Article type : Original Article

Assessing the acceptability, feasibility, and usefulness of a psychosocial screening tool to patients and clinicians in a clinical genetics service in Australia

Katrina Monohan*,[1,2] Rebecca Purvis*,[1,2] Adrienne Sexton,[1,2,3] Maira Kentwell,[1,2] Monica Thet,[8] Lesley Stafford,[4,5,6] Laura Forrest[1,2,7]

1. Parkville Familial Cancer Centre, Peter MacCallum Cancer Centre, Melbourne, Victoria, Australia

2. Parkville Familial Cancer Centre and Genomic Medicine, Royal Melbourne Hospital, Melbourne, Victoria, Australia

3. Department of Medicine (Royal Melbourne Hospital), The University of Melbourne, Parkville, Victoria, Australia

4. Centre for Women's Mental Health, Royal Women's Hospital, Melbourne, Victoria, Australia

5. Melbourne School of Psychological Sciences, The University of Melbourne, Parkville, Victoria, Australia

6. Department of Psychiatry, The University of Melbourne, Parkville, Victoria, Australia

7. Sir Peter MacCallum Department of Oncology, The University of Melbourne, Parkville, Victoria, Australia

8. Melbourne Medical School, The University of Melbourne, Parkville, Victoria, Australia

Running title: Using a pre-appointment psychosocial screening tool in clinical genetics

Corresponding author:
Dr Laura Forrest
Locked Bag 1, A'Beckett St, Melbourne 3000, Victoria, Australia
T: +61 3 8559 6191
E: laura.forrest@petermac.org

Abstract

Increasing demand for clinical genetics services may impact the resources and quality of genetic counseling, potentially impacting patient outcomes. Using a psychosocial screening tool may aid the provision of genetic counseling by reliably identifying patients' psychosocial needs. The Genetic Psychosocial Risk Instrument (GPRI) is a validated genetics-specific screening tool designed to identify psychological risk factors that predict distress in patients having genetic testing. This questionnaire-based study investigated the perceived acceptability, feasibility, and usefulness of the GPRI in patients and clinicians in routine clinical genetics practice.

From December 2018 to January 2019, 154 patients attending an Australian clinical genetics service were invited to complete a paper-based survey that included the GPRI. The GPRI was scored and provided to the clinician for use in the appointment. In February 2019, clinicians completed an anonymous online survey regarding acceptability, feasibility and usefulness of the GPRI. Descriptive statistics, chi-squared, t-tests and regression analyses were used to analyze the patient data, and descriptive statistics were employed for clinician surveys.

A total of 145 patients participated (94% response rate). The average GPRI score was 46.3 (95% CI 43.6—49.0) with 41% of patients meeting the 50-point threshold indicating high risk for psychological distress. The GPRI was highly acceptable to patients, regardless of their level of psychosocial risk. Fourteen clinicians participated (54% response rate): 85% found the GPRI not too time consuming and

86% believed it improved patient care by identifying patient needs. All were willing to use the GPRI routinely.

Use of the GPRI is highly acceptable to patients and clinicians in this setting, assisting in identifying patients at risk for distress, prompting clinicians to address concerns, provide psychosocial support and consider ongoing referral. As 41% of patients' scores indicated a high risk of distress, the GPRI is an important tool for potentially enhancing overall patient outcomes.

Key words: screening tool; distress; acceptability; feasibility; genetic counseling

What is known about this topic:

The increasing demand for genetic testing and counseling has engendered research into the creation of genetics-specific screening tools to better streamline practice. These tools need to be piloted and evaluated in routine clinical genetics practice to assess their implementability.

What this paper adds to the topic:

This paper presents the first study to examine the implementation of the GPRI in clinical genetics practice in Australia and demonstrates that this tool is acceptable to patients and clinicians, feasible to implement on a routine basis, and useful to the clinicians. This study contributes evidence toward the potential of screening tools to improve provision of genetic healthcare.

Introduction

Germline genetic testing has become an integral component in prevention, diagnosis, and treatment for many diseases and conditions. This has resulted in rapidly escalating demand for clinical genetics services and implementation of new models to facilitate genetic testing (James, Mitchell, Bogwitz, & Lindeman, 2013; Kentwell et al., 2017; Stark et al., 2019). Genetic counseling to facilitate the genetic testing process has traditionally been provided by genetic counselors and clinical geneticists. In order to meet demand and address the multidisciplinary nature of

genetic conditions, many genetics services have increased the diversity of medical specialties represented in the clinic, such as oncology, neurology, and cardiology. While service delivery has been evolving, the focus has been on the practical implementation and diagnostic outcomes of genetic testing (Beard, Monohan, Cicciarelli, & James, 2021). However, one of the central tenets of genetic counseling, delivery of psychosocial care (Veach, Bartels, & Leroy, 2007), has rarely been examined when reporting new service delivery models.

