

ORIGINAL ARTICLE

The psychosocial impact of 22q11 deletion syndrome on patients and families: A systematic review

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The 22q11 deletion syndrome (22q11DS) is one of the most common genomic disorders in humans, affecting around 1:2,000 to 1:4,000 people. 22q11DS affects multiple body systems and is associated with multiple physical problems. Given the high rate of physical morbidity associated with the 22q11DS, it was hypothesized that it would exert a high psychosocial impact on patients and their relatives. To investigate this, a systematic review of the literature and narrative synthesis was performed. Three major themes emerged. First, the complex and conflicting emotions experienced by family members resulting from the diagnosis. Second, the pervasive educational and health-care challenges associated with the diagnosis and third that people affected by 22q11DS strived for individualism. The results of this review help to inform clinical management of families with 22q11DS.

KEYWORDS

22q11 deletion syndrome, psychosocial, quality of life, systematic review

1 | INTRODUCTION

DiGeorge Syndrome, alternatively called 22q-deletion syndrome (22q11DS) or Velo-facial syndrome, is a rare genetic deletion syndrome, caused by hemizygous deletion of the q11.2 part of chromosome 22 (Goldberg, Motzkin, Marion, Scambler, & Shprintzen, 1993; Swillen, Vogels, Devriendt, & Fryns, 2000). It is thought to be the most common microdeletion syndrome in man (Shprintzen, 2008), and is estimated to occur in 1 in 4,000 live births (Botto et al., 2003; Devriendt, Mortier, Van Thienen, Keymeulen, & Fryns, 1999; Oskarsdottir, 2004)

Its symptoms are complex and affecting multiple body systems, ranging from intellectual disability, psychiatric illness, cardiac abnormalities, immunodeficiencies to epilepsy (Bales, Zaleski, & McPherson, 2010a; Hallberg, Óskarsdóttir, & Klingberg, 2010). The challenges associated with these complex clinical presentations are likely to have a significant effect on the psychosocial well-being of patients as well as their families. Psychosocial well-being refers to both psychological (emotional) and social well-being of the patient and their families. Chronic stressors, ill mental health, care responsibilities, and challenging family environments are all factors that can impact psychosocial well-being (Clair, Fitzpatrick, & La Gory, 1995; Dawson et al., 2016; Humphrey et al., 2015; Jacob, 2013). Patients with psychiatric diagnoses and

learning disability, for example, often report a disconnectedness from social networks (Goodwin, Alam, & Campbell, 2017); while their parents often report on the pressures of full-time care (Anderson, Elliott, & Zurynski, 2013), increased stress (Briegel, Schneider, & Ko, 2008), and struggling with their child's challenging behavior (Hastings, 2002). Additionally, parents of children with a 22q11DS also report a strain on their relationships as a result of the diagnosis (Okashah, Schoch, Hooper, Shashi, & Callanan, 2015). Due to the complexity of 22q11DS, patients require management by multidisciplinary teams from both primary and secondary care which can be time-consuming; with hospital appointments being stressful experiences. This is therefore likely to further impact the quality of life of both patients and families, and thus have a significant impact on the psychosocial well-being.

While research has sought to better understand the experiences of patients and family members caring for individuals with 22q11DS; conducted research has been very specific, such as focusing on perceptions of oral health of children with 22q11DS or the importance of adding 22q11DS to the Newborn Screening. Until now, no attempt has been made to bring together these research findings to provide a more comprehensive overview of the psychosocial impact of 22q11DS on patients and families. Due to 22q11DS being one of the more common rare diseases, this review is therefore needed to inform health-care

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professionals about the experiences of their patients and their families, areas where support from a medical or psychological point is required and aid their understanding of their role as health-care professionals in care of families affected by 22q11DS.

2 | MATERIALS AND METHODS

Evidence for the psychosocial impact of 22q11DS on patients and families was assessed by conducting a systematic review of published research findings. The protocol for this systematic review was registered on PROSPERO (Reference number: CRD42017078110) on November 8, 2017.

2.1 | Search strategy

Five electronic databases were searched: PubMed, Scopus, Web of Science, PsychInfo, and CiNahl in October 2017. The following 10 combinations of searches terms were used: (DiGeorge OR "Di George" OR 22q*) AND (psychosocial OR social), (DiGeorge OR "Di George" OR 22q*) AND (family OR sibling OR adoles*), (DiGeorge OR "Di George" OR 22q*) AND (patient), (DiGeorge OR "Di George" OR 22q*) AND (adult OR child* OR "young adult"), (DiGeorge OR "Di George" OR 22q*) AND (impact OR burden), (DiGeorge OR "Di George" OR 22q*) AND (car*), (DiGeorge OR "Di George" OR 22q*) AND (psych*), (DiGeorge OR "Di George" OR 22q*) AND (quality of life), (DiGeorge OR "Di George" OR 22q*) AND (experience), (DiGeorge OR "Di George" OR 22q*) AND (life or liv*).

2.2 | Inclusion and exclusion criteria

The following criteria had to be met in order for studies to be included: Only English-language papers which reported on original research were considered; samples had to either include patients with 22q11DS diagnoses or their relatives. Studies were required to have a measure, report, or expression of the psychosocial impact of 22q11DS syndrome. Qualitative, quantitative, and mixed method papers were considered. Reviews and case studies were excluded.

