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ORIGINAL ARTICLE

General Practitioners' knowledge and use of genetic counselling in managing patients with genetic cardiac disease in non-specialised settings

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Abstract There have been over 40 cardiac diseases with a genetic cause identified to date. The management of most genetic cardiac diseases (GCDs) now necessitates multidisciplinary care, including the provision of genetic counselling. This study investigated the knowledge and management of GCDs by General Practitioners (GPs). Questionnaires were mailed out to 685 doctors working in general practice in Tasmania, Australia, with 144 responses (21 %) received. Results showed that the majority (77.8 %) of the responding doctors are managing at least one patient with GCD in their practice. However, GPs identified having limited confidence in the appropriate management of these conditions and indicated that they are very dependent on guidance from a cardiologist, including whether to refer a patient to genetic counselling. To our knowledge, this is the first Australian study that looks at the care of patients with GCD in the primary care sector. The knowledge gained will help us provide more appropriate care for patients who do not have immediate access to specialised services, particularly those outside metropolitan areas, and provides evidence for what resources can be offered to doctors working in general practice to help provide quality care for these patients.

Keywords Genetic cardiac disease · Genetic counselling · Primary care · Patient education

Introduction

Knowledge about cardiac disease is expanding, with over 40 genetic cardiac diseases (GCDs) identified to date (Milewicz and Seidman 2000). Almost all GCDs carry an inherent risk of sudden cardiac death (Nunn and Lambiase 2011). Given the seriousness of this diagnosis, patients with GCD may not only have suboptimal physical health (Hamang et al. 2010; Ingles et al. 2013) but also suboptimal psychological well-being (Hamang et al. 2010, 2011; Ingles et al. 2013; McGorrian et al. 2013) and will often require lifelong follow-up and multidisciplinary care to ensure quality of life. As a part of the multidisciplinary care afforded, the importance of genetic counselling has been widely recognised (Ackerman et al. 2011; Cowan et al. 2008; Hershberger et al. 2009; Hoedemaekers et al. 2010; Sturm 2013; Zodgekar et al. 2011) with all major GCD management guidelines recommending genetic counselling (Ackerman et al. 2011; Zodgekar et al. 2011).

Genetic counselling is the process that can help patients make informed decisions about the implications their diagnosis has on them and their family (Resta et al. 2006). To ensure appropriate genetic counselling, specific GCD centres have been established in Australia and in many developed countries. These clinics provide access to specialists with intimate clinical knowledge of these conditions. In Australia, however, all of these centres are located in metropolitan areas, with many patients in rural and remote areas distanced from these

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centres and unlikely to attend (Ingles et al. 2015). In Tasmania, an island state situated off the south coast of Australia, there are no specialised GCD clinics due to its small population of approximately 500,000, and a population density of 7.5 people per square kilometre (Australian Bureau of Statistics 2015). In place of specialised services, Tasmania has the Tasmanian Genetic Counselling Service, which is run through the Royal Hobart Hospital—the state's tertiary referral centre. This service accepts referrals for *all* conditions, including GCDs. We have no local data on the prevalence of GCD in Tasmania, but extrapolating from other community prevalence studies (Maron et al. 1995; Schwartz et al. 2009; Ward 2000), a conservative estimate would be approximately 8,500 patients with GCD, and by taking into account a more recent assessment of prevalence (Semsarian et al. 2015), may even be as high as 10,000 patients.

In non-specialised settings, GPs play an integral role in caring for patients with GCDs, monitoring response to treatments and titrating these as required, and often providing support not only to the patient but their families. This is particularly important in Tasmania, where patients are more reliant on primary care and are likely to see their GP more often than a specialist (Australian Institute of Health and Welfare 2012). A GPs ability to recognise, manage and refer patients with GCDs and their family members to the appropriate specialists is crucial, and requires appropriate recognition of GCDs and knowledge of first line management strategies including medical management, and benefits of engaging of supporting multidisciplinary services such as genetic counselling (Ingles et al. 2011; Nunn and Lambiase 2011).

