ARTICLE

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Decision-making and experiences of preimplantation genetic diagnosis in inherited heart diseases: a qualitative study

Laura Yeates[1,2,3,4], Kristie McDonald[2,5], Charlotte Burns[2,3,4], Christopher Semsarian[2,3,4], Stacy Carter[6] and Jodie Ingles[1,3,4]✉

Preimplantation genetic diagnosis (PGD) ensures a disease-causing variant is not passed to the next generation, including for inherited heart diseases. PGD is known to cause significant emotional burden, but little is known about how parents experience PGD to select against inherited heart disease. We aim to understand how people with inherited heart disease, and their partners, experience and make decisions about PGD. Participants were recruited from a specialised inherited heart disease clinic. Qualitative semi-structured interviews were conducted with adult participants who had considered PGD. A semi-structured interview schedule explored overall experiences and reasons for undergoing PGD. Broad topics included experience of disease, reproductive history, psychosocial and financial considerations. Interviews were recorded, transcribed verbatim and thematically analysed using a framework method. Twenty participants were included (15 with inherited cardiomyopathy, 3 with inherited arrhythmia syndrome and 2 partners). In contemplating PGD, participants considered 3 main issues: past experience of disease e.g. sudden cardiac death, sport restrictions and clinical heterogeneity; intergenerational responsibilities; and practical considerations such as finances and maternal age. Among those who chose to undergo PGD ($n = 7/18$), past experience of a significant cardiac event, such as family history of sudden cardiac death, was important in the decision process. The decision to undergo PGD for inherited heart disease is complex and influenced by individual values and experience of disease. We highlight key areas where further discussion may assist in PGD decision processes.

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INTRODUCTION

Preimplantation genetic diagnosis (PGD) is a reproductive technique that ensures a genetic condition is not passed to the next generation. PGD is performed after in vitro *fertilisation* (IVF) whereby resulting embryos are tested for the pathogenic variant causing disease in a family, and only those embryos without the pathogenic variant go on to be implanted [1, 2]. Originally, PGD was used for severe or lethal childhood-onset conditions as an alternative to prenatal diagnosis [3–5]. The use of PGD has expanded to include both recessive and dominant single gene disorders, sex-linked disorders, chromosomal rearrangements, aneuploidy and human leukocyte antigen (HLA) matching [2, 6, 7]. High stress levels in couples undergoing PGD have been previously reported [1, 8].

Inherited heart diseases include the primary cardiomyopathies (e.g. hypertrophic, arrhythmogenic and dilated cardiomyopathies) and inherited arrhythmia syndromes (e.g. long QT syndrome, Brugada syndrome, catecholaminergic polymorphic ventricular tachycardia). While distinct diseases, there are a number of common features including largely autosomal dominant inheritance, the need for lifestyle modifications, lifelong medical therapy, and risk of adverse outcomes including heart failure and sudden cardiac death [9–11]. In those aged <35 years, the most common causes of sudden cardiac death are inherited [12] and there is significant psychological burden for the surviving family members [10, 13].

PGD has been available over the last two decades [14]. Over time the awareness of PGD has increased with treating health care professionals. This has grown from previous reports of the option of PGD not being raised with eligible patients outside of specialist IVF clinics [8], to couples exploring their options through pre-conception genetic testing and counselling [15]. A four-step decision making process for couples considering PGD has been proposed, including identify—couples understand the risk of disease and learn of the availability of PGD; contemplate—couples consider and deliberate their options; resolve—couples reach a decision, engage—couples carry out their decision [15]. To date there is little research into how PGD decisions are made specifically in the setting of inherited heart disease and as a result there is little information to guide families considering this option. Here we explore the experience of couples considering PGD in the context of inherited heart diseases.

