Cardiology in the Young

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Communicating with parents of children with trisomy 13 or 18 who seek cardiac interventions

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Brief Report

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Abstract

This case report shares the story of a family who sought care elsewhere after their daughter was denied cardiac surgery in their home state because she had trisomy 18. This case report recommends case-by-case assessment of cardiac surgical interventions for children with trisomy 13 or 18 as informed by review of goals, assessment of comorbidities, and literature-informed practice. Coordinated care planning and interdisciplinary communication are relevant in cardiac surgical considerations for children with these underlying genetic conditions.

Trisomy 13 and 18 were historically considered universally "lethal" and "not compatible with life."[1,2] This viewpoint resulted in lack of conversations about possible cardiac surgical interventions and an expected end of life within the first 2 weeks of life.[3] Ninety per cent of patients with trisomy 18 and 80% of patients with trisomy 13 are concurrently diagnosed with congenital heart disease (CHD), primarily ventricular septal defects.[3] Unrepaired cardiac defects and pulmonary hypertension have been recognised as leading causes of death in children with these trisomy diagnoses.[4–6] In 2019, the 5-year survival rates revealed trisomy 13 at 7.7% and trisomy 18 at 7.7%.[7] Outcomes-based data on preoperative comorbidities have become increasingly available.[8–10] Recognition that patients with trisomy 13 and 18 typically die of cardiac disease, but may survive cardiac surgery and experience a meaningful quality of life,[11–15] warrants case-by-case considerations of cardiac interventions for children with these genetic conditions.[6]

There is a wide spectrum in phenotypic presentation for children with trisomy 13 and 18.[3] Sometimes, these conditions lead to intrauterine fetal deaths. Most babies are born with multiple associated anomalies. Others, though, have less severe disease and few comorbidities. In spite of patients' phenotypic variation, many medical centres have practices to never offer cardiac interventions for patients with these genetic conditions.[16–19] Whether the practice is an actual written institutional policy or a hospital culture or strict preference of the surgical teams,[20,21] some families may hear language of "we never offer interventions for babies with that genetic condition" or "we don't ever do surgery for children with that genetic condition."18 This was similar language heard by parents of children with Down syndrome until the late-1970s.[17,22]

Parents now communicate with each other via social media or online support groups. Thus, they are aware of practices at different centres. They classify some centres as "friendly" and others "unfriendly" to patients with these conditions.[23] Parents then seek care at the "friendly" centres where other families with their child's similar condition received interventions. Hospitals that consider cardiac evaluations or offer cardiac interventions receive a disproportionate amount of requested second and third opinions by distressed parents who often have lost trust in their local centre and sometimes in the medical profession itself. In some cases, there is collaboration and communication of goals between clinicians at either location and in other cases, there is not.

We share a family narrative in this case report with humble recognition that the parent serves as the expert regarding the family's philosophical values, while the medical team serves as the expert of the child's physiological realities and suggest that, based on the phenotypic variation, centres should not have blanket policies regarding cardiac surgeries for children with trisomy 13 or 18. Instead, a thoughtful approach may be case-by-case review of potential cardiac interventions for each child based on that child's unique comorbidities and a review of each family's goals.[24] Such an approach has been considered relevant for "gray zone" biomedical circumstances, that is, situations in which it is ethically justifiable to provide either life-sustaining treatment or comfort care.[25]

Case report

Charlotte (name changed), a thriving and cheerful 2-year-old, was diagnosed with trisomy 18 by amniocentesis in utero. After counselling, Charlotte's parents decided to continue the pregnancy. Similar to other parents with children with this genetic diagnosis,[26] their stated goal was to "meet Charlotte and give her a chance to be part of the family." They were aware that she might die and did not want to use heroic measures to keep her alive. They hoped to take her home and give her the best life possible. Charlotte's mom took extra care during her pregnancy, recognising the days in utero may be Charlotte's only days. The family had decorated a home nursery even though the dire medical warnings left them fully prepared for Charlotte to die in the delivery room.

Charlotte was vigorous at birth. She did not require resuscitation in the delivery room. She was admitted to the neonatal ICU and underwent confirmatory genetic testing. Echocardiogram revealed a moderate-size ventricular septal defect. Brain MRI revealed normal cranial morphology. A sleep study was normal with no apneic episodes. Charlotte went home from the newborn nursery on day 18 with nasogastric tube feeds on room air with supplemental oxygen available at night "for comfort."

