

# Positive and Negative Experiences of Parenting a Pre-school Child with 22q11.2 Deletion Syndrome

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**Abstract** The presentation of 22q11.2 deletion syndrome (22q11DS) is symptomatically variable, presenting diagnostic challenges for paediatricians and anxious uncertainty in parents. The ‘lived’ experience of parenting a small child diagnosed with 22q11DS is unknown particularly how parents make sense, both positive and negative, of their role. A phenomenological study sought subjective interpretations from two fathers and four mothers of a young child (8–36 months) diagnosed with 22q11DS. Using interpretative phenomenological analysis, data were collected through semi-structured interviews and thematically analysed. Four themes embodied uncertainty and fear simultaneously experienced by these parents such as anticipatory traumatic distress, systemic stigma, confusion at professional smoke screens and ‘not knowing’. This cacophony of distressing emotions plagued their unanswered fear, guilt, loss and grief. In seeking hope, their parenting journey was able to be redefined as one of intrapersonal and interpersonal growth from the adversity of 22q11DS. This study provides a lens into the experiences of parents struggling to make sense of their journey with 22q11DS in the early parenting years. It highlights the complex distress of parenting a small child whose development is fraught with uncertainty and where stigma and suspicion are often experienced when seeking support from social and health systems. However, a metamorphosis of personal strengths also emerged facilitated

by hope. Healthcare professionals can best assist through open discussion that acknowledges often limited knowledge, using the guidelines for management of 22q11DS in guiding parents.

**Keywords** Velo-cardio-facial syndrome · IPA · Anticipatory trauma · Psychological growth

## Introduction

The ongoing care required by a child diagnosed with a developmental disability creates chronic levels of stress for many parents (e.g. Stuart and McGrew 2009). Initial diagnostic uncertainty and subsequent prognosis is often exacerbated by the demands on family life from chronic or acute medical issues that may arise associated with the particular disability (Stewart and Mishel 2000). The psychological well-being of parents of children with 22q11.2 deletion syndrome (22q11DS), a poorly understood syndrome, is unknown. Given the wide variability of symptoms possible in the presentation of 22q11DS, parents are likely to experience varying degrees of psychological distress pre- and post-diagnosis while seeking and receiving a diagnosis and raising and caring for their child (Bales et al. 2010). Therefore, it is important to explore the subjective interpretations of parenting a pre-school child with 22q11DS, particularly the positive and negative interpretations of their lived experiences of this phenomenon.

The family social system can be disrupted or incapacitated by family crises (Burr 1973). Crises will occur when stress reaches the peak at which a family can no longer cope (Figley 1998). The most important predictor of parental stress in parents who have a child with an

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intellectual disability is parental negative attribution of their child's behavioural problems or social acceptance (Saloviita et al. 2003). Negative appraisal is associated with higher individual, marital and family burden in parents with a child with autism spectrum disorder (Stuart and McGrew 2009). Conversely, psychological well-being is promoted through positive and problem-focused coping (Pozo et al. 2014). Should families endure a crisis state for a long period of time (as may be experienced when a child has a developmental disability), they are extremely vulnerable to burnout even though a crisis is considered to be temporary (Maslach and Jackson 1982).

Burnout can include oscillation between avoidance and over engagement, listlessness, fatigue and loss of empathy, and those exposed to chronic illness and care of others are particularly susceptible to burnout (Figley 1998). Studies indicate that parents of children with disabilities experience significantly elevated levels of clinical burnout (38%) compared to parents of healthy children (20%; Lindström et al. 2009). Importantly, and despite experienced distress, current research recognises that struggles with adversity can provide a platform for positive change and stress-related growth personally, socially or in terms of coping (Park et al. 1996). For example, Cheshire et al. (2010) found that in parenting a child with cerebral palsy, conscious engagement in positive reinterpretation by parents helped them find meaning, reduce depression and stress.

