HHS Public Access

Author manuscript

Genet Med. Author manuscript; available in PMC 2016 December 01.

Published in final edited form as:

Genet Med. 2016 September ; 18(9): 924–932. doi:10.1038/gim.2015.192.

Adopting Genetics: Motivations and Outcomes of Personal Genomic Testing in Adult Adoptees

Natalie M. Baptista, BSc (Hons)[1,2], **Kurt D. Christensen, PhD**[1], **Deanna Alexis Carere, ScD, CGC**[1,3], **Simon A. Broadley, MD, PhD**[2], **J. Scott Roberts, PhD**[4], and **Robert C. Green, MD, MPH**[1,5,6] **for the PGen Study Group**

[1]Division of Genetics, Brigham and Women's Hospital, Boston, MA, USA

[2]School of Medicine, Gold Coast Campus, Griffith University, QLD, Australia

[3]Program in Genetic Epidemiology and Statistical Genetics, Harvard T.H. Chan School of Public Health, Boston, MA, USA

[4]Department of Health Behavior and Health Education, University of Michigan School of Public Health, Ann Arbor, MI, USA

[5]Harvard Medical School, Boston, MA, USA

[6]Partners Personalized Medicine, Boston, MA, USA

Abstract

Purpose—American adult adoptees may possess limited amounts of information about their biological families and turn to direct-to-consumer personal genomic testing (PGT) for genealogical and medical information. We investigated the motivations and outcomes of adoptees undergoing PGT using data from the Impact of Personal Genomics (PGen) Study.

Methods—The PGen Study surveyed new 23andMe and Pathway Genomics customers prior to and 6 months after receiving PGT results. Exploratory analyses compared adoptees' and non-adoptees' PGT attitudes, expectations, and experiences. We evaluated the association of adoption status with motivations for testing and post-disclosure actions using logistic regression models.

Results—Of 1607 participants, 80 (5%) were adopted. As compared to non-adoptees, adoptees were more likely to cite limited family health history knowledge (OR = 10.1; 95% CI = 5.7–19.5) and the opportunity to learn genetic disease risks (OR = 2.7; 95% CI = 1.6–4.8) as strong motivations for PGT. Of 922 participants who completed 6-month follow-up, there was no

Corresponding author: Robert C. Green, MD, MPH, Department of Medicine, Division of Genetics, Brigham and Women's Hospital, EC Alumnae building, Suite 301, 41 Avenue Louis Pasteur, Boston, MA 02115, (office) 617-264-5834, (fax) 617-264-3018, rcgreen@genetics.med.harvard.edu.

SUPPLEMENTARY INFORMATION

Supplementary information is available at the *Genetics in Medicine* website. Figure S1 presents a flowchart of the PGen Study design and indicates the composition of the baseline and 6-month survey samples. Table S1 and Figure S2 compare the self-reported family health history information of adoptees and non-adoptees. Table S2 presents adoptees' and non-adoptees' views on genetic privacy 6 months following PGT. Table S3, Table S4, and Table S5 compare the demographics of the 6-month survey sample to those who were not retained for follow-up or excluded from the 6-month analyses for all participants, for adoptees, and for non-adoptees, respectively.

significant association between adoption status and PGT-motivated healthcare utilization or health behavior change.

Conclusion—PGT allows adoptees to gain otherwise inaccessible information about their genetic disease risks and ancestry, helping them to fill the void of an incomplete family health history.

Keywords

Adult adoptee; adoption; direct-to-consumer genetic testing; personal genomic testing; family history

INTRODUCTION

The family health history represents a cornerstone of modern medicine,[1,2] but for some patients, this information is unavailable. Direct-to-consumer personal genomic testing (PGT) is one way for these patients to gain personalized information regarding disease risks, inherited traits, pharmacogenomics, and ancestry.[3–5] However, the provision of genetic information directly to customers with limited family history information highlights one of the primary criticisms of PGT: without a family history in which to contextualize PGT results, and without clinician interpretation, consumers may be falsely reassured by low-risk results, or unnecessarily alarmed by results indicating an elevated risk of disease.[6–8]

Adopted individuals constitute one group whose health care may be affected by absent family history information.[9–11] Nevertheless, adoptees can face ethical and practical challenges when attempting to learn genetic risk information.[12] Anecdotal reports from PGT companies and in the media suggests that adoptees have used PGT from Family Tree DNA and 23andMe, Inc. (23andMe) to find biological family members and learn ancestry information.[4,13] Beyond this, little is known about PGT customers who were adopted and there is no consensus on the appropriateness of disclosing genetic results to those with limited family history information. Because adoptees may differ from the general population in their responses to PGT, there is a need for empirical study of how adoptees perceive and utilize PGT results.[10]

Using data from the Impact of Personal Genomics (PGen) Study,[14] a longitudinal study of PGT customers from 23andMe[15] and Pathway Genomics[16] (Pathway), we conducted an exploratory analysis comparing adoptees and non-adoptees who used PGT. We compared cohorts on baseline (pre-results) demographics, psychosocial characteristics, family health history knowledge, and motivations for seeking PGT. At 6 months post-results, we compared adoptees and non-adoptees on PGT-motivated healthcare utilization and PGT-motivated health behavior change. Finally, we analyzed adoptees' perceptions of the value of PGT.

METHODS

Participants

The PGen Study is a collaboration between academic researchers and industry scientists from 23andMe and Pathway Genomics. Details of the study's design and methodology,[14,17] along with other reports from the study,[18–20] have previously been published. Briefly, new customers of 23andMe and Pathway were recruited between March and July 2012. They provided online consent to participate in the study, including consent to link survey responses with PGT results. Prior to receiving their PGT results, 1648 participants completed a baseline survey. Participants were followed for 6 months after receiving their results. A diagram summarizing the PGen Study design, including the timing of measures and exclusions relevant to these analyses, is provided in the supplementary information (Figure S1). The study protocol was approved by the Partners Human Research Committee.

