

Research Article

Exploring Education for Children with 22q11.2 Deletion Syndrome: A Qualitative Study of Mothers Perspectives

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Abstract

22q11.2 deletion syndrome (22q11.2DS) is a complex and widely variable genetic syndrome involving multisystem physical health problems, significant cognitive and psychosocial challenges, all of which may impact upon learning and academic achievement. In the current study, we explored mothers' perceptions of their child's learning within early and primary educational contexts. We conducted 3 online focus groups and 1 online semi-structured interview with a total of 9 mothers of children diagnosed with 22q11.2DS. Mothers were included if their child had a genetic diagnosis of 22q11.2DS and was aged from 3 to 12 years old. The mothers were asked a series of open-ended questions to explore the educational experiences of their child. We adopted an inductive approach to our data analysis and conducted a reflexive thematic analysis resulting in the emergence of 4 major themes (1.) The impact of 22q11.2DS on a child's learning; (2.) The impact of adaptive behaviour skills on their child's learning; (3.) The level of support available within the educational setting and; (4.) Mothers' perceptions about their child's learning. From these themes, the following 12 sub themes were identified: absences affecting learning; the development of specific learning behaviours; the impact of a 22q11.2 DS diagnosis on their child's self-perception; toileting,



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motor issues; communication; sleep; school and teacher support and the presence (or absence) of therapists. Finally, the mothers' confidence about their child's learning; mothers' concerns and comparisons of their child; and mothers' apprehension about their child's future were all identified as sub-themes. Children with 22q11.2DS require increased and specific psychosocial support to allow them greater opportunities to fully participate in their learning environments. Mothers perceived the level of support their child received was greater during pre-school compared to that received at primary school. It was clear that a greater understanding of the needs of children with 22q11.2DS, by educators, was needed. Implications for tailored support for school-based learning and educational transitions for young children with 22q11.2DS are discussed.

Keywords

DiGeorge syndrome; velocardial facial syndrome; education; learning; lived experience

1. Introduction

22q11.2 deletion syndrome (22q11.2DS) is a common microdeletion with an incidence of approximately 1/3000-1/6000 live births [1]. For most individuals, the functional deletion encompasses approximately 50 genes, resulting in a variable within-syndrome phenotype with substantial impact across major organs. Common features of the syndrome include developmental disability, congenital heart defects, palatal defects, immunodeficiency, renal abnormalities, hypocalcaemia, dysphagia, scoliosis, and polyhydramnios [2]. A diagnosis of 22q11.2DS also incurs a significant increased risk of neurodevelopmental conditions such as intellectual disability, autism, and attention-deficit hyperactivity disorder as well as mental health disorders such as anxiety, the presentation of which has been shown to change over time [3]. Individuals with 22q11.2DS also experience an increased likelihood of developing psychotic symptoms during adulthood compared to the general population [3, 4].

Education and learning experiences of children with 22q11.2DS is often a major concern for parents yet has received little focus in the literature. In a questionnaire-based study exploring the key psycho-social issues for 32 parents/caregivers of individuals with 22q11.2DS (aged from 2-23 years old), speech development, relationships with others, focused attention and school difficulties were the most concerning issues [5]. Given these results and the impact of positive schooling experiences on functional outcomes for young people with disabilities [6], exploring parents experiences of learning and early schooling for their child with 22q11.2DS could help to shape improvements in educational opportunities alongside research, that are meaningful for families.

Several characteristics of 22q11.2DS impact upon learning. Firstly, the presentation of intellectual disability (ID) is seen in the majority of individuals with 22q11.2DS with approximately 1/3 of individuals presenting with mild ID [4, 7]. The full IQ profile of individuals with 22q11.2DS is highly heterogenous and has been shown to change over time with the tendency to decline as individuals age [4, 8]. This has significant impacts on learning for school aged children as they navigate through educational contexts that (a) continually demand more abstract thinking, (b) continually demand higher and more complex degrees of independence and planning; and (c)

require more complex problem solving and cognitive skills [9, 10]. As a result, it is common for young people with 22q11.2DS to experience learning delays compared to their peers as they progress through the upper years of primary school in specific areas such as math, reading comprehension, and abstract reasoning [7, 9-11]. All of these factors may impact on young people completing their education, and in light of this, authors of a recent study in the USA examining functional outcomes for adults with 22q11.2DS found that only 51 of 101 adults had completed high school [12].

Secondly, communication and social cognitive difficulties are considered hallmark features of 22q11.2DS and have significant implications for learning and school experience. Young children with 22q11.2DS typically experience delays in pre-linguistic behaviours, first words, and complex expressive language skills [13-15]. Communication difficulties tend to persist into later childhood and adolescence, where impairment is observed in the development of more complex language and conceptual understanding [14, 15] and in the production and comprehension of narratives [16]. A lack of early fundamental expressive language has vast implications for the development of age-appropriate social communication and social cognition such as interpreting, reasoning, and predicting others' emotions and behaviours [17, 18]. As a result, young people with 22q11.2DS experience difficulties in responding to social cues, recognising, and understanding subtle social communication, and initiating and maintaining social communicative exchanges [18]. The presence of on-going communication and social cognitive difficulties can be very challenging for young people with 22q11.2DS who are often described as socially immature, having difficulties making and maintaining same-age peer relationships, being victimised by bullies, and increasingly displaying socially withdrawn behaviours [7, 19].

Lastly, school can be considered a stressful environment for young people with 22q11.2DS where the ever increasing social and cognitive demands may impact upon the development of mental health disorders that typically emerge in young adults with 22q11.2DS [19-21]. We know in the typical population that mental health issues can become more apparent during transitions to higher levels of education as compounding effects of more abstract and challenging cognitive work is introduced in parallel to the demands of more complex social relationships [22]. The difference for those with 22q11.2DS, is the lack or difference in fundamental social, communication and cognitive abilities required to successfully navigate this challenging time [23]. Specifically, the development of anxiety and depression impacts upon communication skills and positive social interactions with family, peers, and teachers [3, 23]. This relationship was explicitly explored by Stephenson et al., [23] who reported that when children with 22q11.2DS experienced comorbid depression and anxiety they also had poorer functional communication skills and reduced functional outcomes in comparison to typically developing peers [23]. The presence of mental health disorders may therefore impact upon psychosocial aspects of school, but how does it impact upon learning? It is well understood that anxiety can overload a cognitive system, thereby reducing processing capacity and performance [24]. To explore this, a group of school aged children with 22q11.2DS were assessed on working memory tasks, identified as a key executive functioning component that is fundamentally important for learning. Authors found that working memory was impaired in the children with 22q11.2DS compared to typically developing peers and the presence of anxiety was found to exacerbate the children's impairments in these tasks [25].

