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Assessment of Parental Disclosure of a 22q11.2 Deletion Syndrome Diagnosis and Implications for Clinicians

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Abstract

Most children with chromosome 22q11.2 deletion syndrome (22q11DS) have an IQ in the range that may allow them to be capable of understanding a genetic diagnosis despite mild intellectual disabilities. However, there are no publications that relate to the disclosure of a 22q11DS diagnosis to the affected child, or the factors that influence parents' disclosure to the child. A pilot study was conducted including eight semi-structured interviews with caregivers of children with 22q11DS, 10 to 17 years of age, to investigate the factors that influence how parents inform their children of the diagnosis. Six of eight participants had disclosed the diagnosis to the child, and most of these parents felt they could have benefited from additional advice from professionals to increase their confidence and success, as well as the child's comprehension of the information. Those who had not informed the child were uncertain about the words to use, how to initiate the conversation, or were concerned about the child's level of understanding. Our results demonstrate that genetics professionals should help prepare caregivers for conversations with their children about the diagnosis of 22q11DS, monitor the understanding of the diagnosis over time, and provide ongoing support.

Keywords

22q11.2 deletion syndrome; Velocardiofacial syndrome; DiGeorge syndrome; information sharing; family communication; genetic counseling; qualitative research

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Introduction

A microdeletion of band q11.2 on chromosome 22 is responsible for over 180 clinical features and is one of the most common multiple anomaly syndromes found in humans, with an incidence of about 1 in 2,000 live births (Shprintzen 2008). There are multiple names for this syndrome, including 22q11.2 deletion syndrome (22q11DS), Velocardiofacial syndrome (VCFS), and DiGeorge syndrome, and the majority of cases (greater than 90%) are *de novo* (reviewed in Shprintzen 2008). Congenital heart defects, palatal anomalies, hypocalcaemia, immune deficiency disorders, learning difficulties, and speech/language delays are some of the more commonly seen clinical features (Bales et al. 2010a; Shprintzen 1978; Shprintzen 1981). Also observed are behavioral, developmental, and psychiatric disorders (reviewed in Shprintzen 2008).

Developmental delays and cognitive deficits are highly prevalent (80–100%) in children with 22q11DS, with the mean IQ being 75. In addition, behavioral problems, poor social skills, language impairments, and speech problems lead to poor academic performance (Gerdes et al. 2001; Lewandowski et al. 2007; McDonald-McGinn & Zackai 2008; Moss et al. 1999; Shashi et al. 2011; Sobin et al. 2005; Swillen et al. 1997; Woodin et al. 2001). Individual assessment and development of educational and therapeutic support programs are recommended as early as possible to help the child reach his potential (Gerdes et al. 2001).

To date there are no studies pertaining to disclosure of a diagnosis of 22q11DS to the affected child by the caregivers. We examined the literature on disclosure of other genetic conditions such as Cystic Fibrosis, carriers of Duchenne Muscular Dystrophy, Familial Adenomatous Polyposis, carriers of Fragile X syndrome, Hemoglobinopathies, Huntington's Disease, Neurofibromatosis, and risk of hereditary cancers to determine if there were themes of communication that would enable us to frame a study to ascertain factors that determined disclosure (or not) of a diagnosis of 22q11DS to an affected child by a parent. Although many of the above conditions affect children, they are dissimilar to 22q11DS since they involve children whose cognition would be expected to be much higher than children with 22q11DS, as well as children who may not be affected, but are carriers (Forrest et al. 2008; Gallo et al. 2005; Gallo et al. 2009b; Gaff et al. 2007; McConkie-Rosell et al. 2009; Metcalfe et al. 2008; Metcalfe et al. 2011; Plumridge et al. 2011; Tercyak et al. 2002). Nonetheless, the findings in these studies can be summarized as follows: the child/young adult being informed wanted the person informing them to be knowledgeable (McConkie-Rosell et al. 2009); parents had differing opinions about the optimal age of the child at which genetic information could be shared (McConkie-Rosell et al. 2002); the decision to share was based on parental assessment of the child's developmental stage, readiness, and interest (Gallo et al. 2005; Metcalfe et al. 2008); and it was recommended that health care professionals provide parents information regarding the genetic aspects of their child's condition, support so they can feel confident about discussing this with the child, and check parents' understanding throughout the child's development to ensure accuracy of information (Gallo et al. 2009a).

