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ORIGINAL ARTICLE

Reflections of parents of children with 22q11.2 Deletion Syndrome on the experience of receiving psychiatric genetic counseling: 'Awareness to Act'

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Abstract

Individuals with 22q11.2 deletion syndrome (22qDS) have a 25%–41% risk for a psychotic disorder. Although early intervention for psychiatric conditions leads to the best long-term outcomes, healthcare providers often provide inadequate information about these issues and psychiatric services are underused by this population. We conducted semi-structured interviews with parents of children with 22qDS a month after they received psychiatric genetic counseling (pGC), to evaluate outcomes and perceived value of pGC with respect to parents' needs. Using grounded theory, we generated a theoretical framework of the process of building parental awareness of psychiatric risks associated with 22qDS and protective and management strategies for mental health (MH). Parents described how after their child's diagnosis with 22qDS, a variety of barriers stalled their building awareness of psychiatric risk and protective/management strategies: dealing with the immediate symptoms of 22qDS; child's young age; parental fear and stigma; and missing MH guidance. These barriers led them to carry the burden of worrying over missing emerging psychiatric symptoms and the stress over advocating for their child's MH. Parents indicated pGC was beneficial in that led them to achieve an 'awareness to act,' feeling confident in being alert and equipped to protect and/or manage their child's MH.

KEYWORDS
22q11.2 deletion, genetic counseling, intervention, mental health, psychiatric illness, velocardiofacial syndrome

1 | INTRODUCTION

22q11.2 deletion syndrome (22qDS) is considered the most common microdeletion syndrome in humans (Shprintzen, 2008), affecting 1 in 2,000 to 4,000 births (Botto et al., 2003; Devriendt et al., 1998; Oskarsdottir et al., 2004). The condition has a variable presentation of a wide range of clinical findings (Shprintzen, 2008), including physical symptoms such as cardiac anomalies, orofacial anomalies, hearing deficits, intellectual or learning disabilities, immunodeficiency, and hypocalcemia, which can predispose to epilepsy (Bales et al., 2010; Hallberg et al., 2010). Individuals with 22qDS also have an increased risk for developing a psychiatric disorder (Hoeffding et al., 2017): The prevalence of anxiety, psychotic, and neurodevelopmental disorders including autism

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and ADHD in youth with 22qDS is substantially greater than in the general population (Antshel et al., 2006; Basset et al., 2003; Jolin et al., 2009; Niklasson et al., 2009; Schneider et al., 2014; Vorstman et al., 2006).

Psychiatric manifestations of the syndrome are known to be of considerable concern to parents of affected children, particularly the chance to develop a psychotic disorder like schizophrenia, which has been estimated at 25%–41%, as compared to a 3% general population risk for the same (Basset et al., 2003; Schneider et al., 2014). However, most parents report that they did not receive information about the psychiatric manifestations of 22qDS from a healthcare provider (Hercher & Bruenner, 2008), and indeed genetics professionals report discussing the psychiatric manifestations of 22qDS less often with families than other aspects of the condition (Martin et al., 2012; Morris et al., 2013; Talcott-Baughman et al., 2015).

A potential way to address concerns about psychiatric disorders among families of children with 22qDS would be to provide psychiatric genetic counseling (pGC). pGC is a psychotherapeutically oriented interaction designed to help people understand—in a personalized manner—the factors that contribute to the development of psychiatric illness, and strategies that can be used to protect mental health (MH) (Austin, 2019). Though pGC has been shown to have positive outcomes for people living with psychiatric disorders *unrelated* to genetic syndromes and their family members, it is neither routinely clinically applied for people who have microdeletion or microduplication syndromes with psychiatric phenotypes and their families, nor have its outcomes been examined in this context (Inglis et al., 2015; Semaka & Austin, 2019). Therefore, we aimed to explore the perceived value of pGC and its fit with the needs of parents of children with 22qDS. Using grounded theory methodology, we sought to develop a theoretical framework that describes parents' perspectives of and experience with MH and care, their knowledge of the psychiatric risks associated with 22qDS, their use of strategies to protect and/or manage their child's MH, and the perceived value of pGC with respect to these issues above.

What is known about this topic

Previous research has documented how the psychiatric risks associated with 22q11.2 Deletion syndrome are inadequately addressed by healthcare providers, including genetics professionals, and as a result, parents are left alone to learn more about the associated mental health problems.

What this paper adds to this topic

This study illustrates how parents of children with 22q11.2 deletion syndrome perceive the experience of receiving psychiatric genetic counseling—it facilitates their awareness of the psychiatric risks and protective and management strategies for the mental health of their child.

2 | METHODS

The study methods detailed in the following sections are consistent with the consolidated criteria for reporting qualitative research (COREQ) checklist (Tong, Sainsbury, & Craig, 2007) and the standards for the reporting of genetic counseling interventions in research and other studies (GCIRS) (Hooker, Babu, Myers, Zierhut, & McAllister, 2017). Completed checklists have been included as supplemental material (Supplement 1 and 2).

2.1 | Grounded theory

Grounded theory is a fitting qualitative methodology for exploring social processes and interactions, such as the process of pGC and the patient-counselor and or parent-child interaction (Corbin & Strauss, 1990; Strauss & Corbin, 1998). Grounded theory generates findings that are rooted in participants' words and underscores aspects of their experience that they find most important. The evidence-based theoretical frameworks developed in grounded theory studies can be used to inform genetic counseling practice (McAllister, 2001, Grubs and Piantanida, 2009).