Thus, the interaction of ID, social-communication difficulties and increased risks of mental health issues means that attending educational environments, and learning, can be challenging for children with 22q11.2DS. We know that the educational profile of children with 22q11.2DS appears to

change over time and that the more complex social demands of subsequent education contexts may impact on mental health [9, 10, 26]. While there is extensive literature regarding the cognitive and psychosocial difficulties children with 22q11.2DS face, there are no studies in the current literature that explore the impact of 22q11.2DS on learning and attending early and primary educational contexts from a lived experience viewpoint. If we are to understand the best way to support and care for those with rare genetic syndromes, all information must be collected, used and valued. This includes expert knowledge from professionals as well as the expert 'lived' knowledge from those who have the syndrome themselves, their families and the key stakeholders in their lives [27]. Exploring parents' lived experiences can inform exactly what the needs are within educational contexts to promote school and learning success [9, 10]. Furthermore, parents' experiences could provide direction to ensure that future research is meaningful and impactful for families and the children themselves.

To explore the lived experience of learning within early and primary educational contexts from a parent's perspective, the present study conducted three focus groups and one semi-structured interview involving 9 mothers of children diagnosed with 22q11.2DS. Discussions were examined using thematic analysis to answer the following questions:

1. What are the main impacts of a diagnosis of 22q11.2DS on a child's capacity to attend an educational setting and learn?
2. What impact does the educational environment have on children with 22q11.2DS and their ability to learn?
3. What are some of the additional challenges impacting the learning of a child with 22q11.2DS?

Results from this study will add to the growing body of evidence working towards best practice in the development of tailored support programs and guidelines ensuring successful educational outcomes for these learners.

2. Method

2.1 Ethical Consent

Ethical approval was obtained from The Human Research Ethics Committee of Newcastle University (HREC: H-2020-0146). Participants were provided with the information statement and consent forms after indicating their interest in participating in the current study. Prior to participating in the discussions, participants completed their individual consent forms and provided these to the research team.

2.2 Participants

2.2.1 Recruitment

Purposeful sampling was used to recruit participants for the present study as we wished to provide a detailed perspective of education as perceived by a specific group; mothers of children with 22q11.2DS [28]. Participants in this study were mothers of pre-school and primary-school aged children who have a genetic diagnosis of 22q11.2DS. Participants received information about the study through social media posts from the 22q Foundation of Australia and New Zealand Facebook

page, on the 'FindLab' web page (through the School of Psychological Science at the University of Newcastle), and via emails from speech language therapists (SLTs) in the Hunter region, Australia.

2.2.2 Demographics

Nine parents (all mothers) were recruited and grouped based upon the age of their children. Two groups comprised of 6 parents with children ranging from 3-5 years of age, and the second group consisting of 2 parents whose children were aged between 8-9 years old. One interview was conducted with a mother of a 9-year-old as she could not attend the focus groups. See Table 1 for an overview.

2.2.3 The Children

All children were from 2 parent families and had (a) a genetic diagnosis of 22q11.2DS, and (b) were aged from 3-12 years old. Specifically, two children were from single children families, and seven children were from multiple sibling families. Families were from a range of rural and metropolitan locations from around Australia and one participant was in a semi-rural location in New Zealand. Six of the children were attending early childhood centres with the 3 others attending primary school settings. One of these primary school children had recently shifted from a mainstream to a special education setting. See Table 1 for an overview.

Table 1 Demographics of mothers, their child with 22q11.2DS, the number of children in the family unit, the mother’s highest education level attained, and details about their child with 22q11.2DS.

Child	Child gender and age	Siblings	Mother highest Education level	Child details	Schooling	Allied Health Involvement
Child 1: Kyran	Male, 3 years old	Only child	Tertiary	Kyran was diagnosed with 22q11.2DS at 10-months old from the results of a Chromosomal micro-array analysis (CMA). Kyran is able to request desired items using one-word labels. His speech is affected by VPI and is significantly hypernasal.	Daycare 2 days per week	Yes (SLT)
Child 2: Kane	Male, 5 years old	3 older siblings	Tertiary	Kane was diagnosed with 22q11.2 DS just before turning 2 from the results of a CMA and a Fluorescence in Situ Hybridization (FISH) test. Kane has a dual diagnosis of autism.	Daycare 2 days per week	Yes (SLT, OT and Physio)
Child 3: Damon	Male, 3-years-old	1 older sibling	Tertiary	Damon who was diagnosed with 22q11.2 DS at 18-months old from the results of a CMA. Damon is able to say many words however has trouble forming two-word sentences.	Daycare 4 days per week	Yes (SLT, OT and Physio)
Child 4: Amara	Female, 2-years-old	2 older siblings	Tertiary	Amara was diagnosed with 22q11.2DS at 4 months old from the results of a FISH test. Amara has a sub-mucous cleft palate. She can use one-word requests consistently. Her speech is hypernasal and difficult to understand.	Daycare 2 days per week	Yes (SLT, play therapist, hydrotherapy with her Physio)
Child 5: Hudson	Male, 4 years old	2 older siblings	High school	Hudson was diagnosed with 22q11.2DS at 6-weeks old from the results of a CMA. Hudson also has sclerocornea, which impedes upon his sight. At 12-months old, Hudson had	Daycare 3 days per week	Yes (SLT, OT and Physio)

				cleft palate surgery. Hudson has a limited vocabulary of approximately 10-words.		
Child 6: Jarna	Female, 4 years old	Only child	Tertiary	Jarna was diagnosed with 22q11.2 DS just before she was 2 from the results of a CMA.	Daycare 2 days per week	Yes (SLT, OT and Physio)
Child 7: Ginny	Female, 9 years old	Only child	Not provided	Ginny was diagnosed with 22q11.2DS at 6 years old from the results of a FISH test.	Primary school 5 days per week	Yes (SLT and Physio)
Child 8: Mako	Male, 8 years old	3 older siblings	Not provided	Mako was diagnosed with 22q11.2 DS at 18 months from the results of a FISH test. He has had also recently been diagnosed with Legg-Calve-Perthes disease.	Primary school 5 days per week	Yes (SLT)
Child 9: Hendrix	Male, 9 years old	1 younger sibling	Tertiary	Hendrix was diagnosed with 22q11.2 DS at 18-months old. Hendrix also has diagnoses of ADHD, autism and hemiplegia.	Primary school 5 days per week	Yes (SLT, OT and Physio and receives support from a psychiatrist)