There are many implications to consider in the disclosure of a genetic diagnosis to a child. Genetic testing and self-awareness of their diagnosis has the potential to alter parent-child bonding, incite a change in self-concept, and cause heightened anxiety, especially if the child is too young or not ready to comprehend the information, and learning that one is affected or is a carrier can alter the child's cognitive and psychosocial development (Fanos 1997). However, if information is withheld from an affected child, there is a risk of creating an environment of secrecy, and a sense of mistrust when the child is informed (Fanos 1997). Informing the child sooner rather than later allows for time to adjust to the knowledge before considering how it might affect their own children (McConkie-Rosell et al. 2002), and

disclosure at a younger age may also allow for the development of more realistic expectations for the child regarding developmental, behavior, or learning delays (Blomquist et al. 1998). Despite the above findings, little is known about the effects of growing up with knowledge of a genetic diagnosis (McConkie-Rosell & Spiridigliozzi 2004).

Furthermore, it is not uncommon for individuals with genetic conditions to also experience behavioral or psychiatric disorders (reviewed in Dykens 2000), which may present additional challenges to the disclosure process. Anxiety, difficulties maintaining attention, perseverative thought patterns, and deficits in social skills are common in children with 22q11DS (Swillen 1997; Swillen 1999; Woodin 2001; Fine 2005; Niklasson 2005). Approximately 20–40% of adolescents and young adults with 22q11DS will develop a major psychiatric illness, most commonly schizophrenia but also bipolar disorder, severe depression, and schizoaffective disorder (Shprintzen 1992; Papolos 1996; Bassett 1998), and parents report this to be their single greatest source of anxiety (Hercher 2008). Parents may fear of the impact of disclosure as an additional environmental stressor on the future development of psychiatric problems.

The diagnosis of any genetic condition in a child can result in families seeking guidance from genetic counselors and other health care professionals for assistance in how to disclose the information to the child. Previous research regarding the complex topic of genetic testing in children has demonstrated that successful genetic counseling requires active partnering between the family and the professional to achieve a common goal: positive adaptation of the child to the genetic information (McConkie-Rosell & Spiridigliozzi 2004). Consideration of how the child may view the information is critical, and the child should be given the chance to respond and have his or her concerns addressed (McConkie-Rosell & Spiridigliozzi 2004). Although we derive knowledge about genetic diagnosis disclosure from other genetic diagnoses where there are published data, as summarized above, there is a need for empiric data on 22q11DS. Although children with this condition have neurocognitive impairments, fifty percent have an intellect in the borderline to normal range, potentially making many of them capable of understanding their genetic diagnosis and its implications. However, many suffer from numerous psychological and psychiatric problems as described above, making it more complex for parents to convey the information and to monitor the consequences of this on their child's psychological well-being. Thus, reports investigating communication of information related to other genetic conditions may be helpful, but do not fit the needs of families with children diagnosed with 22q11DS, making further research essential.

The current study aimed to gain insight into factors related to the decisions of caregivers in disclosing a diagnosis of 22q11DS to their child by interviewing caregivers about their experience. It was hypothesized that the timing and extent of disclosure would be related to the cognitive functioning and developmental age of the child, as well as the caregiver's level of confidence about the information to be shared. Due to the lack of literature available on how parents decide to disclose their child's diagnosis of 22q11DS to family and friends, a secondary objective was to study the factors that influenced how parents shared the diagnosis with individuals other than the other parent.

Methods

Participants

The participants of this study were primary caregivers of eight children between 10 and 17 years of age diagnosed with 22q11DS who are part of a larger longitudinal study currently in progress at Duke University Medical Center that examines risk for psychosis in these children. Background information such as general information about their child's condition,

health care treatment, and strategies for progress and development, was obtained. Demographic data, including current age, age at diagnosis and disclosure, gender and ethnicity were also obtained.

Instrumentation

An interview guide was developed to examine the caregiver's experience of informing others and their child of the 22q11DS diagnosis. This guide was created after reviewing relevant literature and previously developed guides from related studies (Gallo et al. 2001). The guide investigated caregiver decisions regarding how and when to inform the child and others of the diagnosis. A semi-structured interview format was chosen using qualitative interviews with fairly specific questions that have been previously developed (Berg 2001). The guide was refined after each interview to adjust for clarity and appropriate content. IQ information was gathered from the parents' report as part of the interview, and verified by the authors.

