



Rethinking fertility and fatality in Trisomy 13 and 18

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I have not used artificial intelligence in the development of this presentation.

Objectives

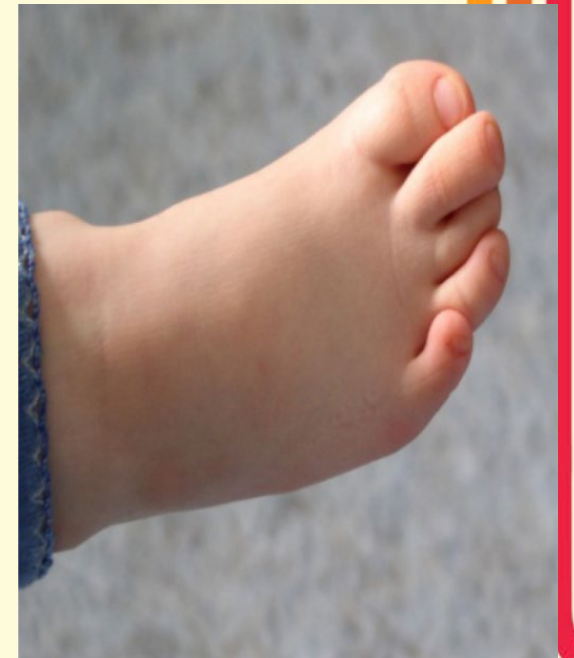
- At the conclusion of this activity, learners will be able to:
 - Understand how outcomes data in Trisomy 13 and 18 is impacted by the approach to care
 - Assess the ethical dimensions of providing intensive medical and surgical care to infants with Trisomy 13 and 18
 - Examine how anti-disability bias shapes physician attitudes towards providing care for infants with chromosomal anomalies
 - Recognize the impact of language and terminology on family experiences

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Trisomy 18

- 1 out of every 1500 pregnancies (Wallace 2018)
 - ~70% result in fetal demise
- Multiple congenital anomalies common (Carvajal 2020)
 - 2 or more systems >50%
 - Cardiac defects in 60-80% (VSD, ASD)
- ~10 survive past 1 year of life (Carvajal 2020)

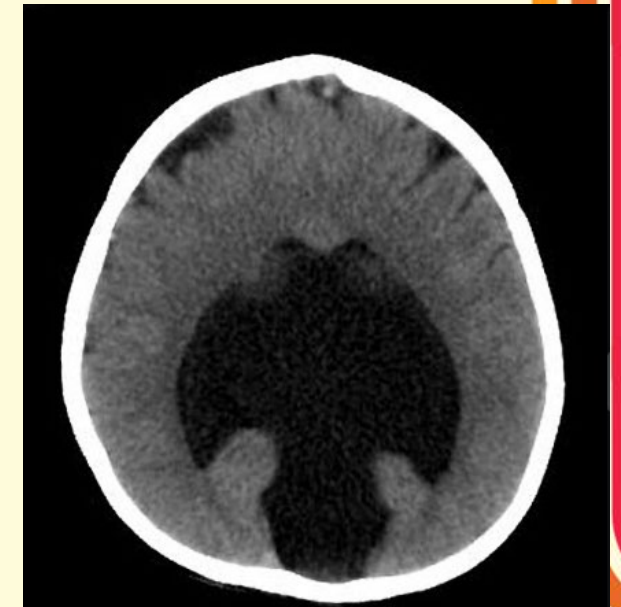


Trisomy 13

- 1 out of every 3800 pregnancies (Wallace 2018)
 - 50% result in fetal demise
- Multiple congenital anomalies common (Carvajal 2020)
 - 2 or more systems >75%
 - Cardiac defects in 60-80% (VSD, ASD)
 - Midline defects
- <10% survive past 1 year of life (Carvajal 2020)



Trisomy.org



Radiopaedia.org

Conventional approach



Neonatal
Resuscitation
Program™

- “Incompatible with life”
- 2005 ILCOR: no resuscitation (updated in 2015)
- 2010 American Heart Association: no resuscitation
- NRP recommendations updated in 2010, ILCOR 2015


