CE 1.5 HOURS Continuing Education

Perinatal Counseling Following a Diagnosis of Trisomy 13 or 18

Incorporating the Facts, Parental Values, and Maintaining Choices

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ABSTRACT

Background: Families with a prenatal diagnosis of trisomy 13 or 18 are told many things, some true and some myths. They present with differing choices on how to proceed that may or may not be completely informed.

Purpose: To provide the prenatal counselor with a review of the pertinent obstetrical and neonatal outcome data and ethical discussion to help them in supporting families with the correct information for counseling.

Methods/Search Strategy: This article provides a review of the literature on facts and myths and provides reasonable outcome data to help families in decision making.

Findings/Results: These disorders comprise a heterogeneous group regarding presentation, outcomes, and parental goals. The authors maintain that there needs to be balanced decision-making between parents and providers for the appropriate care for the woman and her infant.

Implications for Practice: Awareness of this literature can help ensure that prenatal and palliative care consultation incorporates the appropriate facts and parental values and in the end supports differing choices that can support the infant's interests.

Key Words: ethics, palliative care, perinatal counseling, trisomy 13, trisomy 18

The most common chromosomal abnormalities diagnosed prenatally and at birth are trisomy 21 (T21), trisomy 18 (T18), and trisomy 13 (T13).[1] Historically, all 3 of these anomalies were considered either fatal or of extremely poor outcome, and live-born infants were allowed to die naturally.[2,3] Over time, there were medical and legal challenges to the care provided to infants with T21, followed by societal and legal changes that now require parents and physicians to treat infants with T21.[4,5]

While T18 and T13 were thought to remain in the category of lethal conditions, more recently, there have been parental and physician advocates challenging this norm.[4,6-8] T18 has been removed from the list of lethal conditions by the American Academy of Pediatrics' (AAP's) Neonatal Resuscitation Program.[9] Criticisms from these advocates include poor communication and even disrespect, lack of medical knowledge, and that the idea of lethality is a self-fulfilling prophecy.[8,10-12] Some have suggested that perhaps there is a cultural shift for these conditions, similar to what happened with T21, which certainly would lead to changes in how one might consider prenatal and neonatal consultation in these pregnancies.[4,13,14]

The goal of this article is to review some of the ethical and medical literature that can be used to help guide prenatal consultation for a family with a new diagnosis of T18 or T13, with the aim to present a balanced view of the disease processes in order to support and help a family make independent, best decision for their future child.

Some prenatal cases to consider are as follows:

Case 1: A couple presents with a male fetus who on ultrasound (US) scan is revealed to have significant intrauterine growth restriction (IUGR), abnormal hands, cleft lip and palate, ventricular septal defect (VSD), and polyhydramnios and concerns of tracheoesophageal fistula. Genetic testing reveals T18. They have been counseled and accept that their infant cannot survive with the quality of life they would like and are seeking termination of pregnancy.

Case 2: A Catholic couple presents with a female fetus who on US scan is revealed to have IUGR, has abnormal hands, and hypoplastic left heart syndrome (HLHS). Genetic testing reveals T13. They are opposed to termination of pregnancy, want to be reassured that they understand all their infant's conditions and issues, yet are realistic, and acknowledge they do not want to put their infant through any suffering.

Case 3: A couple presents with a female fetus who on US scan is revealed to be small for gestational

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age (SGA) and has a possible coarctation of the aorta on fetal echocardiogram. Genetic testing reveals T18. The family is quiet but shares they are seeking information about all options. They have been in contact with one advocate on social media but are not open about their thoughts on what to pursue postnatally.

Case 4: A couple presents with a female fetus who on US scan has a now resolved cystic hygroma, mild growth restriction, abnormal hands, cleft lip and palate, and a VSD. Genetic testing reveals T18. They have been in contact with one advocate on social media and come in a defensive posture with demands to "avoid discrimination by ignoring the genetic condition" and treat their infant like any other child.

HISTORY AND LETHALITY

If we look back in time at physician attitudes and practices for children with disabilities over 40 to 50 years, perhaps we can learn something. In the 1970s, physicians were willing to defer to parental discretion to refuse treatment on infants with T21 and myelomeningocele.[15,16] In the 1980s, however, there was significant societal change, and limits to parental discretion to refuse treatment of T21 emerged through the "Baby Doe" regulations. However, further questions about more profound disabilities remained,[17] and T18 and 13 were still universally considered a lethal condition.