Procedures

This study was approved by the institutional review board (IRB) at the University of North Carolina at Greensboro and Duke University Medical Center. Eight families were contacted by phone and offered participation in a pilot study examining parental disclosure of a 22q11DS diagnosis; all agreed to participate. Interviews were completed either in person or by phone, recorded, and later transcribed. Transcription of the interviews allowed for coding of themes and content analysis.

Data Analysis

Demographics were analyzed using frequencies and descriptive statistics. Qualitative data were analyzed using the Colaizzi method for phenomenology to determine major themes (Colaizzi 1978). Phenomenology is a method of data analysis that aims to produce a description of the nature of the reality as seen through an individual's experience (Priest 2002). It was concluded this was the most appropriate method of data analysis due to the limited amount of previous research available on this topic, and the limited sample size in this study.

Categories and themes were formed during analysis of the data obtained through each interview. The steps of data analysis included reading each transcript multiple times, extracting significant statements, formulating general meanings, and organizing these meanings into theoretical clusters (Sanders 2003). Two independent members of the study team analyzed the transcripts for themes. Quotes representative of each theme were selected and reported to demonstrate participant experience.

Results

Table I reflects age at which children were diagnosed, age at which they were first informed of their diagnosis, and age at the time of the interview. In regard to school-related information, six out of eight participants (75%) stated their child was currently placed in a mainstream classroom, and seven out of eight (87.5%) stated the use of an individualized education plan (IEP). The IQ range of the children as reported by the participants, and verified by the authors through the larger study in which they participate, was from 60–105. One mother did not recall her child's IQ. Most children (7/8) were followed regularly by a geneticist and at least one other subspecialist. Regarding demographics of the interviewees, all were female, the majority were Non-Hispanic Caucasian (6/8 or 75%), and the majority had at least some college education (6/8 or 75%).

Sharing Information about the Diagnosis with Family and Friends

Questions about sharing information related to 22q11DS were separated into two time frames: when the diagnosis was first made, and the time after this initial period. When asked who they told at the initial time of diagnosis, all participants reported disclosing the diagnosis and sharing information about 22q11DS with family members, including grandparents, aunts and uncles of the child. The children of four participants (50%) were school-age at the time of diagnosis, while the other four were diagnosed at birth or in infancy. All of the participants whose children were school-age at diagnosis mentioned sharing information with the child's school and teachers at that time. Three participants (37.5%) said they told close friends about the diagnosis, and one recalled sharing the information with her church family.

In the time since diagnosis, all participants reported telling teachers at the beginning of each school year, as well as other individuals who care for the child, such as Scout leaders and summer camp staff. Additionally, some participants noted informing their supervisor or co-workers to explain why they may request time to attend medical appointments or other meetings related to their child, or need to answer a personal cell phone in case of an emergency. Participants took different perspectives on who to tell, and cited various reasons for why they decided to tell particular people about the diagnosis (Table II). Themes reflected participants' concerns that people may view the child differently when they are made aware of the diagnosis, as well as their desire for others to learn about 22q11DS to increase their understanding of the child.

Participants were also asked to rate how emotionally difficult it was to share their child's diagnosis with others by placing their feeling on a Likert scale from 1 to 7. Responses are shown in Table I. The median Likert score was 2, with a mean of 3.143. Participants did not report a high degree of emotional difficulty. Participants provided comments explaining why they gave a low or high rating for emotional difficulty of sharing the diagnosis with others (Table II).

Sharing Information about the Diagnosis with Children

The next set of questions addressed the participant experiences of sharing information with the affected child. Six out of eight participants (75%) had disclosed the diagnosis of 22q11DS to the child at the time of the interview. When asked what they had told their children, participants described their experiences as reported in Table II. Of the two who had not disclosed the diagnosis, one family reported regularly talking with their child about the medical aspects of the condition, but did not use any technical labels (such as 22q, VCFS, or DiGeorge syndrome). This participant commented that she feels the child would not understand those words, but said she sometimes tries to explain in more general terms. At the time of the interview, the child was 13 years old with an IQ of 64. In the other family who had not yet informed the child of the diagnosis, the child was 15 years of age with an IQ of 60. When asked about reasons for deciding not to share the diagnosis information for now, the participant mentioned significant uncertainty about what to say to the child.