METHODS

Participants and recruitment

Participants were recruited from a specialised, multidisciplinary tertiary referral clinic in Sydney, Australia. The clinical team includes cardiologists and genetic counsellors with expertise in inherited heart disease. Patients

[1]Cardio Genomics Program at Centenary Institute, The University of Sydney, Sydney, NSW, Australia. [2]Agnes Ginges Centre for Molecular Cardiology at Centenary Institute, The University of Sydney, Sydney, NSW, Australia. [3]Faculty of Medicine and Health, The University of Sydney, Sydney, NSW, Australia. [4]Department of Cardiology, Royal Prince Alfred Hospital, Sydney, NSW, Australia. [5]School of Psychology, The University of Sydney, Sydney, NSW, Australia. [6]Australian Centre for Health Engagement, Evidence and Values (ACHEEV), School of Health and Society, University of Wollongong, Wollongong, NSW, Australia. ✉email: jodie.ingles@populationgenomics.org.au

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Table 1. Participant characteristics.

Participant characteristics	***n* = 20**
Gender	
Female	14 (70)
Male	6 (30)
Clinical status	
Affected	18 (90)
Partner	2 (10)
Mean age ± standard deviation	36 ± 5
Interview type	
Phone	17 (85)
Face to face	3 (15)
Mean interview length, minutes (range)	45 (20-66)
Education	
Postgraduate/University	14 (70)
High School/Diploma	5 (25)
Year 10 or below	1 (5)
Family characteristics	***n* = 18**
Family history of sudden cardiac death	8 (44)
Family history of disease	14 (78)
Families with a child prior to contemplating PGD	6 (33)
Personal history of cardiac arrest or appropriate ICD shock	11 (61)
Other major personal events (stroke, heart transplant)	3 (17)

Data shown as *n* (%) unless otherwise indicated.
ICD implantable cardioverter defibrillator.

and their at-risk family members are seen for clinical diagnosis, management, clinical surveillance and genetic discussions. A purposive sampling strategy was used to recruit individuals with an inherited heart disease who had undertaken PGD, had engaged in discussion about the option of PGD with the clinical team, or had recently had children. Both individuals with disease and their partners were invited to participate. Partners, if they consented to participate, were interviewed separately. Participants were invited to participate via phone with follow-up information sent via email/post if they were interested. During this recruitment phone call an invitation was also extended to their partner. Participants were aged 18 years or older, with sufficient self-reported English skills to participate in the interview. This study was approved by the Ethics review Committee (RPAH Zone) of the Sydney Local Health District (protocol number X17-0009).

Study design

A semi-structured interview schedule was developed, and focused on the participants' experience of disease, reproductive history, information sources around PGD, financial and emotional considerations as well as the experience of going through IVF/PGD itself. The interview schedule was developed based on review of the literature and discussions between the clinical team who have experience seeing this patient group (genetic counsellors, cardiologists and a clinical psychologist). Interviews were conducted by one author (LY) over the phone or in person, depending on the preference of the participant. LY is a genetic counsellor with over 10 years' experience in the cardiac field. Interviews were recorded and transcribed verbatim. Transcripts were evaluated using the framework method [16]. Each transcript was independently reviewed by the primary researcher (LY) and KM, a clinical psychologist. A subset (25%) of transcripts were reviewed by a third author (CB). A code list was generated with each transcript review, the list was revised and discussed between authors (LY, KM, CB) until consensus had been reached. Codes were then grouped into themes and discussed amongst all investigators until a consensus was reached. Each transcript was then coded using the consensus framework. Data were summarised with supporting quotes in an excel spreadsheet.

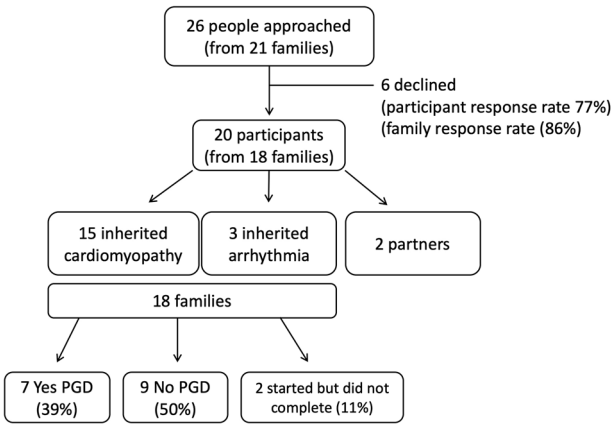

Fig. 1 Flowchart of study participants. Abbreviations: PGD preimplantation genetic diagnosis.