2.2 | Participants

Adults (>18 years old) with a child (<19 years old) diagnosed with 22qDS, who were fluent in English, able to provide informed consent, and had not already had pGC were recruited from the individuals who had been referred to a specialist pGC clinic in Vancouver, British Columbia, Canada (Inglis et al., 2015) and via email/website advertisements to the Rare Disease Foundation and 22qDS support groups in Canada and the United States of America (USA).

2.3 | Psychiatric genetic counseling

A detailed description of pGC is provided in Inglis et al., (2015) and Austin (2019). Briefly, the information-provision component of a pGC session (typically 60 to 90 min in length) involves: using the family history and empiric data to discuss risk for self and/or family members (e.g., children) to develop the indicated condition (Austin et al., 2006, 2008; Peay & Austin, 2011), relaying research-based information about the factors that have been associated with the indicated condition to the participant's family history and their existing explanation for cause of illness (in lay language, and using visual aids (see Figure 1) to facilitate comprehension), and using the information about factors that contribute to the development of illness as a framework to discuss evidence-based strategies for protecting mental health (e.g., sleep, nutrition, and exercise) (Baglioni et al., 2011; Bodnar & Wisner, 2005; Harvey et al., 2018; Lakhan & Viera, 2008).

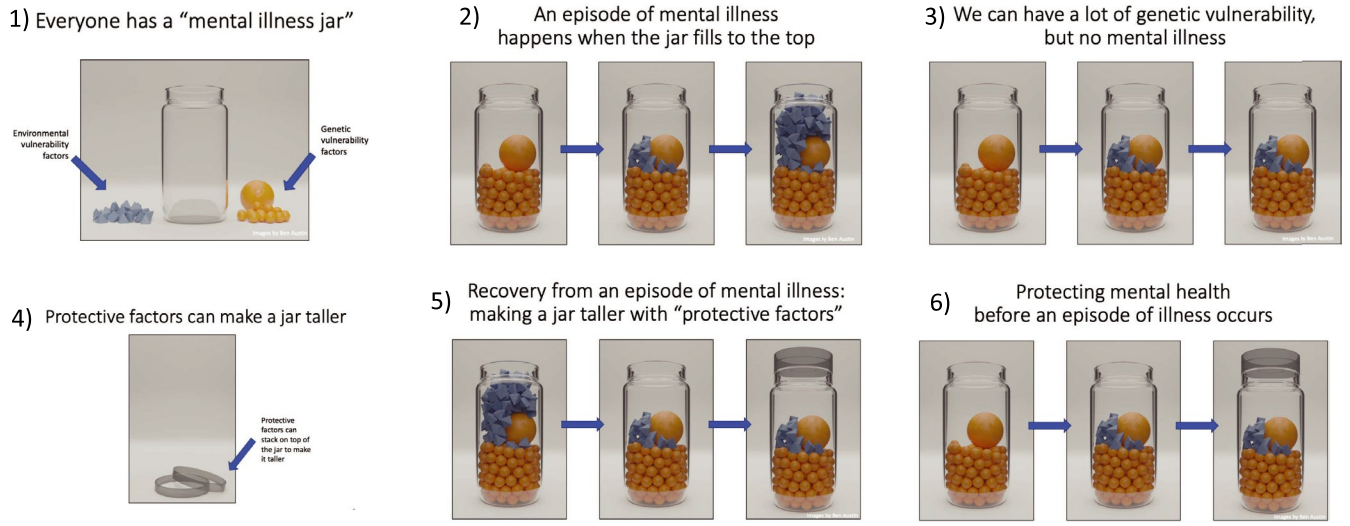

FIGURE 1 The larger '22qDS' ball represents the greater contribution of the 22q11 deletion compared to the other genetic factors (smaller yellow balls) in the chance for an affected individual to develop a mental illness. Adapted by Ben Austin from *How to Talk with Families About Genetics and Psychiatric Illness*. Copyright (c) 2011 by Holly Peay and Jehannine Austin. Used with permission of the publisher, W. W. Norton & Company, Inc. All rights reserved

The genetic counselor also works to uncover and address emotional ramifications (e.g., guilt, shame, stigma, and fear) that may be evoked (Veach et al., 2007).

Much of the information and support provided during pGC is facilitated by using an analogy of a 'mental illness jar', which helps individuals understand how various genetic and environmental factors work together to 'fill up the jar' and cause an episode of MI (Figure 1). The genetic factors are represented by multiple yellow balls, indicating the contribution of polygenic risk in the development of mental illness. A larger yellow ball represents how the 22q11.2 deletion makes a larger contribution toward the development of MI. The MI jar analogy also helps people better understand strategies that can be used to protect MH (Semaka & Austin, 2019). Protective factors, such as sleep, nutrition, exercise, and social support, are discussed as ways to make a person's MI jar larger, thereby increasing its capacity to accommodate environmental stressors before becoming full. In the context of 22qDS, pGC is ideally designed to improve parents' knowledge of psychiatric problems; increase their ability to identify concerns should they arise; and empower them to implement strategies to protect and manage their child's MH.

2.4 | Procedures

Between June 2016 and February 2019, a purposive sample of parents of children with 22qDS was recruited to participate in a study that involved completing a single, semi-structured, telephone-based interview one month after receiving pGC. Participants in this study were provided pGC by one of four Canadian and/or American board-certified genetic counselors (AI, CS, EM, and JA). All counselors were trained and supervised by JA and a pGC checklist (Austin, 2019) (Supplement 3) was applied to ensure competency/consistency and adherence to a pGC protocol. pGC was provided either in person or by videoconference and took place in the specialist pGC clinic or in the team's clinical research space in Vancouver, British Columbia, Canada. There was no fee for participants to receive genetic counseling in the specialist pGC clinic (included in medical service plan coverage for British Columbia residents) or through research participation in this study (grant funded). Participants were provided an honorarium as an incentive to complete demographic surveys and the one month post-pGC interview. The timing of the telephone interview conducted with participants coincided with the one month post-pGC standard clinical follow-up by the genetic counselor (Austin, 2019; Inglis et al., 2015).