In the six families where the child had been informed, the age of first disclosure ranged from 7 to 12 years, with a mean of 9.6 years. IQs in this group ranged from 71–105. Some participants said that the child knew all along, but only began to understand the diagnosis after a certain point when they could start to comprehend the information and its significance. In half of the families, the participant (mother) was the one to initiate the discussions, while the other half of participants reported that the child brought up the topic through questions. All six participants who had disclosed the diagnosis to the child stated that disclosure occurred over several conversations rather than in one larger discussion.

Participants reported that conversations occurred most often when children were struggling with school or on the way to medical appointments.

Motivations and Barriers: Why, When, and What to Say

When asked about their reasons for disclosing the diagnosis to the child, participants expressed various motivations. These ideas included the need to explain things to the child, such as why doctor visits were necessary, or why he or she was having a difficult time in school. Additionally, participants felt it would be important for the child to be aware of his or her own differences and limitations, wanted their child to develop responsibility, and in the future, be able to take care of oneself, go to school, or hold a job. Lastly, participants wanted the child to be aware that the diagnosis was not something to hide or keep secret, and most noted an open communication style. (Table II)

On the other hand, participants stressed that they didn't want the child to focus on the diagnosis or use it as an excuse somehow. Participants also wanted to be certain they could share the information with the child in a way that would be beneficial, but without "say[ing] anything that's going to scare [child], or make [him/her] feel different." In this way, many participants expressed uncertainty about words to use when talking with their children about the diagnosis, both in families that have disclosed the diagnosis and those that have not. (Table II)

All participants, including those who had not yet disclosed the diagnosis to the child, were asked to rate how well-equipped they felt to have these types of conversations with their children, using a Likert scale from 1 to 7, with 1 being not well-equipped at all and 7 being very well-equipped (Table I). The resulting mean was 4.875, with a mode of 5, indicating that parents felt reasonably well-prepared to discuss the diagnosis, but may need additional resources to increase their confidence.

When asked if they had ever received advice about how to tell or talk with the child about 22q11DS, six out of eight participants (75%) responded that they had not. The six participants who did not receive advice from health care professionals included the four who had already disclosed the diagnosis to the child and both parents who had yet to disclose. One participant, who had not yet disclosed the diagnosis to the child, wished she had received more guidance in this area and expressed her frustration (Table II). Another participant, who had already disclosed, recalled receiving valuable advice and described something she found helpful (Table II). When participants were asked about any additional sources of information they found useful for helping the child understand 22q11DS, no sources of this type were reported. Most participants said they had talked with their children using their own strategy, while actively gauging how the child's understanding had changed with age (Table II). Lastly, when asked about the age at which they believe a child should be told about his or her diagnosis of 22q11DS, the majority of participants (7/8 or 87.5%) mentioned that it depended on when the child could comprehend the information, but this was not necessarily at a specific age (Table II). Participants expressed that each child may be ready at a different age, as each child seems to have a different level of understanding, and that "Sharing information with them has to be just according to the child."

Discussion

In the current study it was hypothesized that the timing and extent of disclosure of a 22q11DS diagnosis to an affected child by the caregiver would be related to the cognitive functioning and developmental age of the child, as well as the level of confidence felt by the caregiver in the information to be shared. The data from this study are consistent with our hypotheses because participants reported taking into account the child's ability to understand

and comprehend the information when making decisions regarding disclosure, and additionally those who had not yet disclosed the diagnosis felt ill-equipped to have this type of conversation with the child. Notably, both participants who had not disclosed the diagnosis to the child had not received any guidance regarding this by health care professionals. By interviewing caregivers regarding this topic, it was possible to gain insight into their experiences, and consider how professionals might be able to assist with and encourage well-equipped, successful disclosure of a 22q11DS diagnosis in the future.

Sharing Information about the Diagnosis with Family and Friends

There is no literature currently available on how parents decide to disclose their child's diagnosis of 22q11DS to family and friends. In this study, all participants shared information with their family members at the time of diagnosis, while close friends, teachers, supervisors and co-workers were informed later in some cases. Participants whose children were school-aged at diagnosis reported disclosing to the child's school and teachers at that time. Generally there was a low degree of emotional difficulty in disclosing a 22q11DS diagnosis to these individuals, possibly because they were already aware of the child's learning, social, or behavioral difficulties, and informing those in an educational role about the diagnosis provided an explanation and an avenue for modifying educational strategies. Parents expressed concern that their child would be viewed differently, but ultimately felt that sharing information related to the diagnosis would help others to understand their child. Overall there appeared to be few barriers related to disclosing a diagnosis to family, friends, and others who care for the child.