Personal and Family History

Adoptees and non-adoptees were identified using two items in the baseline survey. Participants were asked, "are you adopted?" and responded "yes" or "no." They also reported whether "desire to learn more about my genetics because I am adopted" was a motivation to seek PGT. Participants who responded "yes" to the first item were classified as adoptees. Participants who responded "no" to the first item, and "very important" or "somewhat important" to the second item, were classified as having an unclear adoption status. Participants with an unclear adoption status who consented to re-contact were re-contacted by phone to confirm if they were adopted, otherwise these participants were omitted from further analyses.

Demographic information was collected via self-report. Self-reported health was measured on a 5-point scale derived from the 36-item Short Form Health Survey ("excellent" = 1 to "poor" = 5).[21] Body mass index (BMI) was calculated using participants' self-reported height and weight. Frequency of exercise was measured using a question adapted from the National Health Interview Survey,[22] where participants reported how many days per week (0 – 7) they performed vigorous exercise for at least 10 minutes. Fruit and vegetable consumption was measured by asking participants to report how many servings of each they consumed on a typical day (0/1/2/3/4/5+), with responses of "5+" recoded as "5." Participants' anxiety and depression levels were assessed using the two-item Generalized Anxiety Disorder (GAD-2) scale[23] and two-item Patient Health Questionnaire (PHQ-2) score,[24] respectively. Higher scores (range = 0 – 6) indicated greater anxiety/depression. A positive emotions score (0 – 6) was calculated as the sum of responses to two Mental Health Inventory[25] items rated on a 4-point scale. Higher scores indicated greater frequency of positive emotions.

Family health history knowledge about first-, second-, or third-degree relatives was ascertained by asking participants whether any of their blood relatives had a history (yes/no) of 15 specific conditions.

Genetic Testing

Exposure to genetic testing and genetics specialists was assessed by asking participants if they had ever met clinically with a genetic counselor or genetics specialist, undergone previous genetic testing other than newborn screening, or previously purchased PGT from a different company. A five-item genetics self-efficacy scale,[18] with a Cronbach's α value of approximately 0.94, was used to calculate a genetics self-efficacy score. Participants rated their agreement with each item using a 7-point Likert scale ("strongly disagree" = 1 to "strongly agree" = 7) and the ratings were summed to give a score ranging from 5 to 35.

Participants reported how important 11 motivations were in their decision to seek PGT using 3-point scales ("very important," "somewhat important," "not at all important"), and provided open-ended responses articulating why they sought PGT. On 3-point scales, participants rated 7 factors in their decision to seek PGT ("considered a lot," "considered somewhat," and "did not consider"), and their interest in learning about 4 types of PGT results ("very interested," "somewhat interested," "not at all interested").

At 6 months post-disclosure, participants were asked with whom they had discussed their results. Participants could choose from family, friends, co-workers, medical professionals (primary care provider (PCP), genetics specialist, or other), and social networking contacts. Healthcare utilization was measured by creating a composite yes/no variable where yes represented a participant who affirmed that their PGT results prompted any of the following: medical tests, exams, or procedures; or consultations with a medical professional. Health behavior change was measured by creating a composite yes/no variable where yes represented a participant who affirmed that their PGT results prompted any change to: diet, exercise, medications (prescription, non-prescription, or alternative), or use of vitamins/supplements. Weekly vigorous exercise, and daily fruit and vegetable consumption, were measured again at 6 months post-disclosure using the same questions employed at baseline.

At 6 months post-disclosure, participants also provided open-ended responses explaining why they thought the PGT experience was valuable or not valuable. A 5-point scale ("not at all" = 1 to "extremely" = 5) was used to assess how valuable participants found their results and how satisfied they were with their decision to seek PGT. Decision regret was assessed using a validated five-item scale (range = 0 – 100),[26] where higher scores indicated greater regret about the decision to undergo PGT.

Data Analysis

We excluded participants from all analyses if they viewed their PGT results prior to completing the baseline survey, had an unclear adoption status, or were missing required baseline data. We excluded from follow-up analyses participants who were missing required 6-month survey data for variables used to determine healthcare utilization or health behavior changes.

Descriptive statistics were computed to summarize the characteristics of the study sample. Data were stratified by adoption status. Categorical variables were compared using Chi-squared tests. Continuous variables were compared using Welch's *t*-tests, which allow for unequal variances within cohorts. Statistical significance was set at $p < 0.05$.

For logistic or linear regression analyses of baseline variables, bivariate or multivariate regression was undertaken as appropriate. To facilitate analysis, motivations were dichotomized to very important versus somewhat/not important, decision-making factors were dichotomized to considered a lot versus considered somewhat/not considered, and informational interests were dichotomized to very interested versus somewhat/not interested. Each of these outcomes was regressed on adoption status using bivariate and multivariate logistic regression.

For logistic or linear regression analyses of 6 month outcomes, bivariate or multivariate regression was also undertaken as appropriate. Discussion of PGT results, healthcare utilization, and health behavior changes were analyzed as dichotomous variables, with regression on adoption status in bivariate and multivariate logistic regression models. Changes from baseline in vigorous physical exercise levels and daily fruits and vegetable consumption were compared by adoption status using Welch's t tests, after confirming normal distributions. Correlation tests were also conducted to assess whether reported PGT-motivated changes in exercise and diet were associated with changes in frequency of vigorous exercise and daily fruit and vegetable consumption from baseline to 6 month follow-up. Satisfaction and value responses were dichotomized to extremely/very versus somewhat/a little/not at all, and differences by adoption status were analyzed using Chi-squared tests. Due to skewed distributions, decision regret was analyzed as a dichotomous variable of scores of 0 and scores of greater than 0. Differences of decision regret by adoption status were analyzed using bivariate and multivariate logistic regression.

