The measurement of patient attitudes regarding prenatal and preconception genetic carrier screening and translational behavioral medicine: an integrative review

Jennifer J. Shiroff, PhD, APN-C,¹ Mathew J. Gregoski, PhD, MS²

¹Jefferson College of Nursing,
Thomas Jefferson University, 130
South 9th Street, Edison Building,
754, Philadelphia, PA 19107, USA

²College of Nursing,
Medical University of South Carolina,
99 Jonathan Lucas Street, MSC 160,
Charleston, SC 29425, USA

Correspondence to: J Shiroff
jennifer.shiroff@jefferson.edu

Cite this as: *TBM* 2017;7:364–370
doi: 10.1007/s13142-015-0346-8

Abstract

Measurement of recessive carrier screening attitudes related to conception and pregnancy is necessary to determine current acceptance, and whether behavioral intervention strategies are needed in clinical practice. To evaluate quantitative survey instruments to measure patient attitudes regarding genetic carrier testing prior to conception and pregnancy databases examining patient attitudes regarding genetic screening prior to conception and pregnancy from 2003–2013 were searched yielding 344 articles; eight studies with eight instruments met criteria for inclusion. Data abstraction on theoretical framework, subjects, instrument description, scoring, method of measurement, reliability, validity, feasibility, level of evidence, and outcomes was completed. Reliability information was provided in five studies with an internal consistency of Cronbach's $\alpha > 0.70$. Information pertaining to validity was presented in three studies and included construct validity via factor analysis. Despite limited psychometric information, these questionnaires are self-administered and can be briefly completed, making them a feasible method of evaluation.

Keywords

Integrative review, Conception, Pregnancy, Genetic testing, Attitude, Patient

BACKGROUND

The use of genetic carrier screening for autosomal recessive traits has become an integral part of prenatal care and the care of individuals who are planning to conceive a pregnancy. However, an estimated 51 % of pregnancies are unplanned, as a result, most people do not seek counseling from their healthcare providers prior to conceiving a pregnancy because they are already pregnant [1, 2]. Rare genetic diseases account for 20 % of infant mortality, making them a considerable public health issue [3]. For example, approximately 1 in every 20 Americans is an unaffected carrier of a cystic fibrosis mutation, amounting to an estimated 12 million people [4]. Roughly 90 % of these carriers are unaware of their carrier status [5]. Internationally, although carrier screening has been extensively studied, it is still largely restricted to patients with family history or partner history [6]. In

Implications

For practitioners: Assessment of patient attitudes will help patients feel more empowered and increase the actual use of genetic carrier testing among patients.

For policymakers: Patient education and community outreach will benefit the welfare of this population and impact genetic carrier testing use.

For researchers: There is a pressing need for reliable and valid instruments to ensure comprehensive measurement of attitudes regarding preconception and prenatal genetic carrier testing.

the USA, all women are now offered some form of genetic screening or prenatal diagnosis during pregnancy [7]. Cystic fibrosis screening for all women of reproductive age regardless of family history was recommended over a decade ago by the National Institutes of Health [8], the American College of Obstetricians and Gynecologists (ACOG), and the American College of Medical Genetics and Genomics (ACMG) [9]. This screening test for cystic fibrosis correctly identifies a mutation 97.9 % of the time [10]. Conversely, the same test correctly identifies when a mutation is absent 99.4 % of the time [10]. In the USA, universal recessive carrier testing options are becoming readily available to patients for a host of single gene conditions. One such example screens patients for over 100 single gene conditions through a single serum or saliva sample [11].

Current challenges encountered with use of genetic carrier screening include false positives, ambiguous test results, overdiagnosis, and incidental findings [12]. However, carrier screening for recessive genetic diseases, combined with education and counseling, can markedly reduce disease incidence [13]. One example of evidence is the success of screening efforts of the Ashkenazi Jewish population for Tay-Sachs

Fig 1 | Literature search workflow

disease. Since 1970, preconception and prenatal screening in this population has reduced the disease incidence of Tay-Sachs disease in the USA and Canada by greater than 90 % [14].

In terms of translational behavioral medicine, the goal of preconception and prenatal genetic screening is to provide patients with information necessary to make informed decisions about their reproduction [15]. As genetic screening options and genomic advances progress, the need for patient support and continued evaluation of how patients are educated regarding risks and benefits of testing are necessary. To adequately educate and counsel these patients, the healthcare provider must understand the patients' educational and psychosocial needs in response to these testing options.