Charlotte did well at home. Her family loved her. At 2 months of life, Charlotte showed signs of heart failure: sweating during feeds, fatigue, pallor, and a wet-sounding cough. Her primary care doctor initiated antibiotics for a presumed pneumonia. During one coughing episode, she turned blue and her father brought her to the ER. She was diagnosed with congestive heart failure.

The parents were informed by the cardiologists that the hospital "does not perform cardiac surgery on babies with trisomy 13 or 18." They recommended comfort care in the anticipation that Charlotte's heart failure would worsen and eventually cause her death. Charlotte's family was encouraged to meet the local palliative care team.

Charlotte's parents were in touch with other parents of children with trisomies on social media. They recognised that their daughter was "treated as a genetic diagnosis" rather than as an individual patient. In an online social media support group, they read of children with trisomy diagnoses who were operated at another hospital. They sent Charlotte's medical records to that hospital and arranged a transfer. She underwent ventricular septal defect closure and gastrostomy tube placement then remained in the neonatal ICU for 3 weeks.

Parent perspective

Our daughter has a special role in our family. She makes our home happy. During the pregnancy, we were told she would not interact with us at all. That is incorrect. She is quite responsive to our voices and our cuddles, and she is a calm happy baby with a happy life. For us, being good parents means fixing the hole in her heart because her life is a life worth living. We cannot just sit around and watch her die. At the same time, we do not want her to have to be hooked to machines long term or to live at a hospital. We want her heart to be fixed so we can return home to enjoy the time we have with her. It does not seem like her time to die yet for something that seems fixable. We were told that we did not need to meet with the heart surgeon. We were told that her genetic condition made her absolutely not a surgical candidate. There was no discussion about her development or about how she had beaten the odds. That hurts. It feels like we were told her life did not have worth. It feels like rejection of her and a rejection of our entire family.

Receiving interdisciplinary team perspective

The surgical decision is based on a thorough review of comorbidities, physiology, and psychosocial factors. A total of 31 children with trisomy 13 or 18 have undergone cardiac surgery at our centre since 2016 (Table 1). Of these, 27 (87%) survived to discharge and 23 (74%) are still alive at the time of paper submission. A consecutive summary of the cardiac diagnoses of 45 patients who were additionally referred in 2019 who did not undergo cardiac surgical intervention is provided in Table 2.

Our paediatric hospital involves the palliative care team in the initial assessment and longitudinal care of all children with trisomy 13 or 18, recognising goals of care and decision-making in these complex cases warrant an interdisciplinary team approach.[22] Our Trisomy Translational Care Team considers surgery for children with trisomy 13 or 18 after careful review of each child by an interdisciplinary team consisting of cardiology and cardiac surgery, neurology, otolaryngology, genetics, and other subspecialty members (Fig 1). A medical social worker, chaplain, and nurse case manager are assigned to each family during the medical review process to foster psychosocial assessments and support. We assess the child and family's ongoing complex care needs and connect families with community-based resources. We help the family identify a caring general paediatrician and a skilled nurse case manager who can care for the child and the family when they return home. In Charlotte's case, as in the case reported by the majority of other parents of children with these genetic conditions denied cardiac evaluation[18] that meant trying to repair the broken trust that they'd experienced with the referring hospital.[27]

Palliative physician perspective

A model of partnership and proactive communication had to be carried forward by the care teams across settings. Our intention was for the family to feel well-held across care locales, and we recognised this could only happen if the home team could be included in shared conversations together. Many teams too often rely on "routing discharge summary notes" via the electronic medical record as a main mode of communication between care settings. The "friendly" team are heroes, save the day and perform an increasing amount of surgeries, while the primary team stays "unfriendly" and are fled by families tagging them as "anti-trisomy" on social media. Re-establishing a dialogue is critical.[28]

In order to provide the best long-term care for Charlotte, we knew that we would have to rebuild trust with doctors at her referring hospital. We set up family meetings with both the home and the referral teams via tele-health. The goals of these meetings were to define parents' understanding of their role, exploring the parents' understanding of Charlotte's diagnosis and prognosis, fostering curiosity about spiritual growth, and managing symptoms while celebrating life milestones. The ultimate goal was to develop a care plan balancing medical intervention escalations with consideration of Charlotte's lived experience. A similar link can be created with any trusting physician team at a referring hospital.