Emergent themes were identified from adoptees' free-form responses describing why they underwent PGT, and whether they found PGT to be valuable. Themes were identified by generating word frequency lists, followed by a key-words-in-context analysis performed by the first author.[27]

Multivariate analyses were adjusted for biological children, PGT company, prior PGT, and demographics found to differ by cohort (age, gender, education, race, ethnicity). Statistical significance was set at $p < 0.05$. Data analyses were performed using R software (version 3.2.0; R Foundation for Statistical Computing, Vienna, Austria).

Code Availability

Computer code used for statistical analyses is available from the corresponding author upon request.

RESULTS

Baseline Survey Sample

The baseline analyses of 1607 participants included 80 adoptees and 1527 non-adoptees. Participants who completed the baseline questionnaire but had an unclear adoption status (n = 24) or missing data for descriptive and motivational questions (n = 17, all non-adoptees), were excluded from the analyses (Figure S1).

Compared to non-adoptees, adoptees appeared to be, on average, younger and less highly educated, with fewer biological children, higher BMIs and a lower daily fruit intake, and were more likely to be customers of 23andMe (Table 1), although the difference in number of biological children was not significant when controlled for age ($p = 0.209$). Adoptees reported fewer positive emotions than non-adoptees in the two weeks prior to completing the baseline survey, while anxiety and depression scores did not differ.

Adoptees reported fewer conditions affecting blood relatives than non-adoptees (mean = 3.3 among adoptees versus 6.8 among non-adoptees, $p < 0.001$) (Figure S2). Conditions for which adoptees most frequently reported having an affected blood relative were substance abuse (40%), cancer (37%), and heart conditions (34%). In contrast, non-adoptees most frequently reported having an affected blood relative with cancer (79%), heart conditions (73%), and high cholesterol (70%). Non-adoptees were more likely to report an affected blood relative on 9 of 15 queried conditions (Table S1, all $p \leq 0.001$). Data were missing for 18 adoptees and 32 non-adoptees.

Motivations and Considerations when Purchasing PGT

Adoptees and non-adoptees were both strongly motivated to purchase PGT because they were curious about their genetic make-up (Table 2). Adoptees were more motivated by their limited family health history and desire to learn their personal disease risk than non-adoptees. Half of adoptees and non-adoptees factored actionability of PGT results into their decision to purchase PGT, and adoptees were less likely to consider genetic privacy (23% of adoptees versus 41% of non-adoptees, OR = 0.4, 95% confidence interval, CI 0.2–0.7, $p = 0.001$). Both groups were keenly interested in learning about ancestry and disease risk, and less interested in learning about carrier status. Bivariate analyses suggested that adoptees were less interested in learning pharmacogenomic information than non-adoptees, although differences were not significant in adjusted analyses.

A number of themes emerged from analysis of adoptees' open-ended responses describing why they sought PGT (Table 3). In all themes adoptees acknowledged their lack of personal or familial information, using language such as "no access to knowledge," and indicated that they had turned to PGT to search for their missing information. Adoptees expressed a longstanding desire to learn about their ancestry: "I have always wanted to know more about my background" and "I have always felt a desire to know where I come from." Adoptees' lack of knowledge about their genetic make-up motivated their search for personal genetic risk information such as one who reported wanting to know "relative risk for diseases and adverse pharmacogenetic interactions." Adoptees wanted to learn familial risks for family planning purposes and to educate offspring, and some wanted to find and contact biological family members: "this service will be a long shot to connect with them."

Six Month Follow-Up Survey Sample

Six month follow-up survey data were analyzed for 51 adoptees and 871 non-adoptees (n = 922), after excluding participants who were missing required 6-month survey data for variables used to determine healthcare utilization or health behavior changes (n = 6 adoptees and n = 97 non-adoptees) (Figure S1). No evidence of differential attrition or exclusion by

adoption status was observed, with 64% of adoptees and 57% of non-adoptees included in analyses ($p = 0.237$).

A comparison of the 922 participants who were included in 6-month analyses and the 685 participants who did not complete the 6-month follow-up survey or were excluded from analyses is presented in a supplementary table (Table S3). Compared to participants omitted from analyses, participants whose 6-month data were analyzed tended to be younger (mean age = 46.6 versus 49.4, respectively, $p < 0.001$), were less likely to have biological children (49% versus 57%, $p = 0.001$), were less likely to be 23andMe customers (62% versus 70%, $p < 0.001$), were more likely to have had previous genetic testing (16% versus 11%, $p = 0.018$), and had higher mean genetics self-efficacy scores (29.1 versus 28.2, $p = 0.003$). Among adoptees, no statistically significant differences were observed between participants whose 6-month outcomes were analyzed compared to adoptees who were not analyzed (Table S4). Among non-adoptees, characteristics of participants whose 6-month outcomes were analyzed and omitted followed patterns observed for the study sample overall (Table S5).

