

# Personalized Medicine: patient & healthcare professionals' attitudes towards pharmacogenetics & the PA role in individualized care

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## Introduction

As advancements are being made in the field of genetics, there has been a realization that in addition to understanding how traits and disorders are passed through a family, one's genetic makeup can be used to create personalized medical plans and revolutionize the approach to the administration of pharmaceuticals. This new approach to pharmacology uses information on genetic variation paired with an understanding of drug interactions and chemistry, in order to predict one's response to different types and doses of drugs. As pharmacogenomics has been investigated, it's been shown that both inherited and acquired variants can help predict the efficacy and toxicity of specific drugs in a patient. The aim is to create individualized medicine, where pharmacological decisions are not made as a generalization for an entire population, but rather in a targeted fashion for a single patient. With the correct implementation, pharmacogenetics could offer individualized, more effective, and safer treatments to patients.

## Purpose

As pharmacogenetics (PGx) emerges, there is a need to comprehend the patient perspective, in order to know their concerns and desires from pharmacogenomic testing. It is also important to understand other healthcare providers' views on pharmacogenomics, in order to understand any barriers there are in implementing pharmacogenomics into clinical practice. With a thorough understanding of the current climate of pharmacogenomics in the healthcare system, physician assistants can understand what roles they can fill and services they can provide in this field as it becomes more validated and mainstreamed.

## Methods

A review of the literature surrounding patients' and healthcare professionals' perception, knowledge, and understanding of pharmacogenomics was conducted, including only research performed in the last 15 years. As a large portion of the research was conducted in a qualitative manner, such as through focus groups, analysis was done in a qualitative manner, examining trends that emerged throughout the literature, in order to propose areas where patients seem consistently to be concerned or hopeful. This was then analysed in the context of healthcare provider's knowledge of pharmacogenomic testing and their comments and concerns regarding the implementation of PGx into current standards of care and clinical practice. Given that the field of pharmacogenomics is only growing and that PAs function as physician extenders with prescribing capabilities, it is only natural that they have a role to play in this field. The current literature involving PAs and pharmacogenomics was reviewed and concepts and suggestions for further implementation of PGx education and future directions were discussed.

## Canadian Law & PGx

**BILL 201**

**THE GENETIC NON-DISCRIMINATION ACT**

**1 Can my insurance company request that I undergo genetic testing?**

- No this is prohibited by the law
- Any person, or company, that you enter into a contract with, or who provides you goods or services, cannot require genetic testing prior to upholding contract terms

**2 Do I have to disclose my genetic testing results to anyone?**

- Not unless you wish to do so
- This includes, but is not limited to: employers, insurance companies, banks, etc
- An exception is provided to health care practitioners and researchers through whom you obtain the results, or who request your results for treatment purposes

**3 Can I be discriminated against for my genes or genetic test results?**

- No this is prohibited by the law
- The Canadian Human Rights Act has now been amended to prohibit discrimination on the ground of genetic characteristics

**4 Can my employer request that I undergo genetic testing?**

- No this is prohibited by the law
- The Canadian Labour Code has now been amended to protect employees from being required to undergo or to disclose the results of a genetic test

