Identification of Support Person Needs During Pre-Symptomatic Genetic Testing for Adult-Onset Diseases

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RESEARCH

Please cite this paper as Conaghan, J; Clinch, T; Howat, A; Crawford, A; Richards, F, Kapp, R; Fleming, J; Barlow-Stewart, K. Identification of support person needs during pre-symptomatic genetic testing for adult-onset diseases. Archives of Healthcare. [2021] 2(1):1-10.

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ABSTRACT

Objective: Pre-symptomatic genetic testing (PST) allows at-risk individuals to confirm whether they have inherited the disease-causing mutation. However, little is known about the experience of those who support consultands through testing. Using Huntington disease (HD) as a model of inherited adult-onset conditions, we have explored the experience of individuals who support consultands through the testing process.

Methods: Telephone interviews were conducted with 14 consultands who undertook PST for HD. Inductive analysis identified support persons' role, experience, needs and follow up as important to consumers. Subsequent online surveys were piloted with 35 consultands and 18 support persons who had undertaken PST.

Results: Our findings demonstrate that consultands' and support persons' experience of PST was mostly positive. Most consultands appreciated the support a significant other was able to provide, and support persons felt they had an important role to play. However, support persons often felt ill-equipped for the role, and some were unprepared for the distress they experienced.

Key Words: Predictive genetic testing; support persons; genetic counseling; service delivery models; genetics services.

INTRODUCTION

Pre-symptomatic genetic testing (PST) for adult onset inherited genetic conditions such as Huntington disease (HD) has enabled at-risk individuals to confirm their genetic status (1, 2). The process of PST aims to create a framework that enables the consultand to discuss the context and rationale for their current thinking about whether or not to undertake PST; to make an informed and independent decision; to provide support to help facilitate adjustment to the result; and to assist the development of subsequent sources of support. The core aspects of PST involve the establishment of a respectful working alliance between the consultand and counselor, and the provision of specific verbal and written information to enhance

knowledge and facilitate the decision-making process (3, 4). As part of both generic and disease-specific guidelines for the provision of PST, individuals at risk for inherited adult-onset disease are also encouraged to bring a support person. In this study, we have focused on the experience of consultands and their support persons using HD as an example of consumer experience of the PST process.

Consultands' feedback on the current guidelines for PST highlight the importance of access to the well-developed counseling skills of those delivering the service, and a flexible approach adjusted to meet the needs of the individual (3-9). Given consultands and their partners are both reported to experience psychological problems in adjusting to the test outcome this would support the implementation of a more tailored approach to take into account the impact on both parties (8, 10). Indeed, Williams et al. (2000) reported on the psychological impact of PST on support persons and found that, while being committed to the role, support persons also found this responsibility difficult and emotionally demanding, and for some the role evolved into subsequent caregiving. Support persons also expressed a need for information and had forthright views about the need for a more flexible testing protocol. These authors therefore recommended increased attention to the needs of support persons.

Ascertaining the views of consumers is seen as an integral aspect of improving the overall quality of patient-centred health care, resulting in services which are better designed to meet consumer needs and achieve better health outcomes (11). In line with this approach, obtaining the views of those who have undertaken PST will provide an insight into their experience, ensuring consumers are an integral part of the evaluation and quality improvement process for genetic services. Relatively little attention has been paid to the perspective and experience of support persons of the testing process or their views on the quality of care (4-6, 12-16). Therefore, the aims of this study were to: (1) enable consultands and their support persons to describe the experience and expectations of PST; (2) identify what support persons considered important in facilitating the PST process; and (3) propose recommendations that may improve clinical practice.

MATERIALS AND METHODS

A pilot qualitative exploration at a single site guided the questions for a quantitative nationwide survey.

Participants and recruitment

Consultands - individuals who had had testing 2007-2012 - were recruited from a specialist PST service for HD in Newcastle, Australia having been purposively identified from the medical records (JC). Sixty-one potential consultands were sent an invitation to participate in a telephone interview, with an information statement and consent form. If a response was not received, a second invitation was sent. Consultands who completed an interview are hereafter referred to as C1s.

