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Parent Narratives Provide Perspectives on the Experience of Care in Trisomy 18

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ABSTRACT

Trisomy 18 syndrome, also known as Edwards syndrome, is the second most common autosomal chromosome syndrome after Down syndrome. Trisomy 18 is a serious medical disorder due to the increased occurrence of structural defects, the high neonatal and infant mortality, and the disabilities observed in older children. Interventions, including cardiac surgery, remain controversial, and the traditional approach is to pursue pure comfort care. While the medical challenges have been well-characterized, there are scant data on the parental views and perspective of the lived experience of rearing a child with trisomy 18. Knowledge of the parental viewpoints can help clinicians guide families through decision-making. Our aim was to identify parents' perspectives by analyzing a series of narratives. In this qualitative study, we collected 46 parent narratives at the 2015 and 2016 conferences of the Support Organization for Trisomy 18 & 13 (SOFT). The participants were asked to "Tell us a story about your experience." Inductive content analysis and close reading were used to identify themes from the stories. Dedoose, a web-based application to analyze qualitative data, was used to code themes more systematically. Of the identified themes, the most common included *Impact of trisomy 18 diagnosis* and *Surpassing expectations*. Other themes included *Support from professionals*, *A child, not a diagnosis*, and *Trust/lack of trust*. We examined the voice and the perspectives of the parents in their challenges in caring for their children with this life-limiting condition. The exploration of the themes can ideally guide clinicians in their approach to the counseling and care of the child in a shared decision-making approach.

1 | Introduction

Trisomy 18 syndrome, also known as Edwards syndrome, is the second most common autosomal chromosome syndrome with a liveborn prevalence of about one in 6000 newborns. As is well known, trisomy 18 is a serious medical disorder due to the increased occurrence of structural defects, especially heart malformations, the high neonatal and infant mortality, and the cognitive and psychomotor disabilities observed in older children (Carey 2012; Cortezzo, Tolusso, and Swarr 2022). Interventions, especially cardiac surgery, remain controversial, and the traditional approach typically is to pursue pure comfort care (Weaver et al. 2018; Pyle et al. 2018). Currently, a rich discussion surrounding the traditional approach is occurring in the medical literature (Pyle et al. 2018; Weaver et al. 2018; Kosiv et al. 2023; St Louis et al. 2024).

In prior years, designations such as "lethal", "fatal," and "incompatible with life" were commonly used when referring to trisomy 18 (and the related syndrome trisomy 13), but because

The authors dedicate the paper to Kris Holladay, founding parent of SOFT, who has inspired parents of persons with trisomy 18 and 13 with her warmth and caring attention for more than four decades.

of increased survival rates reported in more recent population studies, these terms are no longer considered accurate and appropriate (Janvier, Farlow, and Wilfond 2012; Weaver et al. 2018; Carey 2021). In current discussions with parents both prenatally and postnatally, clinicians offer a variety of options to families including pure comfort care provided by trained pediatric palliative care teams, or varying degrees of interventions, including intensive interventions and cardiac surgery (Haug et al. 2017; Cortezzo, Tolusso, and Swarr 2022; St Louis et al. 2024). However, there remains variability among centers with some institutions routinely declining to offer technological interventions and cardiac surgery based on the diagnosis of trisomy 13 or 18. In recent years, numerous authors have challenged the narrative of declining interventions based on the diagnosis alone and suggested that a change in the paradigm is occurring (Carey 2021; Silberberg et al. 2020).

While cardiorespiratory difficulties and congenital defects have been well-characterized, there are scant data on the parental views and perspectives of caring for and rearing a child with trisomy 13 or 18. Knowledge of the parental viewpoints and their lived experience can inform care as clinicians guide and counsel families through decision-making. The aim of the investigation reported herein is to characterize parents' views and perspectives on care and decision-making in trisomy 18 by analyzing a series of parent-reported narratives and identifying common themes.