Interviews were audiotaped, transcribed verbatim, and while they were not returned to participants for comments or correction, they were checked for accuracy by the research team. All interviews were conducted by AS or PC: both identified as female, MSc in genetic counseling trained, board-certified genetic counselors, neither of whom provided pGC to any of the study participants or interacted with participants prior to the study. AS has applied grounded theory methodology within her PhD and postdoctoral research aimed at exploring processes of genetic counseling for neurodegenerative and psychiatric disorders, respectively. PC has clinical research experience in the provision of pGC for patients with treatment-resistant psychosis and their family members. At the time of the study, participants were aware that AS was a postdoctoral fellow and PC was a genetic counselor and Clinical Assistant Professor in the University of British Columbia (UBC) Department of Psychiatry. Participant demographics were also obtained through an interview with a research assistant and MH focused family history information was gathered at the start of the pGC session. Interviews consisted of open-ended questions exploring parents' perspectives on and experience with MI and MH care, their knowledge of the psychiatric

risks associated with 22qDS, strategies they used to protect and/or manage their child's MH, and their perceptions of the pGC they received (Supplement 4). In accordance with grounded theory methodology, the interview guide was refined as data were collected to ensure questions captured emerging and important concepts and to further develop conceptual linkages (Corbin & Strauss, 1990; Strauss & Corbin, 1998). Field notes were made by the researchers after the interviews. Themes were derived from the data and analysis continued until data saturation was reached in all categories, meaning that the collection of additional data did not yield new information or insights. Two participants took part in a member check interview where the final theoretical framework was presented to participants to confirm that their experiences were reflected in the developed theory.

2.5 | Data analysis

We used NVivo 4.0 to store, organize, and manage the interview data. Data analysis involved the constant comparative method in which data from each participant were continuously compared and contrasted. Throughout the analysis, written memos were used to capture decisions regarding the data and the emerging theoretical framework. Analysis began with open coding, a 'line-by-line' technique, where specific ideas or events were given a conceptual label or code (Corbin & Strauss, 1990; Strauss & Corbin, 1998). Using the constant comparison method, the codes were condensed and collapsed into major concepts (themes). Axial coding was then used to establish the properties and dimensions of the major concepts and explore relationships between these themes. In the last stage of coding, the theoretical linkages between concepts were modified and verified and a core concept was identified, which encompassed the major concepts into a cohesive theoretical framework. All themes were derived from the data and minor themes were included in the theoretical model. Authors AS and PC performed all coding. Authors AS, PC, and JA had regular meetings to review the transcripts, codes, and memos. Any discrepancies that arose in the analysis were discussed and resolved.

3 | RESULTS

3.1 | Participant characteristics

Sixteen participants consented to participate in the study. Two were lost to follow-up before their pGC appointment. Fourteen parents completed 13 semi-structured interviews (one mother and father participated together) over the telephone within one month of receiving pGC. All parents completed the telephone interviews from their home and there were no other individuals present during the interview besides the participants and researchers. The median length of the interviews was 50 min (range 19 to 88 min).

Table 1 provides detailed demographic information on the 14 parents who completed the study. All parents reported the highest level of education, having attended college or university. The majority of parents (n = 10, 71%) had a psychiatric diagnosis (depression, bipolar disorder, and/or anxiety) themselves and close relatives (first- and second-degree relatives) with a psychiatric disorder (n = 13, 93%). Three parents (21%) had a family history of schizophrenia. None of the parents in this study had a diagnosis of 22qDS.

3.2 | Index children characteristics

The majority of the 13 index children with 22qDS (7 weeks old to 18 years old at the time of the interview) of the parent participants were assigned male at birth (n = 11, 85%), attended regular (n = 6, 46%), or a special/alternative school (n = 4, 31%) and were diagnosed with a developmental or learning condition (n = 10, 77%). Only four children (31%) had a psychiatric diagnosis at the time of study. All four of these children were diagnosed with a neurodevelopment disorder (ADD, ADHD, or autism spectrum disorder) and/or a mood disorder (depression and/or anxiety). One child had a diagnosis of psychosis. The majority of children (n = 3, 75%) with psychiatric diagnoses were taking psychotropic medications at the time of the study.

TABLE 1 Participant Characteristics: parents of children with 22q11.2 deletion syndrome

Characteristics	Total (N = 14) (%)
Age	*Median:* 40 years (*Range:* 35 – 48)
Sex	
Female	12(86)
Male	2 (14)
Marital Status	
Single (incl. separated, divorced, and widowed)	3 (21)
Married/Partnered	13 (79)
Employment	
Paid work	13 (93)
Stay home parent	1 (7.1)
Race	
White	11 (79)
African	1 (7.1)
Mixed	2 (14)
Relationship to the child with 22q	
Biological Parent	13 (93)
Adoptive Parent	1 (7.1)
Country of Residence	
Canada	5 (36)
USA	9 (64)

4 | OVERVIEW OF THE 'AWARENESS TO ACT' THEORETICAL MODEL

Parents of children who have been diagnosed with 22qDS navigate along a continuum of increasing awareness about the psychiatric risks associated with their child's condition and the strategies they can use to protect and/or manage their child's MH (Figure 2). pGC facilitates parents' awareness, helping them achieve an 'awareness to act' to protect and/or manage their child's MH. The process through which parents developed awareness about the psychiatric aspects of 22qDS and the strategies to protect and/or manage their child's MH occurs over time: with parents initially lacking awareness, then building awareness, and ultimately achieving awareness to act following pGC.