The developmental disability 22q11DS (also known as velo-cardio-facial syndrome [VCFS]) is a unique condition with large interfamilial and intrafamilial symptomatic variability (Shprintzen 2008). More than 180 features are associated with the syndrome including characteristic facial features, congenital heart defects and palatal anomalies (McDonald-McGinn et al. 1999). 22q11DS can manifest through intellectual disability and/or learning problems, with specific cognitive impairments in executive dysfunction (Bish et al. 2005), attention deficits (Niklasson et al. 2005) and social impairments (Shashi et al. 2012). Compared to the general population, people with 22q11DS are at greater risk of experiencing autism spectrum disorders (Fine et al. 2005), anxiety disorders (Fung et al. 2010), mood disorders (Green et al. 2009) and psychotic disorders (Murphy et al. 1999). Despite 22q11DS occurring in 1 in 4000 live births (Oskarsdottir et al. 2004), little is known about the subjective experiences of parenting a child with this syndrome nor the impact on the immediate family functioning. The uncertainty that typically accompanies the diagnostic process and also the prognosis may be heightened in this population as compared to parents with children who are diagnosed very early with a more 'predictable' developmental disability. Therefore, it is important to learn more about parents' lived experience as related to 22q11DS.

Mishel's (1990) reconceptualisation of the uncertainty in illness theory explains uncertainty as the inability to find meaning in illness-related events. Parents who have a child with intellectual disability of unknown origin have elevated emotional strain and regret compared to parents with a known disability or no disability (Lenhard et al. 2005). Parents who perceive less personal control over their child's condition experience greater uncertainty, ambiguity and lack of clarity (Madeo et al. 2012). Conversely, flexibility that can occur through uncertainty in parenting a child with a chronic medical issue can facilitate psychological shifts and adaptability for integrating uncertainty more positively into the individual's worldview (Mishel and Clayton 2008). As yet, there is little to inform systems of care on the experience of families with 22q11DS.

Therefore, the aim of this study was to explore the 'lived' interpreted experiences of parenting a young child with 22q11DS, that is, (a) how the participant made sense of parenting a child with 22q11DS, (b) if and how they have changed and (c) how they expect their future will be influenced by these experiences.

## Method

### Participants

Participants were two fathers and four mothers (including two married couples) who had a young child with de novo 22q11DS. The parents' ages ranged from 29 to 42, and the children's ages ranged from 8 months to 3 years. Participants were recruited via a study information pamphlet through online forums and one healthcare setting. Demographic characteristics of participants and their children are outlined in Table 1. Pseudonyms are used to protect the participants' confidentiality.

### Procedure

Following university ethical approval, potential participants who met the study criteria (a parent of a child with 22q11DS aged 3 years or younger) were sent the study information letter, consent form and demographic questionnaire so that any queries arising from this initial contact could be addressed prior to interview. A time and a place suitable to the final participants were organised for the collection of data, which was conducted using semi-structured interviews concerning the phenomenon under investigation. Written consent was collected prior to commencing the interview. The first author conducted three interviews via telephone and three at participants' homes (as chosen by the participants). The interview sought to investigate both positive and negative

**Table 1** Participant and child characteristics

| Participant | Child  |              |                |        |           |                  |                                    |
|-------------|--------|--------------|----------------|--------|-----------|------------------|------------------------------------|
|             | Gender | Age          | Marital status | Gender | Age       | Age of diagnosis | Developmental ability <sup>a</sup> |
| 'Stephanie' | F      | 42           | Divorced       | M      | 21 months | 9 months         | 3                                  |
| 'Elizabeth' | F      | 29           | Married        | M      | 8 months  | 1 day            | 6                                  |
| 'Eric'      | M      | Not provided | Married        |        |           |                  |                                    |
| 'Deborah'   | F      | 36           | Married        | M      | 3 years   | 9 months         | 3                                  |
| 'David'     | M      | 42           | Married        |        |           |                  |                                    |
| 'Frances'   | F      | 36           | Married        | M      | 2 years   | 8 months         | 5                                  |

<sup>a</sup> The child's developmental ability as rated by the parents on a scale of 1–7, where 1 = severely delayed and 7 = not delayed at all

interpretations of the lived experience of parenting a child with 22q11DS, through tunnelling down to the phenomenon of interest (Smith 1996). The interviews were digitally audio-recorded and lasted between 47 min and 1.5 h. Participants were reimbursed for their time with a \$20 gift card.