In 2015, consultands and support persons were recruited via the Australian state-based HD support organizations. The support person was defined as a spouse, partner, friend, or trusted individual who accompanied a consultand to pre-test counseling and/or result giving sessions. Those who wished to enrol were required to have undergone PST or had supported an individual through testing at an Australian genetics service (2009-2014). The exclusion criteria included: being under 18 years; likely to experience distress as a result of participation; being non-English speaking; having developed symptoms of HD since receiving the test result; and an inability to complete a survey in English. Hereafter, consultands who completed a survey are referred to as C2s; support person are referred to as SPs. For those completing a survey, the participants were notified in the participant information document that completion and submission of an anonymous survey would be taken as their consent for use of their data in the research study as approved by Hunter New England Human Research Ethics Committee (HREC).

Instrumentation

The interview guide was developed by three investigators (AH, JC and KB-S), based on the 1994

international guidelines (17). Questions focused on the C1s' or their reports of the support person's experience and perception of the PST process (Supplementary Information File 1).

Two surveys were developed by TC and KB-S: one for completion by C2s and another for SPs who had attended an Australian genetics service for PST. The questions were informed by the six themes derived from thematic analysis of the qualitative interviews described above. The survey for C2s contained 50 items; the SP survey contained 33 items (Supplementary Information File 2 and Supplementary Information File 3 respectively) including 13 and 9 free text boxes respectively. To maintain C2's confidentiality and privacy, responses from the C2s and their SP were not linked.

Data collection

Those who provided written consent were contacted to arrange a suitable time for a semi-structured telephone interview (AH). Interviews were recorded, transcribed verbatim and de-identified. A code list was developed and refined by three coders (JC, AH and KB-S) and updated using an iterative approach.

For the quantitative arm of the study, members of HD support organizations in Australia were emailed an invitation to participate in a survey, with a follow-up email sent two months later. An invitation and participant information statement including a link to the online surveys (hosted on Survey Monkey®) were also placed in two quarterly HD New South Wales (NSW) newsletters. Hard copies could be requested from the support organizations. A 'snowballing' recruitment strategy was adopted, where on completion of the survey, C2s were asked to forward the invitation email to any family members who had undergone testing or had supported someone through testing. Consent was implied by completion and return of the survey.

Data analysis

Interviews

All coders analyzed three transcripts (inter-coder reliability of > 90% concordance); AH coded the remainder of the transcripts. The data was then analyzed thematically by AH and JC using an inductive approach (18).

Surveys

All online survey responses were downloaded from Survey Monkey® to SPSS Statistics for Windows, Version 22.0 (Armonk, NY: IBM Corp). Descriptive statistics were calculated for Likert scale and tick box responses. Where data were missing, respondents' surveys were still included in the dataset according to the denominator. Quotes from those participants in the interview study are identified as P1; P2 denotes quotes from participants in the survey study.

Hunter New England Human Research Ethics Committee (HREC) and the University of Sydney HREC provided ethics approval for the study.

RESULTS

Sixteen (16/61) individuals who had undertaken PST (C1s) agreed to participate in a single telephone interview. Two withdrew, leaving a final response rate of 14/61 C1s (23%). In total, three were male, half of the participants were married, half had offspring, they all lived in NSW and approximately two thirds had a negative result (see Table 1).

Exploration of consultand experience led to the identification of six themes: support person role and experience, access to services, perspectives of the testing process, information and consultand-centered care, the afterwards, and resources. A number of themes (access to services, perspectives of the testing process, information and consultand-centered care and resources) reflected findings consistent with previous studies. All the C1s interviewed, who attended a specialist PST service for HD in Australia, reported that they were seen promptly, often within a couple of weeks. They indicated that all their needs for information and support were met at the time of testing and felt that their autonomy was respected. They perceived the care as client-centered, with the need for flexibility accommodated in the timing of blood collection and receiving a result. A positive consultand–counselor relationship and the perceived high levels of skill and

competence of the staff favourably influenced the C1s' perceptions of the PST process:

"I've had a fantastic experience from the moment that I walked through the door…I've felt respected, I've felt sensitivity was projected towards the whole process, they were all very caring, care was taken through the whole process, and in the way they presented information." [P1#3, C1, positive result].