PGT-Motivated Actions and Attitudes

PGT results specifically motivated 41% of all participants to utilize a healthcare service, and 56% to change a health behavior (diet/exercise/medications/vitamins or supplements) within 6 months of receiving PGT results. Adoptees and non-adoptees reported similar levels of PGT-motivated healthcare utilization and frequencies of health behavior changes (Table 4). Analyses showed no difference between adoptees and non-adoptees with respect to mean change in vigorous exercise days per week (−0.0 versus 0.1, respectively, $p = 0.511$) from baseline to 6 months, although quantified changes were correlated with reported PGT-motived exercise changes ($r = 0.16$, $p < 0.001$). Analyses also showed no difference between groups with respect to mean change in daily fruit (0.2 versus 0.1, respectively, $p = 0.309$) or vegetable (0.2 versus 0.1, respectively, $p = 0.716$) servings, but again, quantified changes in fruit ($r = 0.08$, $p = 0.012$) and vegetable ($r = 0.14$, $p < 0.001$) consumption were each correlated with reported PGT-motived dietary changes. Adoptees appeared to be more likely than non-adoptees to discuss their results with co-workers (53% of adoptees versus 34% of non-adoptees, OR = 2.1, 95% CI 1.2–3.9, $p = 0.014$), but no more likely to discuss their results with social networking contacts, PCPs, family, or friends (all $p > 0.05$).

The majority of adoptees and non-adoptees considered their PGT results valuable (69% versus 62%) and were satisfied with their decision to seek PGT (78% versus 81%). 71% of adoptees and 60% of non-adoptees scored the minimum for decision regret, and no difference in decision regret was observed between adoptees and non-adoptees in bivariate (OR = 0.6, 95% CI = 0.3–1.1, $p = 0.122$) or adjusted analyses (OR = 0.6, 95% CI 0.3–1.0, $p = 0.074$).

An analysis of adoptees' open-ended responses, describing why the PGT experience was valuable or not valuable is presented in Table 5. Adoptees considered PGT to be valuable because it provided a means of accessing information that was previously difficult for them to obtain. Some adoptees expected to receive more definitive genetic results and others were disappointed that the PGT service had not revealed any close biological relatives.

DISCUSSION

Using data from the PGen Study, we have described the characteristics and motivations of adoptees undergoing personal genomic testing, and how adoptees use their genetic results. A strong desire for information is key to adoptees' beliefs and actions. The information obtained from PGT, including genetic risks, ancestry, and the identities of biological relatives, may be otherwise difficult to obtain for adoptees, whereas PGT provides a convenient and affordable method of access. Our results may help healthcare professionals and policy makers to better understand the desires of adopted patients, and how the provision of genetic information may affect their health. While PGT results did motivate many adoptees to change a health behavior or utilize a healthcare service, they did so no more than non-adoptees.

The adoptees in our sample had less family health history knowledge than non-adoptees, as might be expected. Adoptees experienced fewer positive emotions than non-adoptees in the two weeks prior to testing, but were not more often depressed. A sociopsychological study by Sobol et al.[28] found that adult adoptees who searched for birth parents, compared to non-searchers, were more likely to believe that adoption made them feel different and incomplete. Adoptees may be more emotionally invested in discovering their genetic background than non-adoptees, and deciding to seek PGT may arouse more negative emotions for adoptees who are actively thinking about the family history information they lack.

Both adoptees and non-adoptees were strongly motivated to purchase PGT because of curiosity about their genetics and an interest in learning their risk for specific diseases. Similar motivations of PGT customers have been reported.[5,29] Adoptees were more strongly motivated to learn their genetic disease risks than non-adoptees, and this desire among adoptees may arise from their common void of incomplete family history. A lack of family history information may affect an adoptee's mental health,[11] disease management and prevention,[9,30] and stigmatization by medical professionals.[9] Adoptees may recognize that they are at a health disadvantage because they lack certain family health history information, and are seeking genetic testing to improve their health outcomes.

Numerous blog posts and accounts in the news media have suggested that adoptees use PGT to learn ancestry information and find biological relatives.[4,13] A study by Crouch et al.[30] that explored adoptive parents' attitudes towards whole genome sequencing for their adopted children, coincidently discovered that 3 participants had purchased PGT for their adopted children in an effort to address their child's questions about their background. Our results support the inference that learning ancestry information is a strong motivation for adult adoptees when purchasing PGT. A person's identity can be shaped by their ethnicity and ancestry, and lack of information in this area may be a concern for some adoptees.[11] Adoptees were more likely to be 23andMe customers than Pathway customers, and 23andMe's PGT service may have appealed to adoptees because it offers ancestry information and a method of contacting biological relatives. We expected adoptees to be more interested in learning about ancestry than non-adoptees, but we found that interest in learning ancestry information was not more strongly associated with being adopted.

Some PGT companies integrated notions of autonomy and privacy into their marketing strategies, with advertising campaigns promoting direct, autonomous access to genetic information, while bypassing the healthcare system and avoiding inclusion of results in medical records.[5,31] From this perspective, PGT could be perceived as a service that protects customer privacy. However, some PGT customers may inaccurately assume that confidentiality standards governing physician-patient interactions apply to PGT company-customer relationships,[32] and PGT customers may not be aware of their privacy vulnerabilities. Adoptees were less concerned about the privacy of their genetic information prior to testing than non-adoptees, and may have been more willing to share their results with people not closely related. Adoptees may be less concerned about privacy because they are actively trying to discover their genetic identity; and they may be frustrated about the secrecy surrounding their biological family's genetic information and therefore be advocating for easier access to adoption (and genetic) records.

Our evidence did not suggest that adoptees would be more likely than non-adoptees to base important health decisions on PGT results alone. PGT motivated adoptees and non-adoptees to utilize healthcare services, such as consultations with medical professionals or medical tests, and to change a health behavior, such as changes to medication, diet, or exercise. Other studies have found that customers take similar health-related actions after receiving PGT results.[33,34]

Perhaps because PGT may be the only accessible source of genetic risk and ancestry information for adoptees, the mere opportunity to receive such information was considered valuable. Adoptees who did not find PGT to be as valuable as expected were looking for more definitive information regarding their genetic risks. Su et al.[5] suggested that some PGT users may have overestimated the potential value of PGT. This may also be true of adoptees in our study, in cases where a participant's unrealistic expectations prior to testing contributed to post-disclosure dissatisfaction.

