


Indicators of health in Down syndrome: A virtual focus group study with patients and their parents

Stephanie L. Santoro^{1,2}  | Maria J. Cabrera¹ | Kelsey Haugen¹ | Kavita Krell¹ | Vanessa L. Merker³

¹Division of Medical Genetics and Metabolism, Department of Pediatrics, Massachusetts General Hospital, Boston, Massachusetts, USA

²Department of Pediatrics, Harvard Medical School, Boston, Massachusetts, USA

³Department of Neurology and Cancer Center, Massachusetts General Hospital, Boston, Massachusetts, USA

Correspondence

Stephanie L. Santoro, 125 Nashua St., Suite 821, Boston, MA 02114, USA.

Email: ssantoro3@mg.harvard.edu

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Abstract

Background: Down syndrome has a unique medical and psychological profile. To date, few studies have asked individuals with Down syndrome about their views of health.

Methods: Eight focus groups of 20 parents and 8 individuals with Down syndrome, were conducted virtually via videoconferencing to obtain participants' views of health indicators. Focus group moderators employed some modifications for individuals with Down syndrome, including simplified language and use of graphics. Transcripts were coded using a hybrid inductive/deductive framework and thematically analysed using the Framework Method.

Results: We describe lessons learned in conducting virtual focus groups of individuals with Down syndrome and their parents. Individuals with Down syndrome could describe their views of health indicators and identified many of the same topics as their parents. Both groups discussed physical, mental, and social health components. However, people with Down syndrome gave a more restricted range of examples, but with different nuances than parents.

Conclusion: Participants discussed physical, social, and mental well-being components of health in Down syndrome. Interviewing individuals with Down syndrome in virtual focus groups with appropriate modifications added important self-report health information.

KEYWORDS

Down syndrome, focus group, health status

1 | INTRODUCTION

Health is 'a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity' (World Health Organisation, 1946). Down syndrome (DS) is a genetic condition due to the triplication of chromosome 21 and is associated with multiple medical and psychological comorbidities which can affect all aspects of health. The physical and mental well-being of individuals with DS may be impacted by increased rates of ADHD, autism, and Alzheimer's disease (Hithersay et al., 2019; Palumbo & McDougale, 2018; Rafii, 2022). Social well-being may also differ for people with DS due

to higher health care costs, and increased need for community inclusion and school support due to variable intellectual disability, functional abilities, and independence (Graaf et al., 2019; Krell et al., 2021; Matthews et al., 2018; Santoro, Hendrix, et al., 2022).

As such, a health framework for the general population may not accurately measure the condition-specific health of individuals with DS due to these differences in rates of associated medical and developmental comorbidities compared to the general population (Huber et al., 2011). When piloting the use of global health measures in individuals with DS and their parents, we found limitations in health instruments used for the general population including two questions

on expressing oneself and one's pain having high non-completion rates (Santoro et al., 2021). These questions may be valid for use in the general population (Forrest et al., 2014), but may be difficult for parents of children with DS to answer due to the varying forms of expression and language abilities in people with DS.

Although outcome studies for individuals with DS have focused on health-related quality of life in adults and physician adherence to guidelines (Graves et al., 2016; Jacola et al., 2014; Rafii, 2016; Santoro et al., 2018), or focused on specific aspects of health including components of cognition, sleep, and behaviour (Graves et al., 2016), the overall health status in people with DS has not been directly measured. We began this research within a larger project aimed to develop a measure of health specific to DS with the rationale that health is a measurable concept with defining constructs which may be different for DS in comparison to the general population. We developed a conceptual model of health in DS; our hypothesized constructs (concepts) of health in DS included: physical well-being, mental well-being, and social well-being (Exworthy, 2008; Hosseini Shokouh et al., 2017; Martikainen et al., 2002; Schulz & Northridge, 2004). Through the process of developing and testing our conceptual model, we conducted focus groups (FGs) with individuals with DS and, separately, parents of individuals with DS.

Specifically, in this study we aimed to describe views of health indicators from (1) adolescents and young adults with DS, and (2) parents of individuals with DS. Few studies of individuals with DS reporting their views exist (Santoro, Donelan, & Constantine, 2022); we included the health views from individuals with DS in this study. By conducting this research during the COVID pandemic, we learned lessons about conducting virtual FGs of individuals with DS which could inform other researchers. Our results lay the groundwork to develop a measure of health in DS and might guide researchers studying other genetic syndromes.

2 | METHODS

2.1 | Participant eligibility and recruitment

Inclusion criteria for the study were: (1) either an individual with DS age 13–21 years, or a parent of an individual with DS ages 0–21 years, (2) English-speaking, and (3) access to a computer, internet, and virtual video-technology website. In addition, for all individuals with DS, the research assistant spoke with parents to determine if participants with DS had the skill and expressive language ability to participate in a Zoom discussion. All parents were the primary caregiver of an individual with DS and all individuals with DS were children of participating parents. We chose to focus on adolescents with DS, as there is very little literature on perspectives of adolescents with DS (Santoro, Donelan, & Constantine, 2022) and adolescence is a key time of establishing one's identity, views and beliefs separate to their parents. The lower age limit for individuals with DS (13 years) was chosen to ensure participants with DS could provide informed consent and participate in discussion. As individuals with DS in the

United States often attend high school until age 21 years old, we chose this as an upper age limit to maintain homogeneity in participants and keep focus on the adolescent period rather than adulthood.