### Data Analyses

The analysis was based on procedures described by Smith et al. (2009) (see Table 2). Interviews were transcribed verbatim and analysed one at a time. All data sets were de-identified with a pseudonym. The first stage of analysis involved re-listening to the interview and re-reading the transcript. Next, relevant items and psychological constructs were noted in the margin of the transcript as emergent themes. Higher-order themes and subthemes were listed in a table and grouped based on different aspects of the parent's experience. This procedure was carried out by authors 1 and 2 independently. The subjective interpretation of each author gradually proceeded from descriptive to interpretative then particular to shared (Smith et al. 1999). It was also an iterative and inductive cycle (Smith et al. 2009). Finally, a narrative analytic

account was used to link theory to themes generated through pertinent verbatim extracts from transcript. Conclusions were drawn in terms of the similarities and differences amongst participants. Throughout the process, the researchers consistently referred back to the transcripts and recordings to ensure that they were staying true to the data.

### Results

Four major themes emerged: (1) agony of uncertainty, (2) systemic stigma, (3) the pain and the gain and (4) making sense of this life: growing me. These four themes embodied the uncertainty and fear that at times threatened to overwhelm these participants as related to a diagnosis of 22q11DS. Simultaneously, they experienced future trepidation, systemic stigma, confusion at professional smoke screens and not knowing. Heightened fear is agonisingly packed with distress, grief, guilt and hope. Despite unrelenting distress, they actively defied themselves to reframe fear and negativity allowing unexpected intrapersonal and interpersonal growth from the adversity of 22q11DS (Table 3).

**Table 2** Stages of interpretative phenomenological analysis

| Stage | Description  |
|-------|--|
| 1     | Re-listening, transcription, reading and re-reading  |
| 2     | Developing emergent themes through independent interpretation (e.g., noting)   |
| 3     | Credibility established through robust author discussion   |
| 4     | Repeating stages 1–3 for the other five cases  |
| 5     | Searching for connections across emergent themes, identifying convergence and divergence   |
| 6     | Clustering of the themes 'agony of uncertainty', 'systemic stigma', 'the pain and the gain' and 'making sense of this life/growing me' |
| 7     | Reviewing transcripts to validate interpretations in the results   |

**Table 3** Summary of superordinate themes

| Themes                               | Description   |
|--------------------------------------|---|
| Agony of uncertainty                 | Participants' lives are invaded by grief and sadness, particularly prior to the diagnosis when they are the only one searching for an answer to their child's symptoms. Anxious uncertainty about how the syndrome is/will manifest is constant. Paradoxically, not knowing brings hope for the future. |
| Systemic stigma                      | Healthcare services increase feelings of uncertainty and frustration. Participants must fight to get the support that their child needs.  |
| The pain and the gain                | Conflicting emotions exist simultaneously within participants. They feel loss, fear and distress while at the same time experiencing gratitude, empathy and hope.   |
| Making sense of this life/growing me | Participants actively search for meaning surrounding their child's 22q11DS. They view their challenges as an opportunity to grow, finding new love and meaning in their lives.  |

### Agony of Uncertainty

#### *Grief/Sadness/Loss of Expected Role*

Prior to the diagnosis, participants experience doubt about their child's symptoms. Suspicions invade their lives, and active imaginations conjure up worst-case scenarios, bringing distress and grief:

I felt like I was going insane but I knew there was something that wasn't right, and I was dreaming up terrible things ... We couldn't think of anything to ask other than would he have reduced life expectancy? [Deborah]

Participants describe being voiceless victims. Their instincts are dismissed by healthcare professionals who perceive their training to outweigh parenting knowledge. A sense of helplessness arises:

They didn't want to listen to me. They just... It was appalling. [Stephanie]

Parents are stuck as the lone searcher. They cannot shake the feeling that something is being missed. Frances in particular was disturbed by premonitions:

The number 22 just kept going round and round my head every night. Honestly. It was like 22, 22...like, I know he's got it. [Frances]

In contrast, Elizabeth did not have these suspicions; which brought about shock and sadness after her seemingly healthy child was born. Robbed of the choice and control over her child and her own life, she feels let down and misled:

It was like doomsday to me when he said it... I know that if it had been picked up, I would have chosen to abort, and so life would look very different for us at this point. [Elizabeth]

Although the diagnosis should have been picked up prenatally, careless work meant that it was missed. Elizabeth feels as though she is missing out on the life she has worked so hard to build for herself. She must deal with the consequences of someone else's mistake for the rest of her life.