In terms of support person role and experience, almost half of C1s (6/14, 43%) brought a SP to one or more appointments. Most C1s were in a relationship and reported that they chose to involve their partner as SP, stating that the test result would significantly impact upon their partner. In a few cases, other family members acted as support persons. Three brought their SP to all appointments; one for the second and third appointments, and two had a SP present for the result session only. Those who involved their SP from the initial session reported this was beneficial:

"I think that was what turned it all around for both of us, really…We both thought that the way she could support me was because of that first session really…she would talk me through certain things…That was very positive for both of us, definitely." [P1#7, C1, negative result].

The C1s who involved their support person from the first counseling session did not describe a negative impact of the result on their support person. In contrast, the two C1s who brought their SP only to the result-giving session (both received a positive result) described their partner being shocked and/or overwhelmed by the result:

"I did go with my husband to the result and for some reason I'd just already steeled myself for it…My husband, he cried, poor thing…I think because he hadn't been part of that introductory thing…He was the odd one out…You know because he was walking into a room full of strangers, I wasn't. We'd already established that relationship…I should've involved him more." [P1#3, C1, positive result].

C1s who did not have a partner often did not bring a support person, despite this being recommended, as they did not feel it was necessary.

To further investigate consumer experiences related to support person needs the survey-based arm of the study was undertaken. A total of 53 participants were enrolled: 35 C2s and 18 SPs who had attended genetics services in four states (see Table 1). It was not possible to calculate the response rate for surveys. Overall, the majority of consultands were < 55 years old, whereas the support people tended to be older > 55 years. As in the interview study most participants were female and were located in two states (NSW or Tasmania). The majority of C2s (29/31, 94%) brought a SP to one or more of their appointments. Similar to C1s this was most frequently their partner (21/29, 72%). Most reported that having the SP attend a pre-test appointment was beneficial for themselves (18/27, 67%) and the SP (22/27, 81%); and attendance at the result-giving session was especially reported to be beneficial by both C2s (5/28, 89%) and for SPs (28/28, 100%).

"I wanted to go on my own, but my counselor advised me to take someone. Very glad I did." [P2 #27, C2, negative result].

The median ratings of the service overall by C2s and their SPs were 'excellent' and 'very good' respectively with no significant difference found in the ratings between all scores (Mann–Whitney U z score 1.89, P = 0.588). However, 5/29 (17%) C2s and 5/15 (33%) SPs rated the service as 'fair – poor' (see Figure 1). Indeed, one SP reported, "At times I was made to feel as if my thoughts didn't matter." [P2#25, SP, positive result].

Overall, 8/15 (52%) SPs reported they did not feel supported by the genetics service, and 8/15 (52%) reported undertaking the role of a support person was difficult, with 7/15 (47%) of SPs feeling ill- equipped to fulfil the role.

These feelings were not limited to those who were supporting an individual who received a positive result. Of those who supported a consultand who would not develop HD, one felt ill-equipped, and two others found it difficult.

"I thought I was equipped to be a support person but probably thought that way of thinking the result would be negative." [P2#3, SP, positive result].

Despite some SPs finding the role challenging, most SPs reported attendance at pre-test consultations (11/16, 69%) and at the result session (10/18, 56%) was beneficial for themselves.

"We were given excellent counseling all through from the first session. I appreciated the good advice about possibly caring for my husband for 10-15 years." [P2#20, SP, negative result].

SPs also assessed how they and their consultand adjusted to the test results. For those who supported a consultand who would develop HD, four SPs felt they adjusted 'very poorly' or 'poorly' to the test result (see Figure 2).

DISCUSSION

The majority of consultands and support persons in this study were positive about their experience of PST care and rated its provision by Australian genetics services highly. These views were represented by participants from both the > 55 and <55 year-old age groups, from four Australian States and Territories, and both males and females. Nevertheless, a number of consultands, and their support persons rated the service as 'fair' or 'poor'. This suggests that for some their experience of PST did not meet their values and expectations. It is important to consider and reflect on what might have happened in these instances of lower ratings, highlighting the necessity to constantly think about the engagement process and review the working alliance between counselor, the consultand and their support person. This may involve actively checking in with the consultand and their support person about how they are thinking and feeling about what is happening for them at regular intervals during PST so as to continually facilitate mutual understanding and hence the engagement process (19). Consistent with previous studies, we identified a positive consultand-counselor relationship as a key feature in effective genetic counseling (3, 14). However, based on support person experience, our study identified additional support and information needs for these consumers.