2.3 | Study selection

Overall, 3,442 articles were identified and saved to the reference manager "Mendeley." After duplicate removal 1,964 papers remained. All titles were screened. Papers indicating reports of experiences of 22q11DS patients or relatives, all medically focused papers were excluded (e.g., alleles, organ function, etc.). Consequently, 1,906 articles were excluded and the abstracts of the remaining 58 screened.

Fourteen studies were considered in their full-text version, which were all found to match the inclusion criteria for the review. A 15th paper was identified by the anonymous peer reviewer, also screened and added to the review (Reilly, Murtagh, & Senior, 2015). The process is illustrated in the PRISMA diagram (Figure 1).

No formal assessment of methodological quality of the studies was conducted as a result of the range of included methodologies; the majority of studies were qualitative in nature and there is no widely used tool to assess their quality. Furthermore, due to this review being

planned as a narrative and descriptive synthesis of the psychosocial impact of 22q11DS, no meta-analysis was conducted.

2.4 | Data synthesis

The results in this review are presented in a narrative method (as described in Lamore, Montalescot, and Untas [2017]). One of the researchers (OKV) utilized a table to keep a record of findings from each study, and to highlight arising codes for each article. Two researchers then independently merged the codes into themes (OKV, KV). After discussion and comparison, the presented themes arose. Communalities as well as discrepancies are highlighted in the results.

3 | STUDY CHARACTERISTICS

Characteristics of the 14 studies reviewed are shown in Table 1.

3.1 | Time of conduction

The 14 included studies were conducted between 2008 and 2017 (Bales et al., 2010a; Briegel et al., 2008; Chan et al., 2015; Cohen, McCartney, & Crampin, 2017; Costain, Chow, Ray, & Bassett, 2012; Goodwin, Alam, et al., 2017; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Hercher & Bruenner, 2008; Karas, Costain, Chow, & Bassett, 2014; Klingberg, Hallberg, & Óskarsdóttir, 2010; Martin et al., 2012; Okashah et al., 2015; Phillips, Goodwin, Johnson, & Campbell, 2017; Reilly et al., 2015), with the majority published between 2016 and 2017 ($n = 5$).

3.2 | Country of conduction

The majority of studies were conducted in English-speaking countries: Australia ($n = 4$), Canada ($n = 4$), United States ($n = 3$), and United Kingdom (UK)/Ireland ($n = 2$). The other two studies were conducted in Sweden ($n = 1$) and Germany ($n = 1$).

3.3 | Methods

Six studies were qualitative in nature (Bales et al., 2010a; Goodwin, Alam, et al., 2017; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Klingberg et al., 2010; Phillips et al., 2017), seven used mixed methods (Butcher et al., 2012; Chan et al., 2015; Cohen et al., 2017; Hercher & Bruenner, 2008; Karas et al., 2014; Martin et al., 2012; Okashah et al., 2015), and two exclusively quantitative papers were included (Briegel et al., 2008; Reilly et al., 2015). Qualitative methods used included interviews, surveys, and questionnaires. The most commonly used method analysis for qualitative papers was interpretive phenomenological analysis. One study used retrospective chart review.

3.4 | Samples

Six studies had mixed samples of parents, caregivers, siblings, patients, and professionals ($n = 6$) (Bales et al., 2010a; Cohen et al., 2017; Costain et al., 2012; Karas et al., 2014; Martin et al., 2012; Okashah et al., 2015). Six studies focused on experiences of parents and/or caregivers ($n = 5$) (Briegel et al., 2008; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Hercher & Bruenner, 2008; Klingberg et al., 2010; Reilly et al.,