Things began to change in the early 2000s when the AAP neonatal resuscitation guidelines omitted T18, but not T13, from the list of examples of conditions for which resuscitation is not indicated.[9] By 2008, there was a shift in neonatologists' attitudes regarding delivery room resuscitation, with 44% of providers reporting they would consider initiation of resuscitation for an infant with T18 even with congenital heart disease.[18] The authors stated, "Support for the best-interest standard for neonates is diminishing in favor of ceding without question to parental autonomy. This shift may have profound implications for ethical decisions in the NICU." (pg. 1106, abstract).

One of the reasons for this shift has been parental advocacy, as well as physicians and ethicists arguing that for at least some of these infants and children, lethality is a self-fulfilling prophecy by the medical community.[8,12,19,20] They argue that the concept of "lethal" malformations is imprecise. A "lethal" diagnosis implies an irresistible progression of a disease that inevitably leads to death in the near future. Such "lethal" language implicates that treatment of such a condition is futile or even detrimental, predetermines medical treatment, because it predetermines parental and medical anticipations on the clinical course, "is harmful because it may distract parents from unprejudiced decisions and treatment options," and that there should always be a caveat about confirming the prenatal findings on postnatal evaluation.

These ethicists argue that "lethality" is a normative concept, and the problem with using "lethal" language is that it medicalizes normative ideas of "quality of life," "suffering," "family burden," or "cost."[4,8,12,19] Providers may not be comfortable stating views about "quality of life" and the "value" of children with profound developmental disabilities. Unexamined normative views about children with profound developmental disabilities can influence provider attitudes and information conveyed to parents. Finally, just as the delivery room resuscitation has changed, there are medical reports of intervening and perhaps prolonging the life of some of these children, which begs the question of whether yesterday's lethal may be today's success.[21,22]

ETHICALLY OBLIGATORY OR PERMISSIBLE?

From this one could infer the argument that since society has decided we should offer all these interventions to infants with T21, why not T18? We have heard parents of these children make that claim. We should be treating all with disability or not as equals, otherwise we do have a self-fulfilling prophecy of lethality. What is important to recognize, however, is the societal change in treatment of an infant with T21 is that parental discretion to decide to "not" provide certain medical interventions is revoked. Medical care has become obligatory, which is very different from a parental request for a trial of therapy and whether it is considered ethically permissible to medically intervene. Medical and surgical interventions in the setting of T13 or T18 should not ever become ethically obligatory.[14] The threshold for obligatory treatment over parental objection ought to be high—the benefits should very clearly outweigh the burdens. T18 and T13 are not equivalent to T21 in viability, neurologic potential, and therefore in perceived quality-of-life potential, and the threshold for obligatory treatment is not met.

At the same time, for a treatment to be ethically impermissible, there should be no chance of benefit to the child, or the burdens of treatment should far outweigh the benefits. It does not seem that this threshold is met in all cases as the chance for benefit in at least some cases has been demonstrated.[21-24] If for a given patient a treatment is permissible, even if inadvisable, then parents should be given the choice. If the physicians judge the treatment to be inadvisable, it seems appropriate for the physician to share his or her recommendation on that question with the parents and the reasoning behind that recommendation.[20] In summary, for infants with T13 or T18, no medical intervention should be considered obligatory, some interventions should be considered impermissible, some

interventions might be considered inadvisable, yet permissible, and other interventions might be considered advisable and permissible. Where a particular intervention might fall in this categorization is dependent on the medical or surgical complexity of the intervention and based on the individual parental goals of care.

BEST INTEREST AND PARENTAL DECISION-MAKING

During prenatal counseling, it is common practice to treat the expectants as parents who are making a medical decision for their child, at the same time understand that a pregnant woman has her own health to consider.[25,26] Therefore, a goal for all involved should be to help balance the "best interest" of the future child, the maternal health interests, and the family interests. This means that the prenatal counseling needs to include an assessment of the understanding of the clinical condition and the parental goals for the pregnancy and future child. In the 4 cases discussed earlier, the range of parental expectations and what they see as best interest is broad, and they reached different decisions for the pregnancy and their child. Assessment of the parental view of best interest is critical in guiding the counselor's discussion about options, and while deferral to parental values is reasonable in most cases, the ethical concept of best interest does allow a pediatric provider to override a parent decision.[18,27] The burden of proof does rest on the provider overriding that decision and typically centers around the concept of harm.[28]