Participants were recruited from the Mass General Hospital Down Syndrome Program and the local Massachusetts Down syndrome Congress (MDSC), the LuMind Down syndrome research group, and DS-Connect through electronic posting of study information. Community partners with diverse membership, such as the MDSC Diversity Outreach Task Force and the 21 Shades Family Support Group, also shared study information with their members. This information included a description of the study, and a hyperlink to an electronic screening form which obtained demographic information in REDCap (Harris et al., 2009). The research team then contacted eligible parents to describe the study and schedule focus groups for themselves and, if eligible, their child. In addition to age and sex, demographic details including race, ethnicity, parent's perception of the educational concerns of the person with DS, and parent's perception of the medical complexity of the person with DS were collected.

2.2 | Focus group procedure

FGs were stratified by (1) status as a person with DS or as a parent of a person with DS and (2) age of the individual or child with DS, to focus on the developmental perspective to health.

In preparing to conduct FGs, care was taken to consider the specific needs of individuals with DS; details regarding use of Zoom videoconferencing technology to conduct FGs, specific modifications to accommodate individuals with DS, and feedback on the utility of these modifications are summarised in the Appendix S1.

At the beginning of the FG, each participant gave verbal consent to participate. FGs were moderated by two researchers using a semi-structured discussion guide informed by the WHO definition of health. Participants were asked to discuss the concept of health as it relates to individuals with DS and to identify indicators of physical, social, and mental health (Appendix S1).

All FGs were videorecorded and transcribed.

2.3 | Qualitative analysis

Building on the WHO Definition of health, a conceptual model of health in DS was created covering aspects of physical, social, and mental health thought to be applicable to the majority of individuals with DS age 0–21 years (Bull, 2020; Bull & the Committee on Genetics, 2011). The topics of this conceptual model were used as initial deductive codes; throughout the coding process, the research team inductively added codes and modified code definitions based on thematic content. All FG transcripts were coded independently by four reviewers and then reviewed collectively as a team to reach consensus in coding applications (Hemmler et al., 2022). No new codes emerged by the end of eight FGs, suggesting thematic saturation was achieved. Using the Framework Method (Gale et al., 2013), coded text was

summarised within individual FGs using matrices. Final analysis focused on describing the components of health identified by study participants across FGs and comparing domains identified by individuals with DS to those identified by parents of individuals with DS. Supportive quotes are presented with 'D' for an individual with DS and 'P' for a parent.

The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions as participants did not consent to data sharing. The MGB IRB approved this study.

3 | RESULTS

From November 2020 to March 2021, 8 FGs occurred using video-technology, of which 4 were with parents of individuals with DS age 0–21, and 4 were with individuals with DS ages 13–21 years. Parents ($N = 20$) were generally mothers, mostly white, married, with a college degree or higher, and lived with their loved one with DS (Table 1). Distance from a specialty clinic for DS varied; of the 10 who responded to this question, only 2 were accessing a specialty clinic for DS. Of those that described their recruitment, most (10 of 11) reported learning about the study through social media. Parents reported that their loved ones with DS had an average rating of significant health problems at 2.7, and an average rating of significant educational/learning difficulties at 4.9 (on scale 1 to 7, where 1 = not a problem, 7 = very much a problem).

3.1 | Health concepts in down syndrome

Participants endorsed a range of symptoms and concerns covering each of the physical health (Table 2), social health (Table 3), and mental health (Table 4) components in our original conceptual model except for the mental health domain of alertness. In all three domains, individuals with DS discussed some, but not all, of the health categories in our model (Tables 2–4); parents and individuals with DS identified overlapping topics within coding categories (see text below). Few additional topics not in our original conceptual model were identified (see 'other physical' in Table 2).

3.2 | Views of physical health

Participants endorsed a wide range of symptoms and physical health issues, including well-known comorbidities of DS (Table 2). Physical concerns which were frequently mentioned in all FGs included: musculoskeletal issues (such as muscle strength and low tone), metabolism (such as weight and diet), and activity (such as exercise). Teens with DS identified staying active and exercising as important signs of health in their own lives:

"I feel healthy because I work out every day, like dancing and swimming and running and walking." Person with Down syndrome (D)3, FG2

Similarly, when asked to identify examples of other healthy people, individuals with Down syndrome mentioned superheroes – 'because they're strong' and 'they have more muscles' – and athletes like Michael Phelps who stay fit and eat healthy. One individual with DS described physiological changes with exercise, such as faster heartbeat, that made them feel healthier and connected regular exercise to living longer:

"If you see someone fit, it's when you work out and build extra muscles. And if you stay healthy, stay fit, be strong-type of person that makes you a good-looking and make you stay fit. So they have a longer life." D4, FG4

Parents echoed these themes; one caregiver worried that if their son had a bad diet and gains weight as he ages, he will slow down and be less able to engage in activities. Parents noted interconnections between physical symptoms, such as the interplay of being overweight with other health conditions and how low tone can contribute to constipation, development delay in meeting milestones, fatigue, joint laxity, and endurance. One mother described this as:

"I remember when I learned about hypotonia in the hospital and I'm like, "Well, how do we fix it?" And they go, "You don't. You just build the muscle. He's got to work harder." And I never thought it would affect his chewing, right? Which affects speech. Think about it, how hard is it to make certain sounds if you don't have the right muscle tone in your [face]?" Parent of person with Down syndrome (P)2, FG

Parents' physical health concerns differed based on the age of their child, and parents described changing physical concerns across the lifespan. Oromotor concerns around feeding and picky eating were primarily mentioned in the FGs of parents of children age 0–5 and 6–12 years, but not in FGs for parents of teens and young adults. As one parent of an older child with DS reflected:

"It seems like from zero to two, it's all you worry about is there physical, like what's going on physically, if they can sit up, if they can roll over when they start walking... And now it's more of like a social concerns for me and expressive language and stuff like that." P3, FG1

Parents of teens noted that their activity level slowed down with maturity/during puberty and reflected on the recurrent respiratory infections in childhood which improved over time.