2 | Methods

In this qualitative research study, we collected 46 narratives written by parents of individuals with trisomy 18 during the annual conferences of the Support Organization for Trisomy 18 and 13 and Related Disorders (SOFT), in Salt Lake City, UT, 2015, and Tacoma, WA, 2016. The narratives were obtained using a web-based tool, Sensemaker, that the parents volunteered to take. Sensemaker incorporates the collection of micronarratives or stories, and then through a series of activities, asks the individual telling the story to self-signify its underlying meaning. The participants were prompted to "Tell us a story about your experience." The data presented here are the analysis of the parent narratives. The Institutional Review Board at the University of Utah waived the need for a full proposal based on the recruitment of attendees at the conference and the anonymity of the narratives.

We utilized two approaches to recognize specific themes in the stories: 1. Inductive content analysis: Inductive content analysis utilizes the process of abstraction to reduce and group data so that researchers can answer the study questions using concepts, categories, or themes (Elo and Kyngäs 2008). As a first step, two of the authors (RB, JCC) independently perused the narratives by close reading, identified broad themes from the content of the stories, discussed them together, and arrived at a consensus on how to label the themes. 2. Systematic analysis using Dedoose software (Dedoose: 8.2.142019 available from: https://app.dedoose.com/App/?Version=8.2.14). This program enables the investigator to easily identify and highlight ideas, phrases, and emotions from the language of the story, collect the themes, and categorize the highlighted material as themes. As a result, the program assists the investigators in more thoroughly recognizing and identifying themes present in the content. In the Results below, we have placed the identified themes in italics, and we have not edited the parents' comments.

3 | Results

The narratives ranged from a few sentences to detailed stories of more than 300 words. The ages of the family member with trisomy 18 were not recorded but were often mentioned in the narrative and ranged from 4 months to the third decade. Since the narratives were collected in 2015–2016, they likely reflect the approach to care that was occurring at the time prior to the two papers in 2016 (Meyer et al. 2016; Nelson et al. 2016) that indicated improved 1-year survival in infants with trisomy 18 compared to the population-based studies of earlier times (summarized by Carey 2012; Wu, Springett, and Morris 2013).

Common themes were identified in the stories using both inductive content analysis and Dedoose. The utilization of the Dedoose tool helped recognize additional themes systematically and categorize emotions not initially detected by the reading. See Table 1 which lists these themes and certain emotions expressed by the parents and coded by the investigators.

The two most common themes included *Impact of trisomy 18 diagnosis* and *Surpassing expectations*. Additional themes of

TABLE 1 | Identified themes from close reading and Dedoose (n = number of narratives with identified theme/feeling).

Themes
Impact of trisomy 18 diagnosis ($n = 14$)
Subthemes
Interventions ($n = 2$)
Worthy/unworthy of care ($n = 5$)
Postnatal diagnosis changes care ($n = 4$)
Surpassing expectations ($n = 12$)
Support from professionals ($n = 8$)
A child, not a diagnosis ($n = 4$)
Support from faith/religion ($n = 4$)
Change in attitude ($n = 4$)
Trust/lack of trust ($n = 2$)
Variable Emotions
Joy ($n = 3$)
Avoidant ($n = 2$)
Devastating ($n = 3$)
Hopeful ($n = 3$)
Negative ($n = 2$)

Treating the child, not the diagnosis and *Support from professionals* were prominent.

3.1 | Impact of Trisomy 18 Diagnosis

Various themes describing the *Impact of trisomy 18 diagnosis on care received* were identified throughout the stories. The impact of the trisomy 18 diagnosis was seen in a variety of contexts in the narratives. We have separated these into subthemes as follows:

3.1.1 | Subtheme 1—Interventions

Narratives included examples when the diagnosis of trisomy 18 itself was the basis for not considering cardiac surgery or interventions:

> In the limited interaction we had with the cardiology team they made it clear that surgical intervention for babies with trisomy 18 was not an option.