PGT is unable to replace a comprehensive family history assessment. Studies that have analyzed genetic risk predictions from PGT have found inconsistencies between risk estimates and family medical histories.[6,35] Aiyar et al.[6] suggested that PGT results and family medical histories provide complementary rather than identical risk information, and Bloss et al.[35] concluded that PGT results provided little added value beyond that which could be obtained through personal and family health history information. It remains to be seen whether genetic risk predictions from PGT may be useful in cases where family history information is absent.

Strengths of our study include the longitudinal investigation of a large sample of PGT customers, the wide-range of survey questions, and lack of differential attrition. The PGen Study was not originally designed to compare the experiences of adoptees and non-adoptees who used PGT. As such, limitations of this study include unequal sample sizes of adoptees versus non-adoptees and limited power to detect differences between cohorts. Data were self-reported; and significance levels were not adjusted for multiple comparisons, increasing the risk for false-positive findings. While all adoptees in our sample live in the US, we do not know their place of birth and although likely, we cannot assume they were subject to

restrictive adoption laws. Another limitation is that our assessment of family history knowledge is not as comprehensive as a gold standard family history in clinical practice. Items about PGT-motivated health behavior change did not clearly distinguish whether participants made changes that would improve or harm their health, although data suggest that dietary and exercise changes were beneficial. The PGen Study did not collect sufficient baseline data on prescription medication use and adherence to perform similar correlation analyses to those presented for PGT-motivated dietary and exercise changes. Data were collected regarding the frequency of physician visits at baseline, but we omitted this variable from the analyses because data were missing for 16 (31%) of the 6-month sample of adoptees. Finally, our findings are only generalizable to direct-to-consumer PGT customers similar to those enrolled in the PGen Study, who tended to be well-educated, of high socioeconomic status, and White.

Our results emphasize the need for further study of the long-term health impact on adoptees who receive genetic information, particularly addressing any harms or unjustified health-related actions arising from the disclosure of genetic risk results. Large well-established longitudinal adoption studies may be in the best position to study adoptees who use genetic testing services. These studies could analyze adoptees who have used, or who will receive, PGT or other genetic testing.

In conclusion, we conducted an exploratory analysis comparing adoptees and non-adoptees who used PGT, using data from the longitudinal PGen Study. Adoptees used PGT to gain otherwise inaccessible information about their biological families. PGT allows adoptees to uncover information about their genetic identities, helping them to fill the void of an incomplete family history. Concerns that adoptees may apply too great a weight on PGT results appear to be unfounded, with adopted PGT customers no more likely to act on their results than non-adoptees.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgments

This work was supported by a National Institutes of Health R01 award (R01-HG005092). At the time this work was completed, DAC was supported by a Canadian Institutes of Health Research Doctoral Foreign Study Award. RCG is also supported by NIH grants U01-HG006500, U19-HD077671, R01-HG002213 and the Department of Defense. The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Center for Research Resources, the National Institutes of Health, or the Canadian Institutes of Health Research.

Current members of the PGen Study Group are Robert C. Green, Joel B Krier, Margaret H Helm, Lisa S Lehmann, Harvard Medical School and Brigham and Women’s Hospital; Deanna Alexis Carere, Peter Kraft, Harvard School of Public Health; J Scott Roberts, Mack T Ruffin IV, Lan Q Le, Jenny Ostergren, University of Michigan School of Public Health; Wendy R Uhlmann, Mick P Couper, University of Michigan; Joanna L Mountain, Amy K Kiefer, 23andMe; Glenn D. Braunstein, Pathway Genomics; Scott D Crawford, Survey Sciences Group; L Adrienne Cupples, Clara A Chen, Catharine Wang, Boston University; Stacy W Gray, Dana-Farber Cancer Institute; Barbara A Koenig, University of California San Francisco; Kimberly Kaphingst, University of Utah; Sarah Gollust, University of Minnesota.