#### *In Limbo/What Is This About?*

This theme highlights the uncertainty that emerges from having a young child with such a variable syndrome. Participants wonder how the diagnosis will reveal itself as their child grows. They find it difficult to enjoy the moment as they are caught up in continuous contemplation of what the syndrome is about:

What the fuck's going to happen? I have no idea what VCFS really means... because there's so many varying factors ... So that's where I'm stuck ... I just don't know. I'm in limbo. [Stephanie]

The ambiguity of 22q11DS brings about an anxious need to prepare for any medical, physical or emotional issues that may arise. But how can one anticipate the unexpected? The enormity of battling an unknown enemy is an overwhelming responsibility. Restless questioning and investigation ensue:

What sort of support will he need? Will he need to go to a special school? ... How will he cope? ... Will he develop a mental illness later in life? Will he be able to function in society, and will he be happy?! Will he...be healthy? ... What curveball am I going to get thrown? [Frances]

Interestingly, Eric was the only participant who avoided this rumination. He did not feel the need to learn about the syndrome, which saves him from the tormented enquiry:

We didn't delve too much into the literature and things that were given to us... We know there will be some challenges moving forward as he develops, but it this stage it hasn't really changed our lives at all [Eric]

Mixed up amongst fear of the unknown is a cautious hope. The participants dare to dream that their child will be one who is less affected than most with 22q11DS. They still dream for a normal or even exceptional life for their child:

He'll be missing a piece of his alphabet but don't write him off. [Deborah]

#### *What Is My Child and What Is the Syndrome?*

Participants also question how well they know their child and in knowing, how best to care:

He's not doing the signs of what I've been told and what I've looked up... is it really VCFS doing that, or that because he had such a bad birth?! I'm going back around in circles again. [Stephanie]

They are unsure which joys and challenges are simply part of who their child is or because of the 22q11DS:

That's always something that I struggle with ... with the medical things ... with the feeding issues ... with the sleeping, we know that that is linked into the syndrome. But behaviour I know that they can have ASD, ADHD, anxiety. But what is normal boy, two-year old behaviour anyway? So it's very hard to pinpoint what is...normal or not. [Frances]

#### *Cacophony of Fears*

Due to the uncertainty, the participants are constantly haunted by a cacophony of fears. They grieve for themselves and the life that they dreamed of having. They plead for reason in the chaos, needing an answer to the eternal question "why me?" The perceived injustice of the situations is present and overpowering:

It's very heart-breaking (crying). I found out I'd lost a lot of time with my other child, stressing and worrying about her up until she had her surgeries, so I didn't get to enjoy her as a baby

(crying). And I didn't bond to him as well... and then I felt that (sobs) why has it happened again, to me? [Frances]

David speaks to these feelings, describing the sadness that can catch him unawares. An active campaign of hope and positivity is necessary to avoid wallowing in grief:

If you can't look forward, then it becomes a really, really bad time. I think the black dog will come and sit with you and you'll just continue to dwell in your own self-pity. [David]

The worry never leaves these parents, expanding into guilt about whether they caused the syndrome; despite knowing logically the deletions are *de novo*. David exemplifies this, ruminating over whether the medical treatment that he was receiving at conception played a role:

But there's still – did I cause that? Did my actions cause that? We had no other choice, but I think maybe it's my fault that we ended up in this situation. [David]

#### **Systemic Stigma**

Adding to the frustration of uncertainty is the systemic stigma that pervades the participants' experiences with healthcare services. Doubting their own instincts, the parents feel insecure and apologetic:

I almost felt like I had Munchausen's (both laugh) in and out, in and out, in and out. The child would vomit and then they'd get there and the child wouldn't vomit anymore. [Deborah]

Participants begin to take ownership of their concerns once they realise the seriousness of their child's condition. They feel blocked at every turn and are constantly fighting for support.