This research stemmed from a recognised need to evaluate whether patients with GCD outside of specialised centres are referred to appropriate services in Tasmania. There is limited research conducted in medical practitioners who routinely manage patients with GCDs outside specialised settings and their knowledge of these conditions (Challen et al. 2010; van Langen et al. 2003, 2005), and no research conducted in Australia on these issues. The aim of this study was to assess Tasmanian GPs' knowledge and perceptions of GCDs and the role genetic counselling plays in the management of these conditions. Four main research questions were identified as being important in exploring this aim: What GCDs are being managed by GPs working in General Practice in Tasmania? Do GPs believe it is important to educate patients with GCDs and how confident are they in doing this? What genetic counselling opportunities do Tasmanian GPs access? And finally, how do GPs perceive the importance of multidisciplinary care and support of patients with GCDs?

Methods

Study population

This study was undertaken in the Australian state of Tasmania. Tasmania was considered an appropriate study site because patients with GCD in the state do not have routine access to specialised GCD clinics available in larger urban centres such as Melbourne, Sydney and Brisbane. Rather, Tasmanian patients with GCD typically have to be referred to cardiologists and genetic counselling separately.

The study population was doctors working in general practice in Tasmania. A GP was considered to be a person with a medical degree who had completed at least one year of hospital training but was working outside of the hospital environment (i.e. in a primary care setting) at the time of the survey. The study population was considered to be appropriate for addressing the study's aims, as GPs are most often the initial doctor responsible for making appropriate referrals related to GCDs. A list of GPs in Tasmania was compiled from the 'Tasmanian Health Directory', a publically available database (http://www.tasmedicarelocal.com.au/tasmanian-health-directory). A total of 685 eligible GPs were identified to participate in the study. This may not have comprised all GPs working in Tasmania but is likely to have captured most who were working in a clinical primary care capacity at the time, with the number of GPs identified consistent with published data (Primary Health Care Research and Information Service 2014).

A full ethics application for the project was submitted to the Tasmanian Health and Medical Ethics Committee and approved (Project No. H0011820). All procedures followed were in accordance with the Helsinki Declaration of 1975, as revised in 2008.

Survey instrument

The authors developed the questionnaire as a self-administered cross sectional survey, to be distributed to medical students, general practitioners and cardiologists, as a part of a larger study exploring the care provided to patients with GCD in Tasmania. The original survey was piloted on final year medical students at the University of Tasmania in 2011. This group was chosen as the final survey could be distributed to future cohorts without influencing the quality of the data collected. From the piloting process, changes and further adaptations were made. The final survey comprised a total of 21 questions and used multiple-choice answers (with binary, categorical and ordinal options), ranked answers using a Likert scale, and short answer questions, providing the opportunity to collect both qualitative and quantitative data. The questionnaire included five main sections: basic demographic information (questions 1–8), general questions about patient

education and counselling (questions 9–11), what information and resources GPs use for managing patents with GCD (questions 12–13), whether referral to genetic counselling services is routinely thought about in managing general and patients with GCD (questions 14–19), general statements regarding knowledge of genetics (question 20), and the final question enabled GPs to leave additional comments they deemed relevant to the survey or topic in general (question 21) (see supplementary material). There were nine conditions that were used as examples of GCDs in several questions to see how they would be managed. The nine conditions were chosen based on the conditions affecting the patient population sampled for the overall study, and the conditions that were likely to be the most prevalent in the population.

Procedure

The questionnaire was mailed out as a paper-based survey in February 2014 to the eligible GPs. Participants were given the option to complete the questionnaire online or as a self-administered Internet-based survey. Informed consent was inferred by return of the completed questionnaire. After six weeks, an interim analysis was completed of the returned questionnaires. This initial analysis revealed an acceptable response and question completion rate (21 %). Response saturation was noted to many questions, with the researchers deciding not to do a follow-up with the study sample.