2.2.4 Focus Groups

Two researchers were present during the focus groups and asked a range of open-ended questions to guide discussion from the participants covering their experiences of education for their children. Due to the wide scope of locations of participants and the impact of COVID-19, online sessions via zoom were identified as the most appropriate method. For the present study, the parents' perceptions of their child's learning and the educational environment for their child was analysed. Participants were guided by the two researchers to discuss the following aspects of their child's education and learning: (a) How does the diagnosis of 22q11.2DS impact on your child's ability to attend their educational setting and learn? (b) what impact does your child's educational environment have on their ability to learn?; and (c) what other challenges impact upon your child's learning?

2.2.5 Interview

One parent was unable to attend the focus group sessions but wished to be included in the study. The interview involved one researcher (first author) who guided her to discuss the same aspects of her child's education and learning. Questions asked were the same as in the focus groups as mentioned above.

2.2.6 Data Analyses

To explore parents' perceptions of their child's learning, we conducted a reflexive thematic analysis to address our research questions. Reflexive analysis, developed by Braun and Clark [29, 30], was identified as the most appropriate method by which we could analyse our data as it is an interpretive analysis method that has broad applicability for a range of health and education research. This type of analysis is described as independent from specific theoretical frameworks and epistemology whereby the researcher is responsible for selecting an appropriate theory to underpin their findings and epistemology and to ensure that reflexive analysis accurately aligns within that approach. Thus, the subjectivity of the research team is recognised and valued as an important component within reflexive thematic analysis. To analyse our data, the 6 phases of reflexive thematic analysis, as developed by Braun and Clark [29], were implemented:

1. The familiarisation of transcript data, where patterns in the data begin to emerge. This is accomplished by the reading and re-reading of transcript data.
2. Generating the initial codes, whereby data is labelled and organised into meaningful groups.
3. The construction of initial themes, involving the sorting of initial codes into groups and where meaningful relationships are identified between the codes.
4. The reviewing of potential themes, whereby the researcher ensures there is enough data to support a given theme. In some cases, themes are collapsed or refined.
5. Defining and naming themes. This is where the broader narrative of the data is organised to respond to the research question.
6. Producing the report, where participants accurate information is told in a compelling way to address the research question(s).

Important decisions are required by researchers when conducting a reflexive thematic analysis. One of these is to decide on what counts as a theme. When deciding this, it is recommended that a

theme is chosen based on its 'keyness' in addressing the overall research question; that is, a theme should be chosen based on the ability of that theme to accurately capture what is most important when considering the research question posed [30].

Another key decision that must be made by qualitative researchers is the type of analysis that will be conducted. In the current study, we chose an inductive approach to our analysis whereby our themes were derived directly from our data and were not influenced by the questions asked of the parents during the discussions, nor by our theoretical interest or knowledge in the topic [29].

Credibility and confirmability are important components in qualitative research when ensuring trustworthy data analysis. To ensure this during our data analysis phase, we followed the guidelines provided by Nowell et al. [31]. Credibility refers to confidence in the accuracy of the findings and was addressed via the use of researcher triangulation whereby the data was analysed independently by the first and second authors. During this process, authors compared their preliminary analyses and from these discussions, the themes were refined, ensuring accuracy in our analysis. Researcher triangulation also addressed confirmability of the trueness of the data, that is, the extent to which our findings were derived directly from the data [31]. In reflexive analysis, the subjectivity of the research team is recognised and valued, however personal researcher bias needs to be accounted for and addressed transparently in order to accurately present findings that are representative of our participants lived experience. To address our personal biases from influencing our analysis, we employed two strategies; 1. We regularly held discussions of the 'trueness' of the emerging themes and the impact of our biases amongst all 3 of the authors; and 2. The original transcripts were consistently referred back to, ensuring that our biases did not impact upon our interpretations. As we had 3 researchers with great variation in their personal experience of 22q11.2DS, and with conducting qualitative analyses, we held regular discussions after each focus group and during data analysis to identify how we were interpreting the data and how our experiences were influencing our understanding of the participants lived experiences.

3. Results

As a result of our reflexive thematic analysis, four major themes were identified from the data: 1. 22q11.2DS impact on learning, 2. Impact of adaptive behaviours on learning, 3. Support within educational environment, and 4. Parents feelings about their child's learning. From these major themes, 12 sub-themes emerged to further explore parents' experiences of their child's learning within educational contexts. See Figure 1 for an overview of the themes and sub-themes identified from the data.

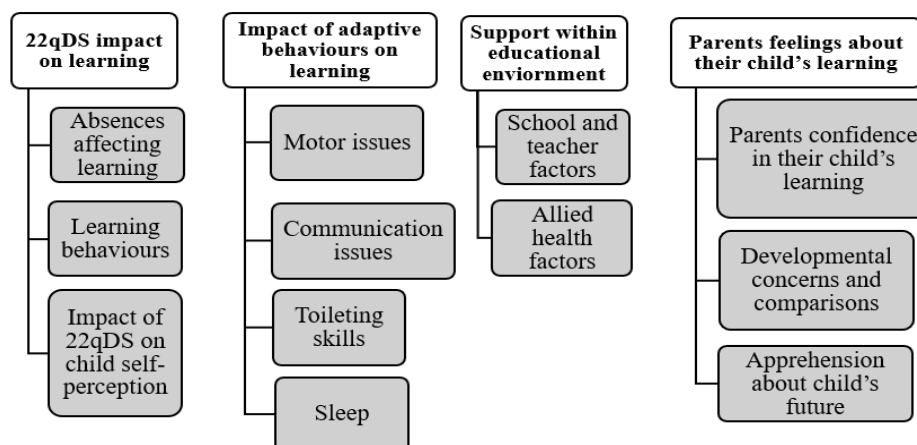


Figure 1 Overview of major themes and corresponding sub-themes.