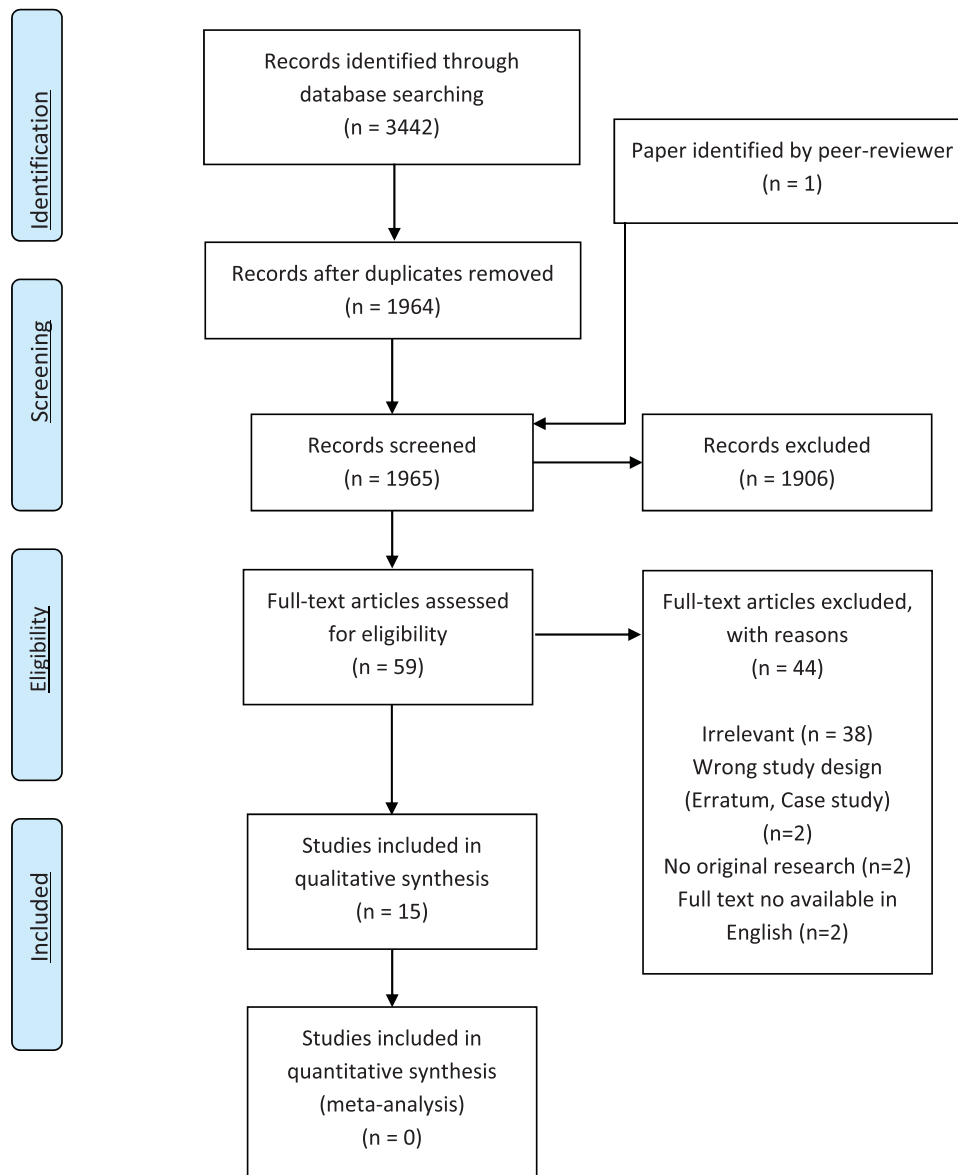


FIGURE 1 Flow diagram of study selection [Color figure can be viewed at wileyonlinelibrary.com]

2015), two studies focused exclusively on patients ($n = 2$) (Chan et al., 2015; Phillips et al., 2017), and one study focused on sibling (Goodwin, Alam, et al., 2017). Sample sizes ranged from 5 to 158; sample size largely dependent on study methodology. Interview-utilizing studies had smaller sample sizes ranging from 5 to 21, averaging 10 participants per study. Samples in studies using mixed methods, such as interviews and surveys/questionnaires, ranged from 34 to 77 with a mean of 57.

4 | RESULTS

Three major themes emerged from the studies included: (1) Conflicting emotions, (2) Challenges associated with health care and educational settings, and (3) Seeking individualism.

4.1 | Theme 1: Conflicting emotions

Thirteen studies revealed negative and positive emotions experienced amongst families, caregivers, and patients (Bales et al., 2010a; Briegel

et al., 2008; Chan et al., 2015; Costain et al., 2012; Goodwin, Alam, et al., 2017; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Hercher & Bruenner, 2008; Karas et al., 2014; Klingberg et al., 2010; Okashah et al., 2015; Phillips et al., 2017; Reilly et al., 2015). Three subthemes were identified: emotions experienced by parents and caregivers, emotional experiences faced by siblings, shared emotional experiences.

4.1.1 | The caregiver perspectives

Eight studies highlighted emotional conflicts experienced by families and caregivers of individuals with 22q11DS, such as feelings of loss, grief, and guilt (Bales et al., 2010a; Briegel et al., 2008; Goodwin, Alam, et al., 2017; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Hercher & Bruenner, 2008; Klingberg et al., 2010; Okashah et al., 2015). Parents' reflections on what life could be like without 22q11DS raised feelings of grief, such as their child losing out on the milestones that a "normal" child is expected to have (Goodwin &