Analysis

A descriptive analysis using frequency tables and percentages was undertaken for all survey responses. When all respondents did not answer questions, this was noted and presented as a comparative percentage between the total respondents to the survey and the respondents to the specific question. Qualitative data was drawn from the responses to short answer questions. The answers were coded by the response number entered into the SurveyMonkey database (e.g. GP1, GP2, etc.), and were analysed using a thematic approach. A thematic approach to analysing qualitative data is a method of reading and identifying patterns, or themes in written data, and was completed using methods outlined by Liamputtong and Serry (Liamputtong and Serry 2013). Two of the authors (JM and JW) had the opportunity to review the qualitative data and agree on the themes presented.

Results

Of the 685 questionnaires that were sent to eligible participants, 144 were returned, representing a response rate of 21.0 %; 127 (88.2 %) surveys were returned as paper-based questionnaires and 17 (11.8 %) were completed online. Response rates varied for questions but yielded a high overall response rate ranging from 80.6 to 100 %.

Demographic questions revealed the gender of respondents was almost equally balanced, with 73 females (50.7 %) and 71 males (49.3 %). The majority of respondents were vocationally qualified GPs (127, 88.2 %), with 16 registrars (GPs in training) (11.1 %) and one resident (0.7 %) also responding. One hundred and four GPs (72.2 %) cited completing their training in Australia. The majority of GPs (79, 54.9 %) were practising in Southern Tasmania, followed by 42 (29.2 %) in Northern Tasmania, and 23 (16.0 %) in North-Western Tasmania. Based on 2010–2011 Tasmanian health workforce figures and the surveyed population, each geographical cohort of GPs was almost equally represented (Southern GP response rate 20.2 %, Northern GP response rate 24 %, and North-Western GP response rate 19.0 %).

How confident are Tasmanian GPs in educating patients about genetic cardiac diseases?

When GPs were asked whether they feel confident in educating patients with GCDs, the majority (51.4 %) of the 140 respondents agreed with the statement, but a significant proportion (29.3 %) were also unsure. When asked specifically about how confident they were in answering patient's questions about GCD, 39.6 % disagreed or strongly disagreed, 32.6 % were unsure how they felt, and 27.8 % agreed or strongly agreed with the statement. When GPs were asked whether they felt confident with the knowledge they have regarding GCDs, 141 responded to the question: 9.2 % of GPs strongly agreed or agreed, 27.7 % were unsure, and the majority (56.0 %) cited that they disagreed or strongly disagreed. Despite the majority of practitioners admitting that they did not feel confident with their knowledge regarding GCDs, the same number of respondents felt differently when asked whether they felt confident with their knowledge in appropriately *managing* GCDs in their clinical practice, with 56.0 % of GPs strongly agreeing or agreeing with the statement, but also a significant proportion (28.1 %) stating they were unsure (see Table 1).

GPs were given the opportunity to provide additional comments in the survey, with 49 GPs providing responses. Analysis revealed that whilst GPs not only recognised that they had limited knowledge about GCD, but some further commented that they would like to know more in order to improve their professional knowledge and practice indicating that education was needed and wished for:

> I've been a GP for 30 years. I've NEVER been to a meeting or educational event about genetic counselling. I'm interested in it to learn. Wouldn't this be a good idea even a monthly newsletter[?] (GP18)

Table 1 Statements regarding respondent knowledge about genetic cardiac conditions and confidence in managing them

