Patient characteristics, experiences and perceived value of pharmacogenetic testing from a single testing laboratory

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Aims: Patients' use of and experience with pharmacogenetic (PGx) testing may be impacted by several factors including patient and provider knowledge, health status, and perceived understanding of results. **Materials & Methods:** We conducted an online survey of individuals who had subscribed to a newsletter service offered by a US commercial PGx testing company, Genelex. **Results:** We find that about half of respondents that had PGx testing reviewed one or more of the lab's web-pages, 43% believed they understood the test results very well, but 40% did not know or could not recall whether their provider had changed their prescription based on the test result. **Conclusions:** There was limited use of the laboratory's online resources by respondents undergoing PGx testing. Increased awareness of the website may improve understanding of test results and facilitate discussions with providers about medication changes.

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The integration of pharmacogenetic (PGx) testing into clinical practice will require educational resources for patients to facilitate informed decision-making, promote provider-patient discussion and understanding of test results and significance to care. As test results may be applicable to multiple occasions when treatment is needed, it is important for patients to understand what PGx testing is and the results, particularly to share them with other providers. Furthermore, improving patient comprehension about PGx results may lead to increased confidence in the safety and effectiveness of medications and improved medication adherence. The additional time needed to consent and/or communicate results to patients during an office visit or by phone may pose challenges for some providers, thereby limiting patient understanding.

Informed decision-making and patient comprehension (perceived or actual) of results have been studied for various types of genetic testing [1–4]. For PGx testing, a study of a direct-to-consumer company reported more than 80% understanding of results for response to statin medications without provider support [5]. A pilot study of pharmacist communication of PGx results as part of medication therapy management was also deemed highly satisfactory but less than half reported understanding of test results [6]. Public awareness of PGx testing has been reportedly low [7]. Popular press on PGx testing tends to present a more favorable review of these applications [8].

To promote provider understanding of PGx, some groups such as the Clinical Pharmacogenetics Implementation Consortium have developed informational resources and tools on the evidence basis for PGx tests and implementation of PGx testing in practice [9,10]. Similarly, some professions have begun to incorporate PGx education in curricula [11–13]. In contrast, to our knowledge, there is no centralized resource or dedicated effort toward developing educational resources for patients about the purpose, benefits and risks of PGx testing, and significance to care. Some PGx testing laboratories include information specifically intended for patients on their website. However, it is not clear if these resources are utilized and how clear and comprehensive they are, and whether new resources and/or improvements are needed to supplement existing resources. In this study, we assessed utilization of online informational materials from a single commercial laboratory and the experience of individuals who had considered or undergone PGx testing.

Methods

We conducted an online survey of subscribers of Genelex's email updates service to ascertain interest, value of testing and utilization and satisfaction with online informational resources about PGx testing. Genelex (<http://genelex.com>), a PGx testing laboratory, provides multiple types of online informational resources to providers and patients, in addition to a detailed lab report and patient card. Test ordering requires provider authorization and results are returned to the provider (Genelex is not a direct-to-consumer testing company). The survey was anonymous; no identifying or private health information was collected. The study was approved by the Duke University Health System Institutional Review Board (#Pro00084184).

Survey development

We created an online 33-question survey to ascertain some general demographics, experience with PGx testing and utilization of online educational resources available on at <http://genelex.com>, and understanding of test results and implications for treatment.

Recruitment & data collection

We used the online survey tool Qualtrics (www.qualtrics.com) to design and administer the survey. Such systems provide several features that allow for custom designs, branching/skip logic, randomization of answer choices, multiple collection features and data output formats. In June 2017, an email invitation was sent by Genelex to all subscribers of the Genelex email updates service with a link to the survey homepage. Individuals can self-subscribe to the email service regardless if they have had testing or not through Genelex (there is no automated subscription). At the time of the survey, the subscriber list included 2242 emails. The survey homepage describes the purpose of the study, risks and benefits and information about the survey. At the bottom of the web page, interested participants were asked to affirm that they have reviewed the information that they are 18 years of age or older, and consent to participate in the survey by clicking on the 'I consent' box. A follow-up reminder was sent approximately 2 weeks following the initial invitation. The survey was open for approximately 1 month. At the end of the survey, respondents were eligible to enter a drawing for a \$100 Amazon gift card.