References

1. Guttmacher AE, Collins FS, Carmona RH. The family history -- more important than ever. N Engl J Med. 2004; 351(22):2333–2336. [PubMed: 15564550]
2. Rich EC, Burke W, Heaton CJ, et al. Reconsidering the family history in primary care. J Gen Intern Med. 2004; 19(3):273–280. [PubMed: 15009784]
3. Roberts JS, Ostergren J. Direct-to-consumer genetic testing and personal genomics services: A review of recent empirical studies. Curr Genet Med Rep. 2013; 1(3):182–200. [PubMed: 24058877]
4. 23andMeBlog. [Accessed October 27, 2015] 2015. http://blog.23andme.com
5. Su Y, Howard H, Borry P. Users' motivations to purchase direct-to-consumer genome-wide testing: an exploratory study of personal stories. J Community Genet. 2011; 2(3):135–146. [PubMed: 22109820]
6. Aiyar L, Shuman C, Hayeems R, et al. Risk estimates for complex disorders: comparing personal genome testing and family history. Genet Med. 2014; 16(3):231–237. [PubMed: 24009002]
7. Heald B, Edelman E, Eng C. Prospective comparison of family medical history with personal genome screening for risk assessment of common cancers. Eur J Hum Genet. 2012; 20(5):547–551. [PubMed: 22215421]
8. U.S. Food and Drug Administration. [Accessed October 27, 2015] Warning letter. 2013. http://www.fda.gov/ICECI/EnforcementActions/WarningLetters/2013/ucm376296
9. Hill CM, Edwards M. Birth family health history: Adopters' perspectives on learning about their child's health inheritance. Adopt Foster. 2009; 33(2):45–53.
10. May T, Strong KA, Khoury MJ, Evans JP. Can targeted genetic testing offer useful health information to adoptees? Genet Med. 2015; 17(7):533–535. [PubMed: 25905442]
11. Corder K. Counseling adult adoptees. The Family J. 2012; 20(4):448–452.
12. Bombard Y, Semaka A, Hayden MR. Adoption and the communication of genetic risk: experiences in Huntington disease. Clin Genet. 2012; 81(1):64–69. [PubMed: 21204796]
13. Swarns, RL. The New York Times. Jan 23. 2012 With DNA testing, suddenly they are family.
14. Carere DA, Couper MP, Crawford SD, et al. Design, methods, and participant characteristics of the Impact of Personal Genomics (PGen) Study, a prospective cohort study of direct-to-consumer personal genomic testing customers. Genome Med. 2014; 6:96. [PubMed: 25484922]
15. 23andMe Inc. [Accessed October 27, 2015] 23andMe. 2015. http://www.23andme.com
16. Pathway Genomics Corp. [Accessed October 27, 2015] Pathway Genomics. 2015. http://www.pathway.com
17. Lehmann LS, Kaufman DJ, Sharp RR, et al. Navigating a research partnership between academia and industry to assess the impact of personalized genetic testing. Genet Med. 2012; 14(2):268–273. [PubMed: 22241103]
18. Carere DA, Kraft P, Kaphingst KA, Roberts JS, Green RC. Consumers report lower confidence in their genetics knowledge following direct-to-consumer personal genomic testing. Genet Med. 2015 advance online publication.
19. Ostergren JE, Gornick MC, Carere DA, et al. How well do customers of direct-to-consumer personal genomic testing services comprehend genetic test results? Findings from the Impact of Personal Genomics Study. Public Health Genomics. 2015; 18(4):216–224. [PubMed: 26087778]
20. Meisel SF, Carere DA, Wardle J, et al. Explaining, not just predicting, drives interest in personal genomics. Genome Med. 2015; 7(1):74. [PubMed: 26269719]
21. Bowling A. Just one question: If one question works, why ask several? J Epidemiol Community Health. 2005; 59(5):342–345. [PubMed: 15831678]
22. National Center for Health Statistics. [Accessed October 27, 2015] Adult physical activity questions on the national health interview survey: 1975–2012. 2012. http://www.cdc.gov/nchs/data/nhis/physicalactivity/pa_questions.pdf
23. Spitzer RL, Kroenke K, Williams JW, Löwe B. A brief measure for assessing generalized anxiety disorder: The GAD-7. Arch Intern Med. 2006; 166(10):1092–1097. [PubMed: 16717171]
24. Kroenke K, Spitzer RL, Williams JB. The Patient Health Questionnaire-2: validity of a two-item depression screener. Med Care. 2003; 41(11):1284–1292. [PubMed: 14583691]

25. Rumpf HJ, Meyer C, Hapke U, John U. Screening for mental health: validity of the MHI-5 using DSM-IV Axis I psychiatric disorders as gold standard. Psychiatry Res. 2001; 105(3):243–253. [PubMed: 11814543]
26. Brehaut JC, O'Connor AM, Wood TJ, et al. Validation of a decision regret scale. Med Decis Making. 2003; 23(4):281–292. [PubMed: 12926578]
27. Ryan GW, Bernard HR. Techniques to identify themes. Field Methods. 2003; 15(1):85–109.
28. Sobol MP, Cardiff J. A sociopsychological investigation of adult adoptees' search for birth parents. Fam Relat. 1983; 32(4):477–483.
29. McGowan ML, Fishman JR, Lambrix MA. Personal genomics and individual identities: motivations and moral imperatives of early users. New Genet Soc. 2010; 29(3):261–290. [PubMed: 21076647]
30. Crouch J, Yu J-H, Shankar A, Tabor H. "We don't know her history, her background": Adoptive parents' perspectives on whole genome sequencing results. J Genet Counsel. 2014:1–11.
31. Hudson K, Javitt G, Burke W, Byers P. ASHG Statement* on direct-to-consumer genetic testing in the United States. Obstet Gynecol. 2007; 110(6):1392–1395. [PubMed: 18055737]
32. Caulfield T, McGuire AL. Direct-to-consumer genetic testing: perceptions, problems, and policy responses. Annu Rev Med. 2012; 63:23–33. [PubMed: 21888511]
33. Kaufman D, Bollinger J, Dvoskin R, Scott J. Risky business: Risk perception and the use of medical services among customers of DTC personal genetic testing. J Genet Counsel. 2012; 21(3): 413–422.
34. Bloss CS, Schork NJ, Topol EJ. Effect of direct-to-consumer genomewide profiling to assess disease risk. N Engl J Med. 2011; 364(6):524–534. [PubMed: 21226570]
35. Bloss CS, Topol EJ, Schork NJ. Association of direct-to-consumer genome-wide disease risk estimates and self-reported disease. Genetic Epidemiol. 2012; 36(1):66–70.