Statement	Strongly disagree	Disagree	Unsure	Agree	Strongly agree	*N*
If a person is diagnosed with a genetic condition, it is important to educate the person about their condition	0 (0.0 %)	0 (0.0 %)	0 (0.0 %)	*39 (27.0 %)*	**104 (73.1 %)**	143
If a person is diagnosed with a genetic condition, it is important to educate their family members about their condition	1 (0.7 %)	1 (0.7 %)	5 (3.5 %)	*58 (40.6 %)*	**78 (54.5 %)**	143
I routinely educate patients and their relatives about their genetic conditions	0 (0.0 %)	9 (6.3 %)	25 (17.6 %)	**78 (54.9 %)**	*30 (21.3 %)*	142
I feel confident talking to patients and their families about their genetic condition (cardiac or otherwise)	2 (1.4 %)	25 (17.9 %)	*41 (29.3 %)*	**58 (41.4 %)**	14 (10.0 %)	140
I feel confident answering a patient's questions about genetic cardiac conditions	4 (2.8 %)	**53 (36.8 %)**	*47 (32.6 %)*	35 (24.3 %)	5 (3.5 %)	144
I feel confident about the amount of knowledge I have regarding GCDs	15 (10.6 %)	**74 (52.5 %)**	*39 (27.7 %)*	12 (8.5 %)	1 (0.7 %)	141
I feel confident that I know enough to appropriate manage GCDs presenting in my clinical practice	15 (10.6 %)	**64 (45.4 %)**	*39 (27.7 %)*	23 (16.3 %)	0 (0.0 %)	141

Bold results indicate largest proportion of responses; italicised results indicate second highest proportional result

N number of respondents to the question, *GCD* genetic cardiac disease

One GP acknowledged lack of knowledge, but offered a solution in how to improve this, in the form of creating clinical pathways to assist in referring appropriately:

> The issue is not so much the knowledge of practitioners but the clarity of the referral systems in which they work. Clinical pathways work would assist in this regard (GP10)

Lack of knowledge, however, was not considered an excuse for some GPs, as they felt that they would still be able to manage these conditions appropriately:

> Not having specific knowledge of every condition does not mean I am not confident that I could find information I needed and refer appropriately, thus managing the conditions appropriately (GP12)

What genetic cardiac diseases are being managed by GPs working in general practice in Tasmania?

GPs were provided with a list of GCDs and were asked to select how many patients of each they thought they had in their patient population. The survey indicated that the three most common GCDs being seen in general practice in Tasmania are hypertrophic cardiomyopathy (HCM), with 86 (62.7 %) out of 137 respondents to the question saying that they had at least one current patient with the condition, followed by familial dilated cardiomyopathy (FDC) with 56 GPs (40.9 %), and Long QT syndrome (LQTS) with 52 GPs (38.5 %).

Do GPs believe it is important to educate patients with GCDs?

Of the 143 respondents to the questions, all GPs (100 %) agreed that it is important to educate patients about their genetic condition, with the majority (95.1 %) also agreeing it is important to educate family members about genetic conditions. When asked whether they routinely educated patients and their relatives, the majority (76.1 %) of 142 respondents responded that they did (see Table 1).

What genetic counselling opportunities do Tasmanian GPs access?

How GPs understood the role of local genetic counselling services and factors influencing their decision to refer patients to

these services were also addressed in the survey. Of the 143 respondents to the question, 86.7 % GPs responded that they had heard about the Tasmanian Genetic Counselling Service. However, there were varying degrees of confidence in respondents' (n=142) knowledge about the service, with only 9.9 % of doctors acknowledging they knew a lot about it, 52.8 % responded they knew a little, 26.1 % only knew it existed, and 11.3 % responded they had not heard about it at all. When asked about referring to the service, and whether patients with [general] genetic conditions were routinely referred, there was more variability. Of 142 respondents, only 37.3 % said that they sometimes referred, 26.8 % GPs responded that they did routinely refer, 14.8 % said that they did not routinely refer, and 10.6 % only referred if the patient asked for it. Eleven respondents (7.7 %) wrote that they had not had the opportunity to refer to the Tasmanian Genetic Counselling Service, as this had not yet been required in their practice. Table 2 shows how likely a GP is to refer a patient with GCD for genetic counselling.

GPs were also asked what influences their decision to refer these patients onto counselling as a short answer response. One hundred and sixteen GPs (80.6 %) identified three main factors influencing further referral: (1) patient's preference/wishes (22.4 %), (2) advice from the cardiologist/specialist (19.0 %); and (3) knowledge of the condition, often referring to their lack of knowledge (17.2 %). Table 3 lists examples of specific responses related to this question.