Data analysis

We generated summary statistics for each question. To assess univariate relationships between demographic variables, use of online educational resources and test experiences, we performed Fisher's exact test of independence. Two-sided p-values for all Fisher's exact tests are reported using a Type I error level of 0.05.

Results

Respondent characteristics

A total of 99 respondents completed the survey; 16 additional respondents partially completed the survey (data not included in analysis). Respondents were 76% female, 93% White, 60% with a Bachelor's degree or higher and spanning all age groups with the largest group in the 50–59 years age range (Table 1).

Health status & experience with medications

47% of respondents described their experiences with medications as poor, often having adverse responses or taking drugs that are not effective. 21% of respondents indicated that they have had some medications that caused side effects and another 20% indicated that they have taken some medications that have not worked. 62% of respondents thought that genes contribute a lot to how a person responds to medications, 27% somewhat and 4% a little.

Use of online educational resources

We asked all respondents to indicate whether they recalled reviewing or accessing any of the online educational materials on the Genelex website. Specifically, we asked about four different web pages: the homepage, and three sections on the dedicated Patient tab ('Talk to Your Doctor,' 'FAQ' and 'High Risk PGx Drug Charts'). A total of 52 respondents reviewed one or more of the Genelex web pages. 92% (48/52) recalled reviewing the Genelex homepage, 69% reviewed the 'Talk to Your Doctor,' 71% reviewed the FAQ page and 63% reviewed the drug chart. 29% reviewed one to two web pages queried; 44% reviewed all four web pages queried. We observed no significant differences with respect to gender, age or education level of the respondents that reviewed one or more

Table 1. Survey respondent characteristics (N = 99).

Respondent characteristic	n (%)
Female	76 (76%)
Age group (years):	
– 18–29	15 (15%)
– 30–39	13 (13%)
– 40–49	18 (18%)
– 50–59	23 (23%)
– 60–69	14 (14%)
– 70–79	12 (12%)
– 80+	4 (4%)
Race [†] :	
– White	93 (89%)
– Black	1 (1%)
– Asian	2 (2%)
– Pacific Islander/Native American	2 (2%)
– Hispanic	1 (1%)
– Other	2 (2%)
Highest education level:	
– High school (no degree)	13 (13%)
– High school diploma/GED	6 (6%)
– Associate degree	19 (19%)
– Bachelor's degree	30 (30%)
– Graduate degree	30 (30%)

[†] Four respondents checked two races.
GED: General education development.

web pages and those that did not. Two of the 52 respondents did not have PGx testing through Genelex nor any other laboratory.

Experience with PGx testing

91% of respondents had a PGx test from Genelex and 2% had a PGx test performed by another laboratory. 48% had testing more than a year ago. 65% of respondents that underwent testing believed that genes contributed a lot to how a person responds and 50% often experience medications that do not work and/or have caused side effects. Of the seven respondents that have not had PGx testing, two indicated that they did not think the test would help them, one indicated that their doctor does not recommend testing and four indicated that their insurance would not cover the cost of testing.

Of the 92 respondents who had PGx testing, 65% indicated that their doctor shared the test results with them (5% could not recall). Overall, 87% of respondents received the results in-person at an appointment scheduled for another reason (47%) or at a follow-up appointment scheduled specifically to review test results (40%). 13% received the results by phone or email. Of the respondents that received the results in-person or by phone (n = 58), the majority discussed options for therapy based on the test results; if any changes to medications were needed based on the results; relevance of test result for other medications (current or future) and interpretation of PGx test results (Table 2).

With respect to respondents' understanding of the test results, 43% understood the results very well, 39% indicated they somewhat understood the results and 11% did not really understand the results. There was no significant association between understanding of test results and review of web pages or education level. 88% received a copy of the test report and 75% received a wallet-sized card of the results.