3.1 22q11.2DS Impact on Learning

Several features of a 22q11.2DS diagnosis were discussed by the mothers as directly impacting their child's learning. Specifically, the ideas and thoughts on how *absences impacting upon learning*, *the impact of a diagnosis of 22q11.2DS on learning*, and *the impact of having 22q11.2DS on self-perception of the child* are outlined below.

Mothers reported that their child often had many absences from school or their learning context due to illnesses, appointments, and hospitalisations as a result of having 22q11.2DS and reported that those increased absences impacted upon their child's learning. Increased absences were described as challenging, but these could not be helped.

'I think for [Child] it was really monumental. You know, each year sort of brought more and more things and, you know, really culminated last year with him missing, you know, months and months of school because he was in hospital... so I think that's had a huge impact on his education.' (Mother of Mako).

'She was out of childcare a bit. She had a lot of ear infections when she was sort of 18 months and then she got grommets and I think that was also related to the 22q... So, I think that probably kept her out of childcare.' (Mother of Jarna).

Mothers discussed recurring and persistent health issues that led to increased absences from their learning environment with one parent specifically highlighting her child's renal failure as a major issue.

'...this kid has a runny nose, he must have had this runny nose for about six months... it did affect, like, you know, we were having, keeping him home...' (Mother of Damon).

'You know, he's got all that other stuff going on in his brain. He's got, you know, dialysis, he's got stomas... because he's at end-stage renal failure.' (Mother of Mako).

Mothers talked about *learning issues* and *learning strategies* that were related to a diagnosis of 22q11.2DS and which impacted upon their child's learning. Specifically, the presence of memory issues and needing more time to grasp concepts were discussed. Furthermore, the idea that these issues could be overcome by repetition were discussed.

'But if it's like he's doing a task and you have to remember something to do the next part of the task, that's difficult.' (Mother of Hendrix).

'So, with [child], I have noticed that she's very much, she's slower with everything, everything takes that little bit longer to learn...' (Mother of Amara).

'... the repetition for us, too, is one of the key things we use to help [child] learn. He is still quite delayed. I think 22q does really impact on his learning.' (Mother of Hudson).

Masking behaviours were often employed by the children so they could 'fly under the radar' of teachers where they would not be noticed when falling behind their peers or when finding school difficult. This was perceived by the mothers as teachers not understanding the extent of the child's delay or the difficulties they faced in the classroom.

'So, she can fly under the radar quite easily, and that's sort of our experience with school at the moment...' (Mother of Ginny).

'And also, he will... he covers himself well too, because they'll ask him if he understands and he'll say he does even if it doesn't, you know what I mean? And then he'll... he's actually got some really good strategies... but he considers them helpful because he can kind of get through.' (Mother of Hendrix).

Mothers also discussed the strategies their child used in their classroom to avoid tasks and to fit in with their peers. Specifically, they talked about their child directly avoiding tasks, procrastinating to avoid completing a task, and copying what they saw their peers doing.

'So, he will avoid stuff. So, it'll be like, "We want you to do this," and he'll like... he'll avoid it and he'll procrastinate, and he'll shuffle around and he'll, you know? He's just like time, he's just watching time tick over until... "Yeah, that worked." You know what I mean? Like, he's getting quite clever at that stuff.' (Mother of Hendrix).

'...They, I don't think they sort of understand the extent of the problem because she's so very good at just fitting in and just checking what everyone else is doing and just doing that...' (Mother of Ginny).

Mothers talked about how their child might not understand what had happened at school, but doing enough for this to go unnoticed by their teachers.

'She'll often come home and report that like she didn't necessarily understand what was going on in the classroom, but she does enough to get by.' (Mother of Ginny).

The impact of having 22q11.2DS on self-perception was discussed by mothers of the older children whereby their child's diagnosis, or knowing they are 'different' from their peers, was starting to be more important to their children. What this meant for their child was highlighted as an area for concern.

'... like he's quite a complex little guy really. Like, he presents kind of calm and fun and kind and sweet, but there's a lot going on inside and there's a lot going on even physically, and he's sort of realising differences and he's trying to manage that... but I don't think he really knows... like he hears it, we talk about 22q, but at this stage he doesn't know what that actually means, like what that has meant for him.' (Mother of Hendrix).

Two mothers talked about their child's presentation of anxiety, how this impacted upon them attending school, what their anxiety looked like at school, and how their anxiety impacted upon their ability to 'fit in' with their peers.

'She's got a lot of anxiety about being different as well, so I think that sort of adds to the stress of trying to fit in in the classroom at school.' (Mother of Ginny).

'So, we're working on some strategies to help him, yeah, and with his anxiety as well... I feel like his anxiety's actually quite high; he worries a lot about a lot of things, but he's the quiet one, you know, he's the freeze version of fight or flight or freeze. So, you might look at him and think he looks perfectly fine, but he's actually freaking out...' (Mother of Hendrix).

3.2 The Impact of Adaptive Behaviours on Learning

The impact of certain adaptive behaviours on attending school and learning were discussed in detail by all of the mothers. Delays and difficulties in *motor skills, communication, toileting* and *sleep* were discussed in regard to how these behaviours had a negative impact upon their child's learning. The impact of low muscle tone on their child's ability to sit at a desk comfortably, was identified as a barrier to children's full participation in their learning. Gross motor issues also made attendance and participation difficult for their child. Motor issues often meant their child was left behind or left out of activities in their place of learning.

'They've got a lot of outside play on the uneven ground, and they go on bush walks and stuff like that. And she's had a gross motor delay and she's low tone and so she, whilst she's participating in some of that, she is behind the other kids so she sometimes doesn't go and do some of the activities because she gets tired more easily... she just, she couldn't do it ... So, she has found childcare a bit difficult in that way.' (Mother of Jarna).

'But because of his low tone as well, sitting at a desk or sitting down writing, he will have no bar or, you know. And that's where the struggles have begun.' (Mother of Kane).

Children's communication difficulties were described as barriers to learning where specific expressive language delays negatively impacted upon children's participation within their learning context. In particular, the mother's perception that the educator was unable to understand their child's communicative attempts were seen as a significant barrier to their child's learning ability.

