CHD Repair in Trisomy 13 or 18

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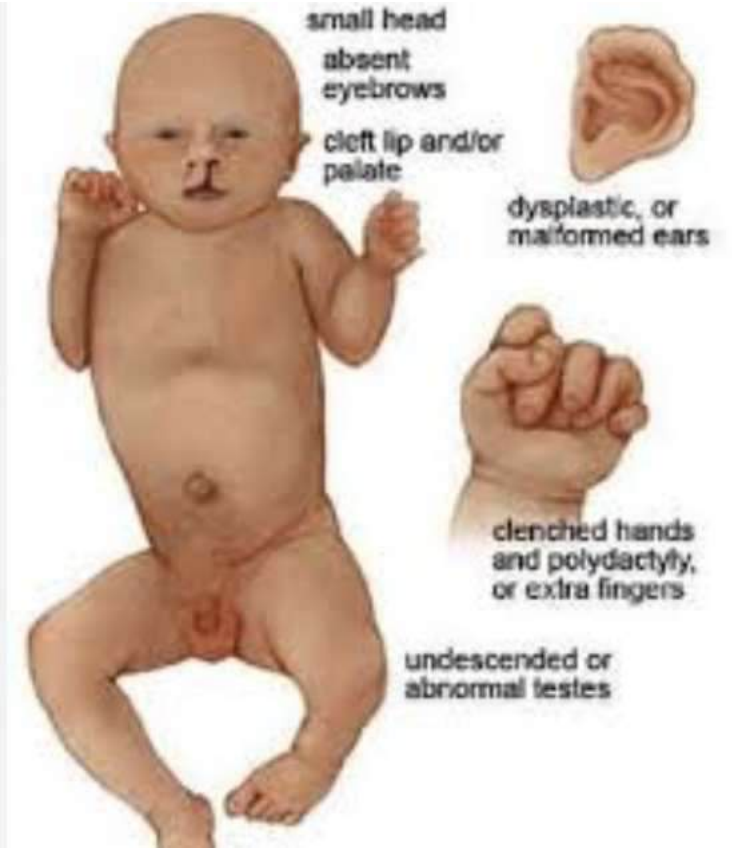
No Disclosures

Trisomy 18: Edwards syndrome

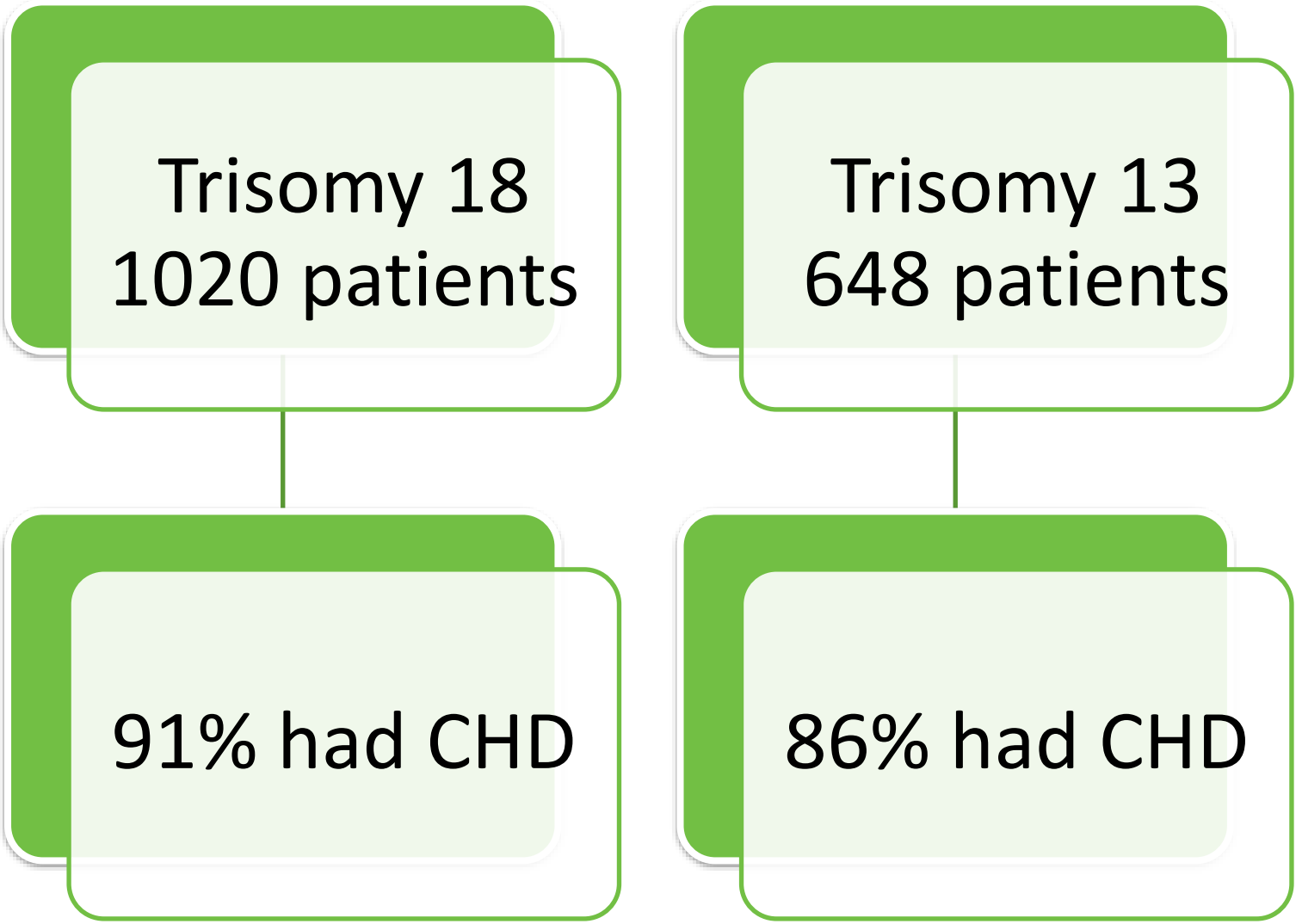
- 1 in 2500 pregnancies
- 1 in 5000-6000 live births
- Types of trisomy
  - Full
  - Partial
  - Mosaic

Trisomy 13: Patau Syndrome



- 1 in 10,000 to 20,000 live births
- 5-10% survival after 1 year
- Types of trisomy
  - Full
  - Partial
  - Mosaic