Both parents and individuals with DS discussed taking steps to maximise health; these modifying behaviours were coded as 'Health-related behaviour'. Parents and individuals with DS valued eating healthy foods and exercising in terms of being physically healthy, and discussed the importance of good hygiene, preventative care including regular blood testing, the value of being outside, and having a good relationship with physicians and involvement in DS-specific communities.

TABLE 1 Demographic traits of 20 parents and their son or daughter with Down syndrome

| Trait | Response | N (%) |
|---|--------------------------------|--|
| Focus group participants: Individual with Down syndrome | | |
| Age (years) | 13–17 | 4 (50) |
| | 18–21 | 4 (50) |
| Gender | F | 4 (50) |
| | M | 4 (50) |
| Race | White | 8 (100) |
| | Black or African American | 0 (0) |
| | Asian | 0 (0) |
| | Indian | 0 (0) |
| Focus group participant: Parent | | |
| Parent's gender | F | 19 (95) |
| | M | 1 (5) |
| Parent's race | White | 17 (85) |
| | Black or African American | 1 (5) |
| | Asian | 1 (5) |
| | Indian | 1 (5) |
| Parent's highest level of education | College or university graduate | 2 (10) |
| | Master's degree | 9 (45) |
| | Doctorate degree | 6 (30) |
| | Missing | 3 (15) |
| Parent's employment | Yes | 12 (60) |
| | No | 5 (25) |
| | Missing | 3 (15) |
| Parent's relationship status | Married | 16 (80) |
| | Divorced | 1 (5) |
| | Missing | 3 (15) |
| Parent's location | Northeast | 7 (35) |
| | Midwest | 4 (20) |
| | South | 3 (15) |
| | West | 2 (10) |
| | N/A, International | 2 (10) |
| Parent's number of children | | Mean: 2.8 Range: 1–6 |
| Combined total household income of parent | | Mean: \$138,500 Range: \$75,000–400,000 |
| Son or daughter with Down syndrome | | |
| Age of son or daughter with Down syndrome (years) | 0–5 | 5 (25) |
| | 6–12 | 4 (20) |
| | 13–17 | 5 (25) |
| | 18–21 | 6 (30) |
| Gender of son or daughter with Down syndrome | Male | 10 (50) |
| | Female | 10 (50) |

TABLE 1 (Continued)

| Trait | Response | N (%) |
|--|----------|--------|
| To what extent does your son or daughter with Down syndrome, in your opinion, have significant health problems? (1 = not a problem, 7 = very much a problem) | 1 | 2 (10) |
| | 2 | 7 (35) |
| | 3 | 4 (20) |
| | 4 | 3 (15) |
| | 5 | 0 (0) |
| | 6 | 1 (5) |
| | 7 | 0 (0) |
| To what extent does your son or daughter with Down syndrome, in your opinion, have significant educational/learning difficulties? (1 = not a problem, 7 = very much a problem) | 1 | 0 (0) |
| | 2 | 1 (5) |
| | 3 | 1 (5) |
| | 4 | 4 (20) |
| | 5 | 5 (25) |
| | 6 | 4 (20) |
| | 7 | 2 (10) |

3.3 | Views of mental health

3.3.1 | Quality of life

People with DS and their parents touched on three components of overall quality of life - feeling happy, fulfilled, and accepted. Individuals with DS stressed the importance of feeling happiness and pleasure as a key part of being healthy. This happiness could come from participating in hobbies (such as dancing or swimming), participating in leisure activities (like listening to music or playing video games), and enjoying favourite foods and drinks. People with DS discussed having a sense of fulfilment from accomplishing tasks and performing well in activities they enjoy. One participant described a ‘good day’ as ‘when you accomplish something that needed to be done’ and another mentioned feeling good when they won challenges in their favourite video game. Finally, people with DS highlighted the importance of feeling welcomed and socially accepted by others. One participant noted ‘I feel happy because I have a community full of people’, and others commented on how they enjoyed spending time with their family, friends, neighbours, and pets.

Parents echoed that being happy is an important part of being healthy, and valued leisure activities and hobbies (like sports, singing and yoga). Parents described a larger hope for their child to participate in and enjoy daily activities despite their medical condition. For example, parents highlighted wanting to ensure their child ‘functions day-to-day and doesn't miss life’ and not letting challenges related to DS ‘set her back in the way she lives her life or how we go about operating’. Parents discussed their role in helping their child feel fulfilled, with one parent noting ‘...that is my