The process of genetic counseling seeks to support individuals and their families prepare for, and navigate, the personal and familial complexities inherent in a genetic test result. These complexities can impact patients' emotional wellbeing causing an increased risk of distress. Quality, patient-centered genetic counseling is effective at addressing this risk. Genetic counseling also facilitates informed decision-making about genetic testing, as well as patient adaptation and empowerment by promoting hope and control after receiving genetic test results (McAllister, Dunn, & Todd, 2011; McAllister et al., 2008). In order to achieve these outcomes, the clinician providing genetic counseling must have expertise and the appropriate skills to accurately identify the patient needs (Patch & Middleton, 2018).

Increased demand for genetic testing, genetic counseling workforce shortages, and access barriers to clinical genetics services have resulted in the need to consider rationalizing service provision and implement new service delivery models that facilitate genetic testing (Nisselle et al., 2019; Tutty et al., 2019). However, any innovative approaches must not compromise the provision of quality genetic counseling as this process has been demonstrated to mitigate suboptimal outcomes of genetic testing and maximize the potential for patient benefits (Patch & Middleton, 2018). Instead, transformative and streamlined approaches to genetic counseling practice are required to ensure patient-centered care prevails (Bowdin et al., 2016; Epstein & Street, 2011).

The implementation of a patient screening tool for use during genetic counseling would reliably identify patients' needs and offers the potential to safeguard the quality of care despite increasing demands upon service delivery. The Genetic Psychosocial Risk Instrument (GPRI) is a 20-item validated screening instrument designed to identify psychological risk factors that predict distress in adult patients

undergoing genetic testing (Esplen et al., 2013). The GPRI demonstrates high reliability and predictive value, where the cut off score of 50 identified 84% of patients who displayed distress after receiving genetic testing results (Esplen et al., 2013). By using the GPRI to reliably identify patients at higher risk for distress, genetic counseling can be tailored directly to patient needs, resulting in quality service provision that incorporates a personalized experience and patient-centered care regardless of the discipline of the health care provider or the service stressors at hand.

Despite the rigorous development and validation of the GPRI making this an ideal tool for identifying patients who are at risk of distress, there is a lack of data on the outcomes of implementing the GPRI routinely into clinical genetics practice. Therefore, the primary aim of this study is to assess the implementation of the GPRI in a clinical genetics service by examining the acceptability, feasibility, and usefulness of this tool to patients and clinicians (Proctor et al., 2011). We also wished to explore whether patients' risk of distress would impact their perceptions of acceptability and feasibility of completing the GPRI prior to their appointment. Therefore, the secondary aim of this study is to compare the acceptability and feasibility of completing the GPRI to patients who are and are not at risk of distress based on their GPRI score. This study is the first stage of a hybrid type 2 effectiveness-implementation trial that ultimately aims to determine whether using the GPRI improves patient outcomes after genetic counseling.

Methods

Study design

This study involved two stages to assess the acceptability, feasibility, and usefulness of implementing the psychosocial screening tool in a clinical genetics service using: 1) a paper-based patient survey, and 2) an online clinician survey. Multisite ethics approval was granted from the Peter MacCallum Cancer Centre Human Research Ethics Committee (protocol reference: HREC/18/PMCC/114).

Setting

Patients were recruited between December 2018 and January 2019 from the Parkville Familial Cancer Centre and Genomic Medicine (PFCCGM), a conjoint clinical genetics service at the Peter MacCallum Cancer Centre and the Royal Melbourne Hospital, Victoria, Australia. Clinicians were recruited from the same clinical genetics service in February 2019.

The PFCCGM is a multidisciplinary clinic where patients are triaged to see one or two clinicians depending on the nature of the appointment and the availability of staff. For example, a patient may see a genetic counselor alone, a genetic counselor and a medical specialist (clinical geneticist, medical oncologist, neurologist or gastroenterologist), or a medical specialist alone. All appointments are expected to include genetic counseling, regardless of which type of clinician provides the consultation.

Participants

Patients were eligible for recruitment if they were attending a new or review clinical genetics appointment for genetic counseling and testing for hereditary cancer, neurogenetics, or hereditary hemorrhagic telangiectasia. These specific conditions were chosen to ensure both cancer and general genetics disciplines were represented in this study. Potential participants were aged 18 years or older, fluent in written and spoken English, and were not approached if they had a known cognitive impairment.