Incidence of CHD in Trisomy 18 and 13



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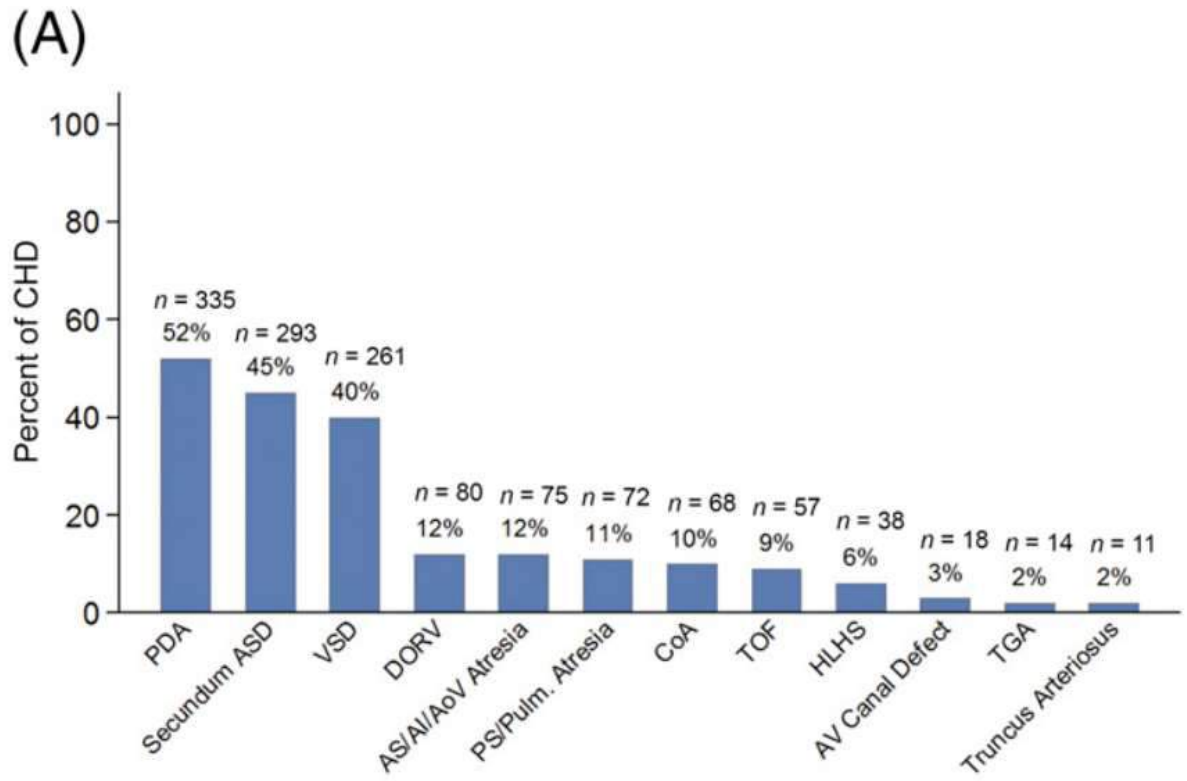
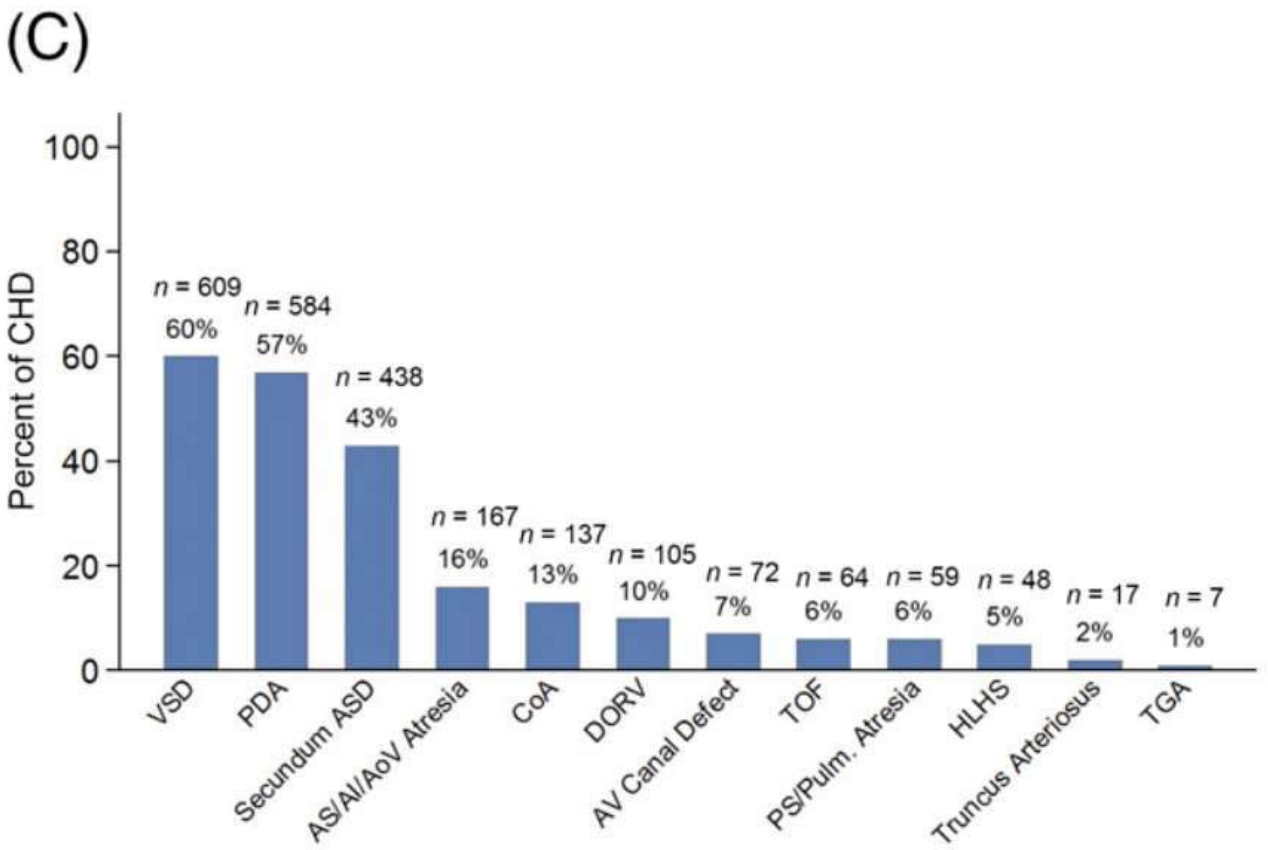
Trisomy 13


(A)
Percent of CHD
100
80
60
40
20
0
n = 335
52%
n = 293
45%
n = 261
40%
n = 80
12%
n = 75
12%
n = 72
11%
n = 68
10%
n = 57
9%
n = 38
6%
n = 18
3%
n = 14
2%
n = 11
2%
PDA
Secundum ASD
VSD
DORV
AS/AI/AoV Atresia
PS/Pulm. Atresia
CoA
TOF
HLHS
AV Canal Defect
TGA
Truncus Arteriosus

Trisomy 18


(C)
Percent of CHD
100
80
60
40
20
0
n = 609
60%
n = 584
57%
n = 438
43%
n = 167
16%
n = 137
13%
n = 105
10%
n = 72
7%
n = 64
6%
n = 59
6%
n = 48
5%
n = 17
2%
n = 7
1%
VSD
PDA
Secundum ASD
AS/AI/AoV Atresia
CoA
DORV
AV Canal Defect
TOF
PS/Pulm. Atresia
HLHS
Truncus Arteriosus
TGA

Trisomy 13 and 18 prevalence and mortality

Abstract

The aim of the study is to determine the prevalence, outcomes, and survival (among live births [LB]), in pregnancies diagnosed with trisomy 13 (T13) and 18 (T18), by con-genital anomaly register and region. Twenty-four population- and hospital-based birth defects surveillance registers from 18 countries, contributed data on T13 and T18 between 1974 and 2014 using a common data-reporting protocol. The mean total birth prevalence (i.e., LB, stillbirths, and elective termination of pregnancy for fetal anomalies [ETOPFA]) in the registers with ETOPFA ($n = 15$) for T13 was 1.68 (95% CI1.3–2.06), and for T18 was 4.08 (95% CI 3.01–5.15), per 10,000 births. The prevalence varied among the various registers. The mean prevalence among LB in all registers for T13 was 0.55 (95%CI 0.38–0.72), and for T18 was 1.07 (95% CI 0.77–1.38), per 10,000 births. The median mortality in the first week of life was 48% for T13 and 42% for T18, across all registers, half of which occurred on the first day of life. Across 16 registers with complete 1-year follow-up, mortality in first year of life was 87% for T13 and 88% for T18. This study provides an international perspective on prevalence and mortality of T13 and T18. Overall outcomes and survival among LB were poor with about half of live born infants not surviving first week of life; nevertheless about 10% survived the first year of life. Prevalence and outcomes varied by country and termination policies. The study highlights the variation in screening, data collection, and reporting practices for these conditions.

Trisomy 13 and 18—Prevalence and mortality—A multi-registry population based analysis

Nitin Goel[1,2], Joan K. Morris[3], David Tucker[2], Hermien E. K. de Walle[4], Marian K. Bakker[4], Vijaya Kancherla[5], Lisa Marengo[6], Mark A. Canfield[6], Karin Kallen[7], Nathalie Lelong[8], Jorge L. Camelo[9], Erin B. Stallings[10,11], Abbey M. Jones[10], Amy Nance[12], My-Phuong Huynh[12], Maria-Luisa Martínez-Fernández[13], Antonin Sipek[14], Anna Pierini[15], Wendy N. Nembhard[16], Dorit Goetz[17], Anke Rissmann[17], Boris Groisman[18], Leonora Luna-Muñoz[19], Elena Szabova[20], Serhiy Lapchenko[21], Ignacio Zarante[22], Paula Hurtado-Villa[23], Laura E. Martinez[24], Giovanna Tagliabue[25], Danielle Landau[26], Miriam Gatt[27], Saeed Dastgiri[28], Margery Morgan[2]

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Birth prevalence for T13 was 1.6 per 10000 births and 4 for T18

Prevalence and Mortality

Median mortality in first week of life for T13 48%

Median mortality in first week of life for T18 42%

One year follow up

- Trisomy 13: 87% mortality
- Trisomy 18: 88% mortality

One-year median survival of 80% for mosaic T13, 70% for mosaic T18

Traditional approach

- Not offered surgery
- Survival in first few months low, unrelated to cardiac surgery

Data on impact of cardiac surgery

Survival of children with trisomy 18 associated with the presence of congenital heart disease and intervention in the Republic of Korea

In Gyu Song[1], Seung Han Shin[2*], Yoon-Min Cho[3] and Youna Lim[4]

Abstract

Background Trisomy 18 syndrome (T18) is the second most common autosomal trisomy and has a high risk of fetal loss and stillbirth. Aggressive surgical treatments for the respiratory, cardiac, or digestive systems of patients with T18 were previously futile, while the results of recent studies are controversial. Over the past decade, there have been approximately 300,000 to 400,000 births annually in the Republic of Korea; however, there have been no nationwide studies on T18. This nationwide retrospective cohort study aimed to determine the prevalence of T18 in Korea and its prognosis according to the presence of congenital heart disease and relevant interventions.

Methods This study utilized NHIS-registered data between 2008 and 2017. A child was defined as having T18 if the ICD-10 revision code Q91.0–3 was reported. Subgroup analysis was performed for children with congenital heart diseases, and survival rates were compared based on the history of cardiac surgical or catheter interventions. The primary outcomes in this study were the survival rate during the first hospitalization period and the 1-year survival

The survival rate in the first admission of children with T18 who did and did not have congenital heart disease was 58 % ad 94%

, with a median
admission of
y. Children with
ho did not.

Conclusions We suggest these data could be used in ante- and postnatal counseling. Ethical concerns about the prolonged survival of children with T18 remain; however, the potential benefits of interventions for congenital heart disease in this population need further study.

Effectiveness of cardiac surgery in patients with trisomy 18: a single-institutional experience

Yosuke Nakai [1], Miki Asano [1], Norikazu Nomura [1], Hidekazu Matsumae [1], Akira Mishima [1]

Affiliations + expand

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Abstract

Background: Surgical repair for cardiac lesions has rarely been offered to patients with trisomy 18 because of their very short lifespans. We investigated the effectiveness of cardiac surgery in patients with trisomy 18. Patients and methods We performed a retrospective analysis of 20 consecutive patients with trisomy 18 and congenital cardiac anomalies who were evaluated between August, 2003 and July, 2013. All patients developed respiratory or cardiac failure due to excessive pulmonary blood flow. Patients were divided into two subgroups: one treated surgically (surgical group, n=10) and one treated without surgery (conservative group, n=10), primarily to compare the duration of survival between the groups.