Through use of televisits with the receiving care team, in these shared communication moments, the family experienced moving

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Table 1. Consecutive summary of operations and STAT mortality category

Primary diagnosis	Primary operation	STAT mortality category
DORV, hypoplastic aortic arch, MV dysplasia	DORV repair, aortic arch repair	4
DORV, MV dysplasia	DORV repair	1
VSD	VSD repair	1
DORV, hypoplastic aortic arch, aortic atresia	Yasui	5
VSD, hypoplastic aortic arch	VSD repair, aortic arch repair	3
AVSD, hypoplastic aortic arch	Bilateral PA band (hybrid stage 1)	4
VSD	VSD repair	1
DORV "tet type"	DORV repair, RVOT myectomy and transannular patch	4
DORV	DORV repair	4
DORV, PA	DORV repair, RV to PA connection (non-valved)	2
VSD, hypoplastic aortic arch	VSD repair, aortic arch repair	4
DORV	DORV repair	4
DORV	DORV repair	4
DORV	DORV repair	4
DORV, hypoplastic aortic arch	DORV repair, aortic arch repair	4
VSD	VSD repair	1
VSD	VSD repair	1
VSD	VSD repair	1
DORV, pulmonary atresia	DORV repair, pulmonary valvotomy	3
VSD, hypoplastic aortic arch	VSD repair, aortic arch repair	4
VSD	VSD repair	1
Multiple VSDs	Multiple VSD repair	3
VSD	VSD repair	1
DORV, PS	DORV repair	4
DORV, cor triatriatum, mitral Stenosis	Cor triatriatum repair, PA band placement	4
DORV "tet type"	DORV repair, RVOT myectomy	4
AVSD, hypoplastic aortic arch	PDA stent with RPA and LPA band	5
Multiple VSDs	Multiple VSD repair	2
DORV	DORV repair	4
PDA	PDA occlusion	2
PDA, tracheal compression	PDA ligation, arteriopexy	2

AVSD = atrioventricular septal defect; DORV = double outlet right ventricle; LPA = left pulmonary artery; MV = mitral valve; PA = pulmonary artery; PDA = patent ductus arteriosus; RPA = right pulmonary artery; RV = right ventricle; RVOT = right ventricular outflow tract.

Table 2. Consecutive summary of cardiac diagnoses for patients not undergoing surgery

Diagnosis	Number of referrals
MV Dysplasia	1
PDA	3
Muscular VSD	1
VSD	21
DORV	5
DORV, PS	3
VSD, CDH	1
AVSD	4
Pulmonary atresia, VSD	1
TGA, pulmonary atresia, VSD	1
Hypoplastic aortic arch, VSD	2
Hypoplastic aortic arch, DORV	1
HLH complex	1

HLH = hypoplastic left heart; TGA = transposition of the great arteries.
Many patients were excluded from cardiac intervention based on their comorbidities (not their cardiac diagnosis). Diagnosis stated reflect those of the referring facility. PDA was present in 21 referrals. Additional VSDs present in six referrals.

beyond a rejection narrative towards partnered perception. Communication between cardiology teams occurred by conference call for continuity in care. Our Trisomy Translational Team shared a summary of our surgical review process and surgical outcomes with Charlotte's home cardiology team for their consideration of future local cases. Charlotte's home surgical team received a phone call update, during which there was mutual regard for her positive outcome and shared recognition that children like Charlotte represent a changing clinical world. On the final day of Charlotte's discharge home, the palliative care team chaplain hosted a "travel benediction" with the intensive care staff and cardiology team members circling Charlotte's hospital bed. The home team joined the group by speakerphone in the blessing of the baby's return to her true home states away.