TABLE 1 Study characteristics & summary of results

Briegel, W.; Schneider, M.; Schwab, K. O.	2008	Germany	n = 77 primary caregivers, support groups	Questionnaires (quantitative)	Statistical analysis	Positive correlation between parental stress and aggressive behavior, social and attention problems in their child with time; life satisfaction is not affected.
Bales, A. M.; Zaleski, C. A.; McPherson, E. W.	2010	United States	n = 21 family members and adult patient (17 mothers; 2 fathers; 1 grandmother; 1 affected adult patient), hospital and clinic patients	Semi-structured in- terviews	Interpretive content analysis	Parents are worried about the child's future medical problems and are stressed because of poor medical management. Difficulties transitioning into adulthood due to social and independence factors. Child's dependency affects parents' freedom.
Chan, C.; Costain, G.; Ogura, L.; Silversides, C. K.; Chow, E. W. C.; Bassett, A. S.	2015	Canada	n = 158 adult patients (75 male; 83 female), clinic	Retrospective Chart Review	Statistical analysis	Not all patients received adequate social support from spouse and/or family. Some families showed signs of estrangement toward the patient.
Cohen, W.; McCartney, E.; Crampin, L.	2017	UK	n = 34 parents, caregivers and patient (25 parents, 8 caregivers, 1 patient), sup- port groups	Observational survey (qualitative & quantitative)	Statistical analysis, content analysis	Lack of knowledge and understanding of 22q11DS in schools leave parents fighting for the child's educational needs. Transition of care and educational services cause anxiety in patients and further challenges parents.
Costain, G.; Chow, E. W. C.; Ray, P. N.; Bassett, A. S.	2012	Canada	n = 73 adult patients and caregivers (53 caregivers, 20 adult patients), clinic	Survey (qualitative & quantitative)	Statistical analysis, content analysis	Without a diagnosis, parents felt frustrated, powerless, and uncertain about the future. With a diagnosis, parents felt relieved and saw improvements in how medical specialists interact with the patients and caregivers.
Goodwin, J.; Alam, S.; Campbell, L. E.	2017	Australia	n = 5 siblings (3 female, 2 male), supporting group	Semi-structured in- terview	Interpretive phenom- enological analysis	Siblings psychological grew to become more accepting and empathetic toward their affected sibling despite being jealous and stressed. Confliction of grief, guilt, sadness, and pride for their sibling and their role as future caregivers.
Goodwin, J.; McCormack, L.; Campbell, L.	2016	Australia	n = 8 parents (6 mothers, 2 fathers), supporting group	Semi-structured in- terview	Interpretive phenom- enological analysis	The diagnosis give mixed feelings of relief, guilt and lost; it brings uncertainty make them the center of judgment. Parents overtime learnt to become more empathetic and accepting; they show gratitude and pride in their child's successes. Anger toward the lack of empathy, knowledge, and care provided by health services. Cravings of respite from being a full-time caregiver.
Goodwin, J.; McCormack, L.; Campbell, L. E.	2017	Australia	n = 6 parents (4 mothers, 2 fathers)	Semi-structured in- terview	Interpretive phenom- enological analysis	Experiences of loss, fear, distress, and uncertainty conflicting with feelings of hope, empathy, and gratitude. Lack of emotional investment from health-care professionals leaves them fighting for support. Parents undergo psychological growth as they make sense of the life and role they were given.
Hercher, L.; Bruenner, G.	2008	United States	n = 41 primary caregivers (40 parents, 1 aunt), support group	Online survey (quali- tative & quanti- tative); semi- structured inter- view	Statistical analysis	Psychiatric illness as a clinical manifestation in 22q11DS causes the most anxiety amongst parents (72.5%); second biggest chief complaint (39.5%). 90% of parents were concerned about stigma associated with mental health. 61.5% (n = 24) of participants used the "Internet/other literature" to seek more information about their child's illness.

(Continues)

TABLE 1 (Continued)

Karas, D. J.; Costain, G.; Chow, E. W. C.; Bassett, A. S.	2014	Canada	n = 73 adult patients and caregivers (53 caregivers, 20 adult patients), clinic	Mail survey (qualitative & quantitative)	Statistical analysis, thematic analysis	Psychiatric and behavioral issues are rated the most challenging aspects of 22q11DS in adulthood. Lack of knowledge about neuropsychiatric problems creates uncertainty and worries for the future. Family relationships are affected; caregivers feel alone, stressed, and overwhelmed. Higher satisfaction of social services shown in adult patients than caregivers; but greater knowledge of, access to social and employment services are needed to improve child's independence.
Klingberg, G.; Hallberg, U.; Oskarsdottir, S.	2010	Sweden	n = 12 parents (10 mothers, 2 fathers), hospital patients, organization	Interview	Grounded theory—hierarchical coding	Lack of information provision further increase parents' struggle to manage oral health which contributes toward unmet dental care. Normalization is expressed by patients and parents.
Martin, N.; Mikhaelian, M.; Cytrynbaum, C.; Shuman, C.; Chitayat, D. A.; Weksberg, R.; Bassett, A. S.	2012	Canada	n = 60 parents, health professionals (3 mothers, 1 father, 56 genetic counselors)	Semi-structured interview; questionnaire	Diekelmann's 7 stages; statistical analysis	Disclosing information about risk of psychiatric illness in 22q11DS from genetic counselors ranged from parents who were not alarmed, relief, shock, and fear to extreme anxiety.
Okashah, R.; Schoch, K.; Hooper, S. R.; Shashi, V.; Callanan, N.	2014	United States	n = 41, parents and adolescent siblings ages of 12–17 (28 parents, 23 siblings), support groups	Survey (qualitative & quantitative)	Statistical frequency, content analysis	Life without 22q11DS: siblings speculate better or different relationship with their affected sibling; more parental attention and time; and experience less worries, stress, and guilt.
Phillips, L.; Goodwin, J.; Johnson, M. P.; Campbell, L. E.	2016	Australia	n = 5 patient (5 female), online forums and one health-care setting	Semi-structured interview	Interpretive Phenomenological Analysis	Desire of normality, social acceptance, and individuation from others and themselves. Accepting difficulties associated with 22q11DS such as learning and social interactions; but have aspirations of parenting and independence for the future.
Reilly, C.; Murtagh, L.; Senior, J.	2015	United Kingdom, Ireland	n = 111 parents (total sample: 381, 29% 22q11DS) more details nr.	Survey	Statistical analysis	Significant restrictions on activities, such as holidays, public transport, or shopping trips, experienced by families with 22q11DS patients compared to the comparison groups (Prader-Willi syndrome, Williams syndrome, and Fragile X syndrome). "General health concerns" was most frequently reported by parents of children with 22q11DS. Worries about future mental health changes were also highest among 22q11DS parents, however, not statistically significant from the other groups.