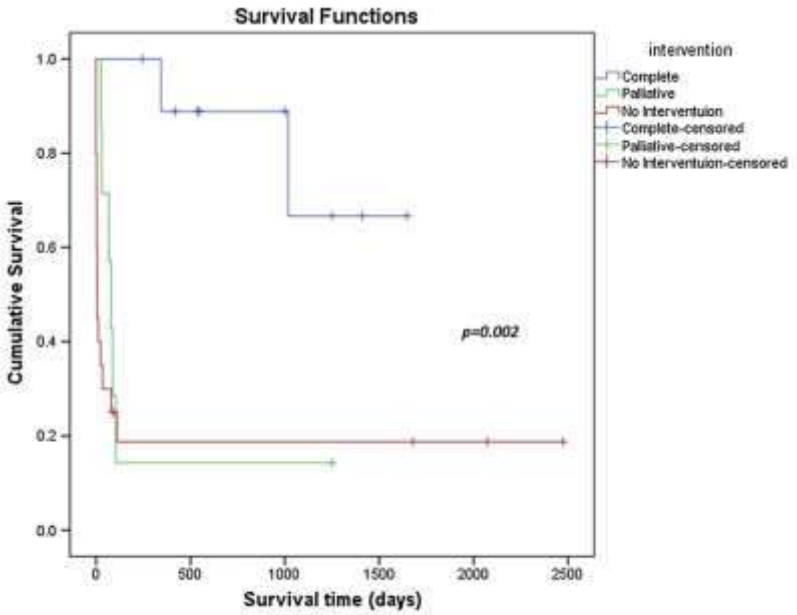
Results: All the patients in the surgical group underwent cardiac surgery with pulmonary artery banding, including patent ductus arteriosus ligation in nine patients and coarctation repair in one. The duration of survival was significantly longer in the surgical group than in the conservative group (495.4±512.6 versus 93.1±76.2 days, respectively; p=0.03). A Cox proportional hazard model found cardiac surgery to be a significant predictor of survival time (risk ratio of 0.12, 95% confidence interval 0.016-0.63; p=0.01).

Conclusions: Cardiac surgery was effective in prolonging survival by managing high pulmonary blood flow; however, the indication for surgery should be carefully considered on a case-by-case basis, because the risk of sudden death remains even after surgery. Patients' families should be provided with sufficient information to make decisions that will optimise the quality of life for both patients and their families.

Factors Influencing Outcomes After Cardiac Intervention in Infants with Trisomy 13 and 18

Renuka Peterson[1] · Nandini Calamur[1] · Andrew Fiore[2] · Charles Huddleston[2] · Kimberly Spence[1]

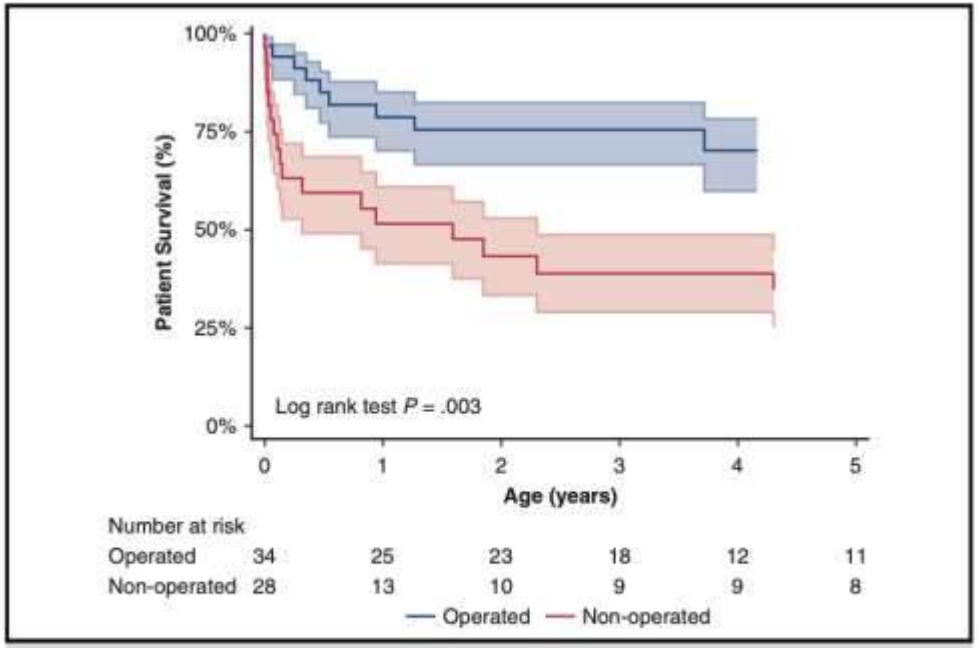
- All palliated patients dead by 813 days
- All complete repair survived early, only 57% of palliated
- Age, weight, home before intervention in complete repair group
- Half needed home oxygen, quarter needed pulmonary vasodilator
- No survivor in PGE dependent group
- Palliated and non intervention group same survival



Operative and nonoperative outcomes in patients with trisomy 13 and 18 with congenital heart disease

Christina L. Greene, MD,[a] Antonia Schulz, MD,[a] Mariana Chávez, MD,[a] Steven J. Staffa, MS,[b] David Zurakowski, MS, PhD,[b] Kevin G. Friedman, MD,[c] Sitaram M. Emani, MD,[a] and Christopher W. Baird, MD[a]

- Almost 40 years of data from 1985 to 2023
- 62 patients (34 operated and 28 non operated)
- Median age of 2.5 months
- Non operative cohort - 65% were stat 1
- 5 in hospital deaths for operated and 7 in non operated group



Survival in patients undergoing operation was significantly higher than those who did not.

PERSPECTIVE

Primary cardiac repair in patients with T13/T18 with CHD is controversial. Here we demonstrate it is safe and effective when standard operative criteria are followed and can greatly improve survival. Despite a high burden of morbidities, most families are satisfied with their child's quality of life. We advocate for early referral for primary cardiac repair in patients with T13/T18 with CHD.

Long term outcome

Jennifer K. Peterson, MS[1], **Lazaros K. Kochilas, MD, MSCR**[2], **Kirsti G. Catton, MSN**[1], **James H. Moller, MD**[3], and **Shaun P. Setty, MD**[1]

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Abstract

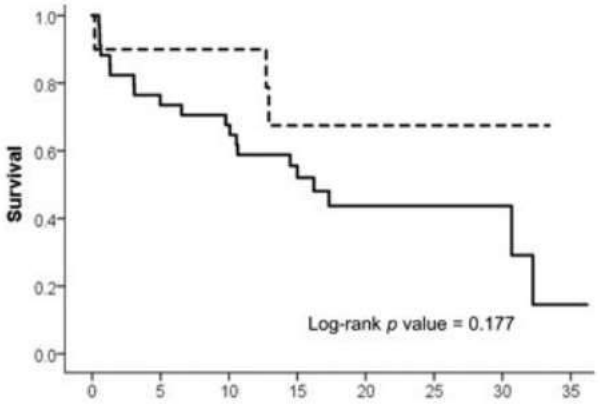
Background—The purpose of this study is to report short and long term outcomes following congenital heart defect (CHD) interventions in patients with Trisomy 13 or 18.

Methods—A retrospective review of the Pediatric Cardiac Care Consortium (PCCC) identified children with Trisomy 13 or 18 with interventions for CHD between 1982 and 2008. Long term survival and cause of death was obtained through linkage with the National Death Index.

Results—A total of 50 patients with Trisomy 13 and 121 with Trisomy 18 were enrolled in PCCC between 1982 and 2008; among them 29 patients with Trisomy 13 and 69 patients with Trisomy 18 underwent intervention for CHD. In-hospital mortality for patients with Trisomy 13 or Trisomy 18 was 27.6% and 13%, respectively. Causes of in-hospital mortality were primarily cardiac (64.7%) or multiple organ system failure (17.6%). National Death Index linkage confirmed 23 post-discharge deaths. Median survival (conditioned to hospital discharge) was 14.8 years (95% CI 12.3–25.6 years) for patients with Trisomy 13 and 16.2 years (95% CI 12–20.4 years) for patients with Trisomy 18. Causes of late death included cardiac (43.5%), respiratory (26.1%), and pulmonary hypertension (13%).

Conclusions—In-hospital mortality for all surgical risk categories was higher in patients with Trisomy 13 or 18 than reported for the general population. However, patients with Trisomy 13 or 18, who were selected as acceptable candidates for cardiac intervention and who survived CHD intervention, demonstrated longer survival than previously reported. These findings can be used to counsel families and make program-level decisions on offering intervention to carefully selected patients.