THE EXPERIENCE OF FAMILIES

Much of what has pushed more medical interventions in this population has been parent advocacy. In one of the first reports on prenatal counseling, the parents shared that practitioner's empathy and language were important.[29] While the parents were satisfied with some and dissatisfied with other practitioners, there was no difference in mean satisfaction scores or the likelihood of being highly satisfied between those families that chose to continue the pregnancy and those that underwent an induced abortion. Both groups reported similar experiences about informed consent for the screening process, delivery of diagnosis, and options given at counseling. The families that opted for induced abortions consistently reported having the support of their providers. For these families, the portrayal of the severe medical consequences justified their decision to end the pregnancy. On the other hand, more than half the families that continued the pregnancy indicated feeling that they were going against the advice of their providers. These families wished that medical information had been more positive and were offended by the phrase "incompatible with life," which often created an environment of distrust with their healthcare providers.

A computer-assisted self-completion questionnaire survey from the social networks of families with live-born children with T13 or 18 provides a similar notion of language and trust being an issue.[23] The respondents are parents of 216 children with full trisomy, of which about half received some level of medical interventions, and of those about half of the children lived beyond a year of age. The parents reported that their children have significant developmental delays but gained milestones over time and that they communicated in some way with their children and understood their needs, and 99% of parents described their child as a happy child.[23] They reported that 63% of parents met a healthcare provider who helped, whereas 37% of parents who chose clinical intervention for their child felt judged. Table 1 displays some of the most common positive and negative comments made by parents and were based on a sense of whether the healthcare providers did or did not see their infant as having value, as being unique, or as being an infant. The parents also reported that their children felt more pain and the parents' lives were enriched. These last few findings could lead one to question the parental decision-making and whether it is centered on the best

TABLE 1. Things to Say and NOT to Say to a Family With a Child With T13 or T18[a]

Things NOT to Say	Things to Say That May Help
• Incompatible with life • Lethal • Would live a life of suffering • Referring to child as an it/that/vegetable/T13/T18 • Would live a meaningless life • Would ruin their marriage • Would ruin their family • This child will hurt you/your family/your children • Waste of money/time/energy • There is nothing we can do for him or her • You can have another one	• May enrich their family • Might have a short meaningful life • Might survive for many days/months/years • Referring to the child by name (even if unborn) • Offering to take pictures (in and ex utero) • Referring to other families or Web sites • Describing not only those organs that had malformations but also those that did not have malformations

Abbreviations: T13, trisomy 13; T18, trisomy 18.
[a]*From reference.*[22]

interest. The goal of pediatric care should not be to increase pain but to enrich the child's life, and often in doing so the parents' lives are enriched. It certainly should not be to increase the pain of a child as a means to an end of being a more enriched parent.

Having a better understanding of the parental perspective can facilitate communication and decision making between providers and parents. While these data provide a lot of information that can be helpful in prenatal consultation, they must also not be overinterpreted. A concern would be to misinterpret these data is to conclude that a majority of pregnant women with a fetus with T13 or T18 are dissatisfied with their healthcare team, because in truth, the data support that most families were satisfied. It is important to hear the voices of these families to improve our counseling ability, yet it must be acknowledged that it is from a certain point of view. There is limited to no data from the viewpoint of those who choose termination of pregnancy. Perhaps, for these families, it is important to hear terms such as "lethal" as it validates why they are making their decision.[29-31] For these families, it is also important to hear that most families in their situation have made a choice similar to theirs; that they do not represent a minority or are "bad parents" because they have made a decision to terminate. The use of these "negative" terms does not necessarily mean that one must view the infant as having no value, being unique, or being an infant. We need to remain cognizant of the language we use in consultation, always reminding ourselves of the respect these mothers and infants deserve no matter what choices they make.

WHAT DO EXPECTANT PARENTS CHOOSE AND WHAT ARE THE PREGNANCY OUTCOMES IN THOSE CHOICES?