In the face of advancing screening technology, in order to meet patients' psychosocial and educational needs, it is beneficial for healthcare providers to be aware of their patients' attitudes towards testing. Accurate assessment of patient attitudes may inform acceptance and uptake of carrier screening. Additionally, identifying patient attitudes of recessive carrier testing will inform evaluation of the clinical utility of carrier testing and the scientific impact attitudes have on reproductive outcomes. While patient attitudes regarding expanded carrier panels have not been examined, there are instruments present in the literature addressing attitudes towards single recessive conditions such as cystic fibrosis and hemoglobinopathies [16]. In order to accurately assess patient attitudes of recessive carrier testing, reliable and valid instruments must be utilized.

PURPOSE

The purpose of this integrative review is to evaluate the availability of quantitative survey instruments to measure patient attitudes regarding genetic carrier testing prior to a conceived pregnancy or during the

prenatal period. The definition chosen for attitude during this review is from the Oxford Dictionary [17] and can be described as "a settled way of thinking or feeling about someone or something, typically one that is reflected in a person's behavior." This broad definition of attitude fits well with the current dearth of evidence regarding preconception and prenatal patients' attitudes about recessive carrier screening; assessment of these attitudes is needed if further steps for translational behavioral medicine facilitation are going to occur. This definition of the concept is well aligned with Ajzen's [18] theory of planned behavior (TPB). For example, the TPB contends attitudes, in part, explain intention and intention explains behavior [18].

TPB has been widely implemented among multiple health behaviors including cancer screening behaviors [19] and genetic screening in the adult population [20]. The TPB contends that a behavior's most contiguous antecedent is the intention to engage in that behavior. Ajzen [18] explains that intention is shaped by three factors: attitudes towards the behavior, subjective norms, and perceived behavioral control.

METHODS

Study design—The methods of this integrative review of the current literature were based on the work of Whittemore and Knafl [21] involving the summary of empirical and theoretical literature to allow for comprehensive understanding of a healthcare issue. This approach was selected due to its utility in incorporating evidence from studies that emerge from diverse methodologies [21, 22]. This systematic approach fosters methodological rigor through problem identification, literature search, data evaluation, and data analysis [21, 22]. The approach involved comprehensively searching the literature to identify studies that focused on patient attitude regarding genetic screening prior to conceiving a pregnancy or during the prenatal period.

Table 1 | Description of identified studies/instruments ($n = 8$)

Instrument/ reference and level of evidence	Theoretical framework	Sample/subjects	Instrument description/ method of measurement	Scoring	Reliability	Validity
Hegwer et al., 2006 3b	Extended Parallel Process Model	$N = 180$ preconceptional Ashkenazi Jewish adult attendees of free education program on Jewish Genetic Diseases	2-part pencil/paper questionnaire evaluating demographics, previous testing status, level of concern/perceived risk 28-item pretest, 19-item post test –5-point Likert scale, true/false	Pretest: demographics, yes/no, 5-point questions, true/false, post test: yes/no, 5-point questions, true/false, knowledge base total/percent correct	N/A	N/A
Ioannou et al., 2010 3b	N/A	$N = 112$ planning a pregnancy or currently pregnant participating in community-based CF carrier screening program (identified carriers and random sample of non- carriers)	Purpose-designed questionnaire exploring: knowledge, recollection/meaning of carrier status, reasons for screening, anxiety, communication of results to family members. 5-point Likert scale, true, false, 3-point answer categories, yes/ no, multiple responses	Reasons for having screening: 5- point Likert scale; knowledge/ carrier status not described; knowledge of CF: true/false/ unsure; anxiety: score 20–70; attitude/screening: yes/no/ unsure; attitudes to family communication: yes/no, number of responses	N/A: anxiety scale (STAI) correlation coefficient $\alpha = 0.82$ (Marteau & Becker, 1992)	N/A; knowledge questions from validated survey used in CF and Tay- Sachs (Gason et al., 2005) (unable to locate original instrument) anxiety: STAI validated in pregnant population with abnormal MSAFP test (Marteau & Becker, 1992)
Lakeman et al., 2009 2b	Theory of planned behavior	$N = 418$ multi-ethnic participants with partner and planning pregnancy	37-item pencil and paper questionnaire evaluating: attitude, social influence, perceived behavioral control, perceived benefits, reproductive consequences of test results, severity of disorders, perceived susceptibility, psychological impact, stigmatization, feelings of eugenics –5-point Likert scale	1 = negative, 5 = positive: psychological impact, stigmatization, feelings of eugenics 1 = positive, 5 = negative	Internal consistency: Cronbach's $\alpha \geq 0.60$ for all scales and 0.85 for attitude scale	Factor analysis with direct oblimin rotation utilized for scales
N/A						N/A