> Our local university hospital said it was not their normal practice to do surgery on T18 children but that we would explore it if needed.

3.1.2 | Subtheme 2—Worthy/Unworthy of Care

Additionally, the theme of *Worthy/unworthy of care* arose in narratives. The following excerpts illustrate this theme:

> They said they would not resuscitate or do any other medical procedures to save her life as they deemed her life to be of no value and were happy to let her die as this condition is fatal. Not compatible with life.

> When discussing my daughter's heart defects with doctors after her birth and diagnosis some were very cold and didn't feel my daughter was worthy of having a surgery to correct her heart defects when she wouldn't live past infancy anyway.

3.1.3 | Subtheme 3—Postnatal Diagnosis Changing Care

Examples of *Postnatal diagnosis changing care* included:

> When we got her results, they questioned whether she should have surgery or not.

> A few days later the FISH test came back and our son was diagnosed with full T18. At the point of full T18 diagnosis, care and effort of the medical team changed regarding our child's treatment.

> Our son was diagnosed with trisomy 18 on day 3 of life. At that point care and treatment was withheld and we were hearing "not for your child" and "incompatible with life."

3.2 | A Child, Not a Diagnosis

Inductive content analysis identified several narratives focused on the theme of being treated as *A child, not a diagnosis* including:

> Even though we had a trisomy 18 child, this Cardiologist was going to treat her as he would a normal child and do what was needed to let her be comfortable and enjoy whatever time we had with her.

> She was the first person in our lives that saw our son as a human being and not a diagnosis, as a part of our family and not a burden, a living baby and not a dying baby.

> I want to focus on him, not his diagnosis.

3.3 | Trust/Lack of Trust

Another theme identified by the readers included *Trust or lack of trust*. The following excerpt illustrates the theme of *Lack of trust*:

> Doctors wanted us to view our child in the same manner as them. This went totally against our moral compass and values. After our son died we were shocked! There was a DNR and comfort feeds in his chart. This was never discussed with us, nor did we consent to it.

3.4 | Surpassing Expectations

The theme of *Surpassing expectations* emerged numerous times in the accounts. The notion here is that the dire predictions of poor outcome for survival led to an expectation that did not occur, and the child did much better than the prediction. Below are excerpts:

> Our daughter was born alive! This was such an accomplishment after all of the negative possibilities we had faced during her pregnancy.

> We were also told that based on his diagnosis he most likely would not survive to the end of the week. He was born on a Monday. By the end of the week the NICU doctors felt we should discuss options for the esophageal atresia and tracheoesophageal fistula with a surgeon.

> She was born with PDA, PFO, bicuspid aortic valve, VSD and ASD. While in the hospital in the days following birth, the doctor thought she was going into the beginning of heart failure, but she is a fighter and has been with me for 16 years now.

3.5 | Changes in Attitude

Lastly, several parents identified a *Change in attitude* during the care processes:

> Our pediatrician called me one evening when our son was 5 years and 5 months old in tears apologizing for everything that had happened because he didn't know there was any hope for kids with this condition.

> So, we then had to go to a medical ethics board to get treatment since his partners were saying no. The board was divided when we went in but after meeting our son, hearing our story and seeing that we were not naïve they agreed 100% to allow treatment.

3.6 | Other Themes

Other themes included *Support from faith or religion* and *Support from certain professionals*. The following excerpts illustrate these themes:

> But with the help of God, proper medical equipment, and lots of love and dedication, my son has overcome eight cardiorespiratory arrests and 10 surgeries. He now is three years and nine months old, he has gastrostomy and tracheostomy, but he is a child very happy and loving.

> We chose comfort care and came home on day 3 with hospice. we found a great ped who suggested we just let our baby tell us what she needed. we worked on weight gain but did not do unneeded tests. we did not see specialists. we just enjoyed her and now she is still with us many years later.