Clinicians were eligible for recruitment if they had been provided with one or more patient-completed and scored GPRI for use in an appointment during the patient recruitment period. Clinicians included genetic counselors, clinical geneticists, medical oncologists, breast surgeons, neurologists, gastroenterologists, and medical trainees. Clinicians who were members of the research team were excluded.

Instrumentation

The patient survey was paper-based and included demographic questions, the GPRI, and purpose-designed questions about the acceptability of completing the

GPRI (see supplement 1 for patient survey). The demographics section included five categorical variables: gender, relationship status, educational level, whether the patient had children, and language spoken at home (English or other) with an open text response option for other. This section also included one continuous variable, age in years, and an open text response for postcode. Items in the purpose-designed acceptability section were drawn from a survey evaluating a psycho-educational oncology intervention (Stafford et al., 2017). This section included ten items with a 5-point Likert scale response (strongly disagree to strongly agree), with four items about how the completion of the GPRI made patients feel emotionally and how it made them feel about their appointment, a fifth item that was open text for participants to provide an 'other' response, another four items about the experience of completing the GPRI, and two items examining preferences of choice whether or not to complete a psychosocial screening tool. There were also three binary responses items investigating patient preferences regarding location of completing the GPRI.

The clinician survey included demographic questions, and purpose-designed questions about the acceptability, feasibility, and usefulness of using the GPRI in clinical practice (see supplement 2 for clinician survey). The clinician survey was hosted online using REDCap electronic data capture tools hosted at the Peter MacCallum Cancer Centre (Harris et al., 2019; Harris et al., 2009).

Procedures

Patients were approached in the clinical genetics service waiting room by a research assistant (MT) and invited to participate. A recruitment list was used to document whether patients were eligible, and whether eligible patients accepted or declined to receive the participant information sheet and the paper-based survey. Patients were only invited to participate once, when attending either a new or a review appointment. Completion of the survey implied informed consent. The research assistant then scored the completed psychosocial screening tool according to the validated method (Esplen et al., 2013) and gave it to the clinician seeing the patient for their appointment. Recognizing that the GPRI identified 84% of patients at risk of distress in the validation study (Esplen et al., 2013), clinicians were encouraged to incorporate the completed GPRI responses and score into their genetic counseling

practise but use discretion and their clinical expertise to assess patients' needs further, if required.

The survey responses were manually entered into REDCap hosted at the Peter MacCallum Cancer Centre for data management and storage (Harris et al., 2019; Harris et al., 2009).

Once patient recruitment was complete, eligible clinicians were invited by email to complete the online survey anonymously.

Data analysis

Data were analyzed using Stata IC 15 (StataCorp, 2017). An exploratory analysis was undertaken to characterize patient demographics, the acceptability, feasibility, and usefulness of the GPRI, and whether there were independent predictors of acceptability. Given the exploratory approach of this study, there were no a priori hypotheses and subsequent empirical evaluation established.

Patient and clinician demographics were described using numbers and percentages. The location of patient appointments were categorized into clinic type regarding whether they were attending an appointment in the familial cancer center (FCC: for hereditary cancer) or adult genetics (AG: for neurogenetics and hereditary hemorrhagic telangiectasia). The characteristics of patients who completed all the GPRI items (enabling scoring) were compared to patients who did not complete all GPRI items (meaning their GPRI could not be scored) using chi squared tests for categorical variables and t tests for continuous variables. Patients were dichotomized into two groups according to their GPRI score (<50 or ≥50) and demographics were compared between the groups using chi squared tests for categorical variables and t tests for continuous variables.

The GPRI score was calculated for each participant by calculating the sum of the raw scores for each item, where the total score ranged from 10 to 100 (Esplen et al., 2013).

The 5-point Likert scales for the questions about patients' acceptability of completing the GPRI were collapsed into three categories: disagree (0), neither (1), or agree (2)

and analyzed with disagree as the reference category. Only patients' responses who had completed all GPRI items were included in this analysis. Pearson's chi squared tests were used to compare the responses in these three categories by the two patient groups according to GPRI score (<50 or ≥50). Ordinal logistic regression was used to examine relationships between patient demographics (including clinic type, appointment type, dichotomized GPRI score, gender, age and whether or not they had children) and patients' responses to the acceptability of completing the GPRI, with $p<0.05$ considered statistically significant.