Maxwell et al., 2011 3b	<i>N</i> = 149 members of the Cystic Fibrosis Association of Australia with personal/family CF history	28 items regarding perceived value, risks/benefits/acceptability of prenatal and preconception screening, 7 items regarding acceptability of termination of pregnancy. –5-point Likert scale	Likert statements reclassified into binary variable: score = 0–14	Internal consistency: prenatal scale Cronbach's α = 0.86, preconception scale Cronbach's α = 0.85
Poppelaars et al., 2004 2b	<i>N</i> = 380 stratified random sample of recently married couples	33-item quantitative questionnaire with scales and single items –5-point Likert scale, yes/no	Individual score for each scale, divided by number of items, ranging from 1–5.	Barriers: α = 0.77, consequences of test results: α = 0.77, discrimination: α = 0.62, susceptibility: α = 0.83, decision making behavior: α = 0.60, preconception behavior: α = 0.61, social influences: α = 0.79
Van Elderen et al., 2010 3b	<i>N</i> = 109 Turkish female immigrants planning to become pregnant	Questionnaire guided interview-measured intention to participate in hemoglobinopathy screening, measures to avoid having affected offspring, sociodemographics, psychological characteristics, knowledge.	Scoring method not described beyond variables dichotomized	N/A
Weinreich et al., 2009 2b	<i>N</i> = 264: 80 general public members, 184 general practitioners/midwives	Structured questionnaire measured knowledge, attitude, social norm, perceived behavioral control, intention to screen. –5-point scale except intention (agree/disagree)	Awareness and understanding: correct/incorrect; attitude, social norm, perceived behavioral control: 5 = positive, 1 = negative; intention: agree/disagree.	Internal consistency: information re carrier status testing α = 0.8; participation in carrier testing α = 0.8; social norm scale α = 0.8; perceived behavioral control α = 0.5, 1 factor extracted; burden of being a carrier α = 0.8; equal access to care α = 0.7
Widayanti 2011 2b	<i>N</i> = 180 Javanese mothers >20 years old	17-item quantitative questionnaire –5-point Likert scale, yes/no/maybe/don't know, yes/no	Attitudes towards carrier testing/receiving information, social norms, perceived behavioral control, perceived stigmatization: 5-point scale; Thalassaemia awareness: 2 = high, 0 = none; future reproductive planning = yes/no	Attitudes re thalassaemia carrier testing α = 0.8, social norms re thalassaemia carrier testing α = 0.9

Exploratory

factor analysis by unweighted least squares and oblimin rotation

Factor analysis using oblimin rotation

The title and abstract of each search result were screened to further identify original scientific research utilizing quantitative measurement of patient attitude regarding genetic carrier screening prior to conceiving a pregnancy or during the prenatal period. If it was not evident from the title whether quantitative measurement was utilized, the full manuscript was obtained and read by the authors to determine relevance to the aim of the review. Studies were included if they were peer-reviewed, written in English, utilized a quantitative measurement instrument to examine patient attitude regarding genetic carrier screening prior to a conceived pregnancy or during the prenatal period, and published between 2003 and 2013. This time frame was included to reflect current science due to how rapidly evidence and technology change in the area of genetics. Studies were excluded if they were reviews, were not original research, or were conducted with a pediatric population. The authors evaluated each study based on the screening and inclusion criteria to establish inter-rater reliability. The level of agreement was 100 %.

Search methods—A comprehensive database search of research studies from January 2003 to the present was conducted in PubMed and MEDLINE (databases of the US National Library of Medicine), Cumulative Index to Nursing and Allied Health Literature (CINAHL), PsycINFO (database of the American Psychological Association), and Nursing Reference Center using the following search terms: preconception, prenatal, diagnosis, genetic testing, patient, and attitude.

Search outcomes—The search produced 344 results, which were screened to determine inclusion according to review criteria. A total of eight studies (eight instruments) were included in the review. This sample size is representative of the dearth of information regarding quantitative measurement of patient attitude of genetic screening prior to a conceived pregnancy or during the prenatal period. Figure 1 summarizes the literature search process.

Quality appraisal—Included studies were appraised according to Oxford Centre for Evidence-Based Medicine Levels of Evidence (CEBM) (<http://www.cebm.net/index.aspx?o=1025> accessed 6/4/13). Individual study results were included in Table 1. Collectively, according to the CEBM, consistent findings of level two or three studies would command a grade of “B” for the included research findings.

Data abstraction—Important characteristics of the included studies are provided in Table 1. Data abstraction comprised the following categories: theoretical framework, sample/subjects, instrument description, scoring, method of measurement, reliability, validity, feasibility, level of evidence, and outcome/findings.