In this study, half of the SPs experienced the role as difficult and felt ill-equipped, including those SPs who accompanied consultands who were confirmed as non-carriers of the HD mutation who would not therefore develop the condition, as reported previously (10). In addition, a number of SPs who only attended the results session were distressed by the outcome of testing. While such distress is expected, this finding suggests that if SP involvement is appropriate, it seems preferable at the earliest possible time, so as to enable their concerns to be addressed and acknowledged (10). Both of these findings add weight to those of Williams et al. (2000) that while SPs are committed and involved in the PST process, they also have a need for information about the relevant condition, its progressive nature, interpretation of results and subsequent caregiving role. No doubt some SPs will be well informed, but for some others this will not be the case. Williams et al., (2000) also found that SPs deal with intense and complex emotions whether the SP is a spouse or a friend. Perhaps information normalizing these emotions and providing some basic guidance about active listening and responding empathically may assist in their preparation for this role. The provision of such information would be a supplement to discussions with the SP about their legitimate concerns and worries for the future.

Of particular interest in this study was the high satisfaction rate with a SP being present at the different sessions, both for the consultand and SP, independent of the test outcome. It appears that some of the contributing factors to this high satisfaction may relate to the level of attention and inclusion given to the SP by the genetic service during the PST process mentioned above. Efforts to make the SP feel a necessary and important part of the PST process may lessen the burden experienced by both the consultand and the SP. This finding seems to correlate with the view that the consultand's satisfaction with a supportive companion is the pivotal ingredient that enhances their ability to face the future, which may not be the case without the presence of this companion (16, 20). Quaid and Wesson (1995) also concluded that wherever possible partners be

included in the PST process in an endeavour to maximise the benefits for both parties.

Research has shown that consultands' partners are very cognizant of the potential impact that the outcome of PST can have, not only for their consultand and offspring, but inevitably for their own expected future in a caring role (10, 20-23). Rolland (1990) also suggested that spouses, in anticipation of future losses, experience a range of intense emotional responses over the course of an illness, which begins with the disclosure of results (24). However, some consultands do not experience their partners as supportive and are therefore reluctant to involve them in the testing process, as previously identified (23, 25). These relationships may not endure after either a genetic or clinical diagnosis, perhaps due to concerns that the partner will be unable to provide the ongoing psychosocial support required. In such cases it would seem counter-productive to insist a consultand bring a partner as a SP. However, the consultand should be encouraged to bring an alternative SP.

Importantly, a key aspect of genetic counseling for PST in HD and other late onset disorders is the provision of information for the consultand as well as their support person (4, 26). In addition, the importance of post-result support for both the Cs and their SPs was emphasised by many participants in this study, as some experienced difficulties adjusting to either a positive or negative result. These findings support provision of follow up for all individuals who are given a genetic test result and for their SPs, regardless of the future implications of the result, given the potential for adverse reactions when follow up is not provided (26, 27). Indeed, it has been suggested that post-test counseling is important to address not only coping and adjustment but also any misunderstanding or misinterpretation of the result, for example, inability to accept a normal result (28). Certainly, the process of PST, including decision-making around testing and return of results for asymptomatic 'well' individuals has far reaching familial implications unlike other testing scenarios in general medicine. It is therefore important to have appropriate support and guidelines in place for Cs, SPs and family members as required.

Limitations

This study was conducted a few years ago with the original intention to evaluate services. Nevertheless, the results have highlighted the needs of support persons in the context of PST, which have received little attention in the literature. Therefore, the findings reported in this study address a gap in the research. and remain relevant to current practice.

The limitations of this study include the small numbers of participants with largely female participation; the findings being limited to the experiences of those who participated; the support persons were from the older age group only; the age groups of the interview study participants was not collected; the participants were largely located in only one State and one Territory (NSW and ACT); and the retrospective nature of the study requiring participants to recall events that happened between one and four years previously. In the interview arm of the study, only one specialist HD service was evaluated. These factors may affect the reliability of the data collected.