How do GPs perceive the importance of multidisciplinary care and support of patients with genetic cardiac diseases?

One hundred and forty-one (98.0 %) respondents provided open-ended answers in regards to who they felt should be involved in providing care for patients with GCD. The cardiologist or specialist (94.3 %) was the person most often mentioned as being most important in the team of GCD care providers. The identification of other disciplines varied, with the geneticist (36.7 %), genetic counsellors (30.5 %) and genetics services (9.9 %) all featuring prominently. Other team members suggested included a nurse (specialist and/or general practice), psychologist, paediatrician and cardiothoracic surgeons.

What is the role of the cardiologist?

GPs clearly acknowledged that they rely on the guidance from the cardiologist, particularly when it comes to recommending genetic counselling. This was particularly evident with one GP, who believed that regardless of the condition, they would first refer to the cardiologist, who can then refer on, if needed:

> More likely to refer to cardiologist who can decide if further counselling is needed (GP25)

This belief was further developed by another GP, who thought that given the cardiologist is more likely to diagnose the condition, and that were also more likely to initiate the referral:

> Many cardiac conditions are complicated and I would expect the cardiologist to refer for counselling if needed (GP45)

One GP mentioned that regardless of what they knew, it was still the role of the cardiologist to refer and that meant that they did not need to know more:

> I wasn't aware of many of these genetic diseases. I feel the expertise is with the cardiologist and they should provide genetic clinic referral or instruct the GP to. I don't feel I need to increase my knowledge in this area (despite being limited) as I would always refer these sorts of cases to a cardiologist. (GP11)

Table 2 Indication as to how likely a GP is to refer these conditions for genetic counselling

Condition	Always	Sometimes	Rarely	Never	Unsure	*N*
FDC	**53 (39.6 %)**	*33 (24.6 %)*	18 (13.4 %)	4 (3.0 %)	26 (19.4 %)	134
ARVC	*41 (31.5 %)*	21 (16.2 %)	7 (5.4 %)	4 (3.1 %)	**57 (43.9 %)**	130
LVNC	*37 (28.5 %)*	16 (12.3 %)	10 (7.7 %)	5 (3.9 %)	**62 (47.7 %)**	130
RC	*31 (23.7 %)*	27 (20.6 %)	17 (13.0 %)	5 (3.8 %)	**51 (38.9 %)**	131
CPVT	*39 (30.5 %)*	17 (13.3 %)	9 (7.0 %)	4 (3.1 %)	**59 (46.1 %)**	128
LQTS	**51 (39.5 %)**	27 (20.9 %)	18 (14.0 %)	4 (3.1 %)	*29 (22.5 %)*	129
BrS	**49 (38.3 %)**	19 (14.8 %)	10 (7.8 %)	2 (1.6 %)	*48 (37.5 %)*	128
HCM	**42 (31.6 %)**	*34 (25.6 %)*	20 (15.0 %)	7 (5.3 %)	30 (22.6 %)	133
BAV	27 (20.6 %)	*28 (21.4 %)*	24 (18.3 %)	11 (8.4 %)	**41 (31.3 %)**	131

Bold results indicate largest proportion of responses; italicised results indicate second highest proportional result

N number of respondents to the question, *FDC* familial dilated cardiomyopathy, *ARVC* arrhythmogenic right ventricular cardiomyopathy, *LVNC* left ventricular non-compaction, *RC* restrictive cardiomyopathy, *CPVT* catecholaminergic polymorphic ventricular tachycardia, *LQTS* long QT syndrome, *BrS* Brugada syndrome, *HCM* hypertrophic cardiomyopathy, *BAV* bicuspid aortic valve

Table 3 Examples of responses given for the question "What influences your choice to refer/not refer the above genetic cardiac conditions to genetic counselling?"