There are several reports in recent years that have addressed prenatal diagnosis, what families choose, and the natural history of obstetrical outcomes should parents choose expectant management that are important for prenatal consultation.[32-37] Ethical counseling requires knowing outcome data as opposed to generalizations of myths (see Table 2). All too often, expectant parents are presented with misinformation from their obstetrician, pediatrician, or perhaps even a neonatologist and maternal–fetal medicine physicians, about when and how these

TABLE 2. Provider and Parent Myths and Facts Regarding T13 or T18[a]

	Myth	Fact
Choice of termination	Most women terminate or Most women want "everything" done	50%-85% of women terminate <5% want "everything done"
Expectant management and in utero demise	"These infants never make it to term"	30%-60% die during gestation, and this risk is evenly distributed throughout the pregnancy; infants with coexisting anomalies and male fetuses with T18 are at greater risk of in-utero demise or stillbirth
Expectant management and live-born	"These infants all die during the birthing process"	Stillborn (death during labor) 20%-25% Live-born 13%-50%
Infant death	All these infants die shortly after birth	50%-65% die within 1 wk of life 60%-90% die within 1 mo of life 75%-90%[b] die within 1 y of life
Interventions and survival	All infants die no matter what we do or Response to interventions is similar to infants without T13 or T18	• Some interventions may prolong life for some of these infants, but numbers are small, long-term survival data are lacking, and quality of life may not improve even after surgery • Mortality, length of stay, and need for medical interventions are much higher • Female gender, higher birth weight, and absence of major surgical anomalies increase chances of survival to hospital discharge
Neurodevelopmental outcomes	They will be a "vegetable," or (more recently) there is wide variation in neurodevelopmental outcomes	Survivors may achieve a few developmental milestones, but there is a narrow, not wide, range of developmental potential, and all of these children have severe or profound neurodevelopmental disabilities

Abbreviations: T13, trisomy 13; T18, trisomy 18.
[a]*Data from references.*[6,20,29-33,38]

infants die. Unfortunately, this leaves the parents either angry and mistrusting or somehow believing their infant is different should their infant live longer than described. A counseling strategy might be acknowledging that for the expectant choice, some infants die in utero and some do not, some infants die during birth and some do not, some infants die shortly after birth and some do not, some die before mothers go home and some do not, a majority of those born alive die before 1 year of age at various times in that first year of life, with a median survival time being 7 to 10 days and 10 to 14 days for T13 and T18, respectively,[32,35] and finally, there are rare cases of those living far beyond a year (see Figure 1).

For some, this information on pregnancy and poor survival outcomes may be enough to decide about the pregnancy and neonatal care. For others, more information on neonatal interventions and outcomes, their potential benefits and burdens, and long-term developmental outcomes will be important to share before making obstetrical or neonatal decisions.

WHAT ARE THE NEONATAL OUTCOMES SHOULD THE PARENTS CHOOSE INTERVENTIONS?

T13 and T18 are heterogeneous conditions. Some live-born children will survive without any neonatal interventions, some children will survive and go home after medical intervention(s), and some children will die despite aggressive medical interventions (avoidable pain and intensive care). While it is hard to tell these 3 groups apart, there has been more research that can help prognosticate. Knowing these data for counseling is critical as some parents appropriately argue that providers do not know the data. A summary of this information is provided in Table 3.