Post discharge survival in intervention group



Trisomy 13: Median survival = 14.8 years (95% CI 12.3-25.6 years)

Trisomy 18: Median survival = 16.2 years (95% CI 12-20.4 years)

Patients at Risk

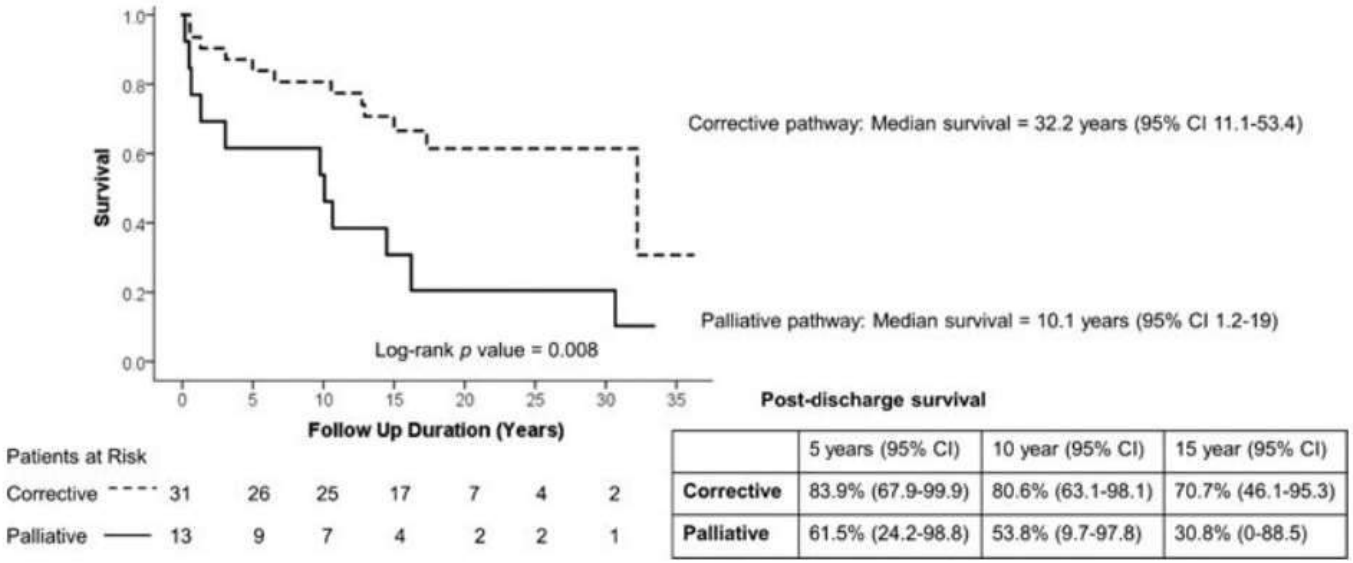
Trisomy 13	10	9	9	5	2	2	2
Trisomy 18	34	25	23	17	7	3	3

Post-discharge survival

	5 year (95% CI)	10 year (95% CI)	15 year (95% CI)
Trisomy 13	90% (64.9-100)	90% (64.9-100)	67.5% (16.4-100)
Trisomy 18	73.5% (54.2-92.8)	67.6% (46.3-88.9)	55.6% (29-82.2)

Figure 1.
Kaplan-Meier conditioned survival following intervention for CHD in Trisomy 13 and 18 patients.

Post discharge survival for corrective versus palliative group

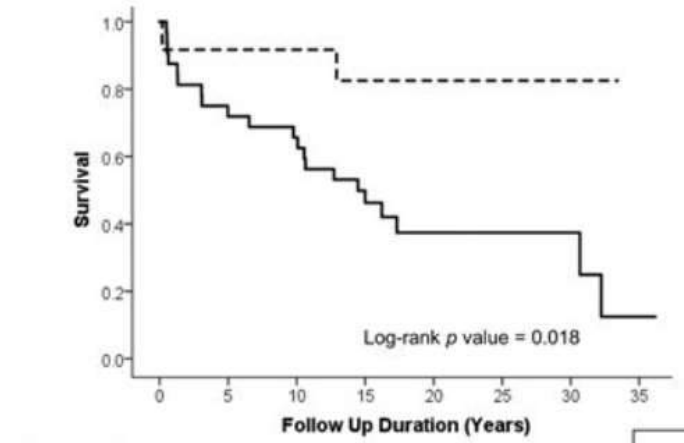


Post-discharge survival

	5 years (95% CI)	10 year (95% CI)	15 year (95% CI)
Corrective	83.9% (67.9-99.9)	80.6% (63.1-98.1)	70.7% (46.1-95.3)
Palliative	61.5% (24.2-98.8)	53.8% (9.7-97.8)	30.8% (0-88.5)

Figure 2.
Kaplan-Meier conditioned survival following intervention for CHD in Trisomy 13 and 18 patients by treatment pathway.

Post discharge survival for mosaics versus non mosaics



Mosaic: Median survival = 16.7 years (95% CI 12.9-23.1 years)

Not mosaic: Median survival = 14.5 years (95% CI 7.7-21.3 years)

Post Discharge Survival

	5 year (95% CI)	10 year (95% CI)	15 year (95% CI)
Mosaic	91.7% (70.9-100)	91.7% (70.9-100)	82.5% (47.2-100)
Not mosaic	71.9% (51.3-92.4)	65.6% (42.9-88.3)	49.8% (22.2-77.4)

Figure 3.
Kaplan-Meier conditioned survival following intervention for congenital heart disease in mosaic and not mosaic Trisomy 13 and 18 patients.

Dilemmas

- Dissimilarities in practice leading to wide variation in reported outcomes
- Surgery is generally offered to older and healthier children
- Cardiac surgery of widely varying risks are often lumped together in individual studies
- Cases where cardiac surgery has been withheld are generally not included in publications

Cardiac Surgery Outcomes in Trisomy 13 and 18 *Cooper et al*

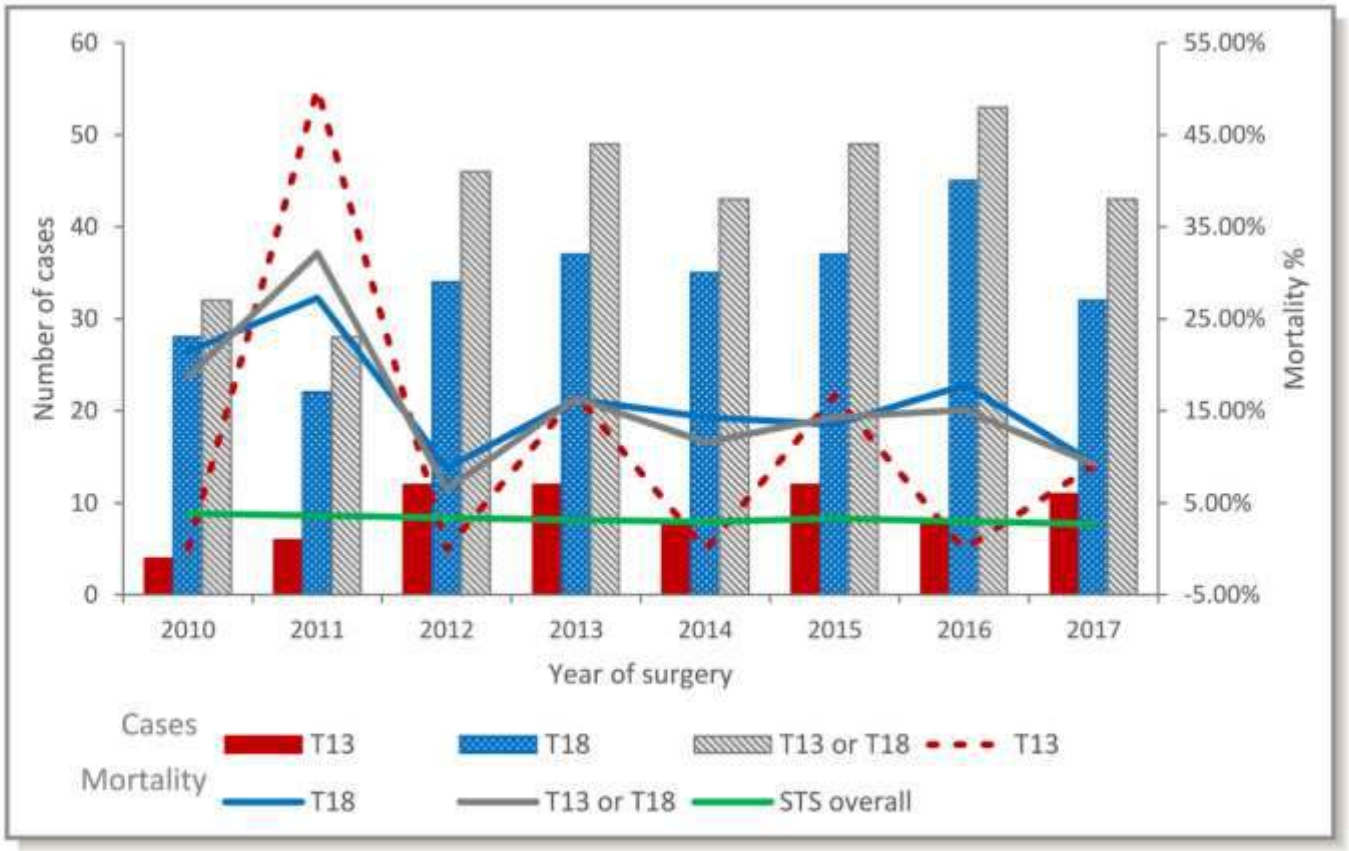


Figure. Annual case volume and mortality rate by aneuploidy diagnosis. National annual Trisomy 13 (T13) and Trisomy 18 (T18) surgical case volume and mortality rate from 2010 to 2017. The overall Society of Thoracic Surgeons (STS) mortality rate is displayed in green for visual comparison.

