Available online at www.sciencedirect.com

Seminars in Perinatology

www.seminperinat.com

Cardiac surgery for children with trisomies 13 and 18: Where are we now?

Annie Janvier, MD, PhD[a,b,c,*], *Barbara Farlow, BEng, MBA*[d,e], *and Keith Barrington, MD*[a,c]

[a]*Department of Pediatrics, University of Montreal, Montreal, Canada*
[b]*Unité d'éthique clinique, Hôpital Sainte-Justine, Quebec, Canada*
[c]*Research Center, Hôpital Sainte-Justine, Quebec, Canada*
[d]*The DeVeber Institute for Bioethics and Social Research, Ontario, Canada*
[e]*Patients for Patient Safety, Ontario, Canada*

ARTICLE INFO

Keywords:
Trisomy 13
Trisomy 18
Cardiac surgery
Congenital anomalies
Clinical ethics
Ethics
Decision-making
Life-sustaining interventions
End of life decisions
Palliative care
Family centered care
Communication
Empathy

ABSTRACT

The objective is to examine whether cardiac surgery should be considered for children with trisomy 13 or 18 (T13 or 18).T13 or 18 were previously referred to as "lethal" conditions due to high mortality rates and severe disability among survivors. In the last decade, investigations have revealed these conditions are heterogeneous, with increasing numbers of studies describing interventions for these children. A number of factors makes the interpretation of reported outcomes after cardiac surgery challenging: (1) dissimilarities in practice lead to a wide variation in reported outcomes after cardiac surgery; (2) cardiac surgery is generally offered to older, healthier children; (3) cardiac surgeries of widely varying risks are often lumped together in individual studies, and (4) cases where cardiac surgery has been withheld are generally not included in publications. It is unclear whether withholding cardiac surgery for some children with a ventricular septal defect will lead to death, or the development of pulmonary hypertension, or if death will occur from other causes. In this article, we describe two children with different clinical situations and examine whether cardiac surgery would benefit them and how to communicate with their families. Cardiac surgery may be beneficial to some children with trisomy 13 or 18, but may harm others. Every child should be approached in an individual fashion and the goals of each family should be addressed. Children who are more likely to benefit from surgery may be older, healthier children without respiratory support. Rigorous and transparent research is needed to identify factors that affect survival in trisomy 13 or 18.

Kayla

Kayla was born at 38 weeks of gestational age with a birth weight of 3150 g. She is the third daughter of Keith and Sally Orion. Sally had a prenatal ultrasound in a small rural medical center, where the results were considered "within the limits of normal." Kayla was born with 12 toes and dysmorphic features. Because of a weak suck, she receives Sally's breast milk mostly through a nasogastric tube. She was transferred to a large pediatric center where a moderate

Abbreviations: T13, trisomy 13; T18, trisomy 18; HLHS, Hypoplastic Left Heart Syndrome; VSD, Ventricular Septal Defect

*Corresponding author at: Department of Pediatrics and Clinical Ethics, University of Montreal, Neonatologist and Clinical Ethicist, Sainte-Justine Hospital, 3175 Chemin Côte-Sainte-Catherine, Montreal (QC), Canada H3T 1C5.
E-mail address: anniejanvier@hotmail.com (A. Janvier).

http://dx.doi.org/10.1053/j.semperi.2015.12.015
0146-0005/

ventricular septal defect (VSD) was found. A diagnosis of trisomy 13 was confirmed 3 days after birth. At 5 days of life, Sally is stable and will be discharged home as soon as her parents are comfortable feeding her.

Daniel

During their first pregnancy, Simone and Gregory Alterbach were told the first trimester screen was abnormal. The diagnosis of trisomy 18 was made at 15 weeks, after an amniocentesis. The couple decided to continue the pregnancy, and named their unborn son Daniel. The bad news continued coming with every appointment: Daniel had esophageal atresia, clubfeet and the latest heart ultrasound revealed a severe form of hypoplastic left heart syndrome (HLHS). Now at 31 weeks of gestation, Daniel only weighs 900 g, showing no fetal growth in the past 2 weeks.