Clinician responses were described using numbers and percentages. Content analysis was used to where a member of the research team (LF) inductively coded open text responses to identify meaningful content and group common responses from the clinician survey.

Results

Patient characteristics

One hundred and forty-five patients (94% response rate) agreed to participate with 116 (80%) completing all items on the GPRI. Most participants were female (63%), attending an FCC appointment (74%) and attending for the first time (60%) (Table 1). No statistical differences were observed when comparing the clinic and appointment type, gender or age for patients who did and did not complete all GPRI items. However, patients who completed all items of the GPRI were more likely to have a tertiary education than those who did not complete all items $\chi^2(1, n=144) = 4.95$, $p= 0.026$.

Patient GPRI responses

The average GPRI score was 46.3 (95% CI 43.6–49.0) with scores ranging from 15 to 86 (Table 2). Cronbach's alpha for the GPRI was 0.80, suggesting good internal consistency. A total of 69 patients (59%) scored less than 50 indicating they were not at risk of distress, and 47 (41%) scored 50 or above indicating they were at risk of distress. When completing the section about personal mental health history, 25% of patients reported a history of anxiety and depression and 14% reported a history of

suicidal ideation. Three times more women scored 50 or above indicating they were at risk of distress compared with men (OR 3.1, 95% CI 1.2—7.5, p=0.02).

Patient acceptability and feasibility

Overall, most patients disagreed that completing the GPRI made them feel upset (70%) or worried about their appointment (55%) (Table 3). Most agreed that completing the GPRI reassured them that the healthcare professional taking their appointment would have a better understanding of their concerns (62%). Further, most agreed that the GPRI was relevant to them (66%) and the majority agreed the GPRI was easy to understand (93%). The majority reported that the GPRI was not too difficult (85%) or took too long (86%) to complete. Finally, most patients disagreed in response to whether they would prefer not to answer any questions about their thoughts and feelings about their genetics appointment (70%).

A greater proportion of patients whose GPRI score was less than 50 disagreed that completing the GPRI made them feel upset compared to patients whose GPRI score was 50 or greater $\chi^2(2, n=112) = 6.18$, p=0.045. Furthermore, a greater proportion of patients whose score was less than 50 were ambivalent about whether completing the GPRI made them feel good compared to patients whose GPRI score was 50 or greater $\chi^2(2, n=113) = 7.62$, p=0.02.

After controlling for demographics, clinic and appointment type, patients who were older (OR 1.04, p=0.02, 95% CI 1.0—1.01), and those whose GPRI score was 50 or greater compared to those whose GPRI score was less than 50 (OR 2.7, p=0.03, 95% CI 1.1—6.3) were more likely to agree that completing the GPRI reassured them that the healthcare professional would have a better understanding of their concerns (p=0.03). There were no other predictors identified for the other acceptability and feasibility items.

Clinician characteristics

Of the 26 clinicians who counseled patients who had completed the GPRI, 14 (54%) completed the online survey. Participants included nine (64%) genetic counselors, two (14%) medical specialists and three (21%) medical genetics trainees. Most (71%) clinicians had less than five years' experience working in a genetics service

and 62% reported seeing the GPRI up to five times during the patient recruitment period. All (100%) stated that they used the completed GPRI tool and GPRI score for every appointment it was provided.

Clinician acceptability, feasibility, and usefulness

Overall, clinicians reported that reviewing the GPRI was not too time consuming (85%) or difficult to interpret (79%). However, eight (57%) clinicians reported that patients completing the GPRI delayed the start of the appointment. As one genetic counselor noted;

> *"(The) screening tool took up to 20 minutes to complete/assess and so I had less time with the patient. Would be useful to be completed prior to the appointment."*
>
> *Participant 2 - Genetic counselor (0-5 years' experience)*

Most believed that using the GPRI helped them to identify patient needs (86%), improved communication with patients (57%) and improved patient care (85%). This improvement of patient care was experienced in different ways including making it easier to identify the patient's key concerns, as another genetic counselor said;

> *"(The GPRI) did sometimes result in a delay but actually may have decreased time spent trying to work out what concerns were in the appointment"*
>
> *Participant 14 – Genetic counselor (0-5 years' experience)*

Another benefit was in prompting questions around patient concerns that the clinician had not prepared for the appointment, with this clinician stating;

> *"I only used the tool if there was a particular flag that I would not normally address"*
>
> *Participant 13 - Genetic counselor (0-5 years' experience)*

The GPRI also facilitated the patients' exploration of their concerns or distress in the appointment, as described by this clinician;

> *"It primes patients to expect to talk about psychosocial issues"*

Participant 12 – Medical Specialist (>10 years' experience)

All clinicians (100%) who completed the feedback questionnaire expressed that they would be willing to use the GPRI as part of routine care. The more experienced clinicians (>10 years' experience, n=3) were more neutral, reporting the tool made no difference to patient care.