TABLE 2 Physical health codes endorsed in eight focus groups of people with Down syndrome and parents of people with Down syndrome

| Code | Topics | Endorsed by... | |
|-----------------|---|----------------|---------------------------|
| | | Parents | People with down syndrome |
| Neurologic | Seizures, infantile spasms | ✓ | Not mentioned |
| Dermatologic | Skin 'break outs' and pimples, alopecia, very dry skin | ✓ | Not mentioned |
| Musculoskeletal | Strength, fitness, low muscle tone | ✓ | ✓ |
| Cardiac | Congenital heart disease, heart murmur, arrhythmia, pulmonary hypertension, need for surgery, need for pacemaker | ✓ | Not mentioned |
| Respiratory | Colds, bronchitis, pneumonia, RSV, asthma, narrow airways | ✓ | ✓ |
| Genitourinary | Chronic renal failure, toilet training | ✓ | Not mentioned |
| Digestive | Constipation, celiac disease, duodenal atresia | ✓ | ✓ |
| Sensory organs | Vision concerns, hearing loss, cholesteatomas, need for glasses, need for ear tubes, need for hearing aid, recurrent ear infections | ✓ | Not mentioned |
| Endocrine | Hypothyroidism | ✓ | Not mentioned |
| Metabolism | Overweight, failure to thrive, glucose issues, type 1 diabetes, fatigue | ✓ | ✓ |
| Activity | Ability to and frequency of exercise | ✓ | ✓ |
| Mobility | Slowing down, sitting up/milestones, strength, balance | ✓ | Not mentioned |
| Sleep | Sleep apnea, getting 'a good night's' sleep, napping | ✓ | ✓ |
| Oromotor | Picky eating; difficulties with breastfeeding, chewing or swallowing; need for gastrostomy tube | ✓ | Not mentioned |
| Pain | Ear pain with ear infections, high pain tolerance | ✓ | Not mentioned |
| Other physical | Dental health | ✓ | Not mentioned |
| Other physical | Medical issues which are not associated with or more prevalent in Down syndrome | ✓ | Not mentioned |

TABLE 3 Mental health codes endorsed in eight focus groups of parents of people with Down syndrome, and people with Down syndrome

| Code | Topics | Endorsed by... | |
|--------------------|--|----------------|---------------------------|
| | | Parents | People with Down syndrome |
| Quality of life | Feeling pleasure/happiness, sense of self-fulfilment, feeling welcomed and socially accepted | ✓ | ✓ |
| Mood/affect | Receptiveness to learning new skills and facing challenges; regulating mood; ability to recognise and communicate about one's emotions | ✓ | ✓ |
| Identity | Pride from ability to set and reach goals; personality/sense of humour; emerging sexuality | ✓ | ✓ |
| Coping | Maintaining health-related routines; being able to manage change/disruptions to routines; developing healthy coping mechanisms | ✓ | ✓ |
| Behaviour | Using behaviour to indicate and cope with feelings; engaging in self-soothing or self-protective behaviours | ✓ | Not mentioned |
| Neurodevelopmental | Learning; independence (including planning and problem-solving skills); developmental progress (including need for therapies) | ✓ | Not mentioned |
| Language | Expressive and receptive language including vocabulary and articulation; use of sign language | ✓ | Not mentioned |
| Alertness | — | Not mentioned | Not mentioned |
| Other mental | — | Not mentioned | Not mentioned |

goal in life, whatever his best is, to get him to his best'. Finally, parents stressed the critical importance of their child developing and maintaining connections with others, with one parent saying, 'it doesn't matter how physically healthy she is, if there's no one to share her life with'.

3.3.2 | Mood/affect

When discussing moods, individuals with DS largely described things that brought them pleasure and joy, as described above. However, one individual with DS did expand on this topic, describing how

TABLE 4 Social health codes endorsed in eight focus groups of parents of people with down syndrome, and people with down syndrome

| Code | Topics | Endorsed by... | |
|-----------------------------|--|----------------|---------------------------|
| | | Parents | People with down syndrome |
| School/work | Having friends and building relationships at school; participating in enrichment activities at school; inclusion with neurotypical peers including school 'mainstreaming' | ✓ | ✓ |
| Community participation | Participating in community activities (through peer groups, hobbies, sports, etc.); Going out to places in community (e.g., park, grocery store); Positive impact of diverse communities | ✓ | ✓ |
| Socialisation | Opportunities to engage with friends, family, and other people in the community either in-person or using technology (i.e., phones, iPad, or social media); avoiding isolation | ✓ | ✓ |
| Relationships | Having meaningful relationships with family members; being able to develop close friendships with and feel included by people without Down syndrome | ✓ | ✓ |
| Resources and opportunities | Access to transportation; access to healthcare and insurance coverage, including knowledgeable paediatricians, specialists, and therapists; safe living situation; access to social support such as relatives nearby and parent support groups; availability of social activities and educational supports | ✓ | Not mentioned |
| Other social | Overscheduling/time limitations; prioritising/age-based differences | ✓ | Not mentioned |

regularly engaging in a fun hobby (dancing) 'makes me happy all the time, especially when I keep positive' about her progress. This allusion to staying positive and making progress in a skill was echoed by a parent who said that if their child is healthy, they are not only in a good mood, but 'in the right frame of mind' – that is, receptive to learning, willing to 'put in the effort' and able to face challenges.