Mccormack, 2017; Goodwin, McCormack, et al., 2017). Parents expressed questions of responsibility for the diagnosis, which led to feelings of self-blame and guilt over potentially failing to protect their children from harm (Goodwin, McCormack, et al., 2017).

Parents also expressed a sense of loss of normality: some mothers, for example, reported an inability to celebrate motherhood as a result of the challenges associated with caring for a uniquely sick baby and caring for a child with increased medical needs (Goodwin, McCormack, et al., 2017).

A number of studies revealed strains on familial relationships due to the severity of 22q11DS (Karas et al., 2014), leading to marital tensions occurring and contributing to divorce (Bales et al., 2010a). They expressed that their child's pain became theirs and felt as though they were lost and entangled (Goodwin, McCormack, et al., 2017). Parents and caregivers found themselves feeling stressed, alone, and overwhelmed by the pressure to deliver constant care (Karas et al., 2014) and yearned for respite (Bales et al., 2010a; Goodwin, McCormack, et al., 2017).

Speculation about the child's future care imposed worries on families (Karas et al., 2014; Reilly et al., 2015), the most prominent one being finite aging, whereby parents believe that one day they will not be healthy enough to take on the role of being principal caregivers (Karas et al., 2014).

Interestingly, one study reported that the life satisfaction of their sample of parents and caregivers' did not seem affected even though changes in a child's behavior have proven to increase stress (Briegel et al., 2008).

4.1.2 | The sibling perspective

Similar to emotional experiences of parents and caregivers, siblings also expressed frustration in their family situations. They shared that having a brother or sister with 22q11DS meant receiving less parental attention, which can cause jealousy and consequently led to guilt and shame knowing that it is not their sibling's fault (Goodwin, Alam, et al., 2017; Okashah et al., 2015). Yet, some considered feelings of guilt and shame irrelevant. In order to cope, siblings developed the habit of inhibiting their feelings and avoiding stressful situations (Goodwin, Alam, et al., 2017).

The reviewed evidence showed that unaffected siblings were well-informed by their parents about genetic, behavioral, and medical information about 22q11DS—despite only 41.7% of parents reported discussing the future care of their affected sibling with their unaffected children (Okashah et al., 2015). For example, children with 22q11DS may need continuous care from a caregiver or family member, in certain cases, siblings (Okashah et al., 2015). Another study reflected on this by showing older sibling's awareness that 1 day they have to take on the role of being the main caregivers when their parents get older (Goodwin, Alam, et al., 2017). Mixed emotions were recorded upon this future responsibility: a sense of sorrow knowing that they will have to sacrifice their time, but also the pleasure of being next appropriate person in line to make their affected sibling happy. In contrast, one younger sibling indicated little responsibility toward their sibling's

future and said they would leave it to their parents (Goodwin, Alam, et al., 2017).

4.1.3 | Shared emotional experiences

However, despite feelings of loss, grief, and guilt, parents and siblings expressed having learnt to come to a rational acceptance of their child's or sibling's diagnosis and the associated limitations (Bales et al., 2010a; Costain et al., 2012; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Phillips et al., 2017).

Challenges experienced in parenting a child (Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017) and caring for a sibling (Goodwin, Alam, et al., 2017) with 22q11DS were viewed as an opportunity to grow psychologically (Costain et al., 2012; Goodwin, Alam, et al., 2017; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Okashah et al., 2015). They also expressed finding themselves to become more patient and empathetic. Parents were able to redefine their purpose in caring for their child and focus on what they are able to do rather than what they lack of (Goodwin & McCormack, 2017). Furthermore, families reported being able to transform feelings of guilt into pride as parents encourage their child to achieve and siblings celebrate their successes (Goodwin, Alam, et al., 2017; Goodwin, McCormack, et al., 2017). Furthermore, one study reported that the majority of parents surveyed felt their child contributed positively to their family by bringing joy and happiness to their lives (69%), and teaching family members to be more patient (57%) and compassionate (51%) (Reilly et al., 2015).

4.2 | Theme 2: Challenges associated with 22q11DS in the medical and social services

Complex clinical manifestations of 22q11DS mean that meeting the needs for every child is a unique and multidisciplinary process requiring the involvement of health, social, and education services (Cutler-Landsman, 2013). Within this theme, three subthemes were identified: road to diagnosis, facilitation of services after the diagnosis, and stigma associated with 22q11DS.