Sharing Information about a Diagnosis of 22q11DS with the Affected Child

In the past (with other genetic conditions) it has been found that parents consider *when* to share information, *what* the child needs to know, and *how much* they feel the child can handle (Metcalfe et al. 2008). Participants in the current study had similar attitudes and took these considerations into account: *how* to talk to their children and *what* to say when talking about the diagnosis of 22q11DS. Metcalfe et al. (2008) found the emphasis of open communication to be a motivating factor for sharing information, and this was also noted by participants in the current study. However, in our study, about half of families waited for the child to ask questions before discussing the diagnosis. Some participants stated that they were always willing to answer the child's questions, but less comfortable initiating the discussion. This could reflect a strategy of waiting for the child to indicate readiness to learn more (McConkie-Rosell et al. 2009).

Parents in our study also reported that disclosure occurred in stages; since many children with 22q11DS experience difficulties in comprehension, information delivered over several occasions with repetition could be more efficacious than one discussion, a fact well-known to the parents based on their day-to-day experiences with their child. The age of the child was an important factor in disclosure, with the median age being 9.6 years. All the parents in this study whose children were diagnosed after the newborn or early childhood periods (n=4) informed their child of the diagnosis at the time it was made. Although we did not directly ascertain information regarding this, we postulate the following reasons for this high rate of disclosure. These children were less severely cognitively impaired and therefore more capable of understanding the diagnosis at that time, borne out by the IQ range in these children of 75–105. Additionally, the events leading up to the family's learning about the diagnosis (i.e. evaluations by medical genetics, blood draw for laboratory testing) during a time the child was capable of understanding provided an opportunity to initiate the discussion with the child, whereas parents who learn of the diagnosis at birth are often managing more urgent medical concerns and may have more difficulty initiating the discussion as the child develops and matures.

Although the two individuals with whom the diagnosis had not been shared had the lowest IQ scores, parents of children with higher IQ scores also reported difficulty sharing the diagnosis. The mother of a child with an IQ score of 103, an average score for a typically developing child, reported the highest level of emotional difficulty and felt ill-equipped for the discussion. The two individuals who did not know their diagnosis yet were also female, but with a small sample size it is difficult to draw conclusions about this, and thus further research is needed.

Although our sample size is small, our survey suggests that parental uncertainty of terminology to use and how to deliver the information were important deterrents to disclosure of diagnosis to the child. Some participants in our study were hesitant about talking with the child because they did not want to scare them or were afraid they would not understand. This reflects a goal of trying to protect the child from potentially painful or threatening information, often provided in uncertain, indirect, and highly complex conversations (McConkie-Rosell 2009). Interestingly, none of the parents cited fear of increasing stress on the child's mental health state as a barrier to disclosure.

The motivating factors for parents in this study to share information with the child were to work towards the goals of coping, responsibility, independence, and self-sufficiency. These are issues of transition for all individuals but are more relevant to children with 22q11DS, due to the medical and psychological problems with which they grapple. Talking with the child about his or her diagnosis, and related strengths and challenges, is a major step for transitioning to independence. Relatedly, providing parents and caregivers with the specific vocabulary rather than general descriptors will be useful in that process. As explained by Reiss and Gibson (2002), adolescents require parental support to attain independent health and social behaviors, and parents, in turn, may require the support of health care providers to aid in negotiating boundaries, setting goals, and developing these skills. Other factors that influenced the parent to share the diagnosis included being able to provide the child with an explanation for their differences and to not hide the diagnosis from their child. Interestingly, none of the parents mentioned recurrence risks of the deletion as a factor that influenced disclosure.

