase parents’ concern about their own or their children’s risk of disease beyond levels conveyed through positive family history of disease.

These findings have several implications. First, like other studies, this study supports the notion that an individual's concern about his or her disease risk is not based solely on a rational interpretation of numerical risk but rather may be affected by a variety of cognitive and emotional factors.\textsuperscript{15} In this study, despite the fact that we provided participants with identical numerical risk estimates, differences in the source of genetic risk information yielded varying concerns about disease risk. Second, these findings highlight the strong influence of family history considerations on individuals’ assessment of their own genetic risk. Even as genetic tests become increasingly available in the current genomic era, physicians must not overlook the influence of family history and, together with patients, should continue strides to track and evaluate family health history.

This study also generated findings that have important implications for genetic risk assessment in pediatrics. Parents generally had the same or greater concern about disease risk for their children as for themselves. However, parents were more likely to report a greater concern about their child’s disease risk compared with their own when that information was generated by a genetic test. These findings suggest generational differences in parents’ concern about their children's genetic risk, which may affect parents' roles as proxy decision makers for their children. Consequently, health care providers must be mindful that parents' concern about their own genetic risk cannot be used as reliable substitutes for their concern about their children's genetic risk.

These findings should be interpreted with some important caveats. First, we did not identify a specific disease in the vignettes. It is likely that the relative influence of family history vs genetic test results on perception of disease risk varies by the disease in question, the clinical scenario, and other real-world factors. However, the goal in this study was to evaluate the independent effect of family history and genetic test results on individuals' perception of genetic disease risk without potential effect modification generated by a specific disease label or other situational factors. We attempted to make the disease scenario as meaningful as possible by presenting a disease that was characterized as having “severe symptoms” as an adult. Moreover, the randomized study design provides internal validity and avoids the selection biases inherent in observational studies regarding specific diseases. As a result, this study provides a basis on which to continue to examine factors that affect parents' perceptions of their own and their children's genetic disease risk.

In this study, we asked participants about their concern for developing a disease. We did so using a scale with a familiar format but that had not been psychometrically validated, and we did not ask about their intentions to act based on this information. However, we focused the analyses on respondents with greater levels of concern because we believed that this group would be most likely to act. Those actions could be positive (eg, engagement in healthy preventive behaviors) or negative (eg, having feelings of anxiety and helplessness). The influences that lead from heightened level of concern to positive or negative actions remain unknown.

We could not examine the effect of child age on parents' perception of their children's genetic disease risk. We expect, however, that the focus on a child's risk for a disease that appears in adulthood may dampen the effect of child age, to some degree.
This survey was conducted using a nationally representative, Web-enabled panel. The participation rate among members of the panel who were contacted was 71.2%. In turn, the makeup of the panel is itself affected by a person’s desire to participate when initially contacted by panel management through random-digit-dialing methods, creating some level of participation bias. After initial enrollment in the panel, participants are not informed of the content of subsequent efforts. Thus, because panel members are not invited in the context of genetic testing research, we believe that it is unlikely that participation affected these results in a meaningful way. Despite their limitations, national surveys using Internet panels recruited through sophisticated methods have made valuable contributions to the literature on a broad variety of topics, including a recent article by Bleakley and colleagues16 regarding public opinions on sex education, work by Davis and Fant9 regarding childhood and adult vaccinations, and work by Klein and colleagues10 regarding patterns of adolescent smoking.

In summary, contrary to the hypothesis, we found among parents that family history as an expression of genetic risk generated greater concern about disease than did genetic test results. We also found that the source of the risk information (eg, family history or genetic test result) did not affect parents’ concern about their children’s disease risk. These findings reinforce the importance of family history in the general public’s assessment of their risk of developing various diseases and emphasize to physicians that they should be mindful of the impact of family history on a patient’s interpretation of risk. Even as technological advances in genetics make genetic tests more easily accessible and more broadly applicable, family history will likely remain a meaningful part of a health evaluation and discussions of disease risk with parents.

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Additional Contributions: Achamyeleh Gebremariam, BS, assisted with the statistical analysis.

REFERENCES