4.2.1 | Road to diagnosis

Patients are diagnosed with 22q11DS at different stages in their lives and early recognition can significantly change the medical management, follow-up and genetic counseling, which are useful for the patient, family, and clinicians (Bassett et al., 2011; Kapadia & Bassett, 2008). Across the included studies, many parents reported feelings of powerlessness and frustration as a result of having to wait years for a diagnosis for their child (Bales et al., 2010a; Cohen et al., 2017; Costain et al., 2012), which resulted in increased uncertainty about the child's future (Bales et al., 2010a; Cohen et al., 2017; Costain et al., 2012; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Karas et al., 2014; Martin et al., 2012). They also described a sense of relief when a diagnosis was given, and perceived that early justification for the child's symptoms can help psychological and clinical aspects of the illness earlier on in life (Cohen et al., 2017; Costain et al., 2012; Goodwin, McCormack, et al., 2017; Martin et al., 2012). Parents have praised the

health care they received: one mother spoke about a pediatrician making the right referral in the process of finding a diagnosis (Bales et al., 2010a); another spoke about improved interactions between medical specialists and caregivers and patients when the clinicians were aware of the diagnosis (Costain et al., 2012). On the other hand, other experience revealed that health-care services have “brushed off” their attempts in trying to seek an early diagnosis for their child and this was prominently found in “inexperienced” and “new parents” (Bales et al., 2010a).

4.2.2 | Facilitation of services after diagnosis

A genetic diagnosis can affect one's lifelong development and understandably, it can be a very challenging and difficult experience for the affected families (Hercher & Bruenner, 2008), thus it is important for professionals to keep the family well-informed, and to facilitate and offer good supporting services.

A common finding concerning the lack of empathy and knowledge around 22q11DS was reported amongst educators and health-care professionals (Cohen et al., 2017; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Karas et al., 2014; Klingberg et al., 2010; Martin et al., 2012; Phillips et al., 2017), this exacerbated the frustration and left many parents feeling like they had to fight for the care and support that their child deserves (Cohen et al., 2017; Goodwin & McCormack, 2017). As a result, many parents and caregivers expressed feeling hopeless and worried. One study reported the majority of their parents (62%) becoming dependent on the Internet and online literature to overcome the lack of information given by health-care professionals and help reduce frustrations (Hercher & Bruenner, 2008; Martin et al., 2012). Lack of advice from dentists made it difficult for parents to keep up with daily oral care for their child, subsequently, patients lacked motivation and energy to maintain good oral health because they did not know its importance (Klingberg et al., 2010). Gaining a better understanding and knowledge about the medical condition was also desired amongst patients, they explained that it would help them clear up the confusion associated with the uncertainty shaped by 22q11DS (Karas et al., 2014).

4.2.3 | Stigma associated with 22q11DS

Further, escalating the frustration of educational and medical services explained above is the perceived stigma associated with 22q11DS. It is thought to be affecting the care participants received (Bales et al., 2010a; Cohen et al., 2017; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Hercher & Bruenner, 2008; Martin et al., 2012).

More generally, parents expressed that stigma associated with the diagnosis of 22qDS, and associated, learning disability made them and their child the center of judgment in schools and health-care settings, they felt labeled and challenged in seeking the right care and support (Bales et al., 2010a; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Hercher & Bruenner, 2008).

Stigma was also experienced in relation to psychiatric illness, as 22q11DS patients are at higher risk for developing psychiatric illnesses, such as mood disorders, psychosis, or schizophrenia (Tang et al., 2014).

The studies report that stigma experienced in relation to mental health, did not only appear to affect societal perceptions but also the possibility of parents unconsciously treating their child differently. This may be partly mediated by the negative portrayal of psychiatric illness in the media (Martin et al., 2012).

4.3 | Theme 3: Seeking individualism

Transitioning from childhood to adulthood and being able to live independently can be challenging for individuals with complex illnesses and disability. Across the studies, it was found that seeking individualism and independence are advocated amongst patients over the course of living with 22q11DS (Bales et al., 2010a; Karas et al., 2014; Phillips et al., 2017), and also a concern for parents of children with 22q11DS (Reilly et al., 2015). Female adult patients are not dissuaded from plans for marriage and parenthood, they express wanting to care for their future children in a manner that will enable them to have normality and acceptance in the world (Phillips et al., 2017). Longing for support of normalization can also be specific, for example, children diagnosed with 22q11DS wanting good oral health care like their peers (Klingberg et al., 2010).

However, studies have shown that achieving individuation can be influenced by various factors. The transition of care from childhood to adulthood is recognized to be difficult from social and medical points of view (Bales et al., 2010a; Karas et al., 2014; Phillips et al., 2017). Findings present the most challenging medical issues experienced in adulthood to be psychiatric and behavioral problems (Karas et al., 2014). In addition, patients with 22q11DS gained greater autonomy as expectations from the society, their families, and themselves increase with adulthood (Goodwin, McCormack, et al., 2017; Karas et al., 2014; Klingberg et al., 2010). Yet, parents felt pressured and frustrated adjusting to these expectations. Parents propose limits and set boundaries in order to provide the right care for their child but find themselves faced with dilemmas (Goodwin, McCormack, et al., 2017; Klingberg et al., 2010). These include parent's struggle to allow children to make mistakes, and relinquishing control in their day-to-day routines such as eating habit. Still, patients stated the importance of family support in emotional and practical aspect of living with the disability, in particular, mothers were seen as the primary supporter (Phillips et al., 2017). Contrastingly, inadequate social support was evident in the family and/or spouse of patients who experienced pregnancy and child birth; some were estranged from their families (Chan et al., 2015).