Practice Implications

Genetic counselors can help with the process of disclosure by aiding parents in their discussion of the diagnosis of 22q11DS with the child, through review of genetic concepts, current issues, and future concerns in a manner sensitive to each family. Health care professionals can encourage discussion about how to share information, and parents can allow the child to be involved in clinic visits so that professionals can be involved in disclosing genetic information (Gallo et al. 2009b). Guidelines for disclosure of a diagnosis of 22q11DS to the affected child have not been published. Both the participants who had not disclosed the diagnosis to the child and the 4/6 who had disclosed reported not having received any guidance from health care professionals. Thus the majority of participants had received no guidance. This finding has been noted previously with other genetic diagnoses (Gallo et al. 2005; Metcalfe et al. 2008). While some families may feel that they are generally knowledgeable and equipped to discuss the diagnosis, advice from health care professionals may boost their confidence and aid in their success. There are published resources available which address the unique challenges of genetic counseling for adults with intellectual disabilities, and many of the suggested approaches may be applied to children (Finucane 1998, 2010). For example, discussion of probability and risk should be minimized, using the child's preferred term for his disability should be considered, and the use of abstract concepts or analogies should be avoided.

Based on our preliminary findings, we offer further guidelines for genetic counselors and points to consider. Since the initiation of the discussion about 22q11DS is difficult for some parents, providing anticipatory guidance before the child is developmentally ready for the discussion may give them time to prepare and increase their confidence level. Professionals can help caregivers learn how to say things in ways that will be most successfully understood by the child (Metcalfe et al. 2008) and, as suggested by the participants in the current study, they can also help by sharing their experiences of what has worked well in the past for other caregivers. They can suggest “the words to use” for parents who are struggling with finding appropriate language, or offer to role-play this conversation with the parents.

Since the increased risk of psychotic illness is shown to be the greatest source of anxiety for parents of children with 22q11DS (Hercher 2008), it is possible that this parental anxiety is an additional barrier to disclosing the diagnosis, but was not detected due to the limited nature of this study. Genetic counselors may engage parents in a discussion of the possible positive and negative consequences of disclosure upon their child’s mental health status. Ongoing and supportive discussion of the diagnosis with the child may increase his awareness of the possibility of mental health problems, and with this knowledge the child may report symptoms earlier if they arise. Based on the child’s behavioral history, families may also be encouraged to pre-arrange supportive therapeutic services in the event that the discussion incites a negative reaction, such as provoking anxiety or damaging self-image.

Finally, some participants included talking to other families of children with 22q11DS as a valuable source of guidance regarding what has worked for them; professionals can facilitate this through support groups and enabling contact with other families with a child with 22q11DS. Thus, genetic counselors have an important opportunity to help families with this difficult task of diagnosis disclosure.

Study Limitations/Strengths and Future Directions

This pilot study assessed parental disclosure of a 22q11.2 deletion syndrome diagnosis and was a qualitative study to learn about participants’ experiences. The limitations were that the study included a small sample size and thus it is difficult to apply conclusions to a larger population of individuals caring for children with 22q11DS. The interviewees are not representative of all families of children with 22q11DS. All of the participants interviewed in this study were mothers, either biological or adoptive, of children diagnosed with 22q11DS; the majority were Non-Hispanic Caucasion; the majority had at least some college education; and none were diagnosed with 22q11DS themselves (in the wider population, 1 in 10 affected individuals will have an affected parent). The challenges faced by an individual with 22q11DS (who likely has cognitive limitations) disclosing the diagnosis to an affected child are greater than those discussed in this manuscript. These families will likely need additional support from counselors during the disclosure process. In addition, questions asked participants to remember specific information and feelings that may have occurred many years prior to the interview, so there may have been recall bias associated with the responses. Also participants may have been more likely to respond in ways they perceived the authors would prefer on particular questions, such as the emotional difficulty rating.

The strengths are that this is the first examination of diagnosis disclosure to children with 22q11DS, and so provided valuable insight on influencing factors. None of the children with 22q11DS had a major psychiatric illness by the time of these interviews, so the parents’ feelings are not influenced by the effects of psychiatric illness. The interview guide developed as part of this study may be used as a framework for collecting information for future studies in this area.

One of the initial challenges we faced when approaching this study was the lack of an appropriate conceptual model to guide our exploration of 22q11DS diagnosis disclosure, since the typical intellect of these children is in the borderline range and many models, such as the concept of open communication discussed previously, presume typically developing children. Although Gallo (2001; 2005; 2009a; 2009b; 2010) has significantly contributed to the literature regarding how families share information with children about genetic conditions and employs the family management style framework (FMSF) model, this does not sufficiently address the unique challenges faced by parents of children with 22q11DS. Parents and caregivers of children with 22q11DS have the need to communicate in a method suitable to a child with cognitive disabilities. They are also trying to balance the desire to share information with their child, with the fear of causing more psychological harm. Since none of the currently existing models appear to fit this population well, these were not tested in our study.

