Cardiac Interventions for Patients With Trisomy 13 and Trisomy 18: Experience, Ethical Issues, Communication, and the Case for Individualized Family-Centered Care

John P. Cleary, MD[1,2], Annie Janvier, MD[3,4], Barbara Farlow, MBA[5], Meaghann Weaver, MD, MPH[6]

online and awareness of variation in approach creates distress for parents and treating teams. It is also possible that parental hopes could be inappropriately inflated by the most positive outcomes shared in support groups. A more rational, patient-centered approach is called for.

Methods

The program committee of NeoHeart 2020—The International Conference of the Neonatal Heart Society identified progress and ethical tension in the care of patients with Trisomy 13 and 18 as an important topic for expert review. The mission of the Neonatal Heart Society includes improving collaboration between disciplines caring for newborns with the cardiopulmonary disease through education and advocacy.

The authors were chosen as recognized leaders with clinical experience and academic work regarding Trisomy 13 and 18 and represented neonatology, cardiothoracic surgery, ethics, palliative care, and parent perspectives. The majority of authors have specifically advocated for heart surgery in patients with Trisomy 13 and 18 in some circumstances. In the session (Trisomy 13/18, Exploring the Changing Landscape of Interventions) the authors reviewed the existing literature on clinical outcomes, ethical constructs, and family surveys along with personal reflections.

Key themes of the session including the evolving approach to heart surgery, defining gray zones, and key concepts in communication are represented in this work along with recommendations to caregivers which reflect consensus of the authors.

Evolving Approach to Infants With Trisomy 13 and 18

A gray zone exists when there is ambiguity as to the best interest or best approach to care. In such a situation, reasonable families might make different decisions regarding care. If care is uniformly harmful or a condition uniformly physiologically lethal even after treatment, then treatment should be considered futile and it should not be offered.[16]

Professionals who have cared for patients with congenital heart disease have experienced the movement of several diagnoses into and out of gray zones. Surgeons routinely balance clinical and ethical risks in deciding when or whether to offer surgery. Complex surgery in patients with Down Syndrome was not routinely performed until the early 1970s.[17,18] Comfort care was the routine approach to the care of infants with hypoplastic left heart syndrome until the 1980s.[19-21] Today, there are few heart lesions for which treatment is clearly physiologically futile. Instead, doctors and other health professionals must help and support families in decisions about whether to pursue surgery with the inclusion of palliative care.[22]

While uncommon, heart surgery for patients with Trisomy 13 and 18 has occurred for many years. Peterson et al[23] performed a retrospective review using the Pediatric Cardiac Care Consortium to identify 171 Trisomy 13 and 18 patients who underwent cardiac surgery between 1982 and 2008. In a hospital, mortality was 28% for Trisomy 13% and 13% for Trisomy 18. Many of these operations were lower risk lesions and in older patients.

The Society of Thoracic Surgeons National Database shows that surgery has been offered under some circumstances at a majority of the 125 member centers.[24] From 2010 to 2017, 343 operations were reported with most centers performing less than 1 case per year and one-third of centers reporting no surgeries. Mortality in this selected group was approximately 15%. Operative mortality was high in comparison to patients without Trisomy 13 and 18.

Domingo et al[25] used the Pediatric Health Information System (PHIS) database to identify 189 patients with Trisomy 13 and 18 who underwent cardiac intervention between 1999 and 2015. Shunt lesions were the most common diagnosis followed by conotruncal defects. Mortality was 29% in Trisomy 13% and 12% in Trisomy 18 patients. Older age at surgery correlated with decreased mortality.

Only a small percentage of potential heart surgeries are performed. Kosiv et al[26] also used PHIS data to identify a cohort of 1668 newborns with Trisomy 13 and 18 born between 2004 and 2015. While congenital heart disease was present in 86% of Trisomy 13% and 91% of Trisomy 18 patients, congenital heart surgery was performed in 7% of patients in each group. Patients who received heart surgery had reduced hospital mortality and improved 2-year survival.