Patients hunger for knowledge and better social and employment services in order to promote their own independence (Karas et al., 2014). A story sharing an individual's happiness by working has been shown by one study (Bales et al., 2010a).

5 | DISCUSSION

The psychosocial impact of an illness refers to its psychological (emotional) and social impact on the patient themselves, and their family members. The current review aimed to provide a comprehensive overview of the psychosocial impact of 22q11DS on patients and

families. Overall, across 15 studies, three major themes were identified (“Conflicting emotions,” “Challenges associated with 22q11DS in the medical and social services,” and “Seeking individualism”) that collectively contribute to the psychosocial impact of 22q11DS on patients and families.

This review found reports of simultaneous positive and negative feelings (Bales et al., 2010a; Cohen et al., 2017; Goodwin, Alam, et al., 2017; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Hercher & Bruenner, 2008; Klingberg et al., 2010; Okashah et al., 2015; Reilly et al., 2015). Caregivers and siblings frequently reported feelings of loss, grief, and guilt not only when caring for their affected child or sibling, but also about becoming more accepting and empathetic. Unique challenges, such as exhaustion from full-time care, frustrations, and uncertainties surrounding the process of diagnosis and service facilitation postdiagnosis were frequently reported. The sibling perspective revealed both feelings of jealousy and avoidance, as well as awareness for future responsibilities as their sibling’s caregivers.

Reports of mixed emotions for parents of children with developmental disability are reported elsewhere and include joy, hope, love, and strength as well as anguish and sorrow (Kearney & Griffin, 2001).

Parents caring for a child with a developmental disability have an increased likelihood to develop anxious or depressive symptoms (Hartling et al., 2014; Miodrag & Hodapp, 2010; Singer, 2006). The uncertainty surrounding the diagnosis and child’s life may be contributing factors to this (Mishel, Padilla, Grant, & Sorenson, 1990). Higher risk of developing mental health problems was also found in siblings unaffected by 22q11DS, but research in typically developing siblings of children with autism spectrum disorders has shown that they manage largely well and are often resilient (Dempsey, Llorens, Brewton, Mulchandani, & Goin-Kochel, 2012; Green, 2013). If stress reaches the peak where a family struggle to cope, crises are likely to occur (Figley, 1998). In the case of caring for a child with developmental disability, like 22q11DS, if the family endures a crisis for a long duration of time, they are therefore exceedingly susceptible to burnout (Jackson & Maslach, 1982). Unsurprisingly, levels of clinical burnout in parents of disabled children are significantly higher (38%) than parents with normally developed children (20%; (Lindström, Åman, & Norberg, 2009). Participants in the included studies reported comparing themselves to other parents their age, feeling lost, overworked, and yearned for respite. Therefore, these parents are a high-risk group for burnout. Figley (1998) specified that burnout may include act of over engagement and avoidance, fatigue, listlessness, and loss of empathy, and those exposed or are involved in chronic illness, are more vulnerable. Therefore, it is important to consider not only parents risking burnout, but also siblings of the affected child too. Findings from the review reinforce this because siblings actively pursued the act of avoidance in stressful family situations associated with 22q11DS. An opportunity arises to explore the cause of this potential hardness in siblings and how positive and negative outcomes are affected by the family system (Goodwin, Alam, et al., 2017).

It could be argued that unpredictable challenges (Rolland & Walsh, 2006) associated with 22q11DS may be impacting the mental health of family members: uncertainty surrounding 22q11DS has evidently

caused anxiety to participants included in this review as well as feelings of guilt and self-blame. However, no included study specifically assessed the mental health of caregivers, parents, or siblings alike, and depressive symptoms were not clearly stated.

The notion of “stigma” within health, social, and educational services was prominent in the results. Indeed, it has been well documented that parents with children with disabilities constantly face stigmatizing encounters as shown by the studies (Bales et al., 2010a; Cohen et al., 2017; Goodwin & McCormack, 2017; Goodwin, McCormack, et al., 2017; Hercher & Bruenner, 2008; Martin et al., 2012). Although parents and caregivers recognize the stigma imposed on them, it remains unclear as to whether they addressed this issue in order to reduce their frustrations. A study by Manago, Davis, and Goar (2017) interviewed parents with children with disabilities found that parents tend to adapt themselves and their families in response to social oppression caused by stigmatization. Choosing to deflect instead of challenging in this instance could be steered by the children’s needs and the practicality of getting with the day without disruptions (Manago et al., 2017), and further evoke psychosocial sufferings in families living with 22q11DS. Implied education of the general public and health-care professionals in order to help diminish stigmatization is therefore much needed (While & Clark, 2010).