This current study could be expanded upon by increasing sample size, as well as the use of a quantitative study design based on these qualitative data. Additional research including the perspectives of fathers, siblings, or other extended family members would be valuable to the field. The development of materials to help guide parents in disclosure of a diagnosis to their child, along with more child-oriented educational materials would be important. Since this study, the authors have partnered with the local North Carolina 22q11DS support group to begin development of these materials. The first booklet “Growing up with 22q,” designed to aid parents in initiating the disclosure discussion, is currently in print (Schoch 2012). Finally, investigating children’s understanding of 22q11DS, the impact of that information, and who they choose to share this information with would also be of interest, as well as studies more specifically addressing how parents or caregivers talk to children about the psychiatric issues related to 22q11DS.

Conclusion

No prior studies exist regarding disclosure of a 22q11DS diagnosis to the affected child, the variables that may influence this process, and the support caregivers may require for successful disclosure. The interview guide developed for this study allowed insights into participant experiences: parents had little difficulty in sharing the diagnosis with family and friends, but more challenges in disclosing the diagnosis to the affected child. We were able to discern parental decision-making factors, as well as concerns and barriers related to sharing information about 22q11DS with their child. Influential factors included the developmental level, understanding, and interest of the child. Barriers consisted of not knowing what words to use or how to initiate the discussion. Results showed that while parents felt reasonably well equipped to have these discussions with their children, they had received minimal guidance regarding how to navigate this challenge, indicating that professionals need to initiate such discussions. Further research is needed to expand upon the knowledge gained through this study. With each experience that is shared, genetic counselors and other health care professionals can continue to advance their ability to aid in the process of successful parental disclosure of a 22q11DS diagnosis.

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Table I

Characteristics of Affected Child and Family

ID	Age of Child at Diagnosis	Child's Age at Initial Disclosure	Gender of Child	Child's Age at Time of Interview	Child's IQ	"Emotional Difficulty" Likert Score	"Well-Equipped" Likert Score
1	Birth	12	Female	15	71	1	5
2	7	7	Male	17	103	7	3
3	2	N/A	Female	15	60	5	2
4	Birth	9	Female	10	73	1	5
5	9	9	Female	10	88	Declined to respond	7
6	Birth	N/A	Female	13	64	5	5
7	10	10	Male	14	75	2	6
8	11	11	Female	13	105	1	6