'I could say that the 22q affects her quite significantly in her learning, like communication, she's behind in her expressive language and she's behind in being able to, like her speech isn't clear because of the 22q... the clearness of the speech, I would say is the 22q thing... The expressive language, I assume that's related to the 22q and that would also affect her learning... Her play is behind, especially her imaginative play.' (Mother of Jarna).

'I guess they learn to communicate in other ways because they haven't got the speech. So, I guess that is a barrier to learning, the speech, because he can't repeat back things...' (Mother of Damon).

Mothers also spoke about the impact of their child's language on social participation within their classrooms whereby they were misunderstood by their peers. This impacted upon their ability to engage in play.

'...that's where a lot of [Child's] frustrations and behaviours were being displayed at preschool because he couldn't communicate what he wanted in amongst his friends and the teachers.' (Mother of Kane).

'...definitely when he was mainstream the kids really struggled to understand him and that led to perhaps feeling isolated or not being played with or having to be the dog always.' (Mother of Mako).

Toileting issues were discussed by mothers as being a concern due to their child's age, and the indirect effects of staying accident free at school on other issues, such as encopresis at night-time.

'[Child]'s not toilet trained. She's four, we've had no luck at all.' (Mother of Jarna).

'So when we linked up with the doctor in the US he got us a course of medication that means that [child] can stay dry and accident-free at school, which was our main aim for her, but the compromise being that she has a lot of encopresis at night time and a lot of waking and a lot of pain from medication.' (Mother of Ginny).

Sleep was found to be an issue for some families with sleep issues and fatigue impacting their attendance at school. One mother described fatigue and managing her daughter's tiredness as a huge issue in their house.

"...we really struggle with her fatigue, and we find it really hard to get a happy balance between when we send her when she's tired and when we just keep her home... Obviously like knowing that if you send her tired she's not going to be productive or achieving really much... Sleep's a huge issue in our house..." (Mother of Ginny).

'... he's got lack of sleep from the machine going off all night long and, you know, just feeling tired generally...' (Mother of Mako).

3.3 Level of Support within Educational Context

The level and types of support that their children received within their education setting was perceived as impactful upon every child's learning. Specifically, the sub-themes of *school and teacher factors* and *allied health factors* were important in mothers' perceptions of the support their child received. Within the school and teacher factors, several minor themes were identified including *Educators taking on information from parents about optimal support methods*, *Support from educators for the child and their needs*, the impact of *how their child was treated in their classroom*, and the *ratio of teacher/educator to support worker*.

3.3.1 School and Teacher Factors

The impact that the school and teachers had on their child's ability to learn and be supported in their place of learning was relevant across both the early childhood learning context and the primary school context. Specifically, mothers talked about their role in educating the teachers about their child, and the importance of educators taking on and adopting relevant information from them to effectively support their child within their learning environment.

'So, I feel like we need... they need more education into, more understanding of how they can communicate with [Child]. So, I need to find a way how I can educate them, how I can give

them more information about, you know, how to communicate with [Child] or the way how he communicates or how he pronounces the words, what he's trying to say.' (Mother of Kyran).

'And they'd also like some more like supports like the physio to go into the centre and, you know, show them what they could be doing to help support [Child].' (Mother of Amara).

Mothers discussed experiences when educators did not provide enough basic support or understanding for their child and their specific needs within their classrooms noting that educators did not appear to spend time learning about their child and their condition. This was described as the most stressful aspect of life for one family.

'I find school probably the most stressful thing in our life... However, we sort of know what works for [Child] and what doesn't, but it's getting that implemented in school. When we've gone to school we find them not extremely helpful... I found them not really super keen to do a lot of that troubleshooting with us.' (Mother of Ginny).

I think the anxiety, they struggle to understand which is why I... I actually put a section in there around his anxiety cues, and for them to try to understand a bit better, because I don't feel like they do quite understand that side of things very well.' (Mother of Hendrix).

How the school or teachers were not able, or willing, to extend their child or support them to further develop their learning was discussed by the mothers. This lack of extending their child in their learning was challenging to deal with and understand.

'I don't think they quite knew how to academically sort of meet him where he was at and try and extend him.' (Mother of Mako).

'...they committed to moving him forward but they never really did. I think they just found it a lot easier for [child] to leave him with those young kids. And so in the six months it was a bit deflating because he really was frustrated, not challenged. It was... yeah, it almost got too hard for them to move him forward. So I found that a bit hard to deal with.' (Mother of Hudson).

One mother specifically spoke about teachers not listening to parents and how that can be detrimental to the child:

'... it's disappointing that they don't... sometimes I think they don't listen enough to parents. Like, we told them about his anxiety and that's it's really important for him that he be with his friends. ...And I don't know why they didn't listen to us, because you know, like we really made it really clear to them that it was very important, and I don't know why they thought they knew better.' (Mother of Hendrix).

One mother talked about communication and undersanding being a reason for moving her son from a mainstream setting to a specialised learning environment whereby the problems experienced in the mainstream school setting were easily overcome in the specialised school setting.

'So I think being in the special school setting has just alleviated all those problems because I can just send a note to school and say, "He has three stomas in the bag, make sure you catheterise him every two hours," and they go yeah, okay, done. You know, it's not a problem. Whereas in a mainstream school it was a real issue...' (Mother of Mako).

Mothers of younger children attending early education centres talked about the support and understanding that was provided by the educators at their child's pre-school/kindergarten noting that their child was well looked after and well supported by educators who understood and met the child's needs.

'We didn't have too many issues with preschool... but I think they understood him because he'd been there already a few years, they understood, you know, his needs and everything.' (Mother of Kyran).

'So [Child's] very, very well looked after. And, you know, when we have our six monthly IEPs, which is his goal setting for the next six months, you know we have up to like eight to 10 people on that Zoom chat all, you know, interested in what [Child's] doing and helping him move forward. So we're very lucky, huge amount of support for him, which is amazing.' (Mother of Hudson).

In comparison, the mothers of primary-school aged children discussed issues with being excluded from discussions regarding the development of their child's Individual Education Plans (IEPs), learning goals, and in general, not understanding the IEP process. They talked about their priorities for their child being missed from IEP documents.