Table 2. Regional Distribution of Operations

Region	Index Cardiovascular Operations, No. (%)	T13 and T18 Operations, No. (%)	T13 Operations, No. (%)	T18 Operations, No. (%)
Northeast	30 260 (15.2)	49 (14.3)	7 (9.6)	42 (15.6)
Midwest	46 782 (23.6%)	88 (25.7)	14 (19.2)	74 (27.4)
South	73 409 (37.0)	142 (41.4)	36 (49.3)	106 (39.3)
West	41 727 (21.0)	59 (17.2)	14 (19.2)	45 (16.7)
Canada	6351 (3.2)	5 (1.5)	2 (2.7)	3 (1.1)

Number of operations performed by the North American Society of Thoracic Surgeons geographic region and Trisomy diagnosis. T13 indicates Trisomy 13; T18, Trisomy 18.

Table 1. Number of Operations Per Center Over the 8-Year Study Period

Total Cases, No.	Sites, No. (%)
1	31 (35.6)
2	14 (16.1)
3	11 (12.6)
4	5 (5.7)
5	6 (6.9)
6	1 (1.1)
7	5 (6.9)
8	3 (3.4)
9	2 (2.3)
10+	9 (10.3)

Number of heart surgery cases being performed in patients with Trisomy 13 and 18 per site over the study period.

Table 6. Preoperative Factors Associated With Operative Mortality

	T13 (n=73)				T18 (n=270)			
	Survivor (n=65)	Mortality (n=8)	*P* Value	Unadjusted OR	Survivor (n=228)	Mortality (n=42)	*P* Value	Unadjusted OR
Mechanical ventilation	11 (17)	5 (63)	0.01	8.2	52 (23)	30 (71)	<0.0001	8.5
Gastrostomy tube	16 (25)	2 (25)	1	1.0	57 (25)	4 (10)	0.027	0.3
Prior cardiothoracic surgery	15 (23)	1 (13)	0.68	0.5	42 (18)	2 (5)	0.024	0.2

Table 4. Surgical Outcomes

	T13	*P* Value to Overall	T18	*P* Value to Overall	T13 or T18	Overall STS Data
	n=73	STS Data	n=270	STS Data	n=343	N=198 185
In-hospital mortality	8/73 (11)		42/270 (16)		50/343 (15)	3.2
STAT 1	0/25 (0)	0.747	11/113 (10)	<0.001	11/138 (8)	0.5
STAT 2	4/14 (29)	<0.001	12/69 (17)	<0.001	16/83 (19)	2.2
STAT 3	0/8 (0)	0.655	2/12 (17)	0.024	2/20 (10)	2.4
STAT 4	4/24 (17)	0.123	14/67 (21)	<0.001	18/91 (20)	6.7
STAT 5	0/2 (0)	0.545	3/5 (60)	0.033	3/7 (43)	15.4
Unknown STAT level	0/0 (N/A)		0/4 (0)		0/4 (0)	N/A
Postoperative LOS, d	13 (6–46)		18 (8–46)		16 (7–46)	7 (4–15)
STAT 1	8 (5–14)		14 (6–34)		13 (6–31)	4 (3–6)
STAT 2	15 (6–46)		12 (6–29)		13 (6–32)	7 (4–15)
STAT 3	14 (8–38)		35 (19–77)		28 (8–66)	8 (5–15)
STAT 4	27 (9–65)		31 (14–65)		31 (13–65)	13 (7–28)
STAT 5	50 (45–55)		51 (36–61)		51 (36–61)	28 (16–51)

Overall take away

- Surgeries on T13 and T18 is performed by most centers across the country
- Operative mortality rate was 15%
- Complication rate was 56%
- Preoperative mechanical ventilation was associated with odds ratio of mortality of >8 for both T13 nad T18 patients
- G tube was associated with better survival in T18 patients, not T 13

Clinical Perspective

What Is New?

- Analysis of the Society of Thoracic Surgeons Congenital Heart Surgery Database provides a national representation of surgery in this population and captures granular data regarding diagnoses and operations performed in patients with Trisomy 13 and Trisomy 18.
- Operative mortality when compared with patients without Trisomy 13 and 18 was high across all surgical complexity categories.
- Preopeative mechanical ventilation is associated with postoperative mortality.

What Are the Clinical Implications?

- Surgical intervention carries a high risk in patients with Trisomy 13 and 18, even those undergoing low complexity Society of Thoracic Surgeons/European Association for Cardiothoracic Surgery Congenital Heart Surgery Mortality category operations.
- Surgical candidacy of patients with Trisomy 13 and 18 who require preoperative mechanical ventilation should be carefully considered.

The American Association for Thoracic Surgery (AATS) 2023 Expert Consensus Document: Recommendation for the care of children with trisomy 13 or trisomy congenital heart defect

James D. St Louis, MD,[a] Aarti Bhat, MD,[b] John C. Carey, MD,[c] Angela E
Laura Miller Smith, MD,[e] Benjamin S. Wilfond, MD,[b] Katherine A. Kosi
Robert A. Sorabella, MD,[g] and Bahaaldin Alsoufi, MD[b]

TABLE 1. Case complexity designations

Mild complexity congenital heart disease
Atrial septal defects
Patent foramen ovale
Persistent ductus arteriosus
Pulmonary valve stenosis
Moderate complexity congenital heart disease
Coarctation of the aorta
Atrioventricular septal defect
Tetralogy of Fallot
Total anomalous pulmonary venous connection
Ventricular septal defect
Sinus venosus atrial septal defect
Ebstein anomaly
Severe complexity congenital heart disease
Functional single ventricle anatomy
Hypoplastic left Heart Syndrome
Tricuspid atresia
Mitral atresia
Double outlet right ventricle
Transposition of the great arteries
Pulmonary atresia with ventricular septal defects
Truncus arteriosus
Pulmonary atresia with Intact ventricular septum

2.1 Recommendation: Mild-to-Moderate Complexity CHD

	COR	LOE
In infants with T13 or T18 and CHD of mild or moderate complexity, who are confined to the hospital or neonatal intensive care unit, cardiac surgery is *reasonable* if the hemodynamic consequence of the CHD prohibits discharge.	IIa	C-LD

2.2 Recommendation: Severe Complexity/ Functional Single Ventricle CHD

	COR	LOE
In infants with T13 or T18 and the diagnosis of CHD of severe complexity or functional single ventricle anatomy, it *might be reasonable* NOT to offer cardiac surgery based on anticipated early and late risks (demographic, physiologic, and clinical).	IIb	C-LD

3.1 Recommendation: Early Elective Repair

	COR	LOE
In infants and children with T13/T18 and CHD of mild or moderate complexity, who are at home or need recurrent hospital admissions, timely elective cardiac surgical repair is *reasonable* to decrease late cardiac morbidity mortality and to improve quality of life	IIa	C-EO

4.1 Recommendation: Extracardiac Defects

	COR	LOE
While associated extracardiac defects such as esophageal atresia and abdominal wall defects are associated with higher morbidity and mortality in infants with T13/T18, repair of CHD of mild-to-moderate complexity *may be considered* once these other defects are adequately addressed.	IIb	C-LD

4.2 Recommendation: Ventilator Dependence and CHD Surgery

	COR	LOE
In infants with T13 and T18 who are ventilator-dependent (with or without tracheostomy) due to central apnea, airway, or lung disease, surgical repair of a congenital heart defect of mild-or-moderate complexity *may be considered*, if the severity of the respiratory disease is not prohibitive. An increased likelihood for long-term tracheostomy and higher mortality risks for these patients should be clearly explained to parents.	IIb	C-LD

5. PALLIATIVE VERSUS DEFINITIVE REPAIR

5.1 Recommendation

	COR	LOE
In infants and children with T13 or T18, surgical repair of CHD of mild-to-moderate complexity is associated with better outcomes than palliation; however, initial palliation (eg, pulmonary artery banding) *is recommended* in those with significant associated morbidity.	I	C-LD

6.1 Recommendation

	COR	LOE
Infants and children with T13 or T18 are prone to develop early pulmonary hypertension due to an intrinsic arteriopathy, among other reasons. Therefore, early cardiac surgery *is reasonable* for mild-to-moderate complex congenital heart defects.	IIa	B-NR

7.1 Recommendation: Multidisciplinary Care Team

	COR	LOE
In the management of children with T13 or T18, *multidisciplinary teams* including palliative care, *are recommended* as a component of an overall comprehensive care plan to enhance decision making for families considering cardiac surgery.	I	C-LD

7.2 Recommendation: Holistic Care Plan

	COR	LOE
Parents carrying a suspected or confirmed fetus with T13 or T18 and the diagnosis of a congenital heart defect *should* participate in the development of a holistic care plan for their child with input from maternal fetal medicine, neonatology, genetics, pediatric cardiology/ cardiac surgery, and other necessary subspecialties involved in the future care of their child. This plan should reflect the parental goals for the pregnancy.	**I**	**C-LD**

PERSPECTIVE

Expert consensus documents are critical to the practice of medicine when peer-reviewed data are insufficient to create clinical practice guidelines. The management of infants with trisomy 13 or trisomy 18 who have an associated congenital heart defect lacks consistency. The variability in practice leads to discontent and distrust among parents and clinicians and to potentially suboptimal patient care.

