TITLE: Genetic counselling as a route to enhanced autonomy: using a sequential mixed methods research approach to develop a theory regarding presymptomatic genetic testing for young adults at risk of inherited cancer syndromes

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Abstract

Undertaking presymptomatic or predictive genetic testing should involve a considered choice. Decisions regarding genetic testing for young adults have to be considered within the context of their key life stage, which may involve developing a career, forming partnerships and/or becoming parents. The aim of this study was to develop a theoretical model regarding the factors involved when young adults (18-30 years) undergo presymptomatic genetic testing for inherited cancer syndromes. The model evolved from synthesis of results of a sequential mixed methods study involving a systematic review, a qualitative study and a quantitative study. The resulting model shows that young adults at risk of inherited cancer syndromes are influenced by others to have testing and come to counselling with their decision already made. However, genetic counselling enhances their feelings of autonomy and integration of their genetic status into their lives. Our theoretical model could be a valid support during the genetic counselling process for young adults

and their parents, as it may sensitize professionals to the specific needs of this population, including education and support to autonomous decision-making. Counselling approaches should be modified in this population: an inclusive, multi-step counselling process is needed, with timing and setting set according to the specific features of this sensitive population.

Key Words

Decision making, genetic counselling, young adults, presymptomatic genetic testing, predictive genetic testing, theory.

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Compliance with ethical standards

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 (5). Informed consent was obtained from all individuals for being included in the study.

Animal studies This article does not contain any studies with animals performed by any of the authors.

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Introduction

Presymptomatic genetic testing can establish if a person at risk carries a gene variant associated with the condition before the development of signs or symptoms of the disease (Skirton et al., 2013). It is available for a wide range of inherited cancer syndromes, including germline BRCA1/BRCA2 variants in hereditary breast-ovarian cancer and DNA mismatch repair gene variants in Lynch syndrome (Hadar et al., 2020; Menko et al., 2019). The results of testing may influence adoption of healthy lifestyles or facilitate early diagnosis and preventive treatment, which have been shown to have clinical benefit in some conditions (Gonzalez et al., 2019; Monaghan et al., 2020). Although genetic counsellors have many roles, including information giving and providing psychosocial support for adjusting to the diagnosis and making decisions regarding testing, prevention and treatment (Middleton et al., 2015; Redlinger-Grosse et al., 2017; Veach et al., 2007), a major role of genetic counselling is supporting clients at risk to make decisions about presymptomatic testing (Skirton et al., 2013). While the definition of YAs varies widely, Rindfuss (1991) defined it as 18-30 years. There is no prescribed age for reaching autonomy (Fuligni, 1998; Rivlis, 1998; Stewart et al., 1999), and this range seemed the most inclusive for our purposes. Presymptomatic testing may facilitate screening, preventive treatment and early diagnosis of YAs at high risk of cancer (Hartmann & Lindor, 2016; Masciari et al., 2008; Villani et al., 2011, 2016). During the transition from adolescence to adulthood, young adults (YAs) (aged 18-30 years) experience key challenges such as moving out of the parental home, completing education, beginning full-time employment, establishing partnerships and becoming a parent (Albritton & Bleyer, 2003; Stern et al., 2010). Decisions made during the process of genetic counselling may influence, and be influenced, by these life events. Cullinan et al (2020) suggest that YAs need to be conscious of potential risks to themselves (e.g. dealing with unexpected consequences of decisions) and their motivations for testing should be carefully explored during genetic counselling. However, genetic counsellors may be concerned about testing YAs because of uncertainty about their

cognitive, psychological, and emotional levels of maturity, which will influence their ability to make autonomous decisions.

Theory generation is an important process in expanding knowledge of a particular health context (Gauffin et al., 2019). Genetic counselling is a relatively recent development in healthcare, and to understand the counselling needs of this population in more depth, we aimed to develop a theoretical model regarding the factors involved in genetic counselling when YAs consider presymptomatic genetic testing. In this paper, 'young adults' refers to people aged 18-30 years (Godino et al., 2016, 2018, 2019) at risk of an inherited cancer syndrome(ICS), while 'parents' refers to parents of young adults at risk.