Table 1

Descriptive statistics of adopted and non-adopted PGen Study participants at baseline

Characteristic	Adoptees (n = 80)	Non-adoptees (n = 1527)	*p*
Demographics			
Age, mean ± SD (range)	44.4 ± 13.6 (20–86)	48.0 ± 15.6 (19–94)	0.025
Female, n (%)	49 (61.3)	918 (60.1)	0.840
Non-white, n (%)	15 (18.8)	237 (15.5)	0.439
Hispanic/Latino, n (%)	4 (5.0)	82 (5.4)	0.886
Education, n (%)[a]			0.002
Less than college degree	30 (37.5)	327 (21.4)	
College degree	21 (26.3)	473 (31.0)	
Some graduate school	26 (32.5)	518 (33.9)	
Doctoral degree	3 (3.8)	209 (13.7)	
Annual household income, n (%)[a]			0.166
< $40 000	10 (12.5)	251 (16.4)	
$40 000 – $99 999	38 (47.5)	575 (37.7)	
$100 000 – $199 999	18 (22.5)	486 (31.8)	
≥ $200 000	12 (15.0)	196 (12.8)	
Unknown	2 (2.5)	19 (1.2)	
Marital Status, n (%)[a]			0.104
Single	24 (30.0)	288 (18.9)	
Long-term partner	8 (10.0)	201 (13.2)	
Married	39 (48.8)	849 (55.6)	
Widowed/Divorced/Separated	9 (11.3)	189 (12.4)	
Biological children, n (%)	33 (41.3)	808 (52.9)	0.042
Health insurance, n (%)	74 (92.5)	1449 (94.9)	0.349
23andMe customers, n (%)	65 (81.3)	987 (64.6)	0.002
Health Status			
Self-reported health (1–5), mean ± SD	2.5 ± 0.9	2.5 ± 1.0	0.518
BMI, mean ± SD	28.3 ± 6.6	26.8 ± 6.0	0.047
Vigorous exercise for ≥ 10 min: mean days/week ± SD	2.0 ± 1.9	2.3 ± 2.1	0.100
Servings of fruit: mean/day ± SD	1.7 ± 1.1	2.1 ± 1.1	0.007
Servings of vegetables: mean/day ± SD	2.3 ± 1.1	2.5 ± 1.2	0.140
GAD-2 score (0–6), mean ± SD[b]	1.4 ± 1.8	1.1 ± 1.6	0.164
PHQ-2 score (0–6), mean ± SD[c]	1.3 ± 1.7	1.0 ± 1.5	0.167
Positive emotions score (0–6), mean ± SD	3.5 ± 1.8	4.0 ± 1.8	0.030
Exposure to genetics			
Have met with a genetics specialist, n (%)	4 (5.0)	127 (8.3)	0.291
Previous genetic testing, n (%)	8 (10.0)	213 (13.9)	0.317
Previously purchased PGT, n (%)	11 (13.8)	154 (10.1)	0.292

Characteristic	Adoptees (n = 80)	Non-adoptees (n = 1527)	*p*
Genetics self-efficacy score (5–35), mean ± SD	27.9 ± 6.1	28.8 ± 5.6	0.186

SD, standard deviation; BMI, body mass index; GAD, generalized anxiety disorder; PHQ, patient health questionnaire; PGT, personal genomic testing.

[a]Chi-squared tests were used to obtain global p values for categorical variables.

[b]The GAD-2 was used to assess a participant's level of anxiety, higher scores indicated greater anxiety.

[c]The PHQ-2 was used to assess a participant's level of depression, higher scores indicated greater depression.

Table 2

Logistic regression analyses of motivations, decision-making factors, and informational interests when seeking PGT, by adoption status

	Adoptees (n = 80)	Non-adoptees (n = 1527)	Unadjusted bivariate analysis		Adjusted[a] logistic regression analysis	
Baseline survey item	**n (%)**	**n (%)**	**OR (95% CI)**	***p***	**OR (95% CI)**	***p***
Motivations considered "very important"						
Curiosity about my genetics	67 (84)	1188 (78)	1.5 (0.8 – 2.8)	0.212	1.3 (0.7 – 2.5)	0.793
Limited information about my family health history	67 (84)	497 (33)	10.7 (6.0 – 20.4)	<0.001	10.1 (5.7 – 19.5)	<0.001
Interest in learning my personal risk of disease	62 (78)	903 (59)	2.4 (1.4 – 4.2)	0.001	2.7 (1.6 – 4.8)	<0.001
Personal interest in genetics in general	38 (48)	828 (54)	0.8 (0.5 – 1.2)	0.241	0.7 (0.4 – 1.1)	0.153
Interest in learning my carrier status	34 (43)	644 (42)	1.0 (0.6 – 1.6)	0.954	1.2 (0.7 – 2.0)	0.473
Desire to improve my health	32 (40)	706 (46)	0.8 (0.5 – 1.2)	0.276	0.9 (0.6 – 1.5)	0.823
Desire to create a better plan for the future	30 (38)	705 (46)	0.7 (0.4 – 1.1)	0.131	0.8 (0.5 – 1.3)	0.385
Interest in my personal pharmacogenomics	28 (35)	600 (39)	0.8 (0.5 – 1.3)	0.444	1.1 (0.6 – 1.8)	0.830
Desire to learn about my genetics without going through a physician	28 (35)	433 (28)	1.4 (0.8 – 2.2)	0.202	1.3 (0.8 – 2.1)	0.262
The service seemed fun and entertaining	27 (34)	551 (36)	0.9 (0.6 – 1.4)	0.672	0.7 (0.5 – 1.2)	0.244
Other members of my family are PGT customers	4 (5)	182 (12)	0.4 (0.1 – 0.9)	0.069	0.4 (0.1 – 1.0)	0.079
Decision-making factors "considered a lot"						
Whether genetic information can inform health-related actions	40 (50)	780 (51)	1.0 (0.6 – 1.5)	0.851	1.1 (0.7 – 1.7)	0.759
The convenience of being tested at home	36 (45)	705 (46)	1.0 (0.6 – 1.5)	0.838	0.9 (0.6 – 1.4)	0.595
How well the results can predict my risk of disease	27 (34)	459 (30)	1.2 (0.7 – 1.9)	0.484	1.2 (0.7 – 1.9)	0.507
Cost of services	23 (29)	453 (30)	1.0 (0.6 – 1.6)	0.861	0.9 (0.5 – 1.5)	0.749
Privacy of my genetic information	18 (23)	620 (41)	0.4 (0.2 – 0.7)	0.002	0.4 (0.2 – 0.7)	0.001
The education materials provided by the company	14 (18)	358 (23)	0.7 (0.4 – 1.2)	0.222	0.7 (0.3 – 1.2)	0.160
The possibility of receiving unwanted information	13 (16)	308 (20)	0.8 (0.4 – 1.4)	0.394	0.9 (0.4 – 1.5)	0.614
Information participants were "very interested" in learning						
Ancestry	66 (83)	1116 (73)	1.7 (1.0 – 3.3)	0.066	1.3 (0.7 – 2.5)	0.405
Risk of disease or health condition	63 (79)	1095 (72)	1.5 (0.9 – 2.6)	0.174	1.6 (0.9 – 2.8)	0.116
Pharmacogenomics	30 (38)	810 (53)	0.5 (0.3 – 0.8)	0.007	0.6 (0.4 – 1.0)	0.063
Carrier status	25 (31)	471 (31)	1.0 (0.6 – 1.6)	0.939	1.0 (0.6 – 1.6)	0.849