What influences your choice to refer/not refer the above genetic cardiac conditions to genetic counselling?	
Patient's wishes	Expectation of patient (GP44)
	Family's concern (GP120)
	If patient asks (GP105)
	Patient demand (GP132)
	Patient preference/knowledge (GP69)
	Patient willingness (GP62)
	Patient's desire/request (GP78)
	Patients wishes after education (GP140)
	Pt acceptability to referral (GP80)
	Pt and family wishes (GP111)
	Pt desire for counselling (GP84)
	Pt knowledge of disease from cardiac specialist Pt desire (GP42)
Cardiologist/ specialist advice	Cardiologist suggestion (GP92)
	I would first check what advice the cardiologist has given (GP115)
	Opinion of cardiologist (GP68)
	Refer to cardiologist: if he recommends further referral or patient were to ask for it I would refer to geneticist (GP119)
	Specialist recommendation (GP142)
	The cardiologist recommendation or my comfort that there is no new genetics information available which this decade of a genetics explosion, is a rare thought (GP75)
	Whether recommended by cardiologist (GP37)
	Would check with their cardiologist prior to doing so (GP118)
	Would rely on opinion from cardiologist (i.e. whether genetic counselling or if pre-pregnancy planning is required or not) (GP112)
Knowledge possessed	Awareness of counselling existence (GP58)
	Full knowledge of the conditions (GP76)
	If I think it could cause sudden death (GP1)
	Knowledge if good/nil if poor (GP18)
	Knowledge of condition (GP120)
	Knowledge of services (GP121)
	Lack of knowledge (GP90)
	Limitations in my own knowledge (GP123)
	May not realise that a condition is genetic (GP135)
	My knowledge of the condition (LACK of knowledge that is) (GP32)
	My level of knowledge as to how strongly they are inherited (GP97)
	Not knowing when to refer (GP50)
	Personal knowledge (GP138)

Pt patient

Discussion

Provision of genetic counselling has become part of the standard of care for managing patients with GCD (Ackerman et al. 2011; Zodgekar et al. 2011). Patients may access this service through specialised GCD clinics, but not all patients have access to such a service. In the state of Tasmania, this is the case whereby patients with GCDs are required to either travel interstate, or, they can access individual services at the discretion of their treating doctors, GPs or cardiologists, who will refer them on, often on an 'as needed' basis. This study was designed to investigate the practices of GPs who may be in this position when managing patients with GCD. Our results indicate that although GPs are aware of their limited knowledge of GCDs, they ensure the patient is appropriately referred to the cardiologist in the first instance. The study reveals, however, that the GP heavily relies on the cardiologist for further management, with few feeling comfortable to refer to other services without prompting.

This study builds on previous studies and provides further information as to what non-specialists consider important in managing these patients. Challen and colleagues (2010) looked at genetic knowledge possessed by GPs—in France, Germany, Sweden, the Netherlands, and the UK—and specifically at the professional responsibility perceived by GPs using GCD (specifically HCM) as an example of a developing genetics area, with the study trying to assess how comfortable GPs would be to counsel patients. They found that the willingness of GPs to engage in genetic counselling depends on the country they practice, but the majority of GPs surveyed did not feel comfortable in engaging in discussions around genetic risk and testing (Challen et al. 2010). This finding is contrary to the fact that GPs need this skill (Guttmacher et al. 2007), and further highlights that a number of factors that influence a GP's approach to genetics issues arising in their practice (Houwink et al. 2011; Scheuner et al. 2014). Whilst our results build on what Challen et al (2010) found, there is an important distinction to be made: the purpose of our study was to look at the role of the GP in the context of providing appropriate multidisciplinary care in this specific patient population, not the expectation that the GP should perform all parts of management and counselling for such conditions.