"What I've noticed is that schoolwork or gym, these two outlets— if she's in a good mood and she's in a proper frame of mind, she performs very well, consistently all the way. Even if I have to give her harder problems, harder questions, she would attempt... So I know if she's in the proper frame of mind, which means she's healthy." P5, FG7

Parents discussed two additional topics: recognising and communicating one's own emotions and regulating mood. Parents felt that communicating about negative emotions was perhaps even more important than recognising the stereotypical positive emotions associated with DS. As one mom described:

"For my daughter, I would say in a stereotypical Down syndrome kind of way, when people talk about, "Oh, they're always happy," in a general frame of mind that is her mode of operation. But she has told us many times over the last 10 months, has used a lot of different emotional words, like, I'm sad or I'm worried or I don't like that. And it's all COVID-related or lack of social interaction related..." P4, FG6

While some children with DS were able to verbalise negative emotions and communicate their needs using language, others could not always communicate their feelings directly. Caregivers said they can tell if their children with DS are happy largely through behaviours – for younger children, whether a child 'smiles a lot and tries to play with us and goofs off' and in teenagers, whether they are 'keeping up with routines' or engaging in enjoyable spontaneous behaviours like singing, cracking jokes, and teasing family members. Similarly, parents described behaviours indicating unhappiness, including 'fussing and [being] grumpy' for a younger child and 'temper tantrums' or being 'irritable or tired at a time she shouldn't be' for older children.

And while caregivers felt they could largely judge their children's emotions through behaviours, it was harder to discern what was causing these negative emotions.

Finally, parents described their children's difficulties regulating emotions in reaction to negative stimuli or experiences. Parents described unhealthy regulation as children feeling overwhelmed, shutting down and not able to engage with others; in contrast, when a child with DS was feeling happy and healthy, 'she's affable; she's engaging; she's completely social'. One parent specifically noted that this acting out through changes in social interaction and behaviour was more noticeable in her child with DS compared to a typically developing sibling:

"We all have these bad days, but this distance between her prime [self] and her really struggling [self] seems greater than in our other child. Self-regulation just sort of goes out the window." P4, FG8

Parents noted that even children who were 'extremely consistent' in their emotions may have specific issues they struggle with,

like being anxious about food restrictions and food monitoring that parents have implemented to help their children maintain a healthy weight.

3.3.3 | Identity

Two young adults with DS talked about the importance of setting and reaching goals towards forming a positive identity. For example, one person described their pride in excelling at their hobbies: '[Dancing] makes me feel like a shining star... It makes me feel proud. I feel like I'm actually reaching for my goals' S4, FG4. Parents agreed that it was important their children had hobbies and artistic outlets to express themselves, and that excelling in these pursuits was a source of pride for their children. A few parents raised additional topics regarding their children's changing identities as they aged. One parent saw their child's emerging sense of humour as a sign of developing personality, while another parent discussed the potential awkwardness of handling their child's pubertal transitions and exposure to sexual content online.

3.3.4 | Routines and coping skills

People with DS discussed routines as a tool for health maintenance. Individuals noted that healthy routines include regularly eating well, being active, and going outside to get some 'sun and Vitamin D', even when it is not circumstances are not ideal (e.g., 'go outside every day even [when it isn't] nice out'). Diligence may be required in keeping up routines to manage co-morbidities like diabetes, where monitoring blood sugar and eating habits is very important. Teens also felt that healthy people have to be able to manage change/disruptions to routines, as highlighted by the COVID-19 pandemic. For example, one teen with DS described navigating remote options for connecting with people and figuring out what tools (e.g., phone calls, texting, social media apps) they liked best to replace in-person socialising.

Parents valued routines to set kids up for success and help them manage transitions. Parents discussed routines both as important mechanisms for maintaining health and as indicators of emotional health, based on whether their child was keeping up with their routines.

Parents agreed that learning how to cope with change is important, and emphasised the importance of developing healthy coping mechanisms and having an 'outlet' to destress. People with DS may use self-talk to process events and coach themselves through new situations. As one parent noted 'even though you think that maybe he is not engaged in what's going on or he's doing his own thing, you'll hear him talk about it later or he'll be processing it later, after the fact'.

One area where coping skills were discussed frequently was around socialisation, as they may have to repeatedly 'put themselves out there' to try to find friends. Interestingly, the adversity of the COVID pandemic (e.g., loneliness from lack of socialisation) forced some kids with DS to verbalise their feelings more and develop better coping mechanisms.

"She's starting to learn her own coping mechanisms. Where maybe in the past, if she was sad or lonely, maybe she would kind of sit in the corner of the couch and get quiet. So if she's quiet, we know something's not right, where now she's been able to verbalize the feelings and learn what to do when you have those feelings." P4, FG6

Parents noted they sometimes had trouble gauging whether their child has developed healthy coping skills. Parents worried about whether some of their child's behaviours were unhealthy coping mechanisms (e.g., having imaginary friends or pretending to talk with people on video games who aren't actually interacting back), and felt that certain coping behaviours like stimming might be an indicator someone is having difficult emotions/feeling less healthy.

3.3.5 | Behaviour

Only parents discussed behaviour and the remaining mental health domains of neurodevelopmental issues and language. Parents viewed behaviour as a general sign of happiness/unhappiness and noted that stark behaviour changes – especially deviating from routines, refusing to do things they normally like, and having outbursts or temper tantrums – are a sign of poor health. While parents noted that more pleasant behaviours usually indicate happiness and good health ('I know he's happy because he'll sing just spontaneously, all day, every day. He'll crack jokes at us, he'll tease us, that's how I know that he's feeling good and his mind is happy.' FG7), kids with DS may have trouble learning what are inappropriate behaviours (e.g., not touching and hugging other people).