PGT, personal genomic testing; OR, odds ratio; CI, confidence interval.

Each dichotomized baseline survey item was regressed on adoption status in bivariate and multivariate logistic regression models.

[a] All models adjusted for baseline age, gender, race, ethnicity, education, biological children, PGT company, and prior PGT.

Table 3

Illustrative quotes from adoptees describing why they underwent PGT

Theme	Quotes
Ancestry (30)[a]	"I was adopted a few days after birth and have no record of the ethnicity of my birth parents. I do not undoubtably look any certain ethnicity but my adoptive parents are both white so I've always stood out. I have always wanted to know more about my background…" (Female, age 24) "The family that adopted me has always had a strong interest in their own personal heritage and genetics. I have no information and I have always felt the desire to know where I come from…" (Female, age 21)
Personal genetic risk (42)	"I am an adopted person with no access to knowledge of my genetic heritage or health background. It is primarily because I want to know something about my own genetic make-up that I have done genetic testing." (Male, age 58) "I am adopted and I have no information regarding family history of illness. I am primarily interested in my relative risk for diseases and adverse pharmacogenetic interactions." (Male, age 23)
Familial risks (5)	"I am adopted and plan on having biological children with my wife. We wanted to get some indication of my family medical history and genetic risk factors before we started the process." (Male, age 29) "I was adopted as a baby and cant get any information on my Bio family. I have been ill most of my life… I wanted to be able to give my children some info so they are aware." (Female, age 47)
Finding biological family members (9)	"I am adopted and have been denied information about my birth family although I have been given limited information about their existence. This service will be a long shot to connect with them." (Male, age 42) "I discovered I was adopted three years ago. I have very limited information on my biological parents… the relative finder portion of 23andme.com is a great feature. I am hoping I have some relatives out there that have used 23andme's service." (Female, age 34)

[a]Numbers in brackets indicate the total number of quotes assigned to each theme. Seventy-five quotes were analyzed and quotes could be assigned to more than one theme. Five adoptees did not provide a free-form response.

Table 4

Logistic regression analyses of PGT results-motivated healthcare utilization and health behavior change, by adoption status

PGT results-motivated action reported at 6-month follow up	Adoptees (n = 51)	Non-adoptees (n = 871)	Unadjusted bivariate analysis		Adjusted[a] logistic regression analysis	
	n (%)	n (%)	OR (95% CI)	*p*	OR (95% CI)	*p*
Healthcare utilization (consultations/tests)	22 (43)	358 (41)	1.1 (0.6 – 1.9)	0.774	1.4 (0.8 – 2.6)	0.267
Health behavior change (medication/exercise/diet)	27 (53)	488 (56)	0.9 (0.5 – 1.6)	0.666	1.0 (0.5 – 1.8)	0.909

PGT, personal genomic testing; OR, odds ratio; CI, confidence interval.

Healthcare utilization and health behavior change were analyzed as dichotomous yes/no variables, with regression on adoption status in bivariate and multivariate logistic regression models.

[a]All models adjusted for baseline age, gender, race, ethnicity, education, biological children, PGT company, and prior PGT.

Table 5

Illustrative quotes from adoptees describing why they found PGT to be valuable or not valuable

Theme	Quotes
Gained otherwise inaccessible information (7)[a]	"There is simply no other practical way to obtain this data. Even though its value in planning is limited and generally contains nothing that requires immediate action, it is still valuable." (Male, age 42) "My medical history was a mystery for the first 20 years of my life. I wanted to see what I could glean from these tests to assist me in maintaining my health and improving it." (Female, age 46) "Gave a lot of information my adoptive family could not" (Female, age 24)
Felt relieved after receiving genetic risk results (2)	"Put me at ease especially about cancer and diabetes tendencies." (Female, age 54) "…where I'm not a carrier for certain traits i felt relieved" (Female, age 37)
Desired more definitive risk information (4)	"…many of the results were not clear cut high or low." (Female, age 60) "Thought it may be more specific and less general" (Male, 56)
Disappointed by the lack of biological family members identified (2)	"…has not yet led me to any close matches. Most are 4th or 5th cousins and without any family history, I can't really tell anything." (Female, age 44) "…I would have liked to have seen more close ancestry matches." (Male, age 50)

[a]Numbers in brackets indicate the total number of quotes assigned to each theme. Twenty-six quotes were analyzed and twenty-five adoptees did not provide a free-form response.