Recruitment of consultands for the survey-based arm of the study was also limited and the snowballing recruitment strategy led to some pairs (consultand and their support person) being recruited. Therefore, there may have been shared narratives from the same consultation, which may not be representative of the broader population of those participating in PST. Similarly, it was not possible to determine if a participant in the interview study had also completed a survey. The response rate for the survey was also not able to be calculated.

Research recommendations

While recognizing these limitations in terms of the generalizability of the findings and the Australian context, they point to the need for an international study to further explore the needs of support persons in the PST process, and an evaluation of interventions to address their needs.

Healthcare implications

Importantly, both a high standard of genetic counseling, which is different from information-giving only,

and the consultand-counselor relationship, are integral to optimal service delivery for PST for an inherited adult-onset disease. Flexibility with the PST protocol may improve consultand and support person experience, including involving support persons throughout the PST process; addressing support person concerns and worries about the future; and information and follow up for consultands and support persons irrespective of the result. Through a more flexible and personalized approach it may be possible for PST services to address individual expectations whilst achieving a good outcome for all parties.

CONCLUSION

The findings affirm the centrality of the patient-counselor relationship and the need for clear and accurate information as foundational aspects in determining satisfaction with PST. The international guidelines recommend individuals undergoing PST should be encouraged to bring a support person to accompany them to all sessions of the PST process (3, 26, 29, 30). Findings in this study suggest it is most beneficial for the involvement of support persons to commence at the initial stages of the PST process. Furthermore, the findings suggest a greater acknowledgement of the role and needs of support persons is required, including relevant information and follow up to more fully support consultands and support persons regardless of the PST result and implications for the development of the genetic condition.

ACKNOWLEDGEMENTS

The authors wish to sincerely thank the research participants, the Australian state Huntington's Associations, and Huntington's NSW for funding the presentation of the findings of the quantitative arm of the study at an annual scientific meeting of the Human Genetics Society of Australasia. This work was conducted to fulfil Amy Howat's and Tenielle Clinch's Master of Genetic Counselling degree requirements. This research did not receive any specific grant from funding agencies in the public, commercial or not-for-profit sectors.

Supplementary Information

The following supplementary information files are available: file 1: interview schedule; file 2: pilot survey for consultands; file 3: pilot survey for support persons.

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PEER REVIEW

Not commissioned. Externally peer reviewed.

Table 1: Demographics and test results of consultands and support persons

		INTERVIEW STUDY	SURVEY STUDY	
		Consultand (C1) N = 14	Consultand (C2) N = 35	Support person N = 18
Age (years)	18-34	Unknown	9	1
	35-54	Unknown	16	5
	55-75	Unknown	8	9
	>75	Unknown	2	2
Gender	Female	11	24	13
	Male	3	11	5
Education	Tertiary		11	4
	Non-tertiary		23	14
State/Territory	NSW/ACT	14	23	7
	TAS		8	11
	QLD		3	0
	WA		1	0
Marital status	Married/partnered	7	27	16
	*Not married/partnered	7	8	2
Offspring	Yes	7	27	15
	No	7	8	2
Genetic result of participant[1]	Positive	5	16	8[+]
	Negative	9	14	4[+]
	MN/RP	0	2	2[+]
Brought a support person	Yes	6	29	
	No	8	2	
	Unknown		5	
Relationship C2/SP	Partner		21	12
	Other		8	6

Positive – ≥ 40 CAG repeats; Negative = ≤26 CAG repeats; Mutable normal (MN) = 27 – 35 CAG repeats; Reduced penetrance (RP) = 36-39 CAG repeats. *Not married = single/divorced/widowed. [+]Result for consultand being supported. One support person did not provide details of age or gender.