Table II

Representative Quotes from Participants

Topic	Quote
(1) Sharing information about a 22q11DS diagnosis with family/ friends/others	"At the initial time of diagnosis, it was just to share our sort of shock. And it was a trauma, that's not too strong of a word, because we were just reeling from it… And he had this great teacher, so we also wanted her support. But I struggled with 'How do I tell?' and 'What do I tell?' because I didn't want anybody to look at my child as defective… So I guess we kept it fairly close for a while." "It is what it is… it is part of who he is. And it explains some things, sometimes, about him. But then the other side of that is, I don't ever want anybody to look at him as defective in any way. Because he's not." "I have concern, I don't want [child] to be labeled like that, like a child with a syndrome. So I am more careful. I don't tell every parent, like all the parents of her friends. Sometimes I still get confused and I don't know how much detail I should give them or not." "We were pretty much just telling whoever. It was no secret… We were just thrilled to finally have answers… And there were so many people somehow connected with him, from school to friends to family, so there were a lot of people we told because so many people had been involved with him already at that point." "For me, you cannot hide it from people… it's just my opinion. Because people can help you when they know. And for us, my husband and me, we always tried to be honest with people, that's what she has, that's what she was born with." "I guess the two things that I have always wanted people to know is, number one, what [child] has, so that they'll know why she's a little quirky and a little different maybe. And number two, I've always wanted to educate people, since we found out, about what it is."
(2) Emotional difficulty of disclosing a diagnosis of 22q11DS to others	Low rating: "I think it wasn't emotionally hard at that point because we were just so relieved to finally have some understanding of what in the world was going on. We had been so frustrated for so long, not being able to figure out what was happening… I think that hardest part for us is really the emotional side of it coming into play now, more so than when we first found out, because [child] is becoming more aware of it." Low rating: "No [it was not emotionally difficult], not at all. I was emotional more in trying to help her… more that I felt sorry for my daughter not to be understood before. More guilty… for not being able to understand her emotional needs before [rather than emotional about telling others]." High rating: "It was like being knocked over by a big wave. I felt so clueless, like how could I have raised this child to this age, and not known. And being so afraid of what it meant for his future."
(3) Sharing information about 22q11DS with the affected child	"Right from the beginning we told him something… When he was little, we just referred to it as his funky chromosome. And 'funky' in our family is just sort of a fun word, no negative connotation, just kind of quirky, maybe even cool." "I just explained it as how God makes us all different, and whereas God gave some people two kidneys, he gave you one kidney. And that doesn't mean you're bad, it just means you're different. It means that… some people can do some things, and you can do other things." "We need[ed] to go see this doctor and that doctor. [Child] had to go do all these things. And we saw no reason to hide from him what was going on, so we started talking to him about it at that point… whenever we had something we thought we could share, in terms of his understanding." "We are just a very open family. We talk openly about anything that's bothering any of us, whether it's me, my husband, or our two children. And we just wanted her to know that she could always talk to us." "I think the biggest thing is 'We want to help you. And when you let us help you, we usually are able to improve things.' And by help, it's not just doing things for him, but it's creating environments that he can be successful and have opportunities in. Like in the classroom, getting him an environment that's going to work. Once we get him there, it's up to him… So we hold him accountable, but we want him to know we are there to help him navigate." "She has to know how to make some healthy decisions, how to take care of herself. I say sometimes 'You know you are getting older, and maybe want to go to college, and you need to learn how to take care of yourself.' We just try to remind her what is part of… a healthy lifestyle. That's why we start talking sometimes, related to being healthy. You just need to be addressing all the issues."
(4) Parental concerns and barriers regarding disclosure of a 22q11DS diagnosis to the child	"You know, we really have not sat down and had a discussion about… 'This is what you have.' We just don't know exactly what to do about that, and so consequently we haven't done anything about it." "My biggest problem is just knowing what words to use because her understanding is limited. I can't say there is something wrong with your genes because she won't know what a gene is. And I just don't know the words to use so it won't be scary to her, so that she won't feel weird or different, and that she will kind

Topic	Quote
	of understand. So I guess that's where I struggle the most, is because you have to do it on her level, and it's hard to know the right things to say." "I don't want [child] to use it as an excuse for things… She'll say 'Well I can't do that because I have VCFS.' That's kind of how she is, she could use that to get her own way, and I don't want her to use it like an excuse. So I just don't know how to handle that yet, and we're working on it." "I guess my primary concern has been his self-image, that it remains just as it would for any other child. I don't want this to define him. But I do want him to have the information he needs to know to make good decisions for himself. And that's a difficult balance to strike."
(5) Receiving advice about how to talk with children about 22q11DS	"I don't feel very well-equipped at all. I sort of feel like I could use help with it… I just don't know what words to use so that she'll be able to understand." "Just knowing how and when to talk about it, and like should someone else be there, like should a doctor be there too? And how to approach it. I suppose with every child, it's so different, the understanding and everything. So I suppose it depends on the child. But that's what I really don't know. I just don't know how to approach it." "The pediatrician was very helpful with that [how to talk to the child]. He gave me the analogy of a pie chart. You know, it's not a big circle that says 22q. It's slices, you know… he's this, he's that. And 22q is a slice in that chart. And that was really useful. That was a very good visual… for me, and I remember sharing it with [child] too, because I thought that was something he could understand as well."
(6) Participant strategies for becoming knowledgeable and preparing for communication about 22q11DS	"I continue to do a lot of reading and looking at different things, trying to learn… because it changes as he gets older… in how things are affecting him. I'm not an expert overall. I just try to be as much of an expert as I can on the specific areas I need to know about." "I've really just had to read and put it in child form for her… I wish there was a book for 22q that an 8 or 9 year old could understand. But I haven't found that book. So I just always read, and anything that needs to be shared with her, I try to put it in children's terminology."
(7) Opinions about when to disclose a diagnosis of 22q11DS to the affected child	"When your child is old enough to understand the first little bit about it, then you tell them. That's my feeling." "I don't know if I could put a year on it specifically. But more… categorizing… it as an ability to understand… when they're recognizing the differences and asking questions, at whatever age that is. It could be 4, it could be 7… I mean the kids vary so much, you know."