Methods

We built our theoretical model from the collective findings of three studies (conducted 2016- 2018) using a mixed-methods sequential exploratory design (Creswell & Plano Clark, 2013). As these have been published, we will not detail the methods here but further information can be found in the three original publications (Godino et al., 2016, 2018, 2019) and a Supplementary File-S1. Detailed material on strengths and weaknesses of the three studies is provided in Table 1.

First, we conducted an integrative review (including both qualitive and quantitative data) of literature focussed on presymptomatic testing of YAs at risk of an autosomal dominant condition (including inherited cancer). Using eight scientific databases, eleven published eligible studies were identified. We extracted information about the period before testing, experience of genetic counselling, parental involvement in decision-making, impact of test result communication and living with genetic risk (Godino et al., 2016).

Second, in a Grounded Theory (Corbin & Strauss, 2014) study, we performed face to face qualitative interviews with Italian YAs to obtain their perspectives regarding undergoing predictive testing for hereditary cancer. Interviews were conducted one month prior to genetic counselling,

two weeks after counselling and six months later. Forty-two interviews were conducted and the constant comparative method was used to analyse the data (Godino et al., 2018).

Third, a cross-sectional survey was prepared, based on results of the first two studies, to explore the psychosocial implications of predictive testing for hereditary cancer in YAs and their parents (Godino et al., 2019). One questionnaire was used to collect data from YAs (152 participants) and a second to survey parents (42 responders). Respondents were chiefly from the United Kingdom, the United States and Italy. The demographic information provided by the study participants is shown in Table S1.

An initial theoretical model was proposed after the first study and modified after each successive study. After the second and third studies, the earlier findings were clarified and sometimes re-organised, whilst new findings were introduced. To prepare the final model, findings of all three individual studies were rigorously reviewed by three of the authors and tables were prepared to aid further analysis and integration of findings into the model. We discussed each step in the model until consensus was reached among the authors as to the robustness of the underpinning material and relevance of each aspect of the model to the provision of genetic counselling for YAs at risk of inherited cancer.

Results

Development of the theoretical model after the integrative review

Young adults had little or no information concerning genetic testing until informed of their potential genetic risk by parents. As a consequence of parental pressure to be tested, YA participants conveyed feelings of disempowerment and lack of control over the testing decision (Figure 1). It appeared that YAs underwent genetic counselling as a necessary precursor to testing: there was no evidence of them declining a test after counselling. Genetic counselling was either expressed as a forum for discussing problems or connected with feelings of disempowerment.

Development of the theoretical model after the qualitative study

The results of our Grounded Theory study indicated that most participants had become conscious of their risk in the year preceding testing. Although some YAs may have had some growing awareness prior to being informed, this was not reported. Young adults at risk of ICSs had made a decision to be tested before genetic counselling and, only after that, did they discover that the purpose of genetic counselling was to help them consider the implications of testing prior to a decision. During the post- counselling interview, YAs displayed new strategies to deal with the situation they were experiencing. A dynamic relationship was identified, linking the decision-making process and the development of their autonomy, and these findings helped us to conclude that the process of genetic counselling enabled them to reflect upon themselves and their lives, as in a mirror.

Final development of the theoretical model after the quantitative study

The last phase of our research confirmed that the decisions of 24.5% YAs at risk of ICSs were influenced by others (mainly parents). Only those who opted to be tested pursued genetic counselling and, although testing requests were usually made by YAs, the majority of parent participants (n=26; 74.3%) felt they had control over the YA's decision and all felt their children should be tested. Overall, some YAs did not understand the implications of testing, but complied with parental pressure.

Our final model shows the journey from knowledge to testing (Figure 2). Initially, YAs are made aware of their risk. They experience pressure from family and others close to them to undergo testing. As a result, they present for genetic counselling, perceiving this to be the route to testing, rather than the forum for discussion of testing options. However, the genetic counselling process enables them to see themselves as independent from others and to 'step outside themselves', as evidenced by their references to themselves in the second person (Godino et al., 2019). While they

continued to proceed with testing, the process gave them a greater sense of autonomy and, as a result, enhanced their ability to integrate the result into their own lives.