Synthesis—Data from the eight included instruments were analyzed and compared according to the abstraction categories. Although data pertaining to the outcome and findings of the included studies were abstracted, this category was not included in the synthesis

as the focus of this review is the instrumentation used to measure attitudes. Table 1 summarizes the pertinent abstracted data.

RESULTS

Included studies were conducted in the Netherlands (50 %), Australia (25 %), Indonesia (12.5 %), and the USA (12.5 %). Half (50 %) of the included studies incorporated the theory of planned behavior as a theoretical framework. One study utilized the Extended Parallel Process Model, while the remaining three studies (38 %) did not identify a theoretical framework.

All instruments were administered to samples that had not yet conceived a pregnancy [23–30]. Sample sizes ranged from $n=109$ to $n=418$ in the included studies. Seven instruments (87.5 %) were self-administered questionnaires while one (12.5 %) was a structured interview based on a questionnaire. Six of the instruments (75 %) were traditional paper questionnaires and one utilized an online survey format (12.5 %). Six instruments (75 %) were administered in a single occurrence. Two instruments (25 %) were administered using a pre/post-test methodology.

Seven instruments (87.5 %) utilized a Likert-type scale to evaluate scores. Other methods of scoring involved dichotomizing responses, combining responses into categories, and utilizing multiple response answers with points allotted for each response. Scoring and measurement characteristics of one instrument were not described. Psychometric evaluation of the included instruments was limited. Only five (62.5 %) of the instruments measured reliability; all five reported Cronbach's $\alpha > 0.70$. One study did not conduct reliability testing on the instrument, but rather discussed the internal consistency of the tool on which this instrument was based. Two studies did not report any reliability data pertaining to their instrument.

Instrument validity was measured in three (37.5 %) of the eight instruments and was completed via factor analysis. One study described factor analysis with direct oblimin rotation performed for each scale [26]. This particular instrument included 10 subscales; however, the authors provided no further discussion of validity assessment [26]. The authors reported 29 % of the variance in intention to participate in currently offered carrier testing was explained by the three subscales pertaining to the TPB (attitude, perceived behavioral control, and social influence) [26]. Another study reported the instrument had been pretested and developed by an expert panel [23], but lacked further description including whether a content validity index was a component in content validity assessment. An additional study was based on a validated survey but not tested for validity, and three (37.5 %) instruments were not evaluated for validity.

Out of the eight studies, only three (37.5 %) of the instruments were evaluated for both reliability and validity. The level of evidence of the included studies was evenly divided between level two (50 %) and level three (50 %), thus, dictating a grade of B for the reviewed literature according to the CEBM criteria.

DISCUSSION

All of the instruments were administered to participants before they conceived a pregnancy [23–30], which is clinically preferential given the preventive care opportunities with genetic screening during this time frame. The authors hypothesize that this is largely due to the benefit of conducting genetic carrier screening prior to conceiving a pregnancy. The authors acknowledge given the realistic prevalence of genetic screening in the prenatal setting, patient attitude regarding genetic testing in this population is of equal concern and needs to be evaluated as well. The authors found the absence of research involving measurement of patient attitudes in the prenatal population disappointing. As this is the first integrative review to examine measurement of this construct in participants who are planning a pregnancy or are pregnant, there are no available reviews for comparison of findings. According to the TPB, attitude greatly impacts intention to participate in a behavior [18], and therefore, directly impacts the clinical utility of genetic testing in individuals who are pregnant or planning a pregnancy. Therefore, the accurate evaluation of the construct of attitudes in this population is extremely important in terms of behavioral translation.

Existing psychometric testing among the reviewed instruments reduces our confidence of the instruments' ability to accurately measure attitudes. A lack of validity impacts the assurance that the instrument is measuring the actual construct. Many (62.5 %) of the included instruments were tested for internal consistency reliability, meaning they had been tested for consistency across multiple items assessed by the tool [31]. However, without demonstrated validity, it cannot be concluded that the instruments were indeed consistently measuring the intended construct. Further, more detailed validity assessment such as confirmatory factor analysis for the three studies including exploratory factor analysis could strengthen confidence in the validity of these instruments. Additionally, thorough validity and reliability assessments of the remaining tools would improve their ability to accurately measure the construct.

Compliance with ethical standards:

Conflict of interest: The authors declare that they have no competing interests.

Declaration of original work: The authors declare that the work described has not been published and is not under consideration for publication elsewhere.

Declaration of overlapping reports: The authors deny any previous reporting of this data.

No animal or human studies were carried out by the authors for this article.

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