3.7 | Variable Emotions

We also recorded various feelings observed in many of the stories. Feelings coded included joyful, avoidant, devastated, hopeful, and negative emotions (see Table 1).

An example of negative emotions included:

> Although he indicated that we had a choice, I definitely had the feeling I was supposed to be able to let the baby go rather than provide heroic measures. I felt like a raw egg that's been thrown on a brick wall. The wall is hard and immovable, and I'm utterly gelatinous like a raw egg. The walls pits in the bricks absorb me into them, and I have no way to change the splat.

Examples of both hope and no hope are below:

> She wasn't unrealistic about her, but at the same time was willing to leave room for hope which we greatly appreciated.

> A long and winding and scary and hopeful road.

> I never heard a word of hope.

4 | Discussion

In this qualitative study, we collected 46 narratives from parents of children with trisomy 18 who were attending the SOFT Conferences in 2015 and 2016. Using two related approaches, we identified various themes in the stories and categorized them into designated groupings that potentially reflect the lived experience and the voices of the parents. We will focus our discussion on three of these themes in more detail as they characterize the salient findings of our study. These notable themes are the *Impact of trisomy 18 diagnosis*, *Treat the child and not the diagnosis*, and *Surpassing expectations*.

4.1 | Impact of Trisomy 18 Diagnosis

Parents shared stories of different instances when the diagnosis of trisomy 18 impacted the care their child received. Unfortunately, many of these stories involved negative attitudes or biases, and failures to align care with the parents' goals. Janvier and Watkins describe how "parents may be forced into a position in which they must fight to obtain interventions for their child, leading to an entrenchment in positions and a cessation of dialogue. An attitude and language of universal futility and lethality for these conditions has unfortunately created many conflicts" (Janvier and Watkins 2013). In some situations, this occurred when the diagnosis was made postnatally, (the theme of postnatal diagnosis changes care), and recommendations changed based on the perspective of the providers. Consistent with parents' experiences, it has been reported that "children who had a postnatal diagnosis and a plan for palliative care were more likely to go home than children with a prenatal diagnosis and a plan for interventions" (90% vs. 58%; $p < 0.01$; still significant after correcting for congenital anomalies, Janvier, Farlow, and Barrington 2016). Having a prenatal diagnosis of trisomy 18 (and trisomy 13) portends lower infant survival (Janvier, Farlow, and Barrington 2016; Kato et al. 2019), and the basis of this observation deserves additional investigation. Mercurio et al. report that current approaches to information-sharing after diagnosis of trisomy 18 may not be as accurate, complete, and unbiased as they should be, and observed that physician arguments for withholding treatment have lost credibility owing to insufficient corroborating evidence (Mercurio, Murray, and Gross 2014).

Themes identified in the parent narratives of our study further reinforce the need to consider the individual child and her problems, rather than anchoring on the genetic diagnosis, when partnering with families to provide goal-directed care for their child.

4.2 | A Child, Not a Diagnosis

Parents impart a positive impact when providers acknowledge their child as an individual. Using their name, seeing beyond the chromosomal abnormality and its prognosis, and sharing in the joy and unique achievements of their child evoked trust in parents. They reported feeling supported in these instances. Several of the narratives explicitly described support from the helping professional, which was also recognized as a consistent theme by Janvier, Farlow, and Wilfond (2012) in their survey of families on social networks. Allowing room for hope was also identified as a source of support in several parent narratives. Hope as a theme and coping mechanism specifically in the parent of a child with trisomy 18 is discussed in a detailed analysis of the narratives in a parent's blog over many years by Szabat (2020). When parents reported that their child was recognized by providers as an individual and an important member of their family, care tended to be better and more satisfactory and aligned with parents' goals.

4.3 | Surpassing Expectations

With the variability of survival and outcomes for individuals with trisomy 18, accurate prognostication is challenging. Parents provided many examples of how their child surpassed expectations—those of medical providers and even their own. Interestingly, parents often described success in achievements that may seem minor to others. This speaks to the difference in perspectives between healthcare professionals and the parents or caregivers of children with trisomy 18. Many parents shared the pride and joy they experienced when their child defied the odds, a meaningful experience to all.