Nelson et al[9] reviewed the outcomes of 328 patients with Trisomy 13 and 18 born in Ontario, Canada between 1991 and 2012. While mean survival was less than 2 weeks, surgical interventions were performed in 24% and cardiac surgery in 14% of patients. Surgical intervention correlated not just with increased likelihood of discharge home and improved 1-year survival but many patients were thriving in the second decade of life. Centers who have historically limited heart surgery in the setting of Trisomy 13 and 18 should at a minimum consider offering surgery to infants who have survived to discharge and are thriving at home.

It is important to acknowledge that the growth in public reporting of congenital heart surgery outcomes could have the unintended consequence of creating conscious or subconscious barriers to offering surgery in patients who may have a limited lifespan. Comorbidities such as genetic syndromes and noncardiac anomalies may not be adequately accounted for and could create an inaccurate description of a hospital's performance.[27] Professionals and advocacy groups should support efforts to improve case-mix adjustment or perhaps to partition Trisomy 13 and 18 outcomes from general comparisons.

Avoiding Harm—the Pendulum Analogy

Improved outcome data have led more centers to rethink their approach and provide complex surgery to patients with Trisomy 13 and 18.[10,28-30] We caution that, similar to a

displaced pendulum, the trajectory moving away from "never offer surgery" for children may risk a turn to the extreme opposite pole to "always offer surgery" for children with these conditions.[31] It would be unfortunate if the family experience moved from barriers to surgery to inflated expectations of benefit or worse shortened lives and reduced time at home. We think that the data justifies an individualized approach based on each patient's condition, associated anomalies, and parental preference. Balancing such information, clinicians and families can weigh the intended goals of the intervention with the feasibility of obtaining that goal.[32] This is ideally accomplished through the inclusion of interdisciplinary teams to foster comprehensive care inclusive of but not limited to the cardiac diagnoses. Janvier et al[8] recently published data obtained from 332 parents of 272 children with Trisomy 13 and 18 documenting that their hopes are generally to give their child a chance, to take their child home, and for their child to have a good life. Their fears include medical complexity, pain, and harm to their child. Parents may have inflated hopes based on beneficial interventions less afflicted children on social networks have received. A family centered or "baby friendly" team must acknowledge that not all interventions are in the best interest of a baby.

The authors endorse the spirit of the report of Neubauer and Boss[32] that babies should not be denied potentially beneficial surgery based solely on the presence of Trisomy 13 or 18 while teams maintain medical and surgical discretion as to the actual benefit of the surgical intervention to the child's well-being. Cardiac surgical teams should always share cardiac outcomes-based insight with families. The goal is to together communicate with families regarding the shifting data including survival paradigms, the benefit-harms ratio specifically in context with the child's concurrent comorbidities, and the family's values or hopes in this context. Extending the pendulum analogy, the pivot away from "never" should be less toward the "always" offer surgery and more toward the "centering position" composed of communication that oscillates about the medical data and the family goals toward a restoring force of trust, honesty, quality care and the best outcome for the child.

Table 1 includes several potential scenarios where the authors suggest that guidance can and should vary from limiting to encouraging surgery.

Guidance to Clinicians

The authors suggest that the care of families whose child has Trisomy 13 and 18 must be individualized and supportive of a range of interventions. A defining feature of a gray zone is that different parents will make different decisions about life-sustaining treatment.[16] Support of families must be individualized. Blanket policies such as "no resuscitation" or "no surgery" may be intended as transparent messaging but can instead lead parents to fight for an intervention they may not have chosen in a more supportive context.

Table 1. Case Scenarios with variable Guidance.