Parents viewed behaviour as intimately connected with the domains of language, mood, and coping. Sometimes children with DS have to communicate through behaviour because they do not have the expressive language skills to be able to articulate what is bothering them. They may instead indicate they are upset with their facial expressions, body language, and behaviour. As one parent described 'We know when he's really upset because he will just physically turn away from you and ignore you or push you off. FG7' Certain behaviours may actually help children cope with difficult feelings though, such as one parent who highlighted their child's co-morbid autism spectrum disorder and use of behaviours like rocking and stimming. Individuals with DS can learn to engage in 'self-protective' behaviours to help prevent health issues from appearing or worsening. For example, one parent noted how their child have learned to sit down to rest while running if breathing is becoming difficult so as not to provoke an asthma attack.

3.3.6 | Neurodevelopmental

Parents had general perceptions of their children's neurodevelopmental functioning based on their cognition and learning (e.g., as

academically challenged, or as 'high-functioning'). To aid learning, some parents recommended a more hands-on learning style that incorporates a fun and playful approach to introducing new concepts, slowly building up to new tasks and interlacing skills their child has already mastered with new tasks, and making sure kids can use their body while learning. Parents emphasised that part of being healthy was becoming more independent, while still appropriately recognising one's limitations. Important indicators of independence were doing tasks alone (e.g., self-care tasks like showering, washing hair, and preparing food); being able to plan and take action to meet their needs (e.g., finding yoga videos and doing them every day, being able to 'take charge of [their] social life') and being able to problem-solve (e.g., finding new people to spend time with after their siblings grow up and move away by signing up for more clubs and activities.) The transition to independence could be difficult for parents sometimes. One parent described a tension between fostering independent decision-making for safety reasons (so that down the line, their child is not '[complying] with what people say just because they say') and needing to overrule their child's decisions on days where their child is 'obstinate' and 'says no to everything'. Finally, parents noted that children with DS may need special services and therapy to help with their developmental progress and may have neurological co-morbidities (like ADHD) with additional symptoms and medication side effects that impact neurodevelopmental functioning.

3.3.7 | Language

Having difficulty communicating was seen as a major challenge by parents, especially if it interfered with daily activities or having friends. Parents noted that their children with DS particularly had trouble with expressive language, including verbalising their thoughts and emotions, articulating words in a way that is understandable to non-family members, and using a vocabulary that is appropriate to their peers (e.g., saying 'It's time to go to the potty' instead of 'I have to go to the bathroom'). Expressive language was especially important for issues relating to communicating emotions, needs, and health. One mom viewed her daughter's ability use language to describe her emotional state and challenges as a sign of being healthy.

"She's used words like lonely or sad or worried. And so, I mean, the best part is for us, she verbalizes her feelings and that I think it's healthy. I think that tells us her mind is healthy because she's able to verbalize her emotions." P4, FG6

Parents thought it was important for their children to express their emotions so that caregivers can help problem-solve and avoid temper tantrums. It can be difficult for parents to assess children's social life if they are non-verbal or have difficulty speaking. Approaches to improve language and communication highlighted by parents included: giving individuals with DS more time to process and

respond to spoken requests, giving multiple choice answers (rather than an open-ended question), seeking speech therapy or biofeedback to improve vocal language, and using sign language to communicate without speaking.

3.4 | Views of social health

3.4.1 | School inclusion

When asked about their social health, participants with DS focused on time spent with friends from school, and enrichment activities like music groups and team sports as signs of social health. As one individual with DS said, 'The main thing is I have friends in my school, my class, my year' D5, FG5. Another individual with DS added that 'less time on your phone and more time doing schoolwork or activities' is a sign of good health. Parents highlighted the importance of school and viewed an interest in going to school/work is an indication of health and happiness: 'When she's healthy and happy, she gets up quick, she gets ready. She wants to go [to school]' P5, FG6.

Parents discussed inclusion in school with neurotypical peers as a sign of health and predictor of future success: 'so that he can excel academically and be whatever he wants to be'. Parents described differences in inclusion by age. Parents of children in daycare mentioned the use of peer modelling and social roleplaying in the classroom to show the importance of inclusivity, and a parent of a kindergartener valued that the teacher set the same expectations and treating for her child with DS as other children. Other parents noted that by third grade, the academic content has more verbal information requiring more time from the teachers to help the child with DS 'keep up'. A parent of a teen in middle school described more opportunities for inclusion in high school than in middle school for her son in a 'special class' with some integration into general education classes. Across FGs, parents expressed a hope for inclusion, for remaining in 'mainstreamed' classes, and for avoiding alienation for their children.

3.4.2 | Community participation

Community participation was viewed as an indicator of good social health by both parents and individuals with DS. As one teen with DS said: 'I feel happy because I have a community full of people. Like my family, my friends, my neighbors from where I live' D3, FG2. The enjoyment derived from community involvement was echoed by parents, one of whom said: 'This is what she wants to do...She wants to be in the community' P5, FG7. Teens focused on the community activities they participated in, such as clubs and sports, as well as activities with friends like playing video games. Parents similarly described the importance of interacting with friends and participating in structured community activities, including local DS peer groups, but noted this may become more difficult as children age and leaves the school environment. Parents indicated the importance of more

unstructured activities that allowed their child to interact with community members, such as playing at the park or going on errands together. Finally, parents discussed how living in a diverse community promoted inclusion and normalised physical differences.

3.4.3 | Relationships

Parents and individuals with DS both talked about the importance of relationships between individuals with DS and others, including their family, friends, those in clubs with shared interests, and pets.