Parents described numerous barriers along the trajectory of their awareness about psychiatric manifestations of 22qDS, including dealing with the immediate symptoms of 22qDS; missing MH guidance; their child's young age; and parental fear and stigma. These barriers stalled parent's awareness, which led to them carrying the burden of identifying emerging psychiatric symptoms and advocating for their child's MH. pGC helped parents achieve an 'awareness to act' to protect and/or manage their child's MH, whereby parents described being alert and feeling equipped to protect and/or manage their child's MH.

4.1 | Lacking awareness

Parents indicated that following their child's diagnosis with 22qDS they were initially unaware of the psychiatric risks associated with the condition, and of strategies that can be used to manage and/or protect their child's MH. Only a small number of parents—those whose child received care at a 22qDS specialist clinic—thought it was possible they learned about the psychiatric risks from a healthcare professional (HCP); although this information was limited to a numerical percentage risk with little if any other information, resources, or support.

> The [HCP from specialist 22qDS clinic] just walked … right into the room where my wife was… staying after giving birth and just sat down and said, "What do you want to know about this?" And we, we talked about it, and got a lot of information but I think the things that we were left wondering about the most were the MH issues, because nobody seemed to have very good answers, very good data on what works in terms of prevention, in terms of treatment, it just seemed like it was the kind of thing you just wait for it to happen and it's [either] going to happen or it's not going to happen.
>
> Participant 9, father of 7-week-old infant

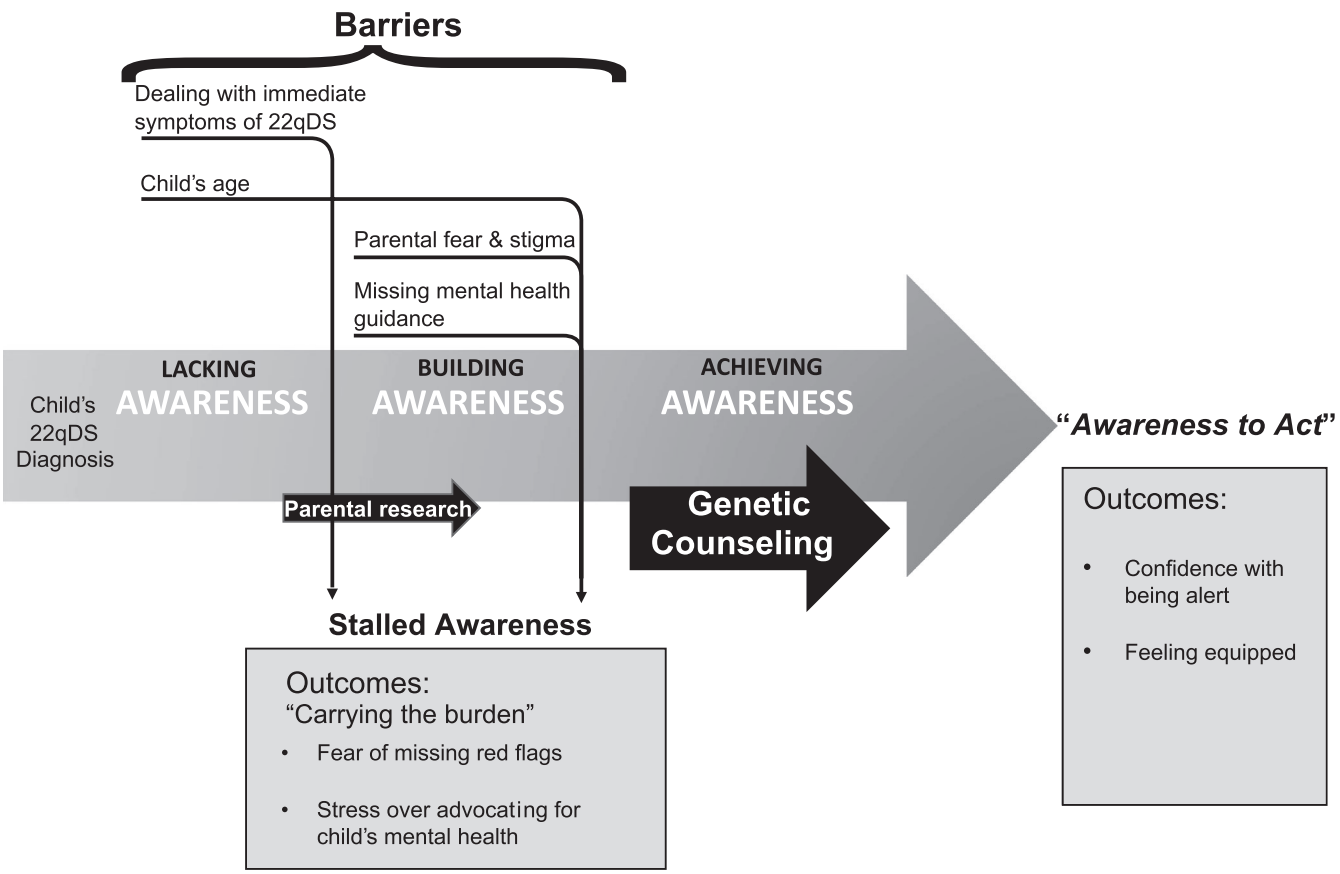

FIGURE 2 'Awareness to Act' Theoretical Model of psychiatric genetic counseling for parents of children with 22q11.2 Deletion Syndrome

4.2 | Building awareness

Parents described 'building awareness' of the psychiatric risks associated with 22qDS over time as their child aged. They described first learning about the psychiatric risks through their own initiative of doing research on the Internet, reading pamphlets and books, talking to other families, or attending community educational events. Despite this building awareness, parents were hesitant to act on this information. They expressed uncertainty, confusion, and fear about the risks, but did not reach out and engage any of the numerous HCP involved in their child's care to have a discussion about the psychiatric risks.