## Patient Perspectives on PGx

| References                            | 1   | 6    | 7                  | 9                    | 10   | 11   | 13   | 18                    | 19   | 23  |
|---------------------------------------|-----|------|--------------------|----------------------|------|------|------|-----------------------|------|-----|
| General Findings                      |     |      |                    |                      |      |      |      |                       |      |     |
| Interest in PGx                       | Low | Yes  | Yes                | Yes                  | Yes  | Yes  | Yes  | Yes                   | Yes  | Yes |
| Understand PGx                        | No  | >75% |                    | No                   | >75% | <50% | No   |                       | <50% |     |
| Understanding of drug response        |     |      | No                 |                      | No   |      | <25% |                       | No   |     |
| Return of results                     |     |      | Familiar, informed | No strong preference |      |      |      | Doctor; not other HCP | GP   | GP  |
| Patient Concerns                      |     |      |                    |                      |      |      |      |                       |      |     |
| Cost                                  | ✓   | X    |                    |                      |      |      | ✓    |                       |      | ✓   |
| Unequal access                        | ✓   |      |                    |                      |      |      | ✓    |                       |      |     |
| Privacy                               | ✓   |      |                    | ✓                    |      | ✓    | ✓    |                       | X    |     |
| Discrimination                        |     |      |                    | ✓                    | X    |      | ✓    |                       | ✓    | ✓   |
| Anxiety or inadequacy                 |     |      | ✓                  |                      |      |      |      |                       | X    |     |
| Role in decisions                     |     | ✓    |                    | ✓                    |      |      |      |                       |      |     |
| Treatment delay                       |     | X    |                    | ✓                    |      |      |      |                       |      |     |
| Inferior treatment or adverse effects |     | X    |                    |                      |      |      |      |                       | ✓    |     |
| 2 <sup>o</sup> findings               |     | X    |                    |                      |      |      |      |                       |      |     |
| Patient Interests                     |     |      |                    |                      |      |      |      |                       |      |     |
| Best drug choice                      |     |      | ✓                  |                      | ✓    | ✓    | ✓    |                       | ✓    |     |
| Improving health outcomes             |     | ✓    | ✓                  |                      |      |      | ✓    |                       | ✓    |     |
| No trial and error                    |     |      | ✓                  |                      |      |      | ✓    |                       | ✓    |     |
| Doctor offered it                     |     | ✓    |                    |                      | ✓    |      |      |                       |      |     |
| Reassurance over current treatment    |     | ✓    |                    |                      | ✓    |      |      |                       | ✓    |     |
| Personalizes healthcare               |     |      | ✓                  |                      |      | ✓    |      |                       | ✓    |     |

## HCP Perspectives on PGx

**HCP Lack of Knowledge**

Expressed more by Primary Care HCPs than those in Genetics/Genetic-related fields<sup>15, 23</sup>

Common suggestion - results accompanied by a statement from an expert in the field with recommendations and original data<sup>8, 20, 23</sup>

- Felt unable to properly interpret results & communicate the disease risk to patients
- Results do not clearly translate into prescribing strategies or risk categories
- Lack of information/resources for HCPs about how to translate metabolic results

**What do Patients Want?**

HCPs overwhelmingly felt that patients would feel pressured to acquire testing for HCP prescribing benefit and assumed most patients did not want any genetic testing<sup>15, 18, 23</sup>

Interestingly - many HCPs commented that patients were the ones bringing them PGx inquiries or results from direct-to-consumer tests<sup>2, 18, 20</sup>

**Access to Care**

Would a certain PGx result make someone ineligible for potentially disease-altering treatment?<sup>25</sup>

Rural HCPs expressed worries about what their access to PGx, specialists, treatment options, counselling for patients & families<sup>7</sup>

**Clinical Utility/Standard of Care**

Many HCPs felt that until PGx testing is incorporated into guidelines, they will continue to use preexisting standards of care<sup>2, 7, 8</sup>

**Warfarin Study: Should Treatment Be Delayed Until PGx Results?**

50% even split between yes and no

**Individualized Healthcare**

Overall positive interest - greatest with GCs, Geneticists HCPs in genetic-related fields (oncology, cardiology) ? Research interest from HCPs > Clinical Interest ?

**Perceived Benefits to Patients:**

- Decreased adverse effects<sup>17, 7</sup>: Less drug toxicity, Able to reach adequate & effective doses faster
- Improved quality of life<sup>2, 4</sup>: Patients feel empowered, Potential improvements in mortality/morbidity

**Who Counsels the Patient?**

Primary Care HCPs felt that they do not have enough time for pre- and post-test counselling - felt it should be a role of genetic-HCPs<sup>15</sup>

Genetic-HCPs felt that whoever orders the test should counsel the patient<sup>15</sup>

**Secondary Findings**

What happens when incidental or ancillary disease information, unrelated to the purpose for which the test was ordered, is found?