Demographic information was also collected at the completion of each interview.

RESULTS

Characteristics of the participants

There were 26 individuals approached from 21 families, and 20 agreed to participate (response rate 77%). Fourteen (70%) of the participants were female, mean age was 36 years (range: 27–48 years) and 14 (70%) had a University-level education. In total 11 (61%) had a personal history of cardiac arrest or appropriate shock from their implantable cardioverter defibrillator and 3 (17%) had experienced a major cardiovascular event such as stroke or heart transplant (Table 1). The 20 participants came from 18 families, 15 had inherited cardiomyopathies and 3 had an inherited arrhythmia syndrome. Two partners consented to participate, and all clinically affected individuals were from separate families. Six of the 18 families had children prior to considering PGD, with clinical diagnosis occurring after the first child, or a gene result not being available prior to having the first child.

Of the 18 families, 7 had undergone PGD (39%). Two families started the process but did not complete it (11%), while 9 (50%) decided not to go ahead with PGD (Fig. 1). Of the 7 families who underwent PGD, four had another reason for considering IVF (multiple miscarriage, late maternal age, need for a sperm donor). Sixty seven percent of households had an income over $104,000 AUD per year (Australian average yearly income $67,012 www.abs.gov.au). There was a family history of sudden cardiac death in 6/7 families who underwent PGD, and 1/11 in the families who did not proceed with PGD. At the time of analysis, 14/18 families have gone on to have at least 1 child.

Overview of results

In considering whether to undergo PGD, participants considered 3 key themes: past experience of disease including risk of sudden death, sports restrictions and clinical heterogeneity; intergenerational responsibilities; and practical considerations such as finances and maternal age. Participants considered whether these factors threatened their imagined future, motivating them to undertake PGD or whether they could incorporate these challenges into their future plans and decline PGD (Fig. 2).

Participants above all wanted a healthy child. For some this was threatened by the risk of an inherited heart disease and became a very strong motivator to proceed with PGD.

> "just not wanting a child to have that burden and just that responsibility of if you're going to have a child and you have the

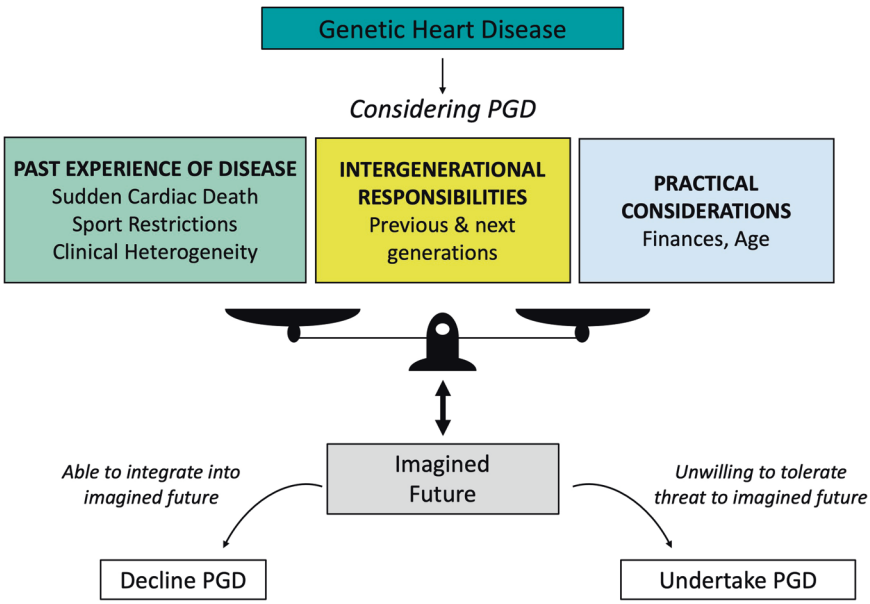

Fig. 2 Summary of themes and the perception on imagined future. In considering PGD participants considered three key themes (in bold) and weighed the impact of these on their imagined future.

