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Review

Striking a balance in communicating pharmacogenetic test results: Promoting comprehension and minimizing adverse psychological and behavioral response

Susanne B. Haga [a,*], Rachel Mills [b], Hayden Bosworth [c]

[a] Institute for Genome Sciences & Policy and Sanford School of Public Policy, Duke University, Durham, USA
[b] Institute for Genome Sciences & Policy, Duke University, Durham, USA
[c] Departments of Medicine, Psychiatry, and Nursing, Duke University, Center for Health Services Research in Primary Care, Durham VAMC, Durham, USA

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ABSTRACT

Objective: Pharmacogenetic (PGx) testing can provide information about a patient's likelihood to respond to a medication or experience an adverse event, and be used to inform medication selection and/or dosing. Promoting patient comprehension of PGx test results will be important to improving engagement and understanding of treatment decisions.
Methods: The discussion in this paper is based on our experiences and the literature on communication of genetic test results for disease risk and broad risk communication strategies.
Results: Clinical laboratory reports often describe PGx test results using standard terminology such as 'poor metabolizer' or 'ultra-rapid metabolizer.' While this type of terminology may promote patient recall with its simple, yet descriptive nature, it may be difficult for some patients to comprehend and/or cause adverse psychological or behavioral responses.
Conclusion: The language used to communicate results and their significance to patients will be important to consider in order to minimize confusion and potential psychological consequences such as increased anxiety that can adversely impact medication-taking behaviors.
Practice implications: Due to patients' unfamiliarity with PGx testing and the potential for confusion, adverse psychological effects, and decreased medication adherence, health providers need to be cognizant of the language used in discussing PGx test results with patients.

Contents

* Corresponding author at: Institute for Genome Sciences & Policy and Sanford School of Public Policy, Duke University, 304 Research Drive, Box 90141, Durham, NC 27708, USA. Tel.: +1 919 684 0325; fax: +1 919 613 6448.
E-mail address: susanne.haga@duke.edu (S.B. Haga).

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1. Introduction

Pharmacogenetic (PGx) testing assesses variation in genes associated with drug response, providing information about a patient's likelihood to respond to a given medication or experience an adverse event. Since the result of a single test can be important for several different medications due to shared mechanisms of drug metabolism, PGx testing may be relevant over the course of a patient's lifetime. In particular, understanding of one's drug predisposition and its impact on dosing or drug selection may result in improved outcomes, medication adherence, and patient satisfaction [1]. Therefore, patient comprehension of the test result becomes both critical but challenging due to a number of factors including ~~to~~ varying provider and patient knowledge and experience with testing, limited time during an office visit for discussion of results, and patient health literacy and numeracy.

The psychological impact of genetic testing has been broadly examined for several conditions. In general, the impact appears to be minimal even for diseases without clinical interventions such as Alzheimer disease [2–4] and even when testing for multiple conditions through panel or array-based testing [5–7]. In contrast, PGx testing is believed to raise even fewer risks of psychological harms and stigmatization or discrimination compared to genetic testing for disease [8–10]; however, no evidence supports this presumption. It is possible that these risks have been underestimated when considering the challenges of effectively communicating results, particularly given the language used to report PGx test results and the potential long-term and recurring use of PGx test results. Furthermore, it is not clear if psychological harms from predictive genetic testing for inherited disease and PGx testing are comparable as individuals with a personal or family history of a disease may cope with test results differently than individuals with no prior personal or family history or expectation for need for testing, as may be the case for PGx testing. Additionally, a patient's psychological response to a PGx test result may be compounded by the nature of the condition for which treatment is needed.

It is anticipated that PGx testing is likely to become more widely used with more than 100 medications containing PGx information in their label and increased development of companion diagnostics [11,12]. Therefore, it is important to consider the effects of PGx testing and the manner in which it is discussed with patients. In this paper, we consider potential risks of the language currently used to report PGx results and suggest alternatives with less risk of psychological effects. To our knowledge, there is little research or literature specific to communication of PGx test results. Therefore, the suggestions proposed in this paper are based on our experiences and the literature on communication of disease-based genetic test results and the broad risk communication strategies. As a best delivery model of PGx testing has not yet been determined, any provider including but not limited to physicians, pharmacists and genetic counselors may consider incorporating these suggestions when using PGx testing.

1.1. PGx testing

Among other factors such as environment, body habitus, and drug–drug interactions, response to medication is affected by variation in genes encoding the drug target or involved in metabolism, transport, and other essential functions. For example, several genes encoding liver enzymes, known as cytochrome P450 (CYP) genes important to the metabolism of many commonly prescribed medications, are highly polymorphic, resulting in a range of enzyme activity levels in patients [13]. PGx testing can provide knowledge about a patient's level of enzyme activity or presence or absence of a genetic variant for a targeted drug can inform medication selection or dosing to improve treatment response or reduce risk of an adverse event [13]. Testing may be ordered when a drug is prescribed (point-of-care) or preemptively [14]. In addition to physicians, nurses and pharmacists may play a role in delivery of PGx testing [15–17], particularly with respect to promoting patient understanding of the test result [18].

Clinical laboratories conducting PGx testing often report the test outcome in the lab report in multiple ways: the molecular genotype (e.g., T/T) and by its allelic abbreviation (e.g., *1/*5) and phenotype (e.g., poor metabolizer). The phenotypic descriptors of the level of enzyme activity used in clinical laboratory reports are standard terminology accepted in the medical research literature and clinical guidelines. Additional text may or may not be included to explain the phenotype: e.g., 'poor metabolizer' status means that the patient has low or no enzymatic activity, whereas an 'ultra-rapid metabolizer' refers to a patient with extremely high metabolic activity ('intermediate' and 'extensive' metabolizer refer to normal or moderately increased levels of enzyme activity, respectively).

2. Concerns with using standard reporting language to communicate PGx results

For communication of any test result, the primary goal is to optimize patient understanding about the test outcome and how the information impacts medical management. However, communicating PGx results may be more challenging than previously considered for many reasons including the type of test outcome (genotype) and interpretation (phenotype), limited patient knowledge about genetics and the role of genes in health, and the differing impact of results for each medication prescribed (e.g., for one medication, dose may be reduced based on the PGx test result, but for another, the dose may be increased based on the same test result). Patient factors such as health literacy [19], genetic literacy [20,21] and numeracy [22] may also affect their ability to understand PGx testing and the results. Additionally, provider knowledge of PGx is one of the most common barriers reported [23–26] and is associated with limited experience with testing [27,28]; similarly, limited knowledge and experience may affect communication about PGx testing [29]. Though participants have reported their informational needs for PGx testing in a research setting [30], to our knowledge, there are no recommendations regarding what information should be discussed with patients (pre or post-testing) and/or the language to be used during these discussions in a clinical setting. Based on our experiences with PGx testing in a primary care setting, we published a paper identifying key information to be discussed pre and post-testing, though did not review in detail the importance and impact of effectively communicating the test result [31]. Given the range of literacy levels of patients and even for those highly literate but unfamiliar with PGx testing, the language used to describe a patient's genotype or phenotype for drug response must be carefully considered not only to promote comprehension, but also to avoid