Parents described the value of sibling relationships and friendships in particular, with one parent saying ‘Actually, it’s probably the number one, more than anything is that need to develop those friendships’ P6, FG7. Parents found it helpful to have another person their child’s age around (e.g., a neighbour or classmate) to guide them at school or when playing. However, a parent of a 0–5 year old worried about genuine their child’s friendships are: ‘Are people inviting them to feel good about themselves or because they want them there?’ There was some worry from a parent about how relationships will look if other kids do not understand how the child with DS talks and behaves (e.g., engages in stimming), and whether other kids will include the person with DS. Despite these worries, parents described their child’s strengths in socialisation, noting how they love to be around people and spread joy to others.

3.4.4 | Socialisation

Building on the theme of relationships, parents, and individuals with DS both noted the importance of opportunities to socialise with a variety of people including friends, family, and people in the community. Socialising in-person as well as through technology were both important. One teen with DS described that they often talk to their friends in-person at school, but ‘sometimes we text at night or send emojis or FaceTime’. However, communication over texting or social media could sometimes be difficult, with one teen with DS describing how miscommunication can occur:

“...my friends is good. Sometimes it’s back and forth on communication and understanding stuff. And almost back and forth with confusion or miscommunication from somewhere. Those two are the main two that I’m kind of struggling at.” D5, FG5

Parents noted the value of social interaction for health and saw their child engaging with siblings and peers as a sign of good health. Parents found it helpful to have another person their child’s age around (e.g., a neighbour or classmate) to guide them at school or when playing.

One parent specified the need of maximising age-appropriate socialisation for her 15-year-old. The parent mentioned that with older care providers, her son ‘clings to them and he enjoys them and

he wants his alone time with them...but I don’t want his best friend to be a 50-year-old woman’ P3, FG6 Parents noted the ways in which social isolation negatively impacts other domains of health:

“My son needs to have interaction with his peers and be outside of the house for him to have a healthy day. When he doesn’t, I notice that his speech regresses, his behaviors increase, defiance definitely increases. But when he’s been able to—that has been really good for him to see some other kids and have a chance to talk with them.” P7, FG7

As one parent noted ‘having a good healthy life means you can’t [sit] in the corner of the couch by yourself’ P6, FG5.

3.4.5 | Resources and opportunities

Parents described the resources and opportunities that contributed to social health, including social and educational programming like Special Olympics teams, community clubs, and local DS support groups. Medical resources, such as affordable insurance coverage, a paediatrician with a child with DS who was a guide for the caregiver, and access to a Down syndrome specialty clinic, were important resources for health. Access to transportation and stable, clean housing were needed to maximise health. Parents particularly noted the importance of providing individuals with DS with opportunities to maximise socialisation. Not having opportunities could hinder social health for teens:

“How do I know her social life is healthy? Actually, knowing it’s not healthy because I get the loneliness that she’ll express.... And partially, it’s fact that she doesn’t drive and she doesn’t have that transportation. She’s limited in terms of social life.” P6, FG7

Parents noted a lack of opportunities in rural locations, and a lack of resources for individuals with DS as they age. One parent of a teen said:

“In elementary school and younger, it was very easy to find activities for him because—what it’s geared towards was those younger kids, the toddlers through elementary school. And when he hit middle school, literally, there was nothing.” P7, FG7

Another parent in that FG echoed this same challenge, noting that for their 18-year-old, there is a ‘more socially distant, bigger gap...just even the resources seem fewer. The community seems more sparse’ P3, FG7.

Comparing views between participants with Down syndrome and their parents

Parents and individuals with DS identified topics across the physical, social, and mental well-being constructs when asked about the

definition of health. In general, parents and individuals with DS discussed similar topics within each construct, with some differences. Regarding physical health, parents identified some categories which were not discussed by individuals with DS, and parents drew more abstract conclusions such as the interconnections between body systems. Parents and individuals with DS identified many of the same components of mental health as contributing to overall health; emphasised the need to be and feel happy, to have goals, and the value of routines. Parents identified some topics which were not discussed by individuals with DS, such as behaviour and language. Parents drew more connections between constructs, highlighting the role of behaviour as a sign of mood, as a form of communication, and a sign of overall health. Parents tended to discuss the range of emotions (happiness to unhappiness) within mental health while individuals with DS focused on happiness and pleasure. Across FGs, parents and individuals with DS highlighted the importance of relationships and socialisation to promote social health, with parents focusing on additional topics not mentioned by individuals with DS, such as: the resources and opportunities to develop those relationships.

4 | DISCUSSION

We conducted FGs of individuals with DS and parents of a son/daughter with DS, through videoconferencing technology with the aim of understanding how health is conceptualised in DS. We found that:

- Health in Down syndrome was conceptualised with physical, mental, and social health components in line with the WHO definition.
- Individuals with Down syndrome can successfully share their views in FGs and describe their health.

We coded FG transcripts to our conceptual model of health in DS, and found that all but one of our constructs/codes (alertness) were identified by either individuals with Down syndrome or their parents (Tables 2–4) and only dental health was added under the physical health domain. Supporting the topics in our conceptual model is of benefit for future work to develop a novel instrument to measure health in DS; capturing all aspects of health will ensure that the item pool for our instrument includes corresponding items. Qualitative concept elicitation is an important and necessary step in developing new patient reported outcome measures (Patrick et al., 2011). These concepts add a rich context to concerns that parents of individuals with DS have regarding mental and social health which might be informative to health care clinicians and important to include in a quality of life measure.