	Case 1	Case 2	Case 3
Scenario	A 3-day old infant with Trisomy 18, ventilator dependence, hypoplastic left heart syndrome with a restrictive atrial septum in the intensive care unit	A 4-month-old infant with Trisomy 13 residing at home with ventricular septal defect tolerating nasogastric feeds with evolving pulmonary hypertension	A 2-year-old with Trisomy 18 residing at home with a gastrostomy and supplemental oxygen, communicates via signs and gestures, unrepaired DORV with PS (TOF type) with cyanosis due to worsening pulmonary outflow gradient
Family perspective	Family seeks a miracle and adamant child will reside independently at home after cardiac surgery	Initial discharge targeted time at home and limiting suffering. Family views baby as happy and responsive to sibling interaction; family desires additional time home together as a family	Family recognizes child has meaning in a family unit and enjoys life; care has already been medicalized
Medical deliberation	Surgical offering would not have a clear benefit to a child; concerning mortality risk; noncardiac comorbidities concerning; family's desired outcome is not realistically conducive to biomedical insight	Surgical offering may have symptoms and survival benefits to a child; ensure that management of pulmonary hypertension has been maximized and reduce likelihood of death related to pulmonary hypertension	Surgical offering would have likely benefit to maintain an already medically and surgically treated child's home-based well-being
Recommendation	Recommend not offering cardiac surgery and interdisciplinary team working with the family to foster meaningful time and recognition of child's value in a family unit	Recommend to consider surgery in context of potential benefit to child and goals of the family unit with interdisciplinary team support	Recommend to offer surgery and work together with interdisciplinary team to maximize home-based developmental supports

Abbreviations: DORV, double outlet right ventricle; PS, pulmonary valve stenosis; TOF, tetralogy of Fallot.

Table 2. Themes in Family Communication.

What to do	What to avoid
Know child's name and use it	Naming by diagnosis or treating like a diagnosis
Demonstrate humility—display uncertainty and partnership	Absolute predictions—when baby proves you wrong trust is broken
Personalize information—share your thoughts openly and be open to learning	Uniformly negative information—closes communication
Balance information—create dialog to define "good life" in families view	Always and Never statements such as "incompatible with life."
Affirm that parents are "good parents" who are doing their best for their child	Judgement—you are in a position to create distress and increase guilt
Be patient—create a supportive environment and affirm that you will work together	Isolating or abandoning parents

Important data generated from the families of children with Trisomy and extreme prematurity confirm that the value of their child must be acknowledged and that a small number of trusted special healthcare providers improved their experience.[7,33,34] While special medical providers are found in all disciplines, clinicians experienced in palliative care and ethics support should be a part of all teams. Supportive providers help families define what their view of a good life might be. They help define wishes and fears generally such as avoiding prolonged hospitalization, allowing time at home, and/or minimizing pain.

Supportive clinicians inspired trust. Parents reported they were knowledgeable, gave accurate information but also knew specific details about the individual child. Supportive clinicians were also humble about the limitations of their knowledge and curious about the family. Parents wanted clinicians to treat their child as an individual of value, not "a trisomy 18." Supportive clinicians recognized the value and the uniqueness of the child by using the child's name, providing information tailored to their specific situation, and recognized the meaning of the child's life, even if short. Phrases such as do everything and do nothing should be avoided.[35,36] Supportive clinicians also provided appropriate hope, such as the hope that parents would spend time with their child—even unborn, that they would meet them at birth, and would be able to care for them for the longest time possible.

Some families might be comfortable weighing statistical data while as many as 40% want direction on how to proceed. Data and support should be adjusted for dose and timing as future decisions such as heart surgery do not need to be confronted during the first contact. Avoid a long menu of advanced planning—some complex decisions can wait or may never be relevant.[8]

Table 2 provides summary guidance in communicating with families.

Summation

Care of families with Trisomy 13 and 18 has moved solidly into a gray zone where all care, including heart surgery, can be considered and individualized in the context of family centered care. To categorically deny interventions to this population is an idea whose time has gone. We must strive to find the "centering position" of the treatment pendulum as surgical outcomes evolve and families are included as partners in care.

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ORCID iD

John P. Cleary https://orcid.org/0000-0001-5036-373X

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