option for [the child] not to have some chronic illness, then how could you go any other way, is the way I look at it." Female PGD50

"the best as you can reduce the chance of your child having the condition. Ah, it feels like you're doing something, it is less of the, genes are just so black and white. What you have got is what you've got and there is nothing you can do about that. So it is nice to feel like you have got a little bit of control, there is not so much helplessness about passing on a condition that can affect someone's life." Female PGD

Below, we consider each of the three themes that motivated prospective parents' PGD decision making.

Sudden cardiac death

Many patients with inherited heart diseases have past experience of sudden cardiac death of a family member. The sudden nature of the events and the slim chance of effective resuscitation add profound uncertainty to prognosis. Such events are rare, but tragic. Participants spoke of the fear of such an event for themselves and for future children, and how this motivated them towards PGD.

"It's just horrible thinking…for a parent to have a child and raise them and see them through to fifteen like my sister and then to lose your child overnight without warning… I didn't want to experience that myself." Female PGD

"but probably the key reason I'm not [declining PGD] is you still have the risk, that you destroy the risk [with PGD] of sudden death, destroy the risk that your kid goes to school one day and doesn't come home." Male PGD

"to definitely have a very strong chance of eradicating a gene that can possibly kill you, kill your child is huge." Female PGD

In contrast, for participants without a lived experience of sudden cardiac death of a family member, the perceived risk of sudden death did not outweigh the burden of PGD.

"I think if it was worse, as you say, like I think if we were really affected or we've lost of family member, that would be very different, but our experience of it, it made IVF, for me, sound not worth it in terms of the trouble and the risk." Female Declined PGD

"[the chance of an inherited heart disease] just wasn't a massive burden and so we just also felt that we were going to be okay. I guess the risk-benefit analysis just simply wasn't –it just wasn't there in the end." Male Declined PGD

Sport restrictions

Some participants felt the impact of sports restrictions on future children would be low. This was based on their own experience of either being diagnosed later in life after their sporting career, continuing to participate despite a recommendation to stop without negative consequences, or on a lack of interest in sport.

"you know, [our child] is likely to have a very normal life and obviously, some limitations for [them] but, yeah, I think we just felt comfortable knowing that we know what we know of the people who've lived with it in our family." Female Declined PGD

In contrast, participants who chose to undertake PGD felt the lifestyle restrictions of a diagnosis, specifically exclusion from competitive sport, would be detrimental to a future child if they were to develop the condition, drawing from their own positive experiences of sport in their own childhoods.

"Sport was a massive part of my life and probably one of my greatest passions…it took a long period of adjustment, almost like a grieving process." Female PGD

"And it's a condition that causes sudden death, so at any time your kid could be running around and could drop over and even if that doesn't happen, you've got that lifetime of worrying that that's about to happen. In your head, imagine

every time they do Cross Country Carnival, you're worrying about that." Female PGD

Clinical heterogeneity

For some participants, the presence and impact of an inherited heart disease did not strongly motivate them to seek PGD, as the impact experienced was small, with both themselves and family members experiencing few symptoms or consequences of their condition. Some considered that in comparison to medical conditions other people may have, theirs was manageable.

"no one [in our family] ever had anything [symptoms] that sort of made them think there was something wrong." Female Declined PGD

"because I've got the thing, my daughter has got it and we know how to manage… it's not likely to present quality of life issues or any major learning disability or something else that could have a traumatic impact on our entire life." Male Declined PGD

In contrast, some participants were highly attentive to the uncertainty around the clinical heterogeneity of disease and the unpredictable impact the condition may have on their future children. For some they deemed this intolerable and they were therefore were motivated towards PGD. For others after consideration they were able to integrate this uncertainty into their imagined future and after consideration declined PGD.

