

Eliciting Narratives to Inform Care for Infants With Trisomy 18

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CLINICAL SCENARIO

In the span of 1 month, our free-standing children's hospital assumed care for 2 infants <4 months of age diagnosed with trisomy 18 from 2 separate out-of-state referring hospitals. One infant had a ventricular septal defect. The other infant had a cleft palate, an omphalocele, and aortic coarctation with atrioventricular septal defect. In both cases, the infants' parents initiated an evaluation and transfer on the basis of what they had read about cardiac care for local infants with trisomy 18 on social media posts by the Support Organization for Trisomy 18, 13, and Related Disorders group. This group maintains a strong social media presence, with parent solidarity across geographic realms. With third-party coverage approval for transport and medical interventions, both families traveled >1000 miles from their homes to pursue cardiac surgical intervention for their infants instead of the locally offered comfort-care approach.

The approach to interventions for infants with trisomy 18 is historically divided across providers and even across hospital policies.¹ Past thought was that trisomy 18 represented a universally fatal condition in infancy and thus any risk, cost, or morbidity associated with cardiac surgery would be intolerably burdensome for a presumably futile outcome. Newer data reveal that trisomy 18 is not uniformly lethal and that prolonged survival is possible.^{2,3} In a large survey of parent support group members, almost all families described a positive view of the quality of life of their

child with trisomy 18 (88%) and report a positive effect on their family life (98%) and marriage (68%).⁴ A tension currently exists between limiting management to comfort, palliative care without surgery, and the offering of interventions, such as corrective cardiac surgery with palliative care for infants with trisomy 18. A survey of 859 neonatologists, geneticists, and cardiologists revealed high variability in clinician support for cardiac surgery for an infant with trisomy 13 or 18, ranging from 32% of cardiologists to 20% of geneticists to 7% of neonatologists.⁵ In a survey of extracorporeal membrane oxygenation (ECMO) medical directors facing 5 scenarios of ECMO candidacy, the lowest ECMO acceptability score was in the scenario of an infant with trisomy 18.⁶ In a 2017 survey of 30 Canadian cardiologists, most (67%) supported comfort care for affected patients with a heart lesion, and none supported surgery for those with complex heart lesions.⁷

Wide variations are now reported in cardiac surgical outcomes for infants with trisomy 18. As a result, outcomes are not homogenous.⁸ Outcomes include 1-year survival rates of up to 75% for select Japanese children with trisomy 18 for whom cardiac surgery was deemed an appropriate option.⁹ Children with trisomy 18 who were selected as acceptable candidates for cardiac interventions and who survived these operations demonstrated longer survival than previously assumed (in-hospital mortality rates: 13%; overall median survival conditioned on hospital



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Dr Weaver designed the study, compared the audio interview content to the typed transcription, coded the interview content by themes in NVivo qualitative software, and drafted the initial manuscript; Ms Austin and Ms Stevenson coconceptualized the paper content; Drs Hammel and Starr reviewed and revised the manuscript; and all authors approved the final manuscript as submitted and agree to be accountable for all aspects of the work.

DOI: <https://doi.org/10.1542/peds.2018-0321>

Accepted for publication May 1, 2018

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PEDIATRICS (ISSN Numbers: Print, 0031-4005; Online, 1098-4275).

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FINANCIAL DISCLOSURE: The authors have indicated they have no financial relationships relevant to this article to disclose.

FUNDING: No external funding.

To cite: Weaver MS, Starr LJ, Austin PN, et al. Eliciting Narratives To Inform Care for Infants With Trisomy 18. *Pediatrics*. 2018;142(4):e20180321

discharge: 16.2 years [confidence interval: 12.3–25.6 years]].¹⁰ Some would argue that technological interventions, including cardiac surgery, not only prolong infant survival but may also improve quality of life in trisomy 18.

Disagreement about the level of cardiac interventions offered may reflect differences in perspective between health care professionals and family members concerning the experience of children with trisomy 18, different knowledge bases (related to medical and surgical feasibility), differing ethical or religious beliefs, misinformation, and cost or insurance coverage. Parents of children with trisomy 18 generally perceive higher quality of life and less pain and suffering than had been predicted by their health care teams.⁴

LISTENING 18 QUALITATIVE INTERVIEWS

When we face uncertainty regarding outcomes, we can remain confidently certain of the value of honest and respectful dialogue. Our team thus designed the LiSTENING 18 (Learning in STory: Elicited Narrative Informs Navigating Genetics for Trisomy 18 in 2018) qualitative interview approach. The qualitative methodology included a sequence of questions designed to explore respondents' experiences, hopes, understanding, and goals. Transcriptions then underwent content analysis by using NVivo software to explore shared themes. This case series protocol was exempted by our institutional review board (IRB) and approved by our medical ethics committee as a means to honor full family partnership through qualitative inquiry. For the voice-recorded interviews, our goal was to meet with the parents of these 2 infants with trisomy 18, who had traveled >1000 miles for cardiac care, and to engage in honest, open dialogue, listening to their stories as

a form of respectful communication to foster care-team understanding. The mothers were interviewed because the fathers remained in their home states for employment. Families were provided with the list of interview questions 48 hours before the scheduled interview and were offered the opportunity to skip questions or not participate; both families eagerly coparticipated in full. Afterward, the written transcript of the interview was shared with each family as a form of dignity therapy so that the family could have access to their own narrative.¹¹ Responses to the interview questions are shared consensually in topical format below:

What Have You Been Told To Prepare for in Terms of Your Child's Trisomy 18 Diagnosis?