- Primary Care HCPs<sup>8, 15</sup>: Patients need to be warned prior to ordering test of all ancillary information risks
- Genetic HCPs<sup>15</sup>: Patients do not always need to be warned of ancillary information risk; genotype is not phenotype (ex: incomplete penetrance)

**Social, Financial & Emotional Burden**

North American HCPs > European HCPs<sup>7</sup>

Primary Care HCPs felt unprepared to deal with the psychosocial aspects of results<sup>18</sup>

- Insurance & Job Discrimination<sup>8, 15, 23</sup>
- Emotional burden on patient & impact on families<sup>15, 20</sup>
- Large cost to the patient and to the system<sup>2, 23</sup>

## PA Role in PGx

# PAs & PGx

## Why PAs Should Care & What They Need to Know

**PAs are a part of PGx targeted interdisciplinary HCP teams**

PGx can only work if the interdisciplinary teams it is targeted towards understand its purpose and work together. PAs as health advocates can assist in educating their team about the benefits of individualized medicine.

**PAs Can Prescribe PGx Tests & PGx Meds**

If within the scope of practice of their supervising physician, PAs are able to refer patients for PGx testing, and can prescribe many of the drugs (ex-warfarin) implicated by PGx test results.

PAs must understand PGx because the impacts of genes may influence a patient's response to medications and why they may have a drug "failure" or drug "toxicity".

**PAs Can Make the Time to Counsel Patients**

One of the most cited reasons by primary care HCPs for not integrating PGx was the lack of time for pre- and post-test counselling<sup>16</sup>: a role that PAs, with their increased patient contact time, can play! A core competency of the PA role is as a patient-centered communicator; PAs can take time to corroborate family histories and explain tests and potential results to patients.

**Advice from a Genetic Counsellor:**

Prior to pursuing any genetic testing, ask yourself these three questions:

- Am I equipped with the proper knowledge & access to resources to give this patient the best care as it relates to this situation?
- Am I prepared to discuss the implications of being tested (or not tested) and the magnitude of the information received from this test's results?
- Am I able to interpret the results in a clinically meaningful manner for myself, and can I explain them to my patient?<sup>19</sup>

**Learn About PGx**

In a US study<sup>8</sup>, PA students underwent a 1 hour laboratory explaining PGx using case studies for determining test candidates and data interpretation. The results: students went from 'not confident' to 'very confident' in their knowledge of genomic medicine & PGx.

Helpful Resources:  
PharmGKB database - <https://www.pharmgkb.org/>  
Canadian Pharmacogenomics Network for Drug Safety - <http://cpnds.ubc.ca/>

## Conclusion

There is a disconnect between patients' desires for PGx testing and the integration of PGx by healthcare providers. As identified by our literature review, patients were most interested in PGx to determine drug responses, and for conditions that are treatable with modern medicine. Despite acknowledgement of the benefits to patient outcomes, HCPs remain reluctant to utilize PGx primarily due to the lack of resources and information available to them. Conversely, a lack of understanding of PGx in patients did not influence their desire to pursue PGx. Further, the Education Committee of the International Society of Pharmacogenomics has called for incorporation of PGx teaching into the core curriculum of HCP schools. It is evident that education of both practicing and studying HCPs of PGx as well as strategies for communicating with patients is essential as we move forward into the age of individualized medicine.

## Future Directions

Of all the studies in this analysis, very few included Canadian patient or HCP populations. Further, only a handful included practicing PAs, primarily in the USA. We would like to expand on this research, given Canada's genetic non-discrimination law, and interview a sample of Canadian HCPs and PA students on their interactions with, and knowledge of genetic medicine and PGx. This study is currently under review by the Hamilton Integrated Research Ethics Board.

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