Discussion

This study is the first to examine the acceptability, feasibility and usefulness of the GPRI within a multidisciplinary clinical genetics service in Australia. The findings demonstrate that patients found completing the GPRI acceptable and that it was feasible to ask patients to complete the GPRI in the waiting room prior to their genetics appointment. Additionally, clinicians felt that the tool was acceptable and useful during an appointment. Exploring the acceptability and feasibility of the GPRI to patients and clinicians was a vital first step in considering whether it is implementable; for example, discovering barriers to use, such as unacceptability to patients or unreasonable demands on clinicians' time or resources, would have prompted a reconsideration. Instead, the findings indicate that this tool is suitable for consideration of routine use in clinical genetics services. Furthermore, the GPRI reliably identified a sizeable proportion of the patients (41%) who were at risk of distress, further demonstrating the usefulness and need for this tool. While most medical professionals are trained to give bad news and counsel patients in distress, genetic counselors receive a unique combination of training in both medical genetics and psychotherapy in order to provide genetic counseling (Sahhar, Hodgson, & Wake, 2013). One of the advantages of having a multidisciplinary genetics service is that each medical specialist brings a unique strength, for example an oncologist providing insight into the treatment implications of genetic testing or a cardiologist interpreting an electrocardiogram in relation to genetic phenotypes. Yet the process of genetic counseling remains critically important when genetic testing is offered as it is demonstrated to decrease anxiety, depression, decisional conflict and condition-specific worry, and improve positive outcomes including empowerment, adaptation, and perceived personal control (Madlensky et al., 2017; McAllister et al., 2011). In acknowledging the role for genetic counselors in improving patients' psychosocial outcomes (McAllister & Dearing, 2015), using the GPRI could be useful for providing

quality and patient-centered genetic counseling regardless of whether patients are attended by genetic counselors or medical specialists.

While patients were generally accepting of completing the GPRI, the identification that older patients found completing the GPRI more reassuring in terms of feeling that their healthcare professional has a better understanding of their concerns, indicates there may be some added utility of this tool within this specific patient subgroup. In other intervention implementation research in clinical genetics, acceptability has not been commonly examined or reported, with more studies focusing on patient-centeredness or uptake (Roberts, Kennedy, Chambers, & Khoury, 2017). In general, utilization of implementation theories and frameworks, including taxonomies of outcomes, are underutilized in the context of translational research in clinical genetics (Morrow, Chan, Tucker, & Taylor, 2021; Paul, Leslie, Trainer, & Gaff, 2018; Roberts et al., 2017). This paucity of data makes it difficult to contextualize this subgroup finding and reinforces the need for more theory- or framework-driven research to better understand how interventions impact different patients and what determinants influence patient, service and implementation outcomes (Davidoff, Dixon-Woods, Leviton, & Michie, 2015).

This study identified that three times more women than men reported being at risk of distress. There are a number of possibilities relating to this finding, with first being the most obvious that women are in fact more likely to be at higher risk of distress than men. However, this assertion is largely unable to be evidenced by the current state of the genetic counseling literature, given studies are dominated by female participants (Wallgren, Veach, MacFarlane, & LeRoy, 2020). As expounded in a systematic review by Lombardi et al. (2019), men only comprised 6% of the sample used to establish the proportion of patients who were distressed, anxious, or depressed after genetic counseling. Another possibility, is that women may be more open to self-reporting their emotions, while also being more emotionally aware compared to men (Barrett, Lane, Sechrest, & Schwartz, 2000). This leads to the prospect that men are not necessarily less at risk of distress than women during the genetic counseling and testing process, but that they may not be as readily identifiable using a psychosocial screening tool that relies on self-report.