"You can do all the [clinical] screening in the world…the uncertainty of the clinical pathway, of a child with the gene, not knowing whether they would express it or not. So I just couldn't face a lifetime of that." Female PGD

"For me, yeah, absolutely, it's been fine, but – yeah, just wondered if they got the worst [heart condition] you could – that you can get, would I still be happy that we' had kids and I think – yes, now I would be, but I wasn't at the start. Yeah. I wasn't sure." Female Declined PGD

Intergenerational responsibility

Participants reasoned intergenerationally, making decisions about PGD partly in reference to their own parents and their existing children, and conscious of the responsibilities that one generation has towards the next. However different participants interpreted these differently.

Participants discussed the emotional burden of decision-making including the impact of the perceived judgement and guilt, towards undertaking or declining PGD, from their family and friends. For some participants, this meant facing that they themselves may not have been born had PGD existed when they are conceived.

"and then my mum gave me a really hard time… she's like if we'd done that, we'd have not had you." Female Declined PGD

"I thought, Gosh, if my mum had to make that decision, I wouldn't be here. And I think I'm contributing [to society] at some – on some level." Female Declined PGD

Other participants felt the responsibility to act both for their potential children and for future generations, and expressed guilt associated with the decision.

"It's different when you have a child and something happens to them, but I became aware of that I was a carrier or I could pass this on, then I have a responsibility, I felt, and I know that was – the risk was too high." Female PGD

"I didn't want to experience that myself [SCD of a child] but also then, her children and her children's children and that would've just continued. If I have decided to go through PGD then that would stop all the other generations from having to make that decision and spare them the difficulty." Female PGD

"the PGD does add another layer 'cause there's this a feeling of guilt – when they say – it didn't happen in our first round, but I was thinking, "What if we only had two embryos and they were both only affected? That's two that if it wasn't for this, we could've implanted." So that kind of feels like – well, you know it's not your fault, but it kind of is your fault." Female PGD

For couples who had already had one child prior to PGD being available to them, the result of the first child's genetic testing affected their decision on whether to go ahead with PGD in subsequent pregnancies. By refusing PGD they were acknowledging the value of their own and their gene positive children's lives. Some felt the need for subsequent children to have "an equal chance" or the same opportunities as the first.

"Once we did get [child 1's] result [positive] we didn't care that much about whether or not our next child was positive…we're not going to manage it any differently." Male Declined PGD

"If it had been prior diagnosed to having [child 1] could've been a different outcome with how we did this process but once we've had [child 1] and fallen in love with him, obviously, it was a no brainer to continue having children, to I guess thinking, you continue rolling that dice." Female Declined PGD

"If [child 1] already had it, would we have taken our chances? Possibly more so – yeah, especially if it's something that they both had. Would it be worth to have two of them with it? I don't know, but at least they'd then have that in common and look at life with defibrillators, all that kind of stuff together, so – yeah, I think it definitely played a part, for sure. Female PGD

Practical considerations

Decisions about PGD for inherited heart disease interacted strongly with more general and practical considerations around fertility and what was financially possible. Participants described the impact their reproductive history – such as miscarriage, termination of pregnancy and advance maternal age – had on their decision-making.

"I think if we'd been able to get pregnant naturally, we would've just done that and tested the baby once it was born, partly because we wouldn't have gone down the track of actually me getting tested because we would have just been pregnant." Female PGD

"I think if I was younger, I would probably – well, I'd do it. I would do it. There'd be no question now. But I think for me, it was – at my age, how many eggs am I going to get? And what

Table 2. Summary of themes with illustrative quotes regarding the impact of IVF/PGD.