Christina: We were basically told to prepare to lose the baby. They told us most people don't make it to birth and the ones that do don't last long. It is hard news to hear when you are pregnant. Birth is supposed to be something that you look forward to [happening]. We hoped to enjoy the experience of pregnancy and the fact that we have a baby girl for the first time. It made me worry everyday whether I am going to wake up and not feel movement? Am I going to wake up and be bleeding? When am I going to lose this loved baby?

Paige: They started telling us this is Trisomy 18 and saying she is not going to live. I was told babies with Trisomy 18 don't live through the pregnancy—most likely. The doctors made it seem like it is very rare for a baby to live with that through pregnancy or even birth and they pass away right after birth. So we were really devastated. I was told to prepare for a miscarriage. I was told my baby would not likely make it. Then we were told we would be lucky if she lived past labor. When I went into labor I was trying to be optimistic that she would make it through, but, I was kind of also

prepared for her not to make it because I know labor is hard on a baby. When her heartrate dropped in labor that was very traumatizing because I thought, "OK, this is it."

What Are You Hoping for?

Christina: We are hoping that she can enhance future options for children with her condition. We are hoping her story shows the world how much children can benefit from getting care that increases their abilities and their opportunities to thrive. She is a very special baby. She is one of those children that will inspire others and even inspire health care teams. I hope that she is able to show people her worth because some people read a diagnosis and a prognosis and make assumptions that don't really reflect individual children. I want her to defy the odds and let people know that there is a range of trisomy that varies so much you can't really make assumptions based on simply a chromosome. You have to learn about that special child and that unique family and then care accordingly. We hope for people to look at her beyond just her trisomy and her prognosis to maximize her chance for today to be a good day for her.

Paige: I would like to be able to bring her home and have a family experience having her home. I am hoping to let her have quality time with us as a family once we are able to get her to that point. I hope one day she will be well enough to go home with us to be a family for as long as she is able.

What Is Your Understanding of Your Daughter's Prognosis?

Christina: My understanding of prognosis is that the odds are stacked against my daughter. She has got a lot of uphill battles to fight. Frankly, we don't know what tomorrow brings. We definitely don't know what next year brings for her health-wise. We are blessed that she is not showing a lot of the significant life-threatening

health issues that a lot of trisomy babies start off with [having]. But that doesn't mean that tomorrow will not be a different story for her and for us. So, in terms of prognosis, we get the big picture prognosis because of her trisomy. We want her to be as comfortable and as healthy as possible for today. We will handle each uphill problem as it comes. As long as she is showing us that she wants to be here and has a will to live and is happy then we are going to support her for that.

Paige: I know it is hard to tell with Trisomy 18. In general, not given all of her additional specific limitations, it is hard to tell. They say days, weeks, months, and rarely a year of life. Then with her heart issues and breathing and bowel it is more impactful for her. Actually, her breathing is my main concern right now. She has the potential to have a complete repair for her heart. You know, they said the heart repair may not actually extend her life greatly. I read a study on Trisomy 18 and 13 babies and young children with and without cardiac repair and even in that study after full cardiac repair, the lifetime of the babies who had full cardiac repair was usually only about 2 full years. They did see that the heart surgery helped in some cases. But, the life span after even a full cardiac repair wasn't given something like 18 years. I know that. I just think every day counts and so even some additional days that are quality days do count as special for a baby and a family.

What Does It Mean for Providers To Care Well for Your Daughter and Your Family, Even Providers Who Maybe Have a Different Perspective on Offering Aggressive Medical or Surgical Interventions?

Christina: Compassion is important in health care. One of the things we are most thankful for is compassionate health care providers. People that really show they care about your child and care about giving them

the best opportunity have blessed us. Compassionate providers have blessed not just our baby but have actually blessed us as a family. It's the health care people who did not write our child off before they met her based just on her trisomy label who helped us. One thing most trisomy families struggle with is having care options immediately limited by this label of trisomy. I have been told by multiple moms of trisomy babies that they were told at the hospital that their only option was hospice. It matters to us that a baby be given every opportunity and that the family's values be discussed and heard. We aren't naïve to prognosis but we also want to hope for our child and to maximize the child's quality of life. It matters when you take the time to listen and to learn about us and to together develop a plan for what care could be for our child. These babies are really loved. If we listen, we can learn from them. I learn from my daughter. I think health providers can even learn from her.