Parents and participants with DS discussed physical, mental, and social factors influencing health. Within physical health, both groups endorsed the value of exercise and movement while parents emphasised weight as being an important indicator of health. Indeed, children and adolescents with DS have an increased likelihood of being overweight (Bertapelli et al., 2016; Ptomey et al., 2020) which can

lead to obstructive sleep apnea and gait issues (Bertapelli et al., 2016). Parents discussion of the value of exercise and weight management shows that factors like education or resource accessibility may have a larger contribution to the increased rates of being overweight in this population than parental unawareness of this issue.

Happiness, social acceptance and engagement in activity and social environments were cited by individuals with DS and their parents. Individuals with DS mention feeling happy as contributing to health and finding happiness in accomplishing their goals and being accepted. The emphasis and value placed on happiness by both cohorts is reinforced by survey findings that report almost 99% of individuals with DS are happy with their lives and individuals with DS have personal satisfaction regardless of functional skills (Skotko et al., 2011). Parents also mentioned the value of having positive coping skills and expression of negative emotions. There is much research on how parents of individuals with DS cope with having a child with disabilities (Darla & Bhat, 2021) and it would be beneficial for future study to expand upon coping strategies endorsed by individuals with Down syndrome.

Regarding social health, both cohorts mentioned the importance of social inclusion and positive relationships. Parents emphasised the social benefit of their child with DS having siblings, and it has been reported that 97% of individuals with DS like their siblings with a majority considering their sibling a good friend (Skotko et al., 2011). Parents had a more holistic view on socialisation, discussing how limited socialisation impacts other facets of health including speech regression and behaviour.

The literature to-date describing FGs of individuals with DS is limited, and parents are often the only participants in research studies. We found that individuals with DS age 13–21 years could successfully participate in discussion about health identifying many of the same constructs of health as their parents. Although modifications were needed for reading level, with mixed benefit of graphic images, videoconferencing technology was successful. We present our full methodology and lessons learned from conducting FGs over videoconferencing, should the pros and cons of this approach be considered by other future researchers (Appendix S1).

Of note, we found the presence of parents in FGs of their son/daughter with DS largely of benefit in clarifying responses to the team from the son/daughter, but in some instances, it added uncertainty about responses due to parents prompting or correcting responses in ways their son or daughter may not have intended originally. Overall, we believe that including individuals with DS and their parents in focus groups together may be beneficial, especially to facilitate the inclusion of individuals with DS who have limited verbal communication ability. Future studies could evaluate the range of communication in individuals with DS, and the impact of including individuals with DS with, and without, their parent present on qualitative research findings. Future research should closely evaluate the use of graphics in reporting outcomes, such as has been studied in self-report of pain by individuals with DS (de Knecht et al., 2013, 2016a, 2016b). The use of graphics and scales has benefit in decreasing the need for verbal communication in reporting, if participants are able to

receptively understand a question and select a response. Although thoughtfulness is needed and modifications may be necessary to conduct self-report research from individuals with DS in practice, we feel that it is the best approach ethically, to ensure that their voice is included in research on DS.

Our study sample may not be representative of all people with DS and their parents. In our study, we focused on participants with DS who could participate in a FG and screened for such during our pre-FG telephone call. However, a wide spectrum of both intellectual disability, and expressive and receptive language, exist for individuals with DS. And, parents may underestimate their children's abilities, especially if they are not familiar with research. Furthermore, skill with technology is also varied, and the use of videoconferencing requires access to Zoom, computer, and Wi-Fi, which may decrease the generalizability of our sample. In this project, only English speakers were included to allow for discussion and dialogue, but in the future, participants who speak other languages could be included. Although we had some participants from diverse backgrounds, it is important to note that the majority of our FG participants were White, and most parents were married with a college degree or higher. This demographic breakdown aligns with research from our clinic population showing that 80% of patients with new patient visits were White, and 92% were not Hispanic (Cabrera et al., 2022). Our FG findings may not represent the views of all adolescents with DS and their parents. In the future, it would be beneficial to have more diversity within FGs, or to organise FGs by race and ethnicity to include the perspective of individuals belonging to other socioeconomic, racial, and ethnic groups and to determine how views of health indicators may vary. In our future work to develop an instrument assessing the health of people with DS, we will prioritise including diverse research participants.

We are hopeful that our FG study demonstrates that individuals with DS can successfully participate in FGs through videoconferencing technology and can be included in research. Future research should continue to collect self-report information when possible and begin to compare and contrast the views of individuals with DS to those of their parents. Parents of individuals with DS who were participants in the FGs provided feedback as to how future FGs may be improved to include individuals with DS. Future study may expand upon these suggestions by testing different methods of including the perspective of individuals with DS, so that researchers may discern what changes or modifications would benefit them when engaging in future FGs.

5 | CONCLUSION

Health in Down syndrome is multidimensional, involving physical, social, and mental well-being. Individuals with Down syndrome shared their views of health indicators in virtual FGs, and identified many of the same health constructs as their parents. Including individuals with Down syndrome in FGs could elevate their voices in research and in health discussions, especially when developing patient-reported outcomes.

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CONFLICT OF INTEREST

Dr. Santoro receives research funding from the LuMind IDSC Down Syndrome Foundation to conduct clinical trials for individuals with Down syndrome and serves on the Professional Advisory Board for the Massachusetts Down Syndrome Congress. The other authors have no conflicts of interest relevant to this article to disclose.

DATA AVAILABILITY STATEMENT

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

ORCID

Stephanie L. Santoro  <https://orcid.org/0000-0002-4172-0288>

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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