Changes in approach to care

- Societal shift towards autonomy
- Expanded access to information
 - Internet
 - Social groups
- Survey of neonatologists (McGraw 2008)
 - 44% would provide resuscitation
 - Beneficence/ non-maleficence vs autonomy





Best interest standard

- Overall benefits to child outweigh burdens of interventions
 - Requires subjective value judgments
 - Who should determine best interests?
- 

Argument for non-intervention

- Unacceptably high morbidity and mortality
- Physician obligations
- Parental guilt

Bias in outcomes data

- Combines palliative and intensive approach
 - Majority of families chose comfort care
- Time bias
 - Changes in management
 - Mortality data from 1976-2014 (Goel 2019)
- Creates self-fulfilling prophecy



Prenatal vs postnatal diagnosis

- 216 infants with Trisomy 13/18 (Janvier 2016)
 - Postnatal diagnosis highest predictor of survival
 - More likely to receive resuscitation
 - Less likely to have major anomalies
- Cause of death in first week of life (Leuthner 2020)
 - Respiratory
 - Withholding artificial nutrition/ hydration
 - Ductal-dependent lesions

Outcomes data: Trisomy 18

	Palliative approach	Intervention approach
Survival at 1 month	33 – 37.2%	83%
Survival at 1 year	3-13.4%	20-84%
Survival at 5 years	12.3%	23%

Adapted from Silberberg et al. *European Journal of Pediatrics* 2020

Parental surveys

- 332 parents of 272 children with T18/13 (Janvier 2020)
 - 40% lived >1 year
 - 104 newborns received comfort care
 - 1/3 survived >1 year
 - 53 newborns received full interventions
 - ½ lived >1 year

SUPPORT ORGANIZATION FOR TRISOMY

SOFT has been empowering families with children diagnosed with Trisomy 18, 13 and related chromosomal disorders for over 40 years.



Surgical outcomes

- American College of Surgeons – NSQIP (Bajinting 2021)
 - 2012-2017
 - 310 patients with T18
 - Gastrostomy tube most common
 - Low morbidity in non-cardiac surgeries
- Cardiac surgery more controversial
 - Complication rate higher
 - Certain patients may benefit



Futility

- No clear definition
- Qualitative versus quantitative futility
- Requires subjective measurement of benefits and harms
- Need to determine goals of treatment

Quality of life



- Another poorly defined concept
- Anti-disability bias prevalent in physicians (Iezzoni 2021)
- Families report their children to be:
 - Happy
 - Able to communicate their needs

Developmental milestones in Trisomy 18 (n=62)

Milestone	Age achieved (months)	Number achieved
Watched toy or face	4.4	57
Smile responsively	4.7	54
Reached for toy	9.6	38
Laughed out loud	13	36
Held head up	9	33

Adapted from Lorenz et al. *Current Opinions in Pediatrics*. 2014

Parental perspectives

- Survey of parents of Trisomy 13/18 (Janvier 2012)
 - 98% say their child enriched their lives
 - 97% report child as being happy
 - 82% felt child had positive impact on siblings
 - 87% were told their child was “incompatible with life”




Importance of language

- Inaccurate statements lead to mistrust
 - Lethal anomaly
 - Incompatible with life
 - Universally fatal
- Acknowledge personhood
- Compassion, humility, non-judgmental



Revisiting ethical framework

- Principles
 - Beneficence
 - Non-maleficence
 - Autonomy
 - Justice?
- 

Justice

- Distributive justice
 - Financial concerns
 - Small number of patients
- Patients in similar situations should be treated similarly
 - Cost considerations in adult ICU
 - Infants with poor prognosis without T13/18

MOC Questions in Slido (3 slides)

- Chapter staff will add these slides to your presentation once

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Question 1 Discussion

- Up to 80% of T13 have CHD
- VSD, ASD, PDA most common
- Often concurrent pulmonary hypertension

Kosiv KA, Mercurio MR, Carey JC. The common trisomy syndromes, their cardiac implications, and ethical considerations in care. *Curr Opin Pediatr.* 2023;35(5):531-537.