While this study has demonstrated that the GPRI is acceptable and useful for clinicians and patients, further consideration needs to be given to practical issues that impact the feasibility of routine GPRI implementation. The high response rate (94%) for this study is likely the result of active recruitment in the waiting room where eligible patients were approached and invited to this study by a member of the research team (research assistant), which is not reflective of how the tool would be implemented routinely. Other mechanisms of providing the GPRI to patients, as observed in other studies; e.g., mailing Patient Reported Outcome Measures (PROMs) to patients prior to a genetic counselling appointment, have resulted in response rates less than 25%, which was reportedly disheartening for clinical staff (Costal Tirado et al., 2017). Clinicians in our study certainly expressed some concern that the time needed to complete and score the GPRI delayed the appointment. We acknowledge it is not feasible in the long-term to have a dedicated staff member assisting patients to complete screening tools and propose that digitalizing the questionnaire so that patients can complete the tool on their smart phone and scores can be automatically calculated may be one way to streamline the implementation into the clinical service. This need for online data collection and analysis was also noted by Costal Tirado et al. (2017) as a potential mechanism to improve response rates.

Study limitations

The limitations of this study include the low numbers of medical specialists and trainees surveyed and, in general, a lack of clinicians with more than five years of clinical genetics experience. Involvement of other adult genetic clinics (e.g., cardiac and general genetics clinics) may have increased the number of medical specialists and trainees to gain better insight into their perspectives of using the GPRI during an appointment. Also, by increasing the number of more experienced clinicians, we may have been able to explore whether the screening tool is more useful for the more junior members of the multi-disciplinary team. Also, a greater breadth of clinics would have allowed for the testing of the GPRI where there are different psychosocial factors (e.g., sudden cardiac death) implicated in other inherited conditions.

The use of a research assistant in this study to invite patients to complete the GPRI in the waiting room, then score their GPRI responses and provide this to the clinician

for the appointment, is not reflective of how the GPRI might be deployed in routine clinical practice. This method may have increased patient response rate, influenced their perceptions of the acceptability of completing the GPRI, and influenced the clinicians' perceptions of the acceptability and feasibility of using the GPRI as they did not have manually score the patients' responses themselves. An alternative method of administering the GPRI is to ask patients to complete the GPRI online, allowing for automation with scoring and distributing the completed GPRI to the clinician.

The exclusion of acceptability and feasibility data from patients who did not complete the GPRI introduces a further limitation to these findings. A larger proportion of these patients did not have a tertiary education compared to patients who completed all GPRI items. This group of participants may have different perceptions about how acceptable and feasible it was for them to attempt to complete the GPRI.

Practice implications

In comparison to other studies that have identified that up to one quarter of the patient population attending clinical genetics services experience clinically significant levels of anxiety or depression (Eijzenga, Hahn, Aaronson, Kluijt, & Bleiker, 2014; Lombardi et al., 2019), the use of the GPRI in this study identified a greater proportion of patients who were at risk of distress. This larger number of patients identified by the GPRI is likely due to the breadth of items included that are specific to the implications inferred by genetic information, including risk to children and other family members, and the potential impact on personal and familial relationships (Esplen et al., 2013). This further perpetuates the relevance and need for a genetic counseling-specific patient screening tool that ensures regardless of whether patients are counseled by medical specialists or genetic counselors, their needs are identified and met.

There is a responsibility in terms of staffing and infrastructure needed to deal with the corollary of identifying patients in need of psychosocial support and also responding to patients' expectations who have been primed through completion of the GPRI that their psychosocial needs will be addressed. This generation of "psychosocial work" through the implementation of the GPRI may prompt a re-think

about the structure of clinical genetics services and how clinics are delivered to ensure patients identified at increased risk of distress and/or with specific concerns identified within the GPRI items have a genetic counselor attend their appointment, provide a follow up telephone service, and/or have a clear referral pathway in place for psychological support.

This study highlights the prevalence (14%) of past or current suicidal ideation in this cohort, which, although not captured in the clinician survey for this study, may raise concerns amongst clinicians regarding their role or obligation to investigate this in the appointment. However, general population-based studies in Australia and the USA have shown that suicidal ideation occurs commonly for some, with 12-month prevalence rates of 2-6%, and in Australia a lifetime suicidal ideation prevalence of 13% (Crosby, Han, Ortega, Parks, & Gfroerer, 2011; Johnston, Pirkis, & Burgess, 2009). Further training for clinicians to increase confidence in addressing these issues may be required, as asking about suicide does not increase risks and is likely to be beneficial, providing opportunity for psychology/psychiatry referral (Dazzi, Gribble, Wessely, & Fear, 2014; Miller et al., 2017). Studies have shown that even brief interventions, such as a telephone follow-up are beneficial in reducing suicide risk (Stanley et al., 2018). Nevertheless, the use of the GPRI had unexpected benefits of identifying patients' psychological history which might usually be missed due to the clinicians' focus on other psychosocial complexities inherent in genetic counseling.