In addition to Challen et al (2010), van Langen and colleagues (2005) performed a survey in which they surveyed all Dutch cardiologists and geneticists in the Netherlands. The Netherlands has a system in which genetic counselling for predictive testing can only be provided by clinical geneticists. Although the study found that cardiologists and

geneticists feel that there is a role for both groups to provide care for such patients, highlighting that multidisciplinary care is appropriate, they found disagreement between the cohorts as to who was in the best position to provide counselling to these patients, and that more education would be appreciated to help manage these patients more appropriately in both groups, but particularly for cardiologists (van Langen et al. 2005). This is likely to be a view held more generally by other medical professionals, including a general practice cohort. The study also reported that the median number of patients with HCM per cardiologist was five, likely indicating that similar to our GP population, although they are being exposed to the condition this small exposure may not be of enough to confidently manage these patients. Similarly, another study also found that even cardiologists are unlikely to refer patients for genetic counselling and testing (van Langen et al. 2003), indicating that although our GPs largely believe that this is the role of a cardiologist, they also may not consider it.

The strength of this project is to provide evidence where there is little and contributes important information on the GP-specialist relationship in places where specialised services are limited. The results are also important given our limited knowledge of the care provided for patients outside of specialist genetic cardiac centres in Australia, as it is likely that many patients encounter this kind of care. The results also provide important information about how GPs perceive their role in managing these patients: the answers received regarding referring to genetic counselling services and multidisciplinary team input suggest that GPs have the skills to adapt to managing more specialised conditions, but may not always do so. It is unclear why some GPs did not include themselves in the multidisciplinary team when asked about managing patients with GCD. It could be due to a misinterpretation of the question, or they may not see a role for themselves given the 'specialised' nature of GCDs, which shows the potential for less than optimal care.

The questionnaire was designed to explore local needs and is not a validated questionnaire, so it will be difficult to compare data from other populations. The study represents the experiences and knowledge possessed by one particular practicing population, making generalizability difficult, but it is also likely that similar issues are being faced by practitioners in other places where patients do not access to specialised cardiac genetics services, and the issues raised are similar to more general cohorts (Houwink et al. 2011). Whilst the response rate was not as high as other studies (Challen et al. 2010; van Langen et al. 2003, 2005), there was a high completion rate. Not unexpectedly, the questions least likely to be answered were short answer questions, particularly the last question where it was optional to provide a comment, and paper surveys were more often fully complete than online surveys. It should also be noted that the focus of the survey on the involvement of genetic counselling in the care of these patients may have influenced some of the answers received; however, many answers received also indicate a lack of awareness and understanding of the role of this service as a part of normal practice.

Whilst these weaknesses are recognised, the information gathered will be useful in considering how these patients should ideally be managed outside of specialised environments. Whilst having dedicated genetic cardiac clinics may not be cost-effective, expanding telehealth or e-health services may be of more use, and should be considered to help provide appropriate care in a timely manner, as well as contributing to GP education seminars and forming clinical pathways for genetic conditions as suggested by many GPs in our survey. Other resources that may also be worthwhile exploring would be the provision of online education, either by establishing a module that GPs could complete as a part of competency training and professional development, or a website that provides information and outlines recommended procedures relevant to the local setting. Any improvement that can be made to help educate GPs in areas where there are no specialised services would help ensure there were no large discrepancies between patients who have and have not accessed specialised services and their health outcomes, although more studies are needed to see what patients with GCDs desire when considering what is quality health care as this would also influence any changes in current practice.

Conclusion

To our knowledge, this is the first study that investigates the knowledge and perceptions of GCD in general practice in Australia and generates insight into how they are managed in non-specialised settings. The study provides insight into what influences GPs to refer patients to genetic counselling and indicates that they rely on guidance from the cardiologist. This study is the first step in identifying barriers to providing recommended care to patients with GCDs, particularly outside of metropolitan centres, and supports the importance of referring patients to clinics that are aware of the needs of these patients. Given the challenges of rural practice, it also highlights the importance and place of continuing professional development for GPs in conditions outside of those that are commonly managed to ensure quality care and the need for clinical referral pathways for patients with GCD outside of specialised centres.

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Compliance with ethical guidelines

Conflict of interest Jessica A. Marathe, Jessica Woodroffe, Kathryn Ogden and Clarissa Hughes declare they have no conflict of interest.

Human rights and informed consent 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 participants included in the study.

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