40% did not know or could not recall if their physician changed the prescription based on the test results, 12% indicated that the dose of the prescribed medication was adjusted based on the results, 30% indicated the prescription was switched to a different medication and 18% did not start on prescribed medication until test results were back. There was no significant association between who could not recall or did not know if their physician

Table 2. Topics of discussion with provider during communication of results (n = 58).

Topic	Yes; my doctor did discuss this	No; my doctor did not discuss this	Do not know/cannot recall
Options for therapy based on the test results	41 (71%)	14 (24%)	1 (2%)
Changes (or no changes) necessary for the medication based on results	41 (71%)	11 (19%)	4 (7%)
Relevance of test result for other medications you currently take or may be prescribed in the future	42 (72%)	9 (16%)	4 (7%)
The specific genetic results/gene changes	19 (33%)	23 (40%)	11 (19%)
A description of what your genetic test results means (for example, that you may break down drugs slowly)	41 (71%)	12 (21%)	3 (5%)

Table 3. Respondent satisfaction with pharmacogenetic testing experience (n = 92).

Question	Extremely satisfied; very helpful; very confident; very likely	Fairly satisfied; somewhat helpful; somewhat more confident; somewhat likely	Uncertain/neutral	Rather dissatisfied; not very helpful; not very confident; not very likely	Extremely dissatisfied; not at all helpful; definitely not more confident; definitely not more likely
How satisfied were you with your decision to take the test?	53 (58%)	19 (21%)	9 (10%)	9 (10%)	2 (2%)
How helpful do you think the test result was to your doctor's decision about your treatment?	35 (38%)	18 (20%)	18 (20%)	13 (14%)	11(12%)
After having the test, did you feel more confident that the medication prescribed would not cause side effects and/or would help your condition, compared to past prescriptions you've received without testing?	31 (34%)	30 (33%)	18 (20%)	7 (8%)	6 (6%)
How likely would you be to have another PGx test if recommended for other medications prescribed to you in the future?	50 (54%)	18 (20%)	15 (16%)	1 (1%)	8 (9%)
Some rows will total more than 100% due to rounding.					

had made a change to their prescription and education level. 41% indicated that the test results had been useful in determining the appropriate medication one- to three-times and 12% in more than six occasions. 26% indicated that the test results had not been useful in determining the appropriate medication.

Sharing of results

25% of respondents indicated that they had shared the test results with a pharmacist. Those who had shared the results were more likely to be 50 years and older ($p = 0.054$); but neither gender, education level nor prior negative experience with medications were associated with sharing of results. Of those who shared the test results with a pharmacist, 48% indicated that the pharmacist responded very positively and reviewed the results, 26% responded negatively (did not understand results, did not know how to interpret, did not have time) and 22% indicated that the pharmacist did not understand the results. 61% shared the results with other doctors that prescribe medications for them. Of those that shared the results with other doctors, 32% indicated that the doctors responded very positively and reviewed the results, 29% indicated that the doctors' did not understand the results and 14% were unsure about the doctors' response.

Satisfaction

58% of respondents were extremely satisfied with their decision to have PGx testing and 21% were fairly satisfied (Table 3). We did not observe any association with between level of satisfaction and gender, age, education level or prior negative experiences with medications. 89% indicated that they trusted the test result; 38% found testing to be very helpful in informing treatment decisions and 20% were uncertain. After having the test, 34% definitely felt more confident that the medication prescribed would not cause side effects and/or would help treat their condition, compared with the past prescriptions received without testing (33% felt somewhat more confident and

20% unsure). 54% indicated that they would be very likely to have another PGx test if recommended for future medications and 20% would be somewhat likely (9% would be very unlikely).