Cardiac Surgery in Trisomy 13 and 18: A Guide to Clinical Decision-Making

Horacio G. Carvajal[1] · Connor P. Callahan[2] · Jacob R. Miller[1] · Bethany L. Rensink[1] · Pirooz Eghtesady[1]

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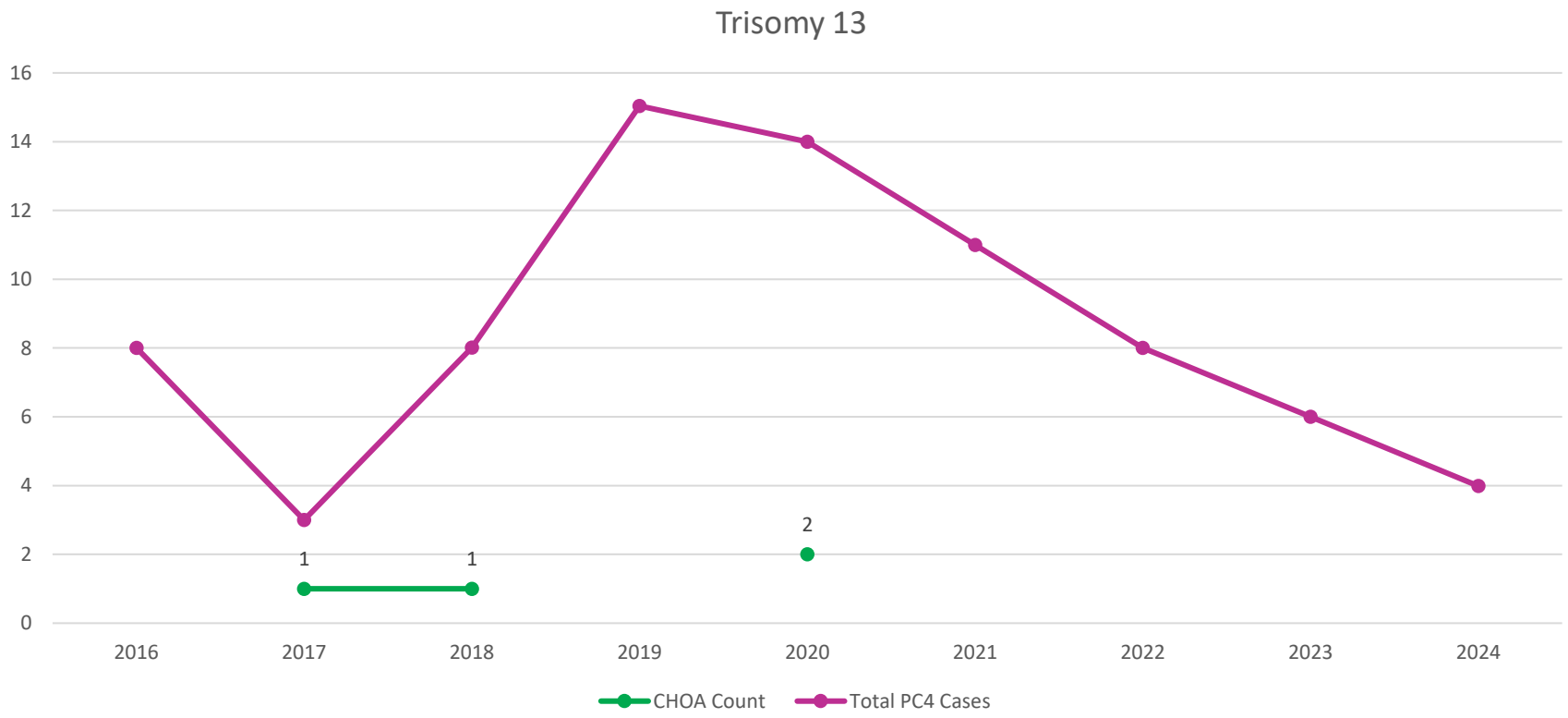
Recommendations

- Improved prognosis
  - Weight >2.5kg
  - Older age at surgery
  - Complete repair rather than palliation
  - No preoperative mechanical ventilation
- Careful management of
  - Preoperative pulmonary hypertension
- Mosaics have longer survival

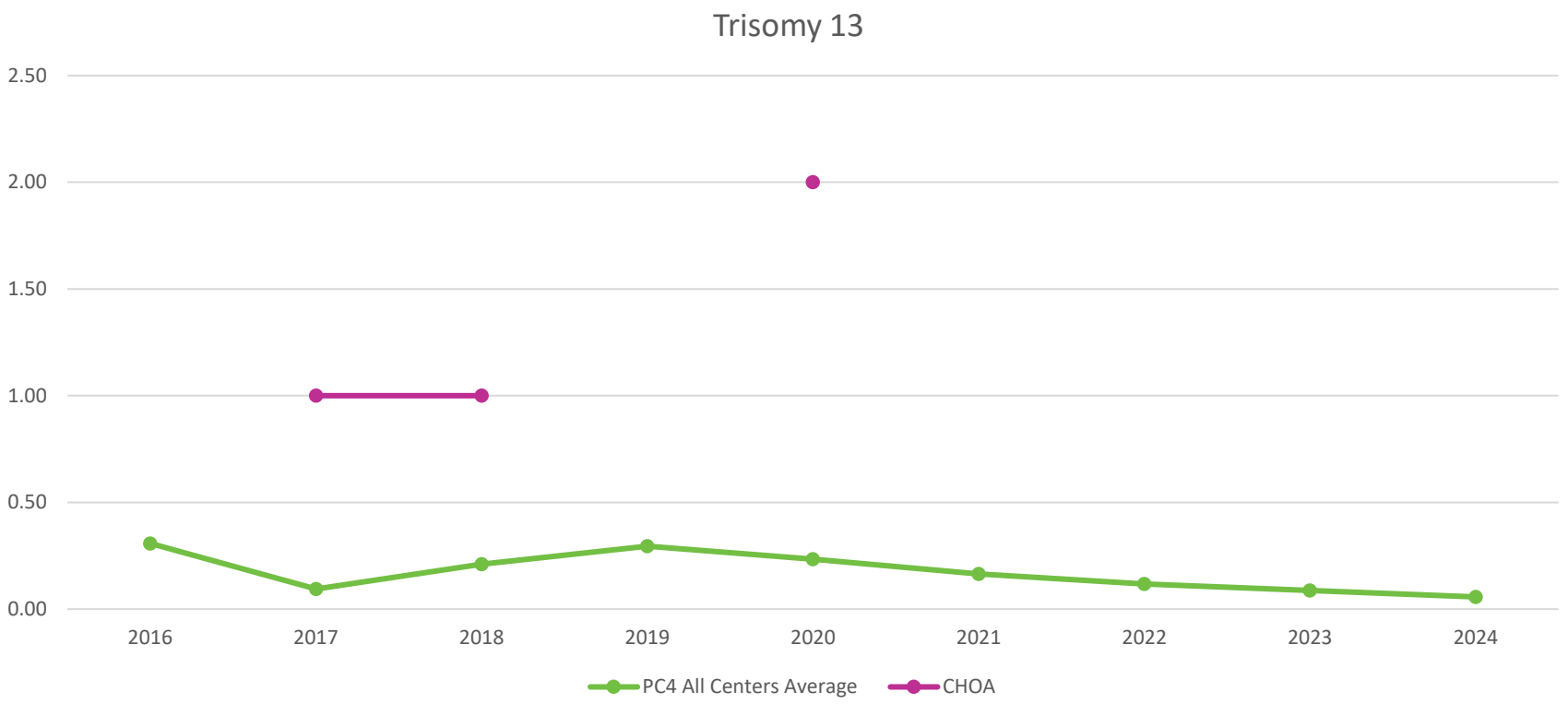
Trisomy 13

	Trisomy 13			
	CHOA Count	Average per Center	Centers Submitting	Total PC4 Cases
2016		2	4	8
2017	1	1	3	3
2018	1	2.67	3	8
2019		1.88	8	15
2020	2	1.4	10	14
2021		2.2	5	11
2022		1.6	5	8
2023		1.2	5	6
2024		1.33	3	4

Trisomy 13


Trisomy 13
16
14
12
10
8
6
4
2
0
1
1
2
2016
2017
2018
2019
2020
2021
2022
2023
2024
CHOA Count
Total PC4 Cases