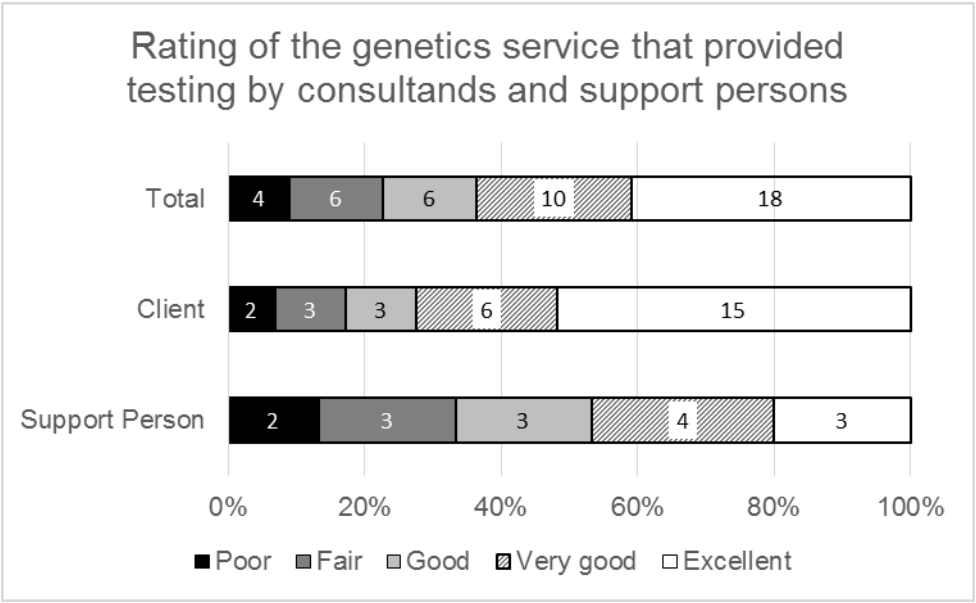

Figure 1: Overall rating of the genetics service by consultands (clients) and support persons.

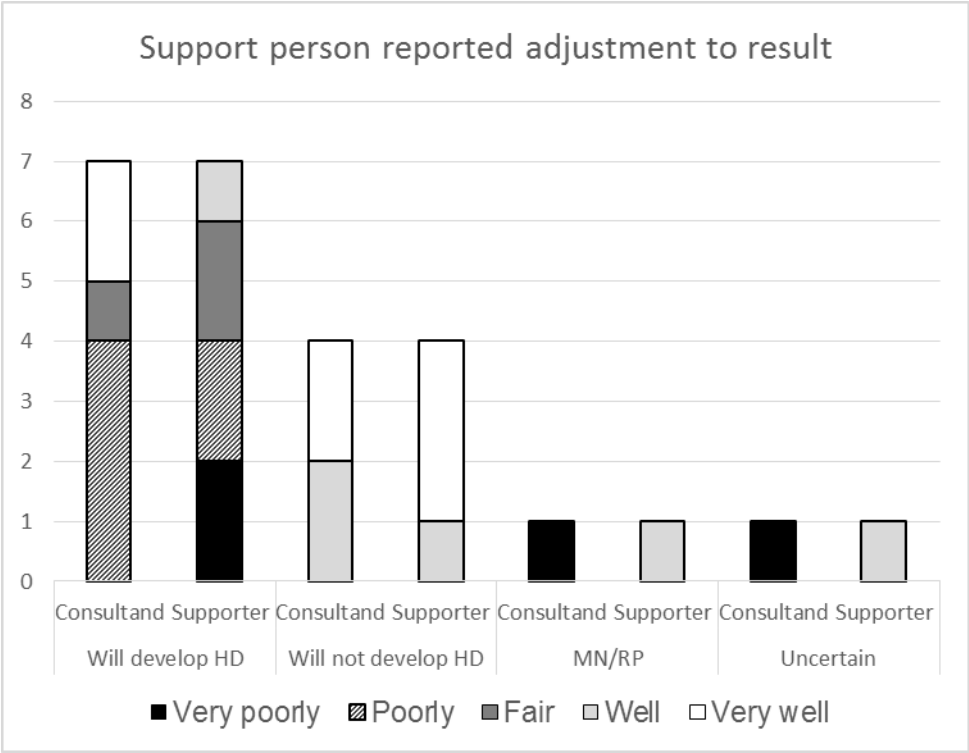

Figure 2: Support person perceived adjustment of consultand and self.