> I think someone said 'don't google this, it will scare you'. And so a couple of times I googled 22q looking for community kind of [things], like I wondered if there were any groups or special fundraisers, and inevitably it always pulls up stuff on schizophrenia so then I'd find myself reading reports and studies that show this increase, like 25% risk of schizophrenia, [and I'm] going "oh my God," and just kind of freaking out.
>
> Participant 8, mother of 4-year-old child with no psychiatric diagnoses

> Unfortunately when [my child] was born and they said DiGeorge to us, we went online and…a Canadian had just released a study … so all the buzz about 22q is about MH. And I'm like oh my god my baby is dying and if he doesn't he's going to be schizophrenic. That's what I thought, based on that. And I didn't have anybody to tell me no that's not what that means.
>
> Participant 13, mother of 13-year-old child with anxiety, depression and ADHD

4.3 | Barriers to parental awareness

Parents described numerous barriers along the trajectory of their developing awareness, including dealing with the immediate symptoms of 22qDS; missing MH guidance; the child's young age; and parental fear and stigma.

4.3.1 | Dealing with the immediate symptoms of 22qDS

Parents described that at the time of diagnosis their child's doctors focused on dealing with their child's most urgent physical health concern, which for the majority of children was a congenital heart defect. Parents explained HCPs then dealt with other pressing physical, cognitive, and developmental symptoms. Parents shared how they were encouraged by their HCPs to focus on issues happening *'now'* instead of worrying about issues that could occur later. Parents shared that this *'wait and see'* approach seemed appropriate and understandable as they themselves were primarily focused on their child's immediate health concerns. In fact, some parents described their conscious choice to deal only with their child's pressing health problems, especially because thinking about the *'180'* different symptoms of 22qDS their child possibly could have was simply overwhelming, suggesting that dealing with the immediate may initially have acted as a parental coping mechanism, but later became a barrier to their awareness.

> Initially I think we focused on the most immediate issues, which were his various heart issues, and then I'm going to say it felt very much like the immediate developmental milestones… they got us into the occupational therapy, speech therapy, physical therapy.
>
> Participant 3, mother of 16-year-old child with depression, anxiety and psychosis

> I guess in one sense it makes sense to not say "here's 180 things that could go wrong with a 22q person and we're checking for all of them" because that would be alarming.
>
> Participant 10, mother of 15-year-old child with autism

4.3.2 | Missing mental health guidance

Parents shared that once their child's immediate health concerns were stabilized, they still did not receive any guidance on their child's future MH. Despite numerous HCPs involved in their child's care, parents indicated they received from them little information, resources, or support on the psychiatric risks associated with 22qDS and strategies that can be used to manage and/or protect their child's MH. Parents further explained they did, however, receive direction on other physical, developmental, and/or cognitive issues associated with 22qDS that may arise for their child in the future. Consequently, parents expressed dissatisfaction and confusion as to why they were never initially *'given the heads up'* about the psychiatric risks, but were given guidance on how to monitor the physical, developmental, and/or cognitive risks of 22qDS in a proactive manner.

> [When] you get a diagnosis like this all of a sudden, you're like 'oh my gosh, [is my child] going to be able to learn to read past the second grade level?' It is such a devastating diagnosis initially [but] I did feel here with the support system I had at the children's hospital it was very clearly explained what some of the expectations were going to be for these children when it came to some of their physical abilities and of course the mental issues, not psychiatric though, but the cognitive and learning and speech [type of] things.
>
> Participant 3, mother of 16-year-old child with depression, anxiety and psychosis

> Like, yeah cause I don't feel like that was really strongly presented to me that there are preventative measures and that, I mean I'm sure there's nothing you can one hundred percent do to stop it but like, just that there are some things you can do. You don't just sit and wait to find out if you have [MI] or not.
>
> Participant 10 mother of 15-year-old child with autism

4.3.3 | Child's age

Parents speculated that a possible reason why their doctors did not explicitly discuss the psychiatric risk and strategies that can be used to manage and/or protect their child's MH was because their child was too young for this to be a relevant health issue. Parents commented on how doctors likely thought such a discussion was premature given that the psychiatric risks were more applicable to adolescents and young adults; and many parents agreed with this rationale. Parents further explained that they *'trusted'* that the doctors would discuss psychiatric risks when appropriate as they have done for other health concerns related to 22qDS.

> Not yet, with him being so young, I was looking for ages and warning signs and my intention was definitely to talk to a professional before his adolescence but I hadn't yet, at this point.
>
> Participant 7, mother of 4-year-old child with no psychiatric diagnoses

As their child aged, parents described dissatisfaction that HCPs did not proactively address the psychiatric risks. Yet when parents learned of the risks on their own accord, they did not bring this risk up with their doctor either. Some parents explained that the fact that their doctors had yet to address the risks reinforced their perception that their child was not yet at the age where MH concerns should be on their *'radar'* and instead they chose to consciously *'tuck away'* this knowledge for when their child was older and they deemed it more relevant.