Discussion

The implementation of PGx testing may pose a learning curve for patients and providers alike until awareness and resources are sufficiently available and testing has become more widespread. While some efforts have been developed to train and support health providers' awareness and appropriate test utilization, it does not appear that equal effort has been devoted to patient education. The combination of several factors – the novelty of PGx testing, lack of patient and provider knowledge and lack of informational resources about testing – may result in a varied experience for patients. In this study, we explored patients' experience with PGx testing, particularly focused on their understanding of the test results, sharing of results and use of the test laboratory's online resources. We found that the majority of respondents were satisfied with their experience with PGx testing and understood the results, although a third could not recall or did not know if their provider changed their prescription based on the test results.

The demographics of patients undergoing PGx testing have not been widely reported; interestingly, our survey respondents represented all age groups and educational backgrounds (although predominantly older and highly educated). Almost half of respondents indicated that they had had poor experiences with medications and 62% believed genes contributed to drug response, two factors presumably contributing to their decision to undergo PGx testing.

With respect to the utilization of the testing website materials, about half of respondents that had PGx testing performed by Genelex reported to have reviewed the laboratory's web page. The limited use of the online resources could have been due to respondents' unawareness that the laboratory had a website and/or that resources for patients were available from the laboratory website. Of those that did review one or more pages on the Genelex website, after the homepage, the majority of respondents reviewed the FAQ page. In addition, respondents may have conducted a general online search for information about PGx testing. However, even if information about PGx testing is found, it may not be comprehensive, accurate or understandable to patients. Providers have also reported struggling to find information online about PGx testing to inform appropriate use [14,15]. Patients with low health literacy may have additional challenges with comprehending information about PGx testing or be less likely to seek health information [16]. We have worked toward developing educational tools for providers to use to help discuss PGx testing [17,18] as well as videos for patient education [19].

Despite providers' reportedly low level of knowledge about PGx testing [20–22], the majority appeared to have discussed several aspects of the PGx results with their patients, with the exception of the specific test result. The broad discussion may be attributed to providers being early adopters of PGx testing and possessing greater knowledge and thus, the results may not reflect the general provider population. Not discussing the actual test result may be due to provider's belief that it was not relevant for patients to know and/or the information was too complex to explain in a limited time. The finding that almost a third of ordering test providers did not share results with the patient is somewhat surprising, given the novelty of test and potential patient interest in learning of their genetic predisposition to experience an adverse event, particularly given that many respondents indicated poor prior experiences with medications. Only a quarter of respondents indicated that they shared results with a pharmacist, but 60% shared the results with other prescribing providers. When respondents shared the results with either pharmacists or prescribing providers, they reported that a similar proportion responded negatively, though prescribing providers had a higher positive response than pharmacists. With 40% of respondents indicating that they either understood the results very well or somewhat, more discussion time, improvement in communication and/or availability of supplemental resources could improve patient understanding.

Some limitations about this study should be noted. The response rate was very low and the population may be a biased pool since individuals chose to self-subscribe to the e-newsletter. The respondents were predominantly White and highly educated and had testing ordered through a single laboratory. We did not gather personal health information such as name of provider and indication for testing. Some responses may be subject to recall bias or poor recollection of events given that some time may have passed between reviewing the website, undergoing testing and receiving the test results. The findings reported here, nonetheless, shed light on the need for more patient support and provider resources to improve patient comprehension of test results.

Conclusion

Online laboratory resources may be helpful to patients considering or who have already underwent PGx testing in understanding the clinical significance of the results. However, not all patients will be aware of their existence and therefore, the test report and/or provider should inform patients about information provided by the laboratory among other resources. In addition, laboratories should insure that the materials are presented and written at a level and style appropriate for patients of all literacy levels to optimize comprehension and utilization.

Summary points

- Ensuring patient comprehension about the purpose, benefits and risks of pharmacogenetic (PGx) testing, and significance to care will be important for ensuring informed consent, improving patient engagement and sharing of test results with prescribers.
- To our knowledge, there is no centralized resource or dedicated effort toward developing educational resources for patients, though some PGx testing laboratories include information specifically intended for patients on their website.
- Our study assessed the use of one PGx testing company's website and found mixed usage of various pages on the website.
- Survey respondents who underwent PGx testing generally reported positive experiences and would undergo testing again if needed.

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