'...I didn't know what an IEP was. It wasn't really explained, it was sort of written and then you're just asked to come in and sign this document. Now that we've been in the school system for a few years and I have a better understanding, I tend to ask for a meeting and go through the IEP and I'll add things and I'll take things out and change things, but originally that wasn't... they'd sort of just write it and ask you to come in.' (Mother of Ginny).

Like, we think the social element of school and the friendships is the most important thing about school... I'm surprised they didn't draft it in there because they know it's a priority for us, and I'd raised over the phone, ... And you know what? It's important for [Child] too, and I know for [Child], the most important thing is his friendships, that to him is the most important thing in life. (Mother of Hendrix).

How their child was treated within the educational environment was important to mothers and impacted upon their child's learning whereby experiences of being treated the same as everyone else, and feeling as if their child was in the 'right place' was viewed as an enabler to positive learning experiences for their child:

'I think that the day care that he goes to are very inclusive, they all love him, they just include him just the same as everybody else.' (Mother of Damon).

'Yeah. I mean, yes, she's just treated like any other child in the centre as far as I know.' (Mother of Jarna).

The ratio of educator to the number of children within the educational setting was discussed. Specifically, the smaller the ratio of children to educator, the more positive the learning experience, whereas mothers discussed issues occurring when the ratio of teachers to children was smaller.

'Yeah. So, and when he comes to 20 odd kids in the class and one teacher, it's proving to be quite difficult and challenging for him.' (Mother of Kane).

'He's in that kind of small, young age group now, so there is a better ratio, like educator per kid in that group.' (Mother of Kyran).

3.3.2 Allied Health Factors

Mothers discussed the critical role of allied health workers (labelled 'therapists') going into their child's learning environment to provide important support for the child and to model good practice for the educators. Therapists were identified as effective communication channels between them and the school.

'But yeah, the physio's worked really closely with the school to get the new table and chair and she works with me really closely, you know, like for shoes and different activities for him, equipment, and, ... We're very lucky with our therapists, they all are really committed and they all love him, they're all really good communicators with me...' (Mother of Damon).

'...they're amazing. They really wrap themselves around [Child]... I mean I have the speech language therapist, you know, weekly, OT weekly... They go above and beyond, like they do amazing stuff with him... And they also supply... education support worker, an ESW, that will go into [Child's] kindy on the other three days and she's just there for three hours a week, just to support him one-on-one at kindy.' (Mother of Hudson).

In contrast, when schools did not allow therapists into the classroom due to policy reasons or due to the impact of Covid-19, this was seen as a missed opportunity for learning. Mothers discussed the school's expectation that therapy would occur outside of the school environment, which wasn't always possible.

'...And the therapists who could work with him, like a speechie or an OT or so on, they weren't allowed into the school for, just a policy reason... It was a Christian school and so he really, he missed out for quite a few years on having therapy because by the time he got home from school he was way too tired to do any therapy or anything...' (Mother of Mako).

'...we haven't, as well, we haven't been allowed to have therapies at school, it's expected that it's out of school.' (Mother of Ginny).

Experiences in navigating the National Disability Insurance Scheme (NDIS: Australia) and working with the school system to access appropriate support for their child was also discussed. Mothers reported issues with understanding what the school's role was in the support their child needed and that they didn't know how to navigate the NDIS system:

'I have found... (child) has been on the NDIS for two years now and I have found since we've been on the NDIS... there seems to be an attitude from the schools that well, your child's on the NDIS now, the NDIS's job is to fix everything, it's not a school responsibility anymore. Yeah. So I'm not sure, that's how I'm sort of left feeling after a lot of the discussions that we have with school.' (Mother of Ginny).

'I think because NDIS don't cover education, it's all a different process. And I think, I don't know. I guess I wasn't sure of the process of how all that funding worked pre starting kindy, because I thought I was on the ball last year getting all the reports and everything, early intervention. And I thought, oh yeah, he's, you know, he's going to get exactly what he needs and what they

can, you know, fund him for, but turns out that I think he needs a lot more and now, so we're kind of stepped back, if you know what I mean.' (Mother of Kane).

3.4 Mothers' Feelings about Their Child's Learning

Mothers discussed their personal feelings of *lack of confidence in supporting their child's learning, developmental concerns and comparing their child to other children, and apprehension about their child's future* as impacting upon their child's learning. A lack of confidence and the idea of 'not knowing' or feeling confused about what stage their child was at cognitively, or where they should be, was discussed as difficult for mothers in supporting their child's learning and in providing information to their child's educators to better support them.

'It's not knowing. I think for me, and, it's just not knowing how much [Child] understands, or what he doesn't understand or where he needs the help... I just feel like I really don't know. Like there's a big question mark.' (Mother of Kane).

'I don't know, is that age or is that a delay or, I don't know. Things like that, it's so hard to benchmark, you don't, yeah... it's hard because, particularly with age but also, I don't really know where he sits on a cognitive level.'... (Mother of Damon).

Mothers discussed how they felt their child was delayed or behind in their learning compared to their peers and at the same time feeling confused about how to measure this, or who should measure this in their child.

'...And I feel like he's probably behind but I don't know, and I'm really sure how to measure that, I guess, or who does.' (Mother of Damon).

'...he's probably developmentally sort of, yeah, a couple of years behind his age I'd say at least... I also found myself throughout the last few years, probably wrongly for me, but finding myself frustrated or impatient with his progress at times, because I mean, not that I was trying to keep him sticking to the set milestones, but because I felt like he, you know, should have got it by now or something.' (Mother of Hudson).

4. Discussion

In the present study we explored the perceptions of 9 mothers relating to the education and learning experiences of their children with 22q11.2DS within early and primary school learning contexts. Overall, the specific aspects of a diagnosis of 22q11.2DS on their child's learning in terms of absences from the educational setting and health concerns that made learning difficult for their child were discussed. Delays or differences in adaptive behaviours and specific behaviours and strategies, such as memory issues, masking, and task avoidance, were perceived as negatively impacting upon their child's learning. It became clear during the focus groups and interview that there were no explicit examples of learning support within educational settings that were designed or tailored to meet the learning needs of their child with 22q11.2DS. The level of support provided by the educational environment was perceived as significantly impactful on learning, particularly the inclusion of therapists and the relationship between educators, parents and the children. Lastly, mental health issues were identified as additional challenges that impacted upon the children's learning.