As life expectancy in most infants with 22q11DS has now reached to adulthood and beyond (Bassett et al., 2009; McDonald-McGinn & Sullivan, 2011); there are increasing numbers of adult patients with 22q11DS. Yet, patient psychosocial perspectives were limited; limited findings highlighted patients’ desires to obtain independence, challenges to achieving individualism, and transitioning into adulthood. Reasons for this may include limitations in patients’ social functioning disproportionate to their capabilities in other aspects of daily living (Angkustsiri et al., 2012; Bassett et al., 2011; Butcher et al., 2012; Ho et al., 2012), difficulty maintaining social relationships (Abery & Fahnestock, 1994; May & Simpson, 2003), and having small social circles (Amando, 1993; Knox & Hickson, 2001). However, these patterns were not necessarily found for all 22q11DS patients included in this review: a number of female patients in (Phillips et al., 2017) express strong desires to marry and become good parents. Although these are encouraging findings, there may be some underlying justifications that need to be considered. Patients living with 22q11DS long for normality and acceptance in the society. Therefore, their desire to fit in may be motivated by social pressure—especially as 22q11DS patients have been found to compare themselves to healthy peers (Phillips et al., 2017). In addition, patients’ awareness in sociocultural barriers to successful parenting were not extensively discussed, these includes: low income, societal judgments, lack of respect, and social support (IASSID Special Interest Research Group on Parents & Parenting with Intellectual Disabilities, 2008). Findings from Chan et al. (2015) indicated these sociocultural barriers, as some patients lacked social support from their families, and some had abusive spouse. It becomes obvious that more research and subsequent understanding of this critical transition from childhood to adulthood from the patients’ perspective is required; such research will also aid the service facilitation and provision.

Given the complexed manifestations of 22q11DS, psychological distress prediagnosis and postdiagnosis was experienced by parents in the process of searching and receiving a diagnosis, this was also found in a study by (Bales, Zaleski, & McPherson, 2010b). Diagnostic certainty is linked to psychological benefit (Graungaard & Skov, 2007; Lenhard, Breitenbach, Ebert, Schindelbauer-Deutscher, & Henn, 2005; Rosenthal, Biesecker, & Biesecker, 2001) in parents, who made up the majority of samples in this review. Meanwhile, from the clinicians' point of view, a diagnosis may equate a platform to improve medical management (Costain et al., 2012). Although there was a sense of validation and relief with a diagnosis, to some degree, better access of services could be reached, but one cannot guarantee perfect care for the affected child.

Despite the challenges associated with 22q11DS, family members in the included studies also recognize the distress as an opportunity to embrace their experiences and grow psychologically. This adaptation is described in the theory of meaning-based coping, where the experience of negative psychological states can motivate people to create positive psychological states (Folkman, 1997). Graungaard and Andersen (2011) stated that other studies have also shown this type of positive reappraisal in parenting a child with severe disability. However, what remains unclear from the included studies is which sources support this positive coping and how it integrates with the conflict of emotions experienced by families and caregivers (Goodwin & McCormack, 2017).

There is a sparsity of papers investigating differences in psychosocial impact between different genetic syndromes. While it was not the aim of this review to address whether 22q11DS has a more profound psychosocial impact than other conditions, one of the included papers (Reilly et al., 2015) assessed the impact of 22qDS in comparison to Prader-Willi syndrome, Fragile X syndrome, and Williams syndrome. Overall, children with 22q11DS were rated as being less challenging than children with the other conditions, the differences however were not statistically significant. However, only cautious, comparative conclusions can be drawn from this one study.

5.1 | Limitations of the review

There are a number of limitations of the review: first, English language-only original studies were included, which may have biased the review. Furthermore, a relevant paper was identified by the peer reviewer that would have otherwise been missed. This suggests a possible limitation of the search strategy.

Second, although the included studies include diverse samples, the patient's perspectives were the least represented group. In addition, the majority were adult-only samples with the exception of one 16-year-old participant. Given the nature of 22q11DS, it is understandable that recruiting patients is more difficult in comparison to recruiting parents, for example. However, higher numbers of participating patients would provide more robust findings.

Third, another imbalance in the sample size was that a higher number of mothers were involved in the research compared to fathers. Some studies did not specify the number of mothers and fathers in the sample "parents", but of those that did, the number of mothers ($n = 40$) were almost five times the number of fathers ($n = 9$). A better

representation of fathers and their role as a parent living with a child with 22q11DS, compared to mothers, is needed.

Most of the studies claimed that family members came to acceptance of living with 22q11DS patients, but such positivity may not be representative for all families. First, this could be justified by strained relationships experienced amongst the participants in the studies: having an abusive spouse and threatened marital status. Second, self-reported data present some limitations, for example, families may have been reluctantly recalled negative events, and exaggerated positive events. Third, occurrence of sample bias was likely as families who had negative experiences may have not volunteered.

Lack of evidence indicative of mental health issues amongst the families could be due to the restrictive format of surveys, questionnaires, and the style of interview used in the studies. One study did not specify the type of interview conducted and described it to be "conversational style" (Klingberg et al., 2010). Whether mental health was caused or became exacerbated in families caring for people living with 22q11DS, it is importance to explore this issue to provide additional support.

6 | CONCLUSIONS

The diverse psychosocial impact experienced by families and patients living with 22q11DS proved to be significant and more apparent in the medical and social management. Vigorous and constructive methodology in research is needed to seek other influencers that contribute to the psychosocial impacts, such as sociocultural factors. In addition, more efforts are required to gain better perspectives from patients and fathers. This review will be a source for clinical applications and help facilitate better services for families and patients affected by 22q11DS.

CONFLICT OF INTEREST

None.

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