I look like this nasty, snarly, foul-tempered, foul-mouthed woman with a baby on her hip... [Stephanie]

The parents perceive that the services created the monster that the services then dread dealing with. Parents must shout to be heard:

You get tired of pushing things. You get tired of when you're walking in and see them rolling their eyes ... you think I'm a bitch because I'm trying to get my son better. [Stephanie]



Stephanie in particular experiences extreme frustration at the authoritative ignorance.

He read the MRI wrong! ... He came to the conclusion that my son was going to be violent, aggressive, non-communicative. [Stephanie]

She believes that she knows more about her child than the counterfeit experts providing care. She cannot understand why healthcare professionals do not have the same emotional investment in her child as she does, finding them as a group to be cold, uncaring and lacking in the practical skills that she perceives to be necessary to help her son:

She didn't look in his mouth to see if he's got a cleft palate ... all they're worried about is the money! [Stephanie]

### The Pain and the Gain

Conflicting emotions exist simultaneously within the participants. For example, Frances feels that she was swindled out of the mothering experience that she desired after already having had a child with medical issues prior to the one with 22q11DS:

I haven't again had that experience of a normal baby... where everything is ok... I felt almost, sort of robbed of that? Of experiencing that... so yea (stops crying)...I- I felt that. But I also felt that I had to try to look at it from all angles and then see. Well my daughter has prepared me. [Frances]

At the same time, she is grateful for this prior learning that equipped her to be an advocate for her affected son. She recognises that the experience that stole something precious from her was actually an opportunity for learning:

It almost prepared me ... what I was going to have to deal with ... I'd sort of become medicalised with her ... that's why I'd been onto it. Because I think that's what prepared me. [Frances]

Although they experience loss, fear and distress, participants actively search for meaning as a result of these feelings and challenges. They acknowledge while they are still unsure of what the significance of the situation is, they are certain that a meaning will reveal itself eventually:

I suppose that's where I'm at, just trying to find the positive... I don't know yet... [Frances]

The pain and the gain is extended through a newfound empathy. The loss of normality in the participants' lives brings about an appreciation for other people's struggles. They channel their pain into empathy, care and advocacy:

I don't look at disability the same way. So I think that that's a really positive thing. You know, I want to try to advocate for children or people with disability. That never would have happened if it wasn't for him, and it's just seeing the world through different eyes. [Frances]

As a health professional, Elizabeth particularly felt camaraderie with similar families, almost as if she is now a member of an exclusive club. She perceives that she has a better understanding of and connection with her clients as a result of her son's 22q11DS:

I've tapped into this amazing secret world that not many people know about, which I can now use to help others, in a much better way. So I feel much better equipped to do the work that I'm already doing. [Elizabeth]

Stephanie experiences a similar positive reappraisal of her situation. Despite the losses she has faced, there is a redefining of gratitude. She experiences glimmers of hope amongst the chaos of care:

I've lost a lifestyle that I loved ... I've now gained a lifestyle that I was never going to ever have. So there are benefits. [Stephanie]

### Making Sense of This Life/Growing Me

#### *Building Relationships*

Similarly, participants search for meaning in relation to 22q11DS. They hypothesise whether the beginnings of growth that they have experienced are the hidden purpose of the situation that they have been thrust into. Redefining love is a salient feature of change. They realise that their priority is now their child/children and start to move to a more selfless, patient and accepting existence. Frances speaks of the new type of love that has entered her life:

You've really got to try to find this deep love to get through the challenging times (laughing)! And that's a positive thing, like really sort of delving deep within yourself to try to find patience and acceptance, and all these things that if he hadn't come into my life, well...I wouldn't have known on the level of depth that I feel and have gone through. [Frances]