4.1 22q11.2DS Diagnosis on Learning

The compounding effects of the health problems associated with a 22q11.2DS diagnosis was reported to have significantly impacted upon the children's attendance and their ability to effectively learn. Health issues related to 22q11.2DS led to significant absence from both early childhood education settings and primary school contexts which led to children missing out on extensive periods of time within their learning environments. Adaptive behaviour issues negatively impacted on their child's ability to learn where physically, their child's low tone impacted upon their ability to sit in standard classroom chairs and concentrate and gross motor delays negatively impacted upon children's ability to fully participate and join in with their peers. Communication issues were perceived by mothers to reduce opportunity to fully engage in their learning environment. All of the children in this study received support from speech and language therapists (SLTs), yet there was no discussion around specific communication support or strategies used within their learning environments to facilitate their daily communication. Delays or, the lack of, effective expressive communication skills were described as barriers to full engagement in the learning environment for all children. Delays and differences in early expressive communication skills are hallmark features of 22q11.2DS and implicate all aspects of functioning [13]. Mothers in this study reported that their child was often unable to communicate clearly with their teachers, their peers and that their ability to express themselves and 'repeat things back' were barriers to their learning. There is a real and significant need for children with 22q11.2DS to be supported in using the evidence-based system of augmentative and alternative communication (AAC) within learning environments to support their expressive language and speech intelligibility [Blinded for peer review]. This is evident in the children discussed here as they all have difficulties in expressive communication including speech delays, unintelligible speech, and difficulties in their speech being understood by teachers and peers. Furthermore, the older children were reported to 'not really understand everything that went on.' at school and have trouble interacting with their peers, lending them to be appropriate candidates for AAC. Sleep issues are also prevalent in individuals with 22q11.2DS, aligning with our findings. Participants in the recent study by Ingram [32] found a high prevalence (85%) of sleep disorders in their sample of 100 participants and showed associations between sleep dysfunction and daytime behavioural challenges. In our study, sleep and toileting abilities were identified as significant factors in some of the children's ability to attend school. A specific link was identified for one child where constipation and bowel issues impacted upon their ability to stay accident free and comfortable at school which was exacerbated by poor sleep due to medication and encopresis at night. Our results showcase the wide impact of adaptive behaviours on effective learning and the ability to attend educational settings comfortably and confidently in those students with 22q11.2DS, a contrasting finding to other literature which does not identify such behaviours as significantly impacting directly upon learning [26]. These issues, that are commonly present in learners with 22q11.2DS, did not appear to be supported by educators in our study and was one of the factors involved described by one mother in moving her son from mainstream to a special education unit stating 'So I think being in the special school setting has just alleviated all those problems because I can just send a note to school... and they go yeah, okay, done. You know, it's not a problem. Whereas in a mainstream school it was a real issue...'.

Specific learning strategies and behaviours were developed by children to fly under the radar of teachers and do 'just enough to get by'. As a result, teachers were perceived as unaware that the

child was struggling or had not understood what was happening in the classroom. Strategies such as masking, or task avoidance to divert attention away from their learning issues were employed. Repetition was another strategy used for their child to overcome memory issues and a 'slow' learning pace. It is well established in the literature that working memory (WM) is impaired in children with 22q11.2DS and that the intellectual profile of children with 22q11.2DS will change over time [20]. We know that a portion of children may develop intellectual skills in a similar way to their peers, a portion may 'grow into deficit' and others may experience significant IQ decline [20, 21]. Coinciding with IQ changes around this age, insight into oneself and relationships with others changes too [22]. These strategies are not unique to learners with 22q11.2DS and are observed in autistic students and those with attention deficit hyperactivity disorder (ADHD), among others. Typically, these strategies emerge in children as they become aware of their differences and attempt to account for, or minimise, their different learning styles and difficulties [33], which can be detrimental to their on-going learning, social experiences and mental health [34]. To reduce the need for these strategies to be employed by children, the learning environment first needs to adopt a fully inclusive and supportive ethos that caters for the diverse learning styles and needs of all children [33]. Masking can be reduced when students feel safe and secure with people around them who meet their emotional needs, and where they can be confident in showcasing their authentic self [33, 34]. Task avoidance can be reduced when children feel confident to ask questions and admit that they do not know how to tackle a task, or when greater flexibility is provided in how and when tasks are to be completed [28]. Furthermore, simple strategies, like breaking down tasks and providing visual cues, can support students with WM issues [35].

4.2 Impact of Educational Environment

School was described as a stressful or difficult aspect of many families' lives. Descriptions like stress and feeling 'deflated' by the educational environment were reported. Feeling like the school were not willing to support the idea of developing a plan or problem solving with the parents for effective learning solutions were reported. One family had moved their son from a mainstream setting to a specialised education setting due to a perceived lack of support and understanding. Support also appeared to differ as the children moved from early education centres to primary school contexts whereby more positive experiences were reported from parents who had younger children. While this reflects the nature of public schooling, whereby early education classes are smaller and well-resourced in comparison to larger classes in primary school settings, parents found that the needs of their children were not always met, and educators were less likely to learn about their child. The importance of educators knowledge about 22q11.2DS children is paramount and empirically supported by a recent study finding that communicating with teachers in educational settings was difficult for caregivers, and that children in educational settings were likely to demonstrate high social anxiety that was not understood by teachers [36]. Herein lies the importance of quality transition programs for children who have disabilities where the progression from early learning centres to primary school education requires clear communication between educators, full collaboration of all education personnel involved, and the transparent sharing of child knowledge by all key stakeholders regarding educational and psychosocial strengths and challenges [37]. Further, mothers discussed the lack of extra support to 'push' their children academically, finding it easier to keep their child at lower levels of education. These findings coincide

with literature demonstrating increased challenges for children with 22q11.2DS as they move through the schooling system. The more cognitively challenging learning environment as well as increasing social demands means that children with 22q11.2DS require a greater amount of support that is specifically tailored to their developing needs [10].