> If you'd asked me I'd probably have said "nooo, [my daughter] is probably good to go." I don't think she's going to develop [MI] – but she could. So, you know, it was in the back, not the forefront, [of my mind] … to make sure that you keep checking.
>
> Participant 14, mother of 15-year-old child with no psychiatric diagnoses

> I think it's possible that if he was eighteen or nineteen and we were talking about mental health concerns; they might have that more on their radar.
>
> Participant 10, mother of 15-year-old child with autism

4.3.4 | Parental fear and stigma

Parents spoke of being aware of the social stigma about MI that exists and shared having their own fears and prejudices regarding MI. Parents often described MI as something *'scary'* or *'terrifying'*, with many parents viewing psychiatric illnesses as the worst symptoms associated with 22qDS. Some parents explained how their own fears and stigma toward MI may have led to denial, which could have acted as a barrier to their awareness of the psychiatric risks and consequently, their willingness to initiate a discussion with their child's HCP about this health concern.

> I felt worried and scared actually, because I don't know anyone with schizophrenia, so I just associate it with people hearing voices and being violent…"Oh my God, what if [my daughter] hurts [my son] when he's sleeping, or what if she tries to hurt us, what if she starts hearing voices, what are we going to do?" I just felt ill equipped; like for everything else I know how to be an advocate and I feel like I can handle the challenges, but with the schizophrenia, that was like I can't. I don't know how to do that. It was just overwhelming.
>
> Participant 8, mother of four-year-old child with no psychiatric diagnoses

> Normally you're scared of – you know these stereotypes, so what your limited knowledge is of it. And what you think his life would be like, right? So immediately you think of quality of life, you think of his socialization, you think of him working, you think of him interacting with other people, you think of him needing medication, you think of the supports he's going to need, you're thinking of how well he's going to be able to navigate through life independently, setbacks, all sorts of things.
>
> Participant 11, mother of 8-year-old child with ADHD

4.3.5 | 'Carrying the burden': Parental outcome of stalled awareness

As an outcome of stalled awareness about the psychiatric risks associated with 22qDS and protective strategies that can be used to manage and/or protect their child's MH (due to the aforementioned barriers), parents described having to *'carry the burden'*, expressing a fear over missing *'red flags'* of MI and the stress over *'advocating'* for their child's MH.

> I do think of it as a challenge I'm not beating. And it's a burden… it's like a chain around my neck because I'm not knowledgeable or smart enough to figure, to

tease those things out. I mean, I just don't have the skillset.

Participant 13, mother of 13-year-old child with anxiety, depression and ADHD

4.3.6 | Fear of missing red flags

Parents spoke of feeling a great sense of responsibility to identify early signs of psychiatric illness in their child. Parents shared that they tried to *'keep an eye out'* for symptoms that could be associated with MI, but had limited awareness about what these so called *'red flags'* may be. Thus, parents expressed fear around missing a sign and wished for guidance from their HCPs regarding the psychiatric risks and early signs of MI. This parental worry also extended to their need to assess the *significance* of a sign or symptom if one was noticed. Parents explained that they struggled to determine whether issues they noted were due to normal development, 22qDS, or early manifestations of MI. While many parents spoke of this self-imposed duty to notice warning signs, many did not consider themselves to be actively *'watching'* their child for signs of MI; instead, parents described it as something they think about in the *'back of their mind'*.

I just remember talking to the school psychologist saying "I feel like [my son] has this anxiety [about a specific circumstance], which is something that is completely out of his control" and it's been daily. And I remember her saying "oh we don't see any of that at school, and being 5 that could be a real natural fear" … I just remember saying "I don't see any harm in us addressing this […] like we're not going to hurt him by giving him coping strategies, and since we know he is at a higher risk for having anxiety, why don't we?

Participant 6, mother of 9-year-old child with no psychiatric diagnoses

4.3.7 | Stress over advocating for child's mental health

Another weight parents described carrying was the need to advocate for their child's MH if they identified a red flag or had a concern. Parents explained that they had to advocate for their child to be assessed or referred to specialists with the appropriate knowledge and experience. Parents expressed frustration and anger at having to *'push'* for referrals and/or assessments and expressed worry that their child would *'fall through the cracks'*. Having to advocate for their child's MH was a source of worry and stress for parents.

We definitely felt like his behaviours were much stronger than typical development. We didn't know what the cause was. And I think around that time we were asking to be assessed for autism as well. We just were like trying to just ask everybody for something because there was obviously something going on, so let's investigate.

Participant 11, mother of 8-year-old child with ADHD

'We really felt that this mental health stuff was more on us and still do. Absolutely.

Participant 13, mother of 13-year-old child with anxiety, depression and ADHD

4.4 | Achieving 'awareness to act': The role of pGC

Prior to pGC, most parents appeared stalled in their awareness process having only the information about the psychiatric risks associated with 22qDS that they had gathered from their own research and limited awareness about the protective strategies that can be used to manage and/or protect their child's MH. The results of this study suggest that pGC can play a significant role in facilitating parents' awareness, moving them further along the trajectory to achieving an *'awareness to act'*.

pGC appeared to be one of the first interactions parents had with a HCP regarding their child's psychiatric risks and management strategies unless their child had a MI diagnosis. Parents discussed how the MH *'jar'* analogy helped them make meaning of all of the *'facts'* they had become aware of regarding their child's psychiatric risks and gain a more cohesive understanding of their child's risks, the causes of MI in general, and the protective strategies that can be used to manage and/or protect their child's MH. pGC appeared to facilitate parents' achievement of an awareness that enabled them to act in ways to protect and/or manage their child's MH.