This openness to love extends beyond their child. Family relationships are built for the better, with the affected child uniting the team towards a common goal. Old wounds begin to heal, with previous conflicts no longer have the significance they once did:

Me and my father don't have a great relationship unfortunately. It's got better. But ... he would give his kidney for my son... my son is everything to him now... You have to make the best of a bad situation. Not saying this is a bad situation, but I'm reconnecting with family ... we're close. [Stephanie]

### *Opportunity Not a Burden*

Instead of viewing the challenges of parenting a child with 22q11DS as a burden, the parents decide that it is an opportunity to become a better person. Whilst disappointment and regret are common emotions, parents do not lose sight of the lessons they perceive that they need to learn:

In my trial of life, it provides a great opportunity ... I get my moments when I ask why...but then I think this has been a really good opportunity to be able to love someone anyway. [David]

Some participants emphasise the spiritual aspect of this 'opportunity' as a chance to grow in their religious faith. Deborah knows that God gave her a child with 22q11DS because she had the resilience and education to manage. As such, she is accepting of her life path:

I think it's because it's part of God's plan. I think that is the only simple answer ... For me personally I can't see that there's been any harm come to it. I can only see good. [Deborah]

Deborah also perceives that even though God gave her struggles, he is there providing the resources for her to handle each situation. Her method of coping through faith gives her strength and happiness:

There's so many things we look back and think, gee that was God's providence in his life or our lives, working together. Other people might look back and think it was fate or whatever, but I think I look back and I see God's hand in all of it. [Deborah]

Other participants did not speak of their journey as a spiritual one; however, they engage in reflection on the meaning of their experiences.

He was a miracle. He was meant to be. I was meant to have him on this journey, whatever this journey is going to be. [Stephanie]

They see that their purpose has been redefined but cannot quite figure out what their new role is. Their new significance is a blurry image that slowly gains focus with time:

I'm still finding my feet. I have no idea what some days what I'm doing. However, when it does come to needing to get stuff done, I feel like I know exactly what I need to ask for...well if that's the one good thing that has come out of it, I'll gladly take it! [Elizabeth]

### *Counting Blessings*

Gratitude is ever present throughout the participants' accounts. They count their blessings and focus on what they do have rather than what is lacking:

We're very aware of how lucky we are to have him. At the moment and how well he's doing—but yea, we love him to bits. Like I said, I think if we had found out sooner he might not be here with us today and now that he is here with us we feel very blessed with having him. [Elizabeth]

Despite their child's differences, they are thankful for the gift of a child and cannot imagine their life without them:

I feel incredibly lucky that I've had the parenting training, the disability training, the medical training I've had...are all culminating beautifully at the moment, and I really feel for the parents that don't have the background I do, because I think navigating through the system would be 100 times harder. [Elizabeth]

This positive assessment also manifests as hope. The child's young age, coupled with a diagnosis that is a "life sentence, not a death sentence" [Deborah], gives the participants hope that their child may live a normal life. Further, they see their role as that of a parent rather than a carer for a disabled child. Although they may struggle along the way, they recognise that things could be worse and are optimistic about the future:

He's not dependent on you 365 days a year, 24 hours a day. He's not going to a nursing home, you know when we die or something like that. It's just a small little hiccup in the genetic pool. [David]

## **Discussion**

This qualitative study identified four major themes representative of these participants' experiences parenting a child under 3 years old with 22q11DS: (a) agony of uncertainty—a

dichotomous experience of grief and wonder; (b) systemic stigma—invalidation and stigmatisation within healthcare services that invalidated parents on their search for a diagnosis; (c) the pain and the gain—simultaneous experiences of conflicting negative and positive emotions; and (d) making sense of this life/growing me—purposeful rumination allowing psychological growth despite distress.

This study highlights the ever-present and ongoing shadow of uncertainty experienced by these participants in parenting a young child with 22q11DS. Felt as being in purgatory without answers, not knowing leads to reflection about what life will bring for themselves and their child in the future. Similar to first responders who experience anticipation of trauma (Van der Kolk, McFarlane, Weisaeth; 1996), these parents are unable to live in the moment and anxiously prepare for symptoms and events that may never appear. This anticipation creates a hypervigilance to potential traumatic events, evoking raised expectation of threat that can negatively impact decision-making (Papazoglou 2012). Fear is constant, and grief about what could have been if it were not for the 22q11DS is not easily subdued.