These ideas were specifically discussed in relation to the development of Individual Education Plans (IEPs) for children who attended primary school in mainstream settings whereby parents felt the teachers did not value their input into the document and did not seek to integrate parents' expert knowledge of their child or their goals. When was adequate support perceived? When therapists (allied health professionals) were allowed access to the educational environment. Our data show that greater access of therapists in the child's learning environment had a positive impact upon the child's learning and the mothers' perception of support. The mothers of younger children discussed the realisation that the support they had received would not be as easily accessible at primary school where the funding delivery of support mechanisms changed. Specifically, issues involving the National Disability Insurance Scheme (NDIS), an Australian based insurance scheme designed to provide monetary support for those impacted by disability, were discussed. Navigating the NDIS and organising and understanding the different roles NDIS providers and the school were now responsible for was confusing for mothers who now needed to navigate the school's attitude around support for their child when they had accessed NDIS funding. In Australia, the NDIS does not directly support education and needs within classrooms, rather this national insurance scheme is designed to provide funds for families to access, for example, specific allied health support for their child with a disability aimed at increasing functional gains [38]. For those who have children with rare genetic syndromes and complex cases, more needs to be discussed in terms of the type of support available, the role of each support agency, and how these specific supports can be utilised to support educational outcomes.

This presents an opportunity for a transdisciplinary approach to better support young children with 22q11.2DS whereby the medical model of 'Patient Journey' could be adopted [27]. In this model, professional and expert knowledge is integrated with the patients lived experience and expertise to develop a 3-stage framework covering clinical presentation and diagnosis; personal needs and challenges identified by the patients; and mutual goals to improve care. Adopting such a model could create a strategic and supportive relationship between educators, allied health professionals and families of children with 22q11.2DS. For the mothers in our study, feeling that their child was understood, treated equally, and cared for in their learning environment was associated with descriptions of positive relationships with educators. Establishing such positive relationships was particularly important for all mothers as many reported feeling confused around their child's learning abilities and the level at which their child was functioning in comparison to their peers.

4.3 Additional Factors Impacting on Learning

Two mothers of primary aged children specifically identified a change in their children where perceptions of their differences were beginning to impact upon their schooling and learning. In these children, anxiety around school was becoming more of an issue in relation to toileting at school, when isolated from peers and when feeling unsure about what was happening at school. Although these points raised by the mothers are varied, they point to a similar theme regarding

peer status and group belonging. Peer status becomes more important for children as they transition from pre-school environments to primary school [22]. Peer acceptance, belonging and peer relationships become increasingly important as children progress through primary school and into high school environments, and when children feel isolated or rejected, school and learning environments can become very stressful places [18, 22, 39]. This is particularly significant for young people with 22q11.2DS as the compounding impact of delayed or different early communication skills and missed social skill opportunities, children with 22q11.2DS often struggle to make and maintain positive peer relationships [17], are more likely to experience social isolation, and are at greater risk of experiencing bullying and victimisation compared to their typically developing peers [39]. Bullying at school is yet another way to increase stress in learners with 22q11.2DS, and is associated with poorer functional outcomes and a greater risk of mental health issues [40]. Providing opportunities to develop fundamental social-communication skills and coping strategies through targeted early intervention in early childhood centres may positively impact upon the development of early social competency [41] and enable these young children to develop and maintain early peer relationships, all factors that are protective against mental health issues across the lifespan [23, 42].

4.4 Limitations

The results from the current study should be considered with caution due to several pertinent limitations; 1. our sample included children from both primary school-age and pre-school age, and 2. only 9 mothers were included in our focus group. In order to better represent this cohort of young children, a greater and more even sample of children within each age group is needed. As discussed, 22q11.2DS is a heterogenous syndrome that brings with it additional co-morbidities. This heterogenous profile was evident in our small sample yet, a larger sample would also contribute to a greater understanding of this varied profile on educational outcomes for this group of learners. Additionally, our participants were all mothers, limiting our findings to one parental viewpoint. Although it is evidenced that mothers tend to fulfil a primary role in supporting the education of children [43], perspectives from both parents would enrich these findings. Future research should therefore investigate a larger, more representative sample of children within the pre-school and primary-school age group, and actively seek to recruit equal numbers of mothers and fathers to ensure an accurate view of 'parents experiences of education for their child' is explored. Investigating teacher perception on educating children with 22q11.2DS would be valuable in determining the teachers experiences of educating 22q11.2DS students and where they see the need for professional learning or additional support to ensure they are effective educators for these children. Exploring this would likely lead to more collaborative approaches in designing and delivering more specific and appropriate support packages to these learners that capture their physical, psychosocial and intellectual needs.

4.5 Conclusion and Future Directions

Future research must work towards a common framework for providing safe, inclusive and successful learning environments for children with 22q11.2DS across all stages of education [44]. From the current findings, it is clear that support plans for children with regular absences must be developed, safe and inclusive educational environments created, support with early childhood to

school transitions provided, explicit social-communication skill development that focuses on developing and maintaining peer relationships is implemented, and mental health awareness and support should be included in these support packages [45]. Due to the heterogeneity in intellectual profiles of children with 22q11.2DS, and as learners with 22q11.2DS are often good at employing strategies to fly under the radar of their teachers, their learning difficulties may go unnoticed. Therefore, it is recommended to regularly assess learners with 22q11.2DS to adapt to their learning needs and provide the right support at the right time [9, 10]. Furthermore, IQ scores should be monitored as children progress through their education, particularly as social interactions and social demands become more complex, due to the known risks of mental health issues associated with significant IQ decline [8]. One emerging theme from this study was the importance of building supportive and trusting relationships between educators, parents and their children. These relationships are built through the actions of listening to and integrating ideas and knowledge from parents, actively including parents in goal setting and educational decisions for their child in a collaborative way and supporting their children within classrooms. The importance of collaborating and integrating all relevant information, i.e., from teachers, from parents and allied health professionals, is important as knowledge and expertise in rare genetic syndromes is scarce, meaning that children with rare genetic syndromes and their families will often feel uncertain and alone [27]. With the right resources and teacher support at the right time, combined with an integrated network of care, children with 22q11.2DS can experience educational success, providing them and their families with a rich and positive school experience. By collaborating with families and learning from their experience, we can reach this goal.

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Author Contributions

LR designed and led the focus groups and the interview, provided primary analysis of the transcribed data, wrote the draft manuscript and managed the edited document. TL assisted with the focus groups and interview, provided robust discussions and analysis of the transcribed data, and edited the draft manuscript. LEC supported the design of the interview questions, was involved in the delivery of the focus groups, provided discussion of the emerging themes from the data, and provided edits on the draft manuscript.

Competing Interests

Authors report no conflict of interest.

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