Conclusion

While this narrative represents one case, the principles extend to families of fragile children, seeking cardiac interventions for their loved child. The medical community is increasingly encountering situations in which interventions can both benefit and harm fragile children and families can opt in or refuse these innovations. When paediatric care centres categorically refuse to offer such interventions, or when they universally provide them, they may unintentionally undermine the practice of shared decision-making.[29] The care model for children with trisomy 13 and 18 is evolving, as families identify a potentially meaningful quality of life enhanced by coordinated, personalised medical care for the child.[18,26]

When a cardiac intervention is likely to correct an anomaly and send the patient home, the intervention can be viewed as beneficial, even in a fragile child with a serious condition. It was in Charlotte's case. She just celebrated her third birthday. Her follow-up cardiology appointment this month revealed that her future cardiac visits can be spaced annually as per her local cardiologist. She remains at home on room air, enjoying oral feeds with tube feed supplements,

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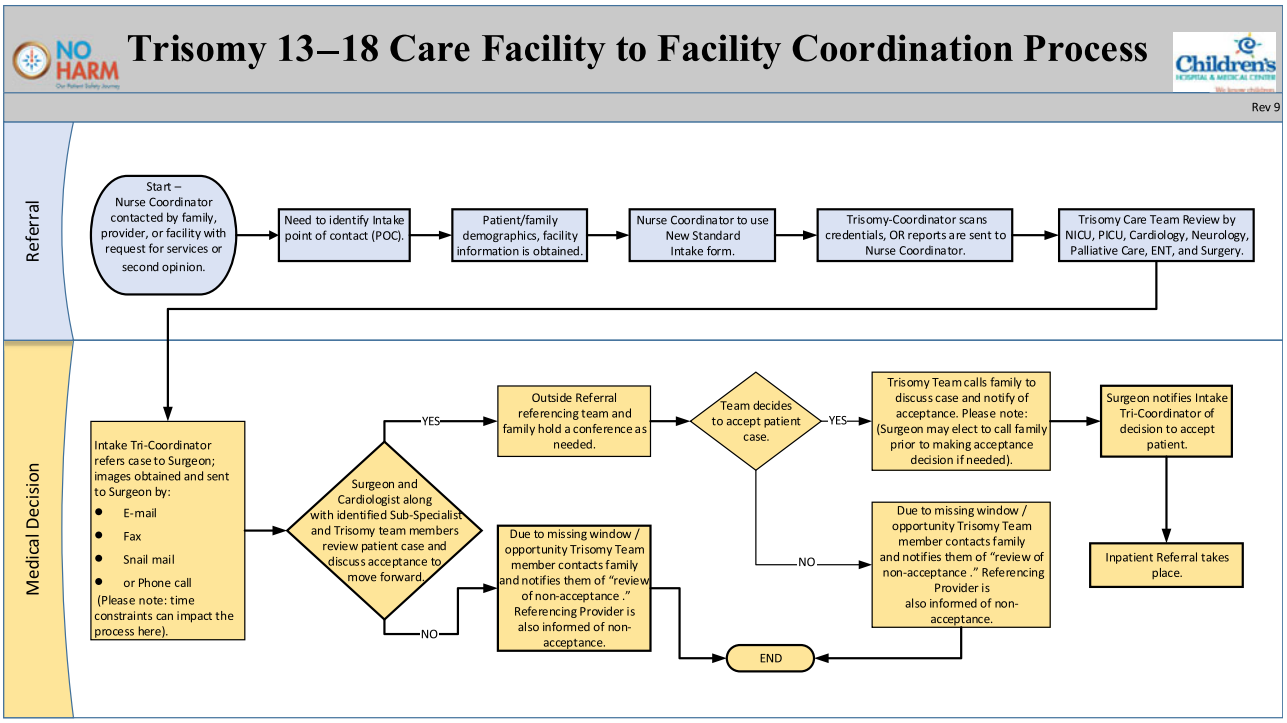

Figure 1. Trisomy 13 to 18 care facility to facility coordination process. ENT = ear, nose, and throat clinical team (otolaryngology); NICU = neonatal intensive care unit; PICU = pediatric intensive care unit.

and says words. She is taking steps in her walker. Her parents are dedicated and an inspiration. They drove 17 hours to attend the neonatal ICU reunion. Her cardiologist called our Trisomy Translational Team a few months after Charlotte's return home to share that their team had successfully completed the institution's first cardiac corrective surgery on a patient with trisomy 18. Babies like Charlotte make us humble and help us improve our care.

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Ethical standards. The authors assert that all procedures contributing to this work comply with the ethical standards.

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