Discussion

The traditional model of genetic counselling has emphasised that it is a process involving information giving which enables counsellees to consider genetic testing in a non-coercive environment (Resta et al., 2006). Our model challenges the conventional wisdom that the decision-making process is central to genetic counselling, at least in this younger age group. We suggest in this situation that the role of genetic counsellor is to use counselling skills to enable the YA to recognise their own autonomy and take responsibility for their testing decision, which has been shown to influence psychological adaptation in other health settings (Deadman et al., 2001). Our theoretical model demonstrates an inclusive, multi-step counselling process is needed, with timing and setting set according to the specific features of this sensitive population. Counselling should involve supporting the YA to explore their own attitudes to testing, their personal motivations and likely responses to results. We would argue that these steps are essential to ensure valid, informed consent is obtained.

Since the publication of our systematic review dates back almost six years, we updated the review, as reported in Supplementary File-S2 and Table S2. In recent papers findings concerning other conditions, such as Huntington disease and cardiomyopathies, are mainly consistent with our model, as many young adults, even though not all, had decided to be tested prior to genetic counselling (Forrest Keenan et al., 2015; Gong et al., 2016; Lewit-Mendes et al., 2018; MacLeod et al., 2014; Wassall et al., 2017). Gong et al. (2016) and Lewit-Mendes (2018) also showed that many YAs felt they were helped to mature as a result of the genetic counselling and presymptomatic testing process, although some reported negative impact of genetic counselling, including a lack of emotional support during the process (Forrest Keenan et al., 2015). Another issue raised by our

findings is the lack of use of genetic services by those who decide not to be tested, indeed in our cohorts, only those who had decided to be tested contacted genetic services. It is suspected therefore that some YAs who do not wish to be tested may not be aware that information on prevention and cancer screening can be offered, regardless of a genetic test result. Scarce prior knowledge of young at risk individuals is also supported by Young et al. (2019) showing that some young adults (18-25 years), who had not yet attended a genetic clinic, expressed the need for basic genetic information. The wish to reduce uncertainty about genetic status has been shown to be a motivating factor for seeking genetic counselling and testing across a range of conditions (Skirton, 2006) and may prompt a rapid decision to be tested in those who have a low tolerance of ambiguity. Most of the YAs in our cohort had only recently become aware of their risk and this, as well as family influence, may have been a factor in their decisions.

In order to reach the group of YAs who wish to access information without necessarily wanting to be tested, the supportive and educational function of genetic services should be publicized, especially the fact that it is relevant even to those who do not wish to be tested. A review by Menko et al. (2019) confirmed that relatives of those at risk influence their access to information, testing and screening and authors have suggested that genetic services should contact those at risk directly. However, genetic services have been based on a non-coercive approach with an emphasis on freedom of personal choice to pursue genetic counselling or testing or not (Resta et al., 2006), so have traditionally not contacted patients directly. Family members know the 'at risk' person and may be better able to judge the most appropriate time and way of giving the information, but as we have shown, they can also influence the 'at risk' person's decisions. More research is needed to see if a direct approach from health services would be generally acceptable and more beneficial.

While we collected data across a number of different cultural settings, one of the limitations of our study was that responses were restricted to those who could speak either Italian or English. The parents in our study were not necessarily those of the YAs who took part, so it was not possible to

compare accounts of YAs with those of their own parents. However, the individual studies were robustly conducted and the synthesis of results was undertaken rigorously by five researchers who had different cultural and professional backgrounds. This study focussed on clients in the YA age group, and may not be applicable to older counsellees. The model requires further testing in YAs. Cultural norms in the participants' countries may also have influenced the results. Further research is needed to test whether the theoretical model does apply to a wider cohort of clients accessing genetic counselling, particularly those who may be heavily influenced by others.

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Animal studies This article does not contain any studies with animals performed by any of the authors

Availability of data and material The data generated during and/or analysed during each study phase are available from the corresponding author on reasonable request.

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Consent to participate The authors consent to participate.

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