There is a common thread throughout these stories: the parent view that the diagnosis of trisomy 18 in and of itself impacted the approach to care that the child received. The themes of the *Impact of trisomy 18 diagnosis*, *Treat the child and not the diagnosis*, and *Surpassing expectations* all reflect this notion. These themes together represent what Attride-Stirling (2001) calls organizing themes that taken together are a global theme, i.e., "central tenet." We suggest that care be taken to recognize the impact that this "central tenet" may have on our ability to truly partner with parents in a shared decision-making process to align their goals with the care their child receives.

4.4 | Literature Review

Other investigators have explored parent perspectives in trisomy 18, often using a mixed methods approach with reporting of analyses of narratives that are evoked in open-ended survey questions. In their survey study of parents using social networks, Janvier, Farlow, and Wilfond (2012) documented that children with trisomy 18 (and trisomy 13) often transformed the lives of those around them. This seminal work and the related papers that followed (Janvier, Farlow, and Barrington 2016; Guon et al. 2014) provide substantial insights that include the parents' voice on the issues of care and the meaningful lives of their children. The survey study of Tsurusaki et al. (2013) also documented the positive feelings of parents in caring for their children with trisomy 18 while concurrently recognizing the medical challenges and the parental anxiety over possible death. Bruns and colleagues contributed two papers between 2010 and 2014 using a parent-reported dataset that also reflected positive views of parents of children with trisomy 18 despite the medical complexity. Arthur and Gupta (2017) interviewed seven families, whose child had trisomy 18 and one whose child had trisomy 13, using open-ended questions designed to examine their experiences with medical care. A number of themes were identified, most interestingly that their children "transform the lives of others" and the families are motivated to tell their story and as stated in the title of the paper "carry their torch." Weaver et al. (2018) documented the perspectives of two mothers of infants with trisomy 18 and summarized their interviews by saying "We benefit from exploring the family's past and current understanding of their child's condition…and actually engaging in a story not of 'a trisomy baby' but rather of a story of a unique, fiercely loved child held within the context of family unit." Szabat (2020) analyzed the experience of one mother through her blog which recounted the story of her son with trisomy 18 and the medical challenges over 8 years. The global theme that Szabat recognized was hope. The nuanced discussion of hope in this article, the author states, "may expand knowledge on this issue helping HCPs (health care professionals) to better understand the parents' experience of care." Parents perspectives shared in these stories highlight key elements in providing family-centered care. These include providing accurate information while allowing room for hope, respecting the child as an individual and as part of the family, and striving to provide compassionate care without bias.

4.5 | Limitations

Our study has limitations. There may be selection bias and confirmation bias in the study sample. Parents who attend a conference may not represent all parents of children with trisomy 18. Presence at a national meeting may reflect more available resources and parents who leaned toward the decision of intervention for their children. Confirmation bias, the tendency of people to favor information that confirms their existing beliefs, could be a factor in parents perceiving that their child is happy or experiencing a good quality of life.

In conclusion, our goal was to examine the voice, the perspectives, and the viewpoints of the parents in their challenges in caring for children with trisomy 18. The recognition and exploration of the themes can ideally guide clinicians in their approach to counseling and support in the care of the child and their parents. This can help facilitate a dialogue regarding the notion of treating a child as a child and not a diagnosis.

Author Contributions

Bierer and Carey conceived the idea of the study; all the authors helped design the methods of the study. Bierer collected the data; all the authors contributed to the analysis of the data. Bierer and Carey drafted the paper, and all the authors read and edited the final copy.

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Conflicts of Interest

The authors declare no conflicts of interest.

Data Availability Statement

The data that support the findings of this study are available from the corresponding author upon reasonable request.

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