Research recommendations

We hypothesize that implementing the GPRI routinely into clinical genetics care is likely to positively impact patient outcomes during and after genetic counseling and testing. By reliably identifying patients at high risk of distress and using the completed GPRI to identify areas of patient concern, counseling can be efficiently tailored to address patient needs, resulting in a more personalized patient experience. This may result in improvements in patient empowerment, adaptation, perceived personal control, over and above standard genetic counseling appointments where the GPRI is not used. This study presents baseline data that has been used to support the design for a hybrid type 2 effectiveness-implementation trial. By determining that implementing the GPRI is acceptable,

feasible, and useful in clinical genetics practice, other implementation domains including adoption, fidelity, costs, penetration, and sustainability, can be examined in this larger trial (Landes, McBain, & Curran, 2020). The effectiveness component of this trial includes a randomized control trial to determine whether using the GPRI compared to not using the GPRI improves patient outcomes. The hybrid type 2 trial will be rolled out in the same conjoint clinical genetics service in Australia from 2021-2025.

Conclusion

This study has demonstrated that the GPRI is an acceptable, feasible and useful risk-screening tool within an Australian clinical genetics service. The GPRI reliably identifies patients at high risk of distress, assisting in the provision of timely psychosocial support and, potentially, better patient care. Potential benefits for the routine implementation of the GPRI in clinical genetics practice may include personalizing the genetic counseling session through reliable identification of patient-reported needs, ensuring consistency in genetic counseling practice regardless of experience or the type of clinician providing the genetic counseling, assisting in genetic counseling clinician training to ensure a workforce who are well versed in the psychosocial complexities that are presented by a diverse and nuanced patient cohort. Evidence is now needed to establish whether the simple idea of implementing the GPRI in routine practice will indeed improve patient care beyond usual genetic counseling practice.

Author contributions

All the authors made substantial contributions to the conception and design of the work, and drafting and revising this work for important intellectual content. Monica Thet also made a significant contribution to the data acquisition and Laura Forrest to the analysis and interpretation of the data. Further, Laura Forrest confirms that she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Acknowledgements

The authors would like to thank Emily Higgs for her contribution to the preliminary idea of implementing a psychosocial screening tool into the clinical genetics service. We also thank Paul James for his support as Director of the Parkville Familial Cancer Centre for this research. Dr Laura Forrest was in receipt of a postdoctoral fellowship from the National Breast Cancer Foundation (PF-14-009; 2014—2020), Australia, while this research was conducted. Dr Forrest now holds a mid-career fellowship from the Victorian Cancer Agency (MCRF20012; 2021—2025), Australia, that funds the hybrid type 2 effectiveness-implementation trial to test the use of the GPRI to improve patient outcomes after genetic counseling.

Compliance with ethical standards

1. Conflict of Interest

Katrina Monohan, Rebecca Purvis, Adrienne Sexton, Maira Kentwell, Monica Thet, Lesley Stafford and Laura Forrest declare that they have no conflict of interest.

2. Human Studies and Informed Consent

Approval to conduct research involving human subjects was obtained by the Peter MacCallum Cancer Centre Human Research Ethics Committee. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

3. Animal Studies

No non-human animal studies were carried out by the authors for this article

4. Data Availability Statement

Research data are not shared.

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Table 1 Comparison of characteristics of patients who completed all GPRI items with those who did not in the clinical genetics department at the Peter MacCallum Cancer Centre and the Royal Melbourne Hospital, Australia

Characteristic		Completed GPRI n (%)	Did not complete GPRI n (%)	Total n (%)	p value
Clinic type	FCC	88 (77)	19 (63)	107 (74)	0.14
	AG	28 (23)	10 (37)	38 (26)	
Appointment type	New	73 (63)	14 (47)	87 (60)	0.09
	Review	42 (37)	16 (53)	58 (40)	
Gender	Female	71 (62)	20 (67)	91 (63)	0.62
	Male	44 (38)	10 (33)	54 (37)	
Education	Tertiary	48 (42)	6 (20)	54 (38)	0.03
	Non-tertiary	66 (58)	24 (80)	90 (62)	
Age (years)	mean (95%CI)	45.0 (42.2 - 47.9)	50.3 (44.7 - 55.9)	46.1 (43.6 - 48.6)	0.10

GPRI: Genetic Psychosocial Risk Instrument; FCC: Familial Cancer Centre; AG Adult Genomics.