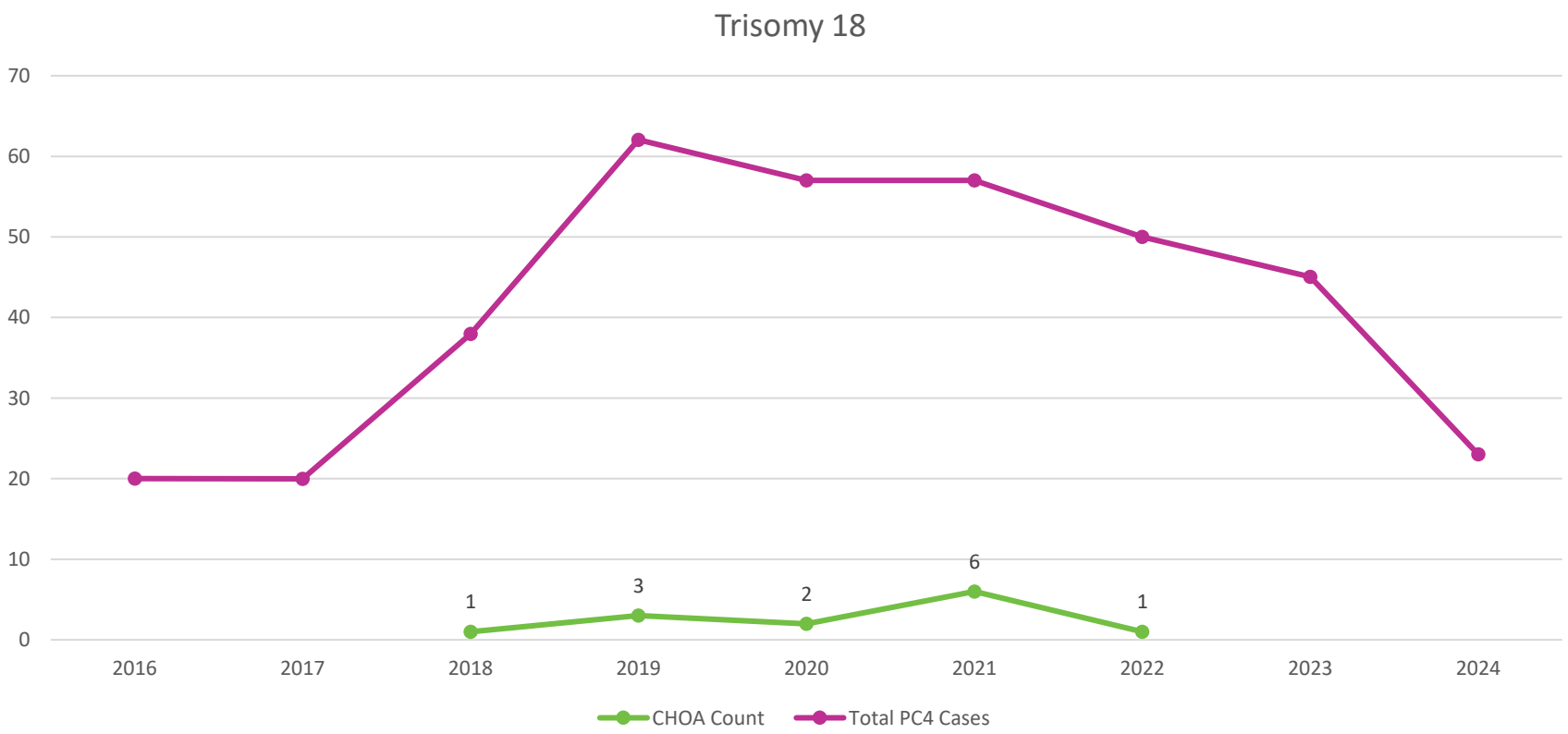
Trisomy 13: average per center



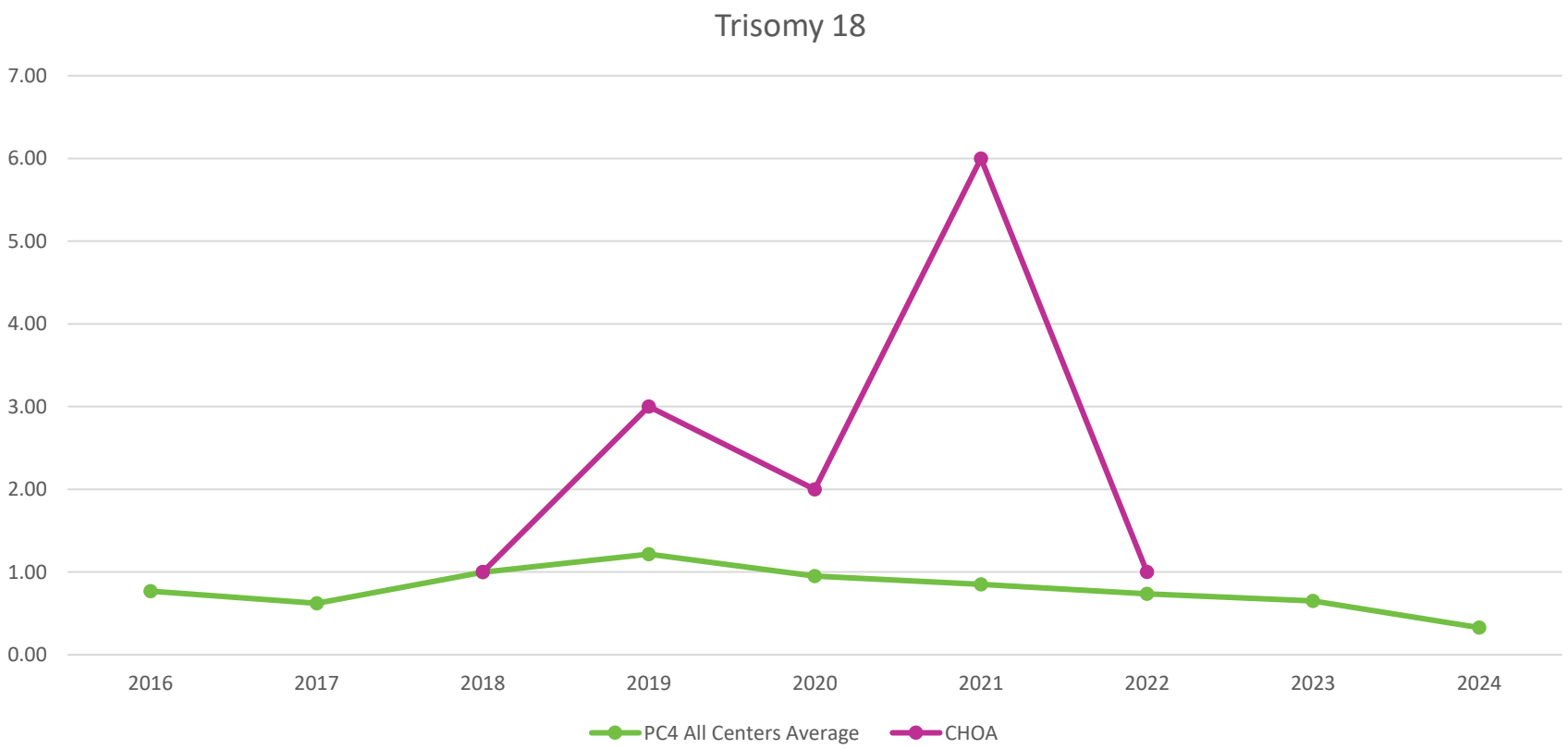
Trisomy 18

	Trisomy 18			
	CHOA Count	Average per Center	Centers Submitting	Total PC4 Cases
2016		2	10	20
2017		2.22	9	20
2018	1	2.71	14	38
2019	3	3.65	17	62
2020	2	3	19	57
2021	6	2.28	25	57
2022	1	2	25	50
2023		2.65	17	45
2024		1.77	13	23

Trisomy 18


Trisomy 18
70
60
50
40
30
20
10
0
2016
2017
2018
2019
2020
2021
2022
2023
2024
1
3
2
6
1
CHOA Count
Total PC4 Cases

Trisomy 18


Trisomy 18
7.00
6.00
5.00
4.00
3.00
2.00
1.00
0.00
2016
2017
2018
2019
2020
2021
2022
2023
2024
PC4 All Centers Average
CHOA

Family experience

The Experience of Families With Children With Trisomy 13 and 18 in Social Networks

AUTHORS: Annie Janvier, MD, PhD,[a] Barbara Farlow, BEng, MBA,[b] and Benjamin S. Wilfond, MD[c]

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KEY WORDS
trisomy 13, trisomy 18, life-sustaining interventions, quality of life, parental opinions, ethics, end of life decision-making

ABBREVIATION
T13-18—trisomy 13 and trisomy 18

All authors contributed significantly to the elaboration of the questionnaire, the analysis of the data, and the preparation of the manuscript.

WHAT'S KNOWN ON THIS SUBJECT: Trisomy 13 and 18 are conditions with 1-year survival rates of less than 10% and have traditionally been treated with palliative care. There are increasing reports of ethical dilemmas caused by parental requests for clinical interventions.

WHAT THIS STUDY ADDS: Parents who belong to social networks report an enriching family experience and describe surviving children as happy. Many of these parents describe challenging encounters with health care providers.

abstract

CONCLUSIONS: Parents who engage with parental support groups may discover an alternative positive description about children with T13-18. Disagreements about interventions may be the result of different interpretations between families and providers about the experiences of disabled children and their quality of life. *Pediatrics* 2012;130:293–298

harder than they expected. Despite their severe disabilities, 97% of parents described their child as a happy child. Parents reported these children enriched their family and their couple irrespective of the length of their lives.

CONCLUSIONS: Parents who engage with parental support groups may discover an alternative positive description about children with T13-18. Disagreements about interventions may be the result of different interpretations between families and providers about the experiences of disabled children and their quality of life. *Pediatrics* 2012;130:293–298

Ethical cosmos

RESEARCH Open Access

"She was finally mine": the moral experience of families in the context of trisomy 13 and 18– a scoping review with thematic analysis

Zoe Ritchie[1*], Gail Teachman[2], Randi Zlotnik Shaul[3,4] and Maxwell J. Smith[1]

Abstract

Introduction The value of a short life characterized by disability has been hotly debated in the literature on fetal and neonatal outcomes.

Methods We conducted a scoping review to summarize the available empirical literature on the experiences of families in the context of trisomy 13 and 18 (T13/18) with subsequent thematic analysis of the 17 included articles.

Findings Themes constructed include (1) Pride as Resistance, (2) Negotiating Normalcy and (3) The Significance of Time.

Interpretation Our thematic analysis was guided by the moral experience framework conceived by Hunt and Carnevale (2011) in association with the VOICE (Views On Interdisciplinary Childhood Ethics) collaborative research group.

Relevance This article will be of interest and value to healthcare professionals and bioethicists who support families navigating the medically and ethically complex landscape of T13/18.