Theme	Quotes
"All consuming" impact of IVF/PGF	"I think one of the difficulties is, because you are injecting most days, and you are feeling uncomfortable, it just keeps it so in your mind every day you are aware of it" Female PGD "There's a lot of stress involved with pressure I suppose, but then that's not just the PGD but because of the IVF" Female PGD "I'd say frustration that it was such an intensive process and we didn't want it to fail and have to go through it again. I know for me, it wasn't that big a deal, but there was a little bit that it played on my mind that if it didn't work the costs would build up… and it would just take forever" Male PGD
Time taken	"it probably took longer in total than I had expected, test for this, test for that and then you think you're done and then there's - Oh no you've got all this other stuff to do – you can't actually do this yet" Female PGD
Low success rate	"You've done the waiting and then they say "Oh Sorry – none of the, all of them are either affected by one [chromosome abnormalities] the other [heart condition] or both" that's when you start thinking oh gosh, maybe we're being too fussy" Female PGD "The pressure, just the worry that it wouldn't work… every time it didn't work I felt like I was costing our family a lot of money by not producing, and also it was my gene, it wasn't my husband that had brought this to our relationship" Female PGD

PGD preimplantation genetic diagnosis, *IVF* in vitro fertilisation.

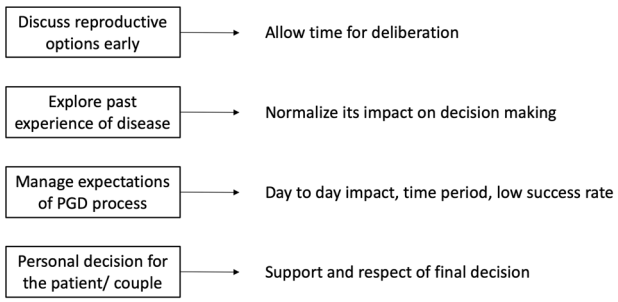

Fig. 3 Key points of dicussion regarding preimplantation genetic diagnosis (PGD) for clinicans with patients who have inherited heart diseases.

am I going to do if that's the viable one and I have to say no? That's a massive decision I prefer – that decision would be out of my hands and I either go through with it or not go through with – not have a successful pregnancy or an unsuccessful pregnancy." Female Declined PGD

"I also was concerned that it would take a long time….Okay, well we got to do this and we got to do that," then maybe it doesn't work, and then suddenly it's like five years and then I'm older, it's going to get harder to have kids. The other worry was that we'd stuff around and then miss the window". Female Declined PGD

For the majority of couples, finances were considered as challenging in decision-making, but were not deemed overly prohibitive.

"It probably only influenced our decision in so far as that we had to acknowledge at the beginning that there were not unlimited funds available to us." Female PGD

"And I guess we thought of it as a – you're paying upfront to save yourself a lot of medical expenses in the future, potentially." Female PGD

"I think if I required it then cost wouldn't be an issue because if that what was needed, then you would want to go through it." Female Declined PGD

Impact of undertaking PGD

Although not unique to inherited heart disease, three subthemes that are commonly raised in research about the impact of IVF/PGD were also raised by the participants in this study who underwent PGD (Table 2). First, participants spoke of the "all consuming" day to day impact of PGD due to the process of IVF, not only in attending the numerous appointments and undertaking hormone injections but also the constancy of the process in their mind. Second, they reported disappointment in the time it took to set up the test, and that after a period of deliberation in making the decision to go ahead they had not comprehended the time it took for the test to be set up. Finally, the impact of a low success rate and the prospect and burden of having to undertake another round of IVF/PGD were important considerations.

DISCUSSION

We explored the decision-making process and experiences of couples considering PGD in the setting of an inherited heart disease. Our findings were consistent with the decision making process outlined by Hershberger et al. and highlight key themes considered in the contemplate step of decision making [15]. In contemplating whether to undertake PGD, participants considered their imagined future, and the impact a diagnosis of an inherited heart disease may have. Participants emphasised three key issues in their decision-making process, negotiating whether or not the threat to their imagined future was significant enough to warrant undertaking PGD.

Overall, the key drivers of the decision to pursue PGD related to a participant's lived experience of the inherited heart disease, and the perceived threat this posed to their imagined future of having healthy children. Whether a child who carries a pathogenic variant will develop disease, and their future prognosis, is entirely unknown. For those who had already experienced the worst outcome, the sudden cardiac death of a young family member, this uncertainty around whether a child would develop disease and how severely, weighed heavily and ultimately motivated the decision to "eradicate" the genetic variant. Those couples whose past experience was more benign seemed more able to tolerate this threat to their imagined future and declined PGD.