Well [pGC] changed [things] in the sense that it's brought [MH issues] to the surface again. And to be more aware of things that we're seeing and maybe not to ignore some things if we're seeing something. And to seek help if we need it because sometimes we sort of get caught up in our own daily routine and things like that.

Participant 11, mother of 8-year-old child with ADHD

I think it's important for people to know that knowledge is power and without [pGC] and the information that I've been able to gain here, people moving forward are not going to have complete knowledge. And therefore, not have the appropriate power to do the best for their child.

Participant 16, mother of 6-year-old child with no psychiatric diagnoses

5 | PARENTAL OUTCOMES OF AWARENESS TO ACT

As an outcome of achieving awareness to act about the psychiatric risks associated with 22qDS and protective strategies that can be used to manage and/or protect their child's MH, parents described '*being alert*' and '*feeling equipped*'.

5.1 | Confidence with being alert

With increased awareness parents described being consciously alert to signs or symptoms of MI in their child. They explained knowing what signs or symptoms to watch for and being more attentive to their child's behavior. Parents expressed feeling more confident in their ability to identify *'red flags'* and consequently reported feeling less worry and distress about their ability to notice any signs or symptoms should they arise.

> Trying to just accept him for who he is and he may have some setbacks and as a family we need to be able to manage that and cope with that and support him. And so [pGC] was just a reminder about that, that he's unique and he may have some different challenges but to be aware of that and always to be alert.
>
> Participant 11, mother of 8-year-old child with ADHD

> [My son] definitely struggles with anxiety, and he has a lot of fears… and I feel like we've launched strategies to try and work through those things… [while] trying to be alert for any new problems going on too.
>
> Participant 4, mother of 7-year-old child with no psychiatric diagnoses

5.2 | Feeling equipped

With increased awareness parents also discussed feeling more equipped to manage and/or protect their child's MH. Parent's felt more prepared with *'tools'* they could adopt to protect their child's MH such as learning different communication or behavioral strategies (i.e., ways to handle fears, and outbursts) and establishing a routine to help reduce stress associated with the unexpected events and/or transitions. They explained being more aware of the need to engage in protective strategies with their child and described this need as something that they carried in the forefront of their minds. Parents also expressed that the jar model was a helpful communication *'tool'* for them to explain *'what MH is'* to their children, which is a critical first step to facilitating their child's awareness and ultimately engagement in protective strategies for their own MH.

Parents also described feeling more equipped to access MH care should they become concerned about their child's MH. They explained *'making a plan'* about which HCP they would contact if they were worried about their child's MH (i.e., their pediatrician or family physician); and/or where they would go if they had an immediate concern about their child displaying symptoms of psychosis and/or if they posed harm to themselves or others (i.e., the ER). Parents also shared how they were more mindful of the importance of establishing a connection with MH services (i.e., psychologist or psychiatrist) even in the absence of any MH concerns so that these services would be in place and would provide a *'baseline'*. Parents talked about feeling equipped with the *'right language'* to be able to communicate with their doctors to ultimately articulate and advocate better for services for their children. Feeling more equipped helped alleviate parents' worries and empowered them in their ability to manage and/or protect their child's MH.

> The [pGC] that I recently had with the [genetic counselor] gave me better tools and made me less anxious. You know, it kind of made me feel like okay, even if he does get schizophrenia, which was always my biggest concern, I felt better prepared to help him as much as possible.
>
> Participant 7, mother of 4-year-old child with no psychiatric diagnoses

> I think [people who don't get pGC] are missing out on a lot of useful information; a lot of preventative things that they can do, and so their kids' health might suffer for longer than it has to because they might not be aware that there's services and there's options that [my daughter] can access and there are tools and ways to help with psychiatric disorders, that it's not hopeless, that they can do something about it, and that they can be okay.
>
> Participant 8, mother of 4-year-old child with no psychiatric diagnoses

> [pGC] allowed me to articulate better. Like what I was trying to ask for. Like I was already asking for it and I was already explaining what I needed but I think that there's always like a more educated professional than myself that gives me the right language and I feel like once I have that language it's like a key that you just unlock a door.
>
> Participant 13, mother of 13-year-old child with anxiety, depression and ADHD

6 | DISCUSSION

This is the first study to explore outcomes and perceived value of pGC with respect to needs of parents of children with 22qDS. The Awareness to Act theoretical framework generated shows that parents experience numerous barriers, which limit their awareness

of the psychiatric risks associated with their child's condition and protective strategies for MH. International clinical practice guidelines for 22qDS recommend regular psychiatric assessment, as early intervention for psychiatric conditions led to the best long-term outcomes (Bassett et al., 2011); yet previous research shows psychiatric services are underused by individuals with 22qDS (Young et al., 2011, Tang et al., 2014), and parents typically first learned of the psychiatric risks associated with 22qDS from non-medical sources (Hercher & Bruenner, 2008), with the internet most often being the first source of information (van den Bree et al., 2013). Our study supports these findings; parents did not learn about the psychiatric risks associated with 22qDS from a HCP involved in their child's care and received little information, resources, or support regarding MH. With limited awareness of the psychiatric risks and protective strategies, parents are left carrying a burden, fearing that they may miss symptoms of psychiatric illness and experiencing stress over advocating for their child's MH. Considering that high levels of anxiety in children with 22qDS have been suggested to be a predictive factor for transition to psychosis (Gothelf et al., 2013), this further underscores the role for pGC to equip parents with an awareness of how to engage in protective MH strategies to help reduce stress for their child with 22qDS. Thus, parents perceived pGC as a helpful clinical intervention, facilitating their awareness so that they could act in ways to protect and/or manage their child's MH.