Table 2 Comparison of participant demographics by GPRI score <50 (not at risk of distress) and ≥50 (at risk of distress) in the clinical genetics department at the Peter MacCallum Cancer Centre and the Royal Melbourne Hospital, Australia

Pair-wise comparisons	Demographic variable	GPRI Mean (95% CI)	p value	GPRI<50 n (%)	GPRI≥50 n (%)	p value
Site	FCC	45.5 (42.6 – 48.4)	0.3	54 (61)	34 (39)	0.5
	AG	48.8 (42.1 –		15 (54)	13 (46)	

		55.4)				
Appointment type	New	46.9 (43.4 – 50.3)	0.6	42 (57)	32 (43)	0.4
	Review	45.3 (40.9 – 49.7)		27 (64)	15 (36)	
Gender	Male	40.5 (36.3 – 44.7)	<0.01*	33 (73)	12 (27)	0.02†
	Female	50.0 (46.7 – 53.2)		36 (51)	35 (49)	
Partnered	Yes	47.1 (43.9 – 50.3)	0.4	45 (57)	34 (43)	0.5
	No	44.8 (39.7 – 49.9)		23 (64)	13 (36)	
Education	Non-tertiary	46.5 (42.7 – 50.3)	0.8	40 (60)	27 (40)	0.9
	Tertiary	45.8 (42.0 – 49.6)		29 (60)	19 (40)	
Children	Yes	47.9 (44.6 – 51.3)	0.1	39 (54)	33 (46)	0.1
	No	43.6 (39.1 – 48.1)		30 (68)	14 (32)	
Age (years)		Range		18 - 80	23 - 76	
		Mean (95% CI)		45 (41 - 49)	46 (42 - 50)	0.8

*$t(114) = -3.58$, $p<0.01$; † $\chi^2(1, n=116) = 5.85$, $p=0.02$

GPRI: Genetic Psychosocial Risk Instrument; FCC: Familial Cancer Centre; AG Adult Genomics.

Table 3. Responses to acceptability and feasibility of completing the GPRI comparing patient groups according to GPRI cut off score

		Disagree (%)	Neither (%)	Agree (%)	p value
Completing the GPRI…					
Made me feel upset (n=112)	**GPRI<50**	52 (77.6)	12 (17.9)	3 (4.5)	0.045
	GPRI≥50	26 (57.8)	12 (26.7)	7 (15.6)	
Made me feel good (n=113)	**GPRI<50**	9 (13.2)	49 (72.1)	10 (14.7)	0.02
	GPRI≥50	15 (33.3)	22 (48.9)	8 (17.8)	
Made me worried about my appointment (n=113)	**GPRI<50**	41 (61.2)	22 (32.8)	4 (6.0)	0.1
	GPRI≥50	21 (45.7)	17 (40.0)	8 (17.4)	
Reassured me the healthcare professional I am seeing today will have a better understanding of my concerns (n=115)	**GPRI<50**	6 (8.7)	24 (34.8)	39 (56.5)	0.2
	GPRI≥50	1 (2.2)	13 (29.3)	32 (69.6)	
Overall, the screening questionnaire:					
Was relevant to me (n=115)	**GPRI<50**	5 (7.3)	22 (31.9)	42 (60.9)	0.2
	GPRI≥50	4 (8.7)	8 (17.4)	34 (73.9)	
Was easy to understand (n=116)	**GPRI<50**	3 (4.4)	2 (2.9)	64 (92.8)	1.0
	GPRI≥50	2 (4.3)	1 (2.1)	44 (93.6)	
Was too difficult to fill in (n=116)	**GPRI<50**	60 (87.0)	4 (5.8)	5 (7.3)	0.4
	GPRI≥50	39 (83.0)	6 (12.8)	2 (4.3)	

Took too long to fill in (n=115)	**GPRI<50**	61 (89.7)	4 (5.9)	3 (4.4)	0.3
	GPRI≥50	38 (80.9)	7 (14.9)	2 (4.3)	
I would prefer to…					
Not answer any questions about my thoughts and feelings about my genetics appointment (n=114)	**GPRI<50**	48 (70.6)	19 (27.9)	1 (1.5)	0.3
	GPRI≥50	32 (69.6)	11 (23.9)	3 (6.5)	
Be offered a choice whether I answer any questions about my thoughts and feelings about my genetics appointment (n=114)	**GPRI<50**	14 (20.3)	29 (42.0)	26 (37.7)	0.5
	GPRI≥50	10 (22.2)	14 (31.1)	21 (46.7)	

GPRI: Genetic Psychosocial Risk Instrument

Supplementary information files

A: Patient survey

B: Clinician survey