Keywords Bioethics, Decision-making, Goals of care, Values, Neonatal, Neonatal intensive care, Palliative care, trisomy 13, trisomy 18

- Families experience the child and quality of life much different than the providers

Mortality and Resource Use Following Cardiac Interventions in Children with Trisomy 13 and Trisomy 18 and Congenital Heart Disease

Liezl Domingo[1,2] · John C. Carey[3] · Aaron Eckhauser[4] · Jacob Wilkes[5] · Shaji C. Menon[1]

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Table 2 Outcomes and resource use following cardiac intervention

	Trisomy 13 (n = 49, 25.9%)	Trisomy 18 (n = 140, 74.1%)	P value
Age at initial cardiac intervention (weeks) (median, IQR)	10.7 (1.29, 26.57)	19.4 (7.79, 42.93)	0.02[a]
In-hospital mortality	14 (28.6%)	17 (12.1%)	0.01[b]
Cardiac intervention related in-hospital mortality	11 (22.4%)	13 (9.3%)	0.003[a]
Intraoperative mortality	1 (2%)	1 (0.7%)	0.43[b]
Total hospital length of stay (days) (median, IQR)	29 (13, 43)	18 (7, 42.50)	0.051[a]
Adjusted total charge (per $1000) (median, IQR)	305.99 (154.46, 676.55)	252.78 (110.25, 497.92)	0.14[a]

Additional cost/resource utilization

Table 4. Adjusted Excess Cost per Case Associated With Specific Complications

Complication	n (%)	Excess Cost/Case	Lower 95% CI	Upper 95% CI
Tracheostomy	29 (0.2%)	$179,350	$132,958	$237,769
Mechanical circulatory support	263 (2.1%)	$68,964	$53,020	$87,475
Respiratory	953 (7.5%)	$67,149	$53,720	$82,386
Renal failure	138 (1.15)	$65,042	$45,232	$89,152
Reoperation/reintervention	662 (5.2%)	$57,137	$47,419	$67,866
Neurologic	209 (1.6%)	$50,649	$29,498	$77,724
Infectious	511 (4.0%)	$49,968	$41,464	$59,298
Cardiac arrest	246 (1.9%)	$42,366	$29,271	$57,739
Phrenic/recurrent laryngeal nerve injury	241 (1.9%)	$37,271	$23,370	$53,958
Pleural effusion/chylothorax	959 (7.5%)	$30,356	$24,132	$37,132

PASQUALI ET AL
EXCESS COSTS AFTER CONGENITAL HEART SURGERY

Ann Thorac Surg
2014;98:1660–6

Table 5. Adjusted Excess Cost per Case Associated With Prolonged Length of Stay

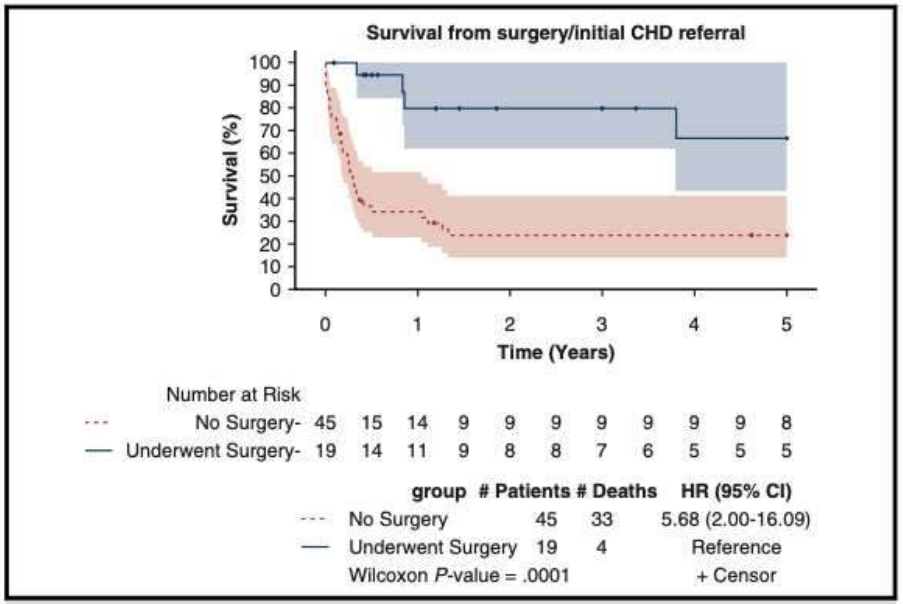
Operation	Additional Days > Median	Excess Cost/Case	Lower 95% CI	Upper 95% CI
ASD repair	+1	$1,912	$−2,007	$6,484
	+2–3	$5,723	$1,014	$11,268
	+4–7	$17,203	$9,212	$27,034
	>7	$123,896	$79,100	$187,877
VSD repair	+1	$5,476	$2,092	$9,180
	+2–3	$10,661	$6,010	$15,857
	+4–7	$22,783	$18,553	$27,355
	>7	$101,246	$86,279	$118,081
TOF repair	+1	$12,130	$6,045	$18,951
	+2–3	$16,469	$10,629	$22,930
	+4–7	$33,004	$26,832	$39,711
	>7	$134,040	$105,467	$168,062
Fontan	+1	$11,096	$6,904	$15,588
	+2–3	$16,062	$11,152	$21,357
	+4–7	$26,845	$18,788	$35,827
	>7	$112,903	$93,718	$134,623
BDG/ hemi-Fontan	+1	$11,543	$7,524	$15,870
	+2–3	$17,919	$13,378	$22,813
	+4–7	$32,759	$27,922	$37,917
	>7	$160,597	$127,862	$199,534
CAVC repair	+1	$16,904	$10,474	$24,023
	+2–3	$25,534	$20,709	$30,691
	+4–7	$35,073	$28,809	$41,839
	>7	$178,721	$152,682	$208,114
ASO	+1	$19,007	$9,905	$28,899
	+2–3	$39,090	$31,162	$47,515
	+4–7	$57,517	$46,172	$69,775
	>7	$189,085	$153,923	$229,216
Truncus repair	+1	$51,597	$8,245	$108,255
	+2–3	$54,637	$22,981	$92,717
	+4–7	$61,825	$30,748	$98,800
	>7	$340,298	$268,188	$425,369
Norwood	+1	$43,789	$9,783	$84,405
	+2–3	$77,795	$38,108	$125,230
	+4–7	$79,185	$46,383	$117,072
	>7	$288,601	$235,308	$348,987

Take home

- Role of cardiac surgery/interventions in T13-18 patients continues to evolve
- There is probably a subgroup that would benefit. Selection is important.
  - Mosaics?
  - Who were able to go home?
  - Corrected extracardiac anomalies?
  - Corrective pathologies?
  - Non PGE dependent?
- Evaluate with cardiac catheterization prior?
- Holistic approach at prenatal care and input from family helpful for counselling and decision making

Cardiac surgery in children with trisomy 13 or trisomy 18: How safe is it?

Joshua M. Rosenblum, MD, PhD, Kirk R. Kanter, MD, Subhadra Shashidharan, MD, Fawwaz R. Shaw, MD, and Paul J. Chai, MD



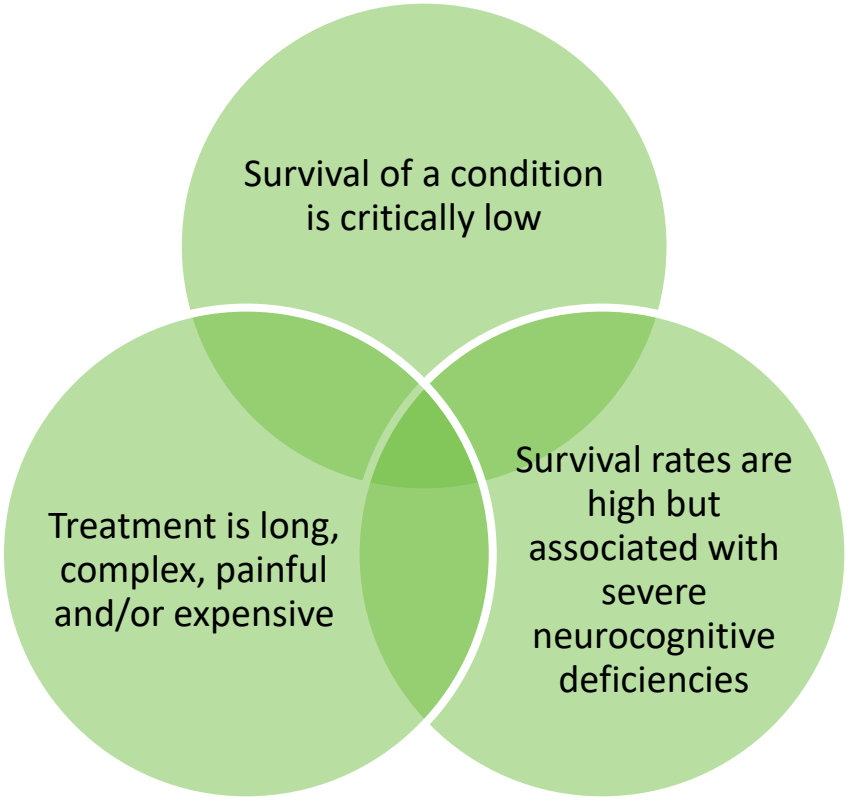
Survival in patients with trisomy 13/18 with repaired and unrepaired cardiac disease.

Our approach

- In general kids who were home before surgery
- In general kids not on invasive mechanical ventilation
- Lower complexity, not requiring multiple surgeries
- Two ventricle circulation

- Over time, we have been more lax

“State of persistent ethical ambiguity”



Ethicist John Lantos: stable grey zone

Children's Healthcare of Atlanta

EMORY UNIVERSITY

Questions???