Question 2 Discussion

- NRP removed T18 from list of lethal conditions in 2010
- >50% of infants will have anomalies in 2 or more systems
- Gastrostomy tube placement is the most commonly performed surgical procedure in this population and is generally well-tolerated
- Central apnea is a common cause of death in patients of all ages with Trisomy 18

Leuthner SR, Acharya K. Perinatal counseling following a diagnosis of trisomy 13 or 18: incorporating the facts, parental values, and maintaining choices. *Adv Neonatal Care.* 2020;20(3):204-215.


Carvajal HG, Callahan CP, Miller JR, Rensink BL, Eghtesady P. Cardiac surgery in trisomy 13 and 18: a guide to clinical decision-making. *Pediatr Cardiol.*

Bajinting A, Munoz-Abraham AS, Osei H, Kirby AJ, Greenspon J, Villalona GA. To operate or not to operate? Assessing NSQIP surgical outcomes in trisomy 18 patients. *J Pediatr Surg.* 2021;56(3):565-568.2020;41(7):1319-1333.


Question 3 Discussion

- >75% of infants with T13 will have multiple congenital anomalies
- Midline defects common
 - Omphalocele
 - Cleft palate
 - Holoprosencephaly

Pereira EM. Trisomy 13. *Pediatr Rev.* 2023;44(1):53-54.



Implications for practice

- Non-directive counseling
 - Reconsider ethical framework
 - Advocate for patients and families
- 

References and additional reading

- Wallace SE, Gilvary S, Smith MJ, Dolan SM. Parent perspectives of support received from physicians and/or genetic counselors following a decision to continue a pregnancy with a prenatal diagnosis of trisomy 13/18. *J Genet Couns.* 2018;27(3):656-664.
- **Carvajal HG, Callahan CP, Miller JR, Rensink BL, Eghtesady P. Cardiac surgery in trisomy 13 and 18: a guide to clinical decision-making. *Pediatr Cardiol.* 2020;41(7):1319-1333.**
- **Janvier A, Farlow B, Barrington KJ, Bourque CJ, Brazg T, Wilfond B. Building trust and improving communication with parents of children with Trisomy 13 and 18: A mixed-methods study. *Palliat Med.* 2020;34(3):262-271.**
- McGraw MP, Perlman JM. Attitudes of neonatologists toward delivery room management of confirmed trisomy 18: potential factors influencing a changing dynamic. *Pediatrics.* 2008;121(6):1106-1110.
- Goel N, Morris JK, Tucker D, et al. Trisomy 13 and 18-Prevalence and mortality-A multi-registry population based analysis. *Am J Med Genet A.* 2019;179(12):2382-2392.

References and additional reading

- Janvier A, Farlow B, Barrington KJ. Parental hopes, interventions, and survival of neonates with trisomy 13 and trisomy 18. *American J of Med Genetics Pt C*. 2016;172(3):279-287.
- **Leuthner SR, Acharya K. Perinatal counseling following a diagnosis of trisomy 13 or 18: incorporating the facts, parental values, and maintaining choices. *Adv Neonatal Care*. 2020;20(3):204-215.**
- Bajinting A, Munoz-Abraham AS, Osei H, Kirby AJ, Greenspon J, Villalona GA. To operate or not to operate? Assessing NSQIP surgical outcomes in trisomy 18 patients. *J Pediatr Surg*. 2021;56(3):565-568.
- Iezzoni LI, Rao SR, Ressler J, et al. Physicians' perceptions of people with disability and their health care. *Health Aff (Millwood)*. 2021;40(2):297-306.
- Janvier A, Farlow B, Wilfond BS. The experience of families with children with trisomy 13 and 18 in social networks. *Pediatrics*. 2012;130(2):293-298.
- **Pyle AK, Fleischman AR, Hardart G, Mercurio MR. Management options and parental voice in the treatment of trisomy 13 and 18. *J Perinatol*. 2018;38(9):1135-1143.**

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