Paige: Caring well means not being immediately like, "Well, she has Trisomy 18, so we can't do that and we can't do this and by the way she will never do that and she won't ever do this." Caring well means listening with openness and working with what is best for her and even hearing family goals. Caring well means don't put a family's hopes down just because of a chromosome. You can be realistic and we can be realistic but maybe there are some hopes or goals that can be achieved and caring well means hearing and helping, as able. I think caring best is done by first listening and then by not putting every child under the same category. Babies with Trisomy 18 are all so different even though they have the same genetic diagnosis. I think a lot of the kids, even the ones with full trisomy, can thrive for the number of days they are given if they are given the proper medical treatment. I do

believe there are a lot of trisomy children that have died before their actual time and now grieving parents and hurt families because they couldn't get forms of care or they couldn't find medical providers agreeable to partner in options.

DISCUSSION

Trisomy 18 is a genetic condition that historically had a perceived dismal outcome. This syndrome has a spectrum of phenotypical presentation and lacks clarity of a best chance of outcome. Although there is certain comfort in uniform standards on how to approach biomedical diagnoses, the reality is that families will make a variety of choices from terminating a pregnancy to advocating for extraordinary and all interventions. Pediatric medical ethicist John Lantos calls this "the grey zone" in which family choice is codeterminative.¹²

Families with infants prenatally diagnosed with trisomy 18 defined special care teams as those who "gave balanced and personalized information, respected their choice, and provided support."¹³ When we struggle as health professionals, we benefit from listening. We benefit from not just telling families about the prognosis but also from exploring the family's experience in receiving prognostic information. A family's previous history of prognostication or receiving bad news may have been a traumatic revelation or a bombardment of agenda (such as one family of an infant with trisomy 18 reported being asked >15 times over the span of 3 weeks about their child's do not resuscitate status). We benefit from exploring the family's past and current understanding of their child's condition, realistically understanding the risk-benefit ratios for interventions, and actually engaging in a story not of "a trisomy 18 baby" but rather the story of a

unique, fiercely loved child held within the context of a family unit.

CONCLUSIONS

In caring for these 2 infants, we bore witness to families who traveled despite the financial strain and relational impact (unpaid leave even with home rent to pay, siblings only knowing their sister through FaceTime, grandparents saying goodbye to a loved grandchild without certainty of a future reunion, marital units physically separated during prolonged hospitalization).

These initial 2 families reached out directly to our cardiac surgical team for a case review and evaluation. As a result of our experience in caring for these 2 families, our center's care for patients with trisomy 18 has dramatically changed. We formed a trisomy 18 interdisciplinary panel at our institution with neonatal, intensive care, cardiac, surgery, otolaryngology, palliative care, social work, and case management members. Before establishing this panel, our study team hosted a hospital-wide, interdisciplinary journal club across medical subspecialists to foster open dialogue among providers regarding coordinated care for future families

of infants with trisomy 18 who are seeking cardiac surgical options. In addition, the study team is now organizing a nursing education initiative on family experiences for infants with genetic diagnoses.

The 2 fathers were not approached for this Learning in Story: Elicited Narrative Informs Navigating Genetics for Trisomy 18 in 2018 protocol because our IRB and ethics committees did not give permission for telephone or remote interviews. Our study group has just received IRB permission to begin a project called PAGES (Paternal Approach to Goals, Experiences, and Support) to further explore paternal perspectives for all pediatric-palliative-care patients. Our center has been selected as host site for the annual Support Organization for Trisomy 18, 13, and Related Disorders conference.

Both infants underwent cardiac surgery. With each of these 2 families' permission, we continued open communication with the local home-palliative care teams for coordination of palliative care across settings longitudinally. One infant returned home and is thriving with ongoing local palliative-care-team support. The other infant died of sepsis, but the parents continue to

receive bereavement care, and they exemplify "peace at having pursued life-honoring care interventions to include surgery."

A trained practice across diagnoses is to listen for good intention. There is a certain honor to ongoing humility and curiosity. When there is a difference in perspective among health care team members or even among family members and care providers, we start by listening. Asking each other what we have experienced, what we hope for, what we understand, and how we define care is a form of clarity. Thus, when we listen to valued partnership with families, we work toward patient-centered care and shared decision-making pathways.¹⁴⁻¹⁶

ACKNOWLEDGMENTS

We thank Ms Priya Maillacheruvu for her assistance in interview recording and Ms Sara Woodworth for her assistance with interview transcribing.

ABBREVIATIONS

ECMO: extracorporeal membrane oxygenation
IRB: institutional review board

POTENTIAL CONFLICT OF INTEREST: The authors have indicated they have no potential conflicts of interest to disclose.

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Pediatrics 2018;142;

DOI: 10.1542/peds.2018-0321 originally published online September 6, 2018;

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