The decision to undergo PGD is a dynamic one and, as described by Hershberger et al., couples often take some time to come to a decision and can oscillate between options [15]. At present there is little understanding of the factors weighing in to the decision to undertake PGD for inherited heart diseases, and

our findings provide a road map to support couples considering this option (Fig. 3).

A significant driver for couples considering PGD, and unique in the setting of inherited heart disease, is the risk of sudden cardiac death. Although sudden death is a rare event, its impact is significant and lifelong. The unpredictable nature of a sudden cardiac death event was a key motivator in undertaking PGD as participants felt the ongoing worry of such an event was a significant threat to their family's future. In contrast, a number of participants described the impact of the disease to be not enough to warrant the invasive process of PGD. In these situations, participants reconsidered how their future life might look, concluding that should significant burden come with a diagnosis in their child, they had the capacity to deal with such adversity.

Intergenerational responsibility and relationships played a part in the decision-making process in considering the opinions and choices of the previous generation as well as the next generation. Factors deemed key in the decision to pursue PGD included whether the couple had previous children. Couples who already had children with inherited heart disease prior to PGD being available ultimately preferred not to proceed with PGD, wanting to give each child an "equal chance". With variable expression being a hallmark of inherited heart diseases, our findings highlight the importance of exploring a patient's past experience of disease and how these experiences are shaping their decisions. Managing uncertainty is challenging in any medical setting, but especially for couples making decisions about potential children. In this scenario they consider the uncertainty of the future, knowing that electing to conceive naturally may result in a child who suffers the most serious disease outcomes, but taking a proactive approach, via PGD, may ultimately limit a pregnancy occurring at all.

Similar to previous studies [1, 5], participants described the significant emotional impact of undertaking IVF/PGD. While in theory PGD is a straight forward process to ensure a pathogenic gene variant is not passed onto the next generation, in reality, it is an invasive time-consuming and emotionally demanding process [5]. For the cardiology treating team it is important to manage expectations of the impact of the process of undertaking PGD when initial discussions on PGD as a reproductive option are raised. While this is a topic that will likely be covered by the IVF/PGD providers, our findings illustrate many participants felt ill-prepared for the process and raises an obvious area for improvement and the benefits of early information.

There are some important limitations to consider in our study. Participants were recruited through a tertiary referral specialised inherited heart disease clinic and may not be the indicative of the general inherited heart disease patients who are cared for in the community. Seventy percent of participants had university or postgraduate education, which may have biased our sample towards people with high health literacy who investigated reproductive options.

CONCLUSION

The decision to undergo PGD for inherited heart disease is highly personal and shaped by individual values and experience of disease. Unique to inherited heart disease is the impact of the risk of sudden cardiac death. The role of the specialised multidisciplinary team including genetic counsellor, clinical geneticist, cardiologist, psychologist, and general practitioner is to ensure that all options available to an individual or couple contemplating children are discussed to ensure an informed decision. We provide a road map for discussions with couples contemplating PGD in the setting of inherited heart disease which includes managing expectations of the process of PGD.

DATA AVAILABILITY

The datasets generated and/or analysed during the current study are available from the corresponding author on reasonable request.

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AUTHOR CONTRIBUTIONS

Concept and design: JI and LY. Aquisition of data: LY. Data analysis: LY, KM, and CB. Interpretation: LY, KM, CB, CS, SC, and JI. Drafting of manuscript: LY. Critical review of manuscript: LY, KM, CB, CS, SC, and JI. Final approval: LY, KM, CB, CS, SC, and JI.

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COMPETING INTERESTS

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ETHICS

This study was approved by the Ethics review Committee (RPAH Zone) of the Sydney Local Health District (protocol number X17-0009).

ADDITIONAL INFORMATION

Supplementary information The online version contains supplementary material available at https://doi.org/10.1038/s41431-021-00963-1.

Correspondence and requests for materials should be addressed to Jodie Ingles.