The compounded impact of unavoidable stressors (e.g. difficulty with healthcare services) leaves these parents psychologically vulnerable. However, all of the participants of this study recognised their own fragility to stress and as such took active steps to embrace the positive aspects of their experiences. Contradictory emotions (e.g. shame and pride) were experienced simultaneously and work to evoke purposeful rumination to positively reframe their struggles. The opportunity for the co-existence of distress and psychological growth is welcomed with participants embracing 22q11DS as an opportunity for growing in their spirituality, feeling humbled and uniting as a family. This mix of emotions has been shown in other studies of families affected by disability. For example, Kearney and Griffin (2001) found that parents experienced anguish and sorrow alongside hope, love, strength and joy. A meta-analysis of benefit finding after trauma showed that higher levels of benefit finding are associated with more intrusive and avoidant thoughts about the stressor (Helgeson et al. 2006), again highlighting the complex and conflicting emotions of parents with children with 22q11DS.

Despite being in the early stage of their journey with 22q11DS, the risk of burnout is recognised as very real for these parents. All showed a willingness to actively utilise their experiences with 22q11DS as a springboard for psychological well-being. This supports Folkman's (1997) theory of meaning-based coping, where negative psychological states associated with significant stress may motivate people (either consciously or unconsciously) to create positive psychological states in order to gain relief. Hastings and Taunt (2002) have also suggested the usefulness of conceptualising positive perceptions as a style of coping in parents who have a child with a developmental disability. This type of positive

reappraisal has been demonstrated in other studies where a parent has a child with a severe disability (Graungaard et al. 2011). Parents turned their experiences into resources such as engaging with hope. As yet, it is unclear which factors promote this positive coping and how it is integrated with the conflicting emotions that the participants described.

The uncertainty that has dominated these parents' experiences also aligns with the reconceptualised uncertainty in illness theory (Mishel 1990). The amount of time since a stressor has passed has been shown to influence benefit finding (Helgeson et al. 2006). Complexly, parents who have a child with 22q11DS are on a continuum of stressors and not limited to one event. The syndrome can manifest in both chronic and critical ways, with uncertainty plaguing parents throughout their child's life. These participants showed a desire to integrate the continuous uncertainty into their lives by reorganising their beliefs to avoid a chronic anticipatory state of distress associated with expectation of predictability (Mishel and Clayton 2008).

In particular, Mishel (1990) notes that while reorganising, people can turn uncertainty from an aversive experience to an opportunity. This was present in our sample, where participants consciously referred to their child and the associated challenges as an opportunity rather than a burden. Ironically, the uncertainty brought hope for a happy future. Further, despite the children's young age, participants were already describing the beginnings of growth (e.g. building relationships, gratitude), which is the desired outcome in Mishel's (1990) theory. Longitudinal studies would be useful to examine if and how parents resolve the uncertainty of 22q11DS and which factors promote specific outcomes, such as psychological growth.

## Limitations

It is important to consider the results within the context of its limitations. As a qualitative study, rather than seeking to generalise findings to all parents who have a child with 22q11DS, we sought detailed insights of this specific homogenous sample of parents with children under the age of 3. Due to the double hermeneutics employed in IPA, the researchers' biases could have impacted on the study both positively and negatively. However, we took steps (e.g. audit trail, robust discussion) to attend to credibility and worthiness of the study at every step of the analytic process. Therefore, despite the limitations, this study provides a valuable contribution to the knowledge of 22q11DS and a basis for further research into the experiences of parents and families throughout the life of a child with 22q11DS including how they manage continued syndromic uncertainty and/or diagnosis of associated features (e.g. autism) at different stages of the child's development.



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**Compliance with Ethical Standards** No funding was required or given for this study.

**Ethical Approval** All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards.

**Informed Consent** Informed consent was obtained from all individual participants included in the study.

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