Although parents acknowledged there is good support around the physical, developmental, and cognitive issues their child may experience as a consequence of 22qDS, parents felt they missed guidance from their HCPs on MH. Previous studies have reported psychiatric disorders are discussed less often than the other features of 22qDS in a medical genetics setting (Morris et al., 2013; Martin et al., 2012; Talcott-Baughman et al., 2015). Barriers, such as HCPs dealing with immediate symptoms, combined with the child's young age, provide potential explanations for the gap in MH information, resource, and support. Furthermore, a previous survey study of medical geneticists that identified that psychiatric disorders were discussed less often than the other features of 22qDS when the geneticists had higher levels of stigma toward psychiatric disorders (Morris et al., 2013). Future research involving qualitative interviews with HCPs would be useful to further explore reasons for this missing MH guidance.

Parents' fear and stigma surrounding MI acted as a barrier to their awareness and prevented them from reaching out to HCPs about the psychiatric risk. Recent research on patient's perspectives of the process and outcomes of pGC indicated that people with MI experience less guilt, shame, and blame regarding the cause and management of their MI following pGC and were empowered to act in ways to protect their MH (Semaka & Austin, 2019). Similarly, parents in our study shared that pGC heightened their awareness, leading them to feel more alert and equipped, which alleviated their fears and empowered them in their ability to identify, manage, and/or protect their child's MH.

Our study shows that for our participants, psychiatric genetic counselors were the first HCPs to explicitly address the psychiatric risks associated with 22qDS, and provide information, resources, and support, which allows parents to be proactive regarding their child's MH. Therefore, the provision of pGC could potentially play a key role in closing the aforementioned gap in MH guidance for these families. A systematic review of the psychosocial impact of 22qDS on patients and families showed parents thought HCPs lacked knowledge and empathy around 22qDS (Vo et al., 2017) and expressed frustration for having to fight to receive MH information, resources, and support for their child (Cohen et al., 2017; Goodwin & Mccormack, 2017). Our study data echo these findings and support the inclusion of pGC in the clinical plan for medical genetics follow-up after the initial diagnosis of 22qDS.

6.1 | Research and practice implications

This study provides evidence suggesting that pGC leads to positive outcomes for parents of children with 22qDS through increasing their awareness of the associated psychiatric risks and MH concerns, thereby helping parents feel more alert to potential emerging signs or early symptoms of MI and equipped to manage and protect the MH of their child. At the time of this study, there was no published clinical protocol outlining the genetic counseling for families affected by 22qDS (Hart et al., 2016). The findings of this research could help inform the development of a clinical protocol, highlighting the role of pGC to ensure that families affected by 22qDS are informed about the psychiatric risks associated with 22qDS and provided MH guidance and support.

The interpretation of these study findings should take into consideration some of the limitations of this research. We did not conduct a baseline interview prior to pGC; therefore, parents were recalling their experiences of learning about the psychiatric risks associated with 22qDS prior to them commenting on what information, support, and resources were provided to them through pGC. Furthermore, although one month post-pGC parents endorsed that pGC facilitated for them an awareness to act in a proactive manner to protect their child's MH, we were unable to explore whether parents acted in this manner beyond this period of time. The two parents from our study who participated in member check interviews, five months and three years post-pGC, both continued to express that pGC was pivotal for them in instilling a proactive approach to protecting the MH of their child with 22qDS. Future research should explore in greater detail parents' actions regarding their child's MH following pGC.

Another important consideration is that the majority of parents in this study were highly educated, employed, and white; thus, the perceived experience of pGC for parents facing socioeconomic and racial disparities may be different. Additionally, participants in this study were comprised of a self-selected group of mostly mothers (n = 12) of children with 22qDS. One of the two fathers who had participated in this study was present during the same interview with the mother of the child with 22qDS. For this reason, it is possible that the Awareness to Act theoretical model of

pGC may not be generalizable to fathers (n = 2) of children with 22qDS. Interviews with a larger number of parents to include parents from more diverse ethnic and socioeconomic backgrounds, more perspectives from fathers, and cover a greater range of ages of children with 22qDS would also be helpful in confirming this theoretic model.

7 | CONCLUSION

The findings of our research demonstrate that pGC facilitates for parents an 'awareness to act' on the psychiatric risks associated with 22qDS, which enables them to feel prepared to act in ways to protect and/or manage their child's MH. As such, pGC was perceived by parents as a valuable clinical intervention in promoting their sense of agency in reducing the risk of psychiatric problems and improving outcomes for their child with 22qDS.

AUTHOR CONTRIBUTIONS

PC, AS, and JA contributed substantially to the study concept and design. RB contributed substantially to the data acquisition. PC and AS drafted the initial version of the manuscript. PC, AS, RB, and JA had access to all data in the study and take responsibility for the integrity and accuracy of the data analysis. All authors contributed to data interpretation and revising the work critically for important intellectual content. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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COMPLIANCE WITH ETHICAL STANDARDS

CONFLICT OF INTEREST

PC, AS, RB, CS, AI, MM, and JA declare no conflict of interests.

HUMAN STUDIES AND INFORMED CONSENT

Institutional Review Board approval was received from the UBC and Children's & Women's Hospital Research Ethics Boards (H14-00256). 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 prior to their inclusion in the study.

ANIMAL STUDIES

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

DATA SHARING AND DATA ACCESSIBILITY

The data that support the findings of this study are available on request from the corresponding author. The data are not publically available due to privacy or ethical restrictions.

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SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section.

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