Introduction

Trisomy 13 and trisomy 18 are the second and third most common aneuploidy, after trisomy 21. These syndromes are frequently referred to as being "lethal" because of their poor outcomes.[1] The risk of spontaneous abortion and in utero death during a pregnancy is high.[2] Children born with trisomies 13 and 18 have low survival rates: the majority of articles report a 1-year survival of around 10%.[2–4] Survivors have serious neurodevelopmental disabilities.[3–7] Some children do not have "full" trisomy 13 or 18, but instead have one of many variants of these chromosomal conditions, with outcomes that may be much less severe.[8,9] Because of these adverse outcomes, a tradition of withholding and withdrawing of interventions has existed for trisomies 13 and 18[10,11]; but over the past decade, investigations have shed a different light on these conditions. A 12-year review of hospitalizations for American children with trisomy 13 or 18 revealed that an unexpected number of children of all ages are receiving interventions, with many survivors, leading authors to conclude that "universal application of the term 'lethal' to the diagnoses of trisomies 13 and 18 is not appropriate."[12] In Japan, the approach to cardiac interventions for children with these conditions has been different for many years, and the 1-year survival rates have been reported to be as high as 75%.[13–15]

We have investigated the experience of 332 parents who live(d) with children who have (had) trisomy 13 or 18.[16,17] In contrast to the experience providers often predicted, parents in this study described their experience as positive, irrespective of longevity. Parents acknowledged the many challenges of caring for children with special needs, but overwhelmingly described them as happy people who enriched their families.[16,17] As a result of these publications, we are frequently asked for our opinion regarding interventions for children living with these conditions. Decisions regarding whether to surgically intervene or when to initiate, withhold or withdraw life-sustaining interventions in vulnerable patients are among the most difficult decisions in medicine. The interpretation of risks, benefits, and outcomes are value dependant. In the remainder of this article, we will examine the literature pertaining to cardiac surgery for children with trisomies 13 and 18, analyze both the cases presented and offer recommendations on how to communicate with parents in these situations.

Cardiac surgery for children with trisomy 13 or 18: Which statistic should we use?

What kind of survival statistics should we use to analyze the case of Kayla and Daniel in order to decide what is in the best interest of these children and optimize communication with their parents? Good decision-making starts with good facts, but good facts are often hard to find when trisomies 13 and 18 are concerned. Policy statements and the opinions of the medical team will have a direct impact on the information parents receive, which may influence the survival of their children. Policy statements in North America and Europe usually recommend against life prolonging interventions for these conditions.[18,19] On the other hand, other countries have a more interventionist approach to these conditions. For example, in Japan, medical interventions, including cardiac surgery, are not rare. This important dissimilarity in practice leads to a wide variation in reported outcomes. While the majority of population studies report survival rates beyond 1 year of 10% or less for T13 or 18[3,6,7]; Japanese publications reveal 1-year survival rates as high as 75% when surgery is provided to select infants for whom surgery was deemed to be a reasonable option.[13] These variations in outcomes do not reflect a difference in financial incentives, available resources, technology, or surgical approaches. Rather, this outcome variation reflects a difference in values. Institutions that value consideration of parental goals and engage in shared decision-making are likely to appreciate the spectrum of trisomies 13 and 18, evaluate children in an individual fashion, and offer interventions when they are deemed appropriate; these values have a direct impact on survival statistics. When examining statistics to make decisions for a child with trisomy 13 or 18, we have to consider that statistics might reflect not only survival of the genetic condition, but also the ethos of healthcare providers. Until very recently, these children routinely received comfort care and generally died shortly after birth. In fact, trisomies 13 and 18 are the only conditions where neonatal survival has overall decreased, despite improving neonatal outcomes. For example, an American population study[6] reported a significant decline in 1-month survival of trisomy 13 newborns: 1-month survival of 47% in the 1980s (from 40% in the 1970s) plummeted to 17% in 1990–1999. This may be due to an increase in prenatal diagnosis, which then led to an increase in the proportion of children who had interventions withheld at birth.

Several articles describe the outcomes of children with T13 or 18 after cardiac intervention.[13–15,20–22] When surgery is offered to children whom authors describe are deemed to have a reasonable chance of survival and discharge, the rates of survival to discharge and survival beyond 1 year can be significant (Table). As can be seen in the Table, many children, with different cardiac malformations, weights and associated anomalies are often lumped together and become the denominator (number of cardiac surgeries). This may not