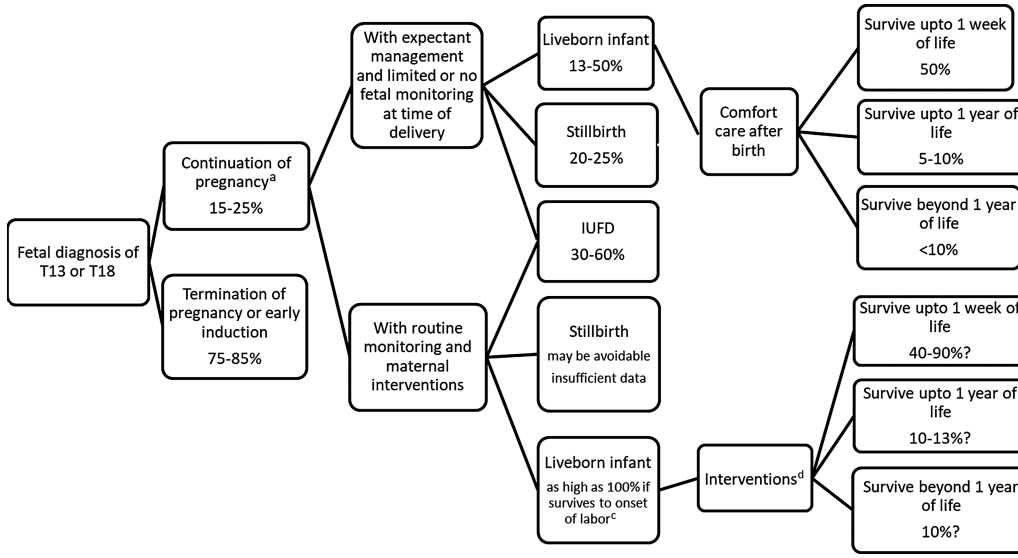
Natural History

The natural history in population-based studies has been used widely to provide the survival rate of 10% or the mortality rate of 90% at 1 year of age.[35,46-49] The denominator used is usually the number of live-born infants. Importantly, these studies were conducted at a time when there were no interventions offered to these infants. As more infants with these conditions receive interventions, these numbers may change.[50] Some of the parent-based reports have suggested higher survival rates, with numbers as high as 50% for those who received "full intervention" surviving beyond 1 year of life, but these have their own selection bias.[23]

Neonatal Interventions

One of the first reports on outcomes when offering standard intensive care treatments to infants with these genetic disorders was reported in 2006 from

FIGURE 1



Pregnancy decisions and possible outcome trajectories following a prenatal diagnosis of T13 or T18. From references[32, 33, 37, 39-41] T13 indicates trisomy 13; T18, trisomy 18; IUFD, intrauterine fetal demise (defined as fetal demise prior to the onset of labor). [a]Pregnancy decisions vary depending on maternal age, detection in the first versus second trimester, insurance status, state abortion laws, and presence of other anomalies [b]Rates of stillbirth and IUFD are combined in some studies. From references[29, 32, 33, 34.] [c]Only 1 study has examined differences in outcomes depending on the level of maternal–fetal monitoring. [d]Outcome after receipt of interventions may depend on individual patient factors such as birth weight, gender, coexisting type, and severity of anomalies.

TABLE 3. Outcomes for Infants With T13 or 18 Who Received Interventions[a]

Study	Methods	Outcomes
Graham et al (2004)[45] US, Canada, Europe	35 patients with T13 or T18 who underwent cardiac surgery in the Pediatric Cardiac Care Consortium database	• 90% patients survived to hospital discharge • Those who required mechanical ventilation prior to surgery were less likely to be weaned from ventilation at discharge • Long-term survival and quality of life not determined
Kosho et al (2006)[21] Japan	24 patients with full T18 Infants received a wide range of interventions including ventilation, cardiovascular medications, GI but not cardiac surgical procedures	• 1-y survival was 25% • Only 1 patient was still alive at the age of 3 y • 5 patients were discharged home; rest died in the hospital • Long hospital stays for most and many postoperative complications
Kaneko et al (2008)[24] Japan	17 patients with T18 who underwent cardiac surgery	• 82% patients survived to hospital discharge
Costello et al (2015)[43] US	Single-center review of 16 cases Comparison of expectant management vs surgical management of congenital heart disease	• Mortality was 29% in the surgical group vs 50% in the expectant management group (not statistically significant) • Patients in the expectant management group were sicker
Imataka et al (2007)[38] Japan	44 patients with T18 at a single center, many of whom received mechanical ventilation, GI surgical procedures, and some cardiac surgical procedures during a 20-y period	• Improved survival in the contemporary group (who received more interventions) in the early infancy period (<1 y), but after 1 y, survival in both groups similar (10% vs 12.5%)
Nelson et al (2016)[22] Canada	428 patients Linked administrative database	• 12%-20% survival at 1 y of age • 10%-13% survival at 10 y of age • 70% survival 1 y after surgical procedure (including ENT, GI, cardiac surgeries)
Acharya et al. (2017)[42] US	841 patients with T13 or T18 in the Pediatrix NICUs, many of whom received medical interventions	• For infants admitted to NICUs, 40% survived to NICU discharge • Low birth weight, male gender, and presence of surgical anomalies were associated with higher mortality • Infants who received (or required) more interventions had higher mortality
Kosiv et al (2017)[44] US	1020 patients with T13 or T18 in the PHIS database	• Infants who received cardiac surgery had lower in-hospital mortality than those who did not • Higher birth weight, older age, and female sex were associated with a lower mortality after surgery • Most common surgical procedures were VSD, ASD, and PDA repairs

Abbreviations: ASD, atrial septal defect; GI, gastrointestinal; NICU, neonatal intensive care unit; PDA, patent ductus arteriosus; PHIS, Pediatric Health Information System; T13, trisomy 13; T18, trisomy 18; VSD, ventricular septal defect.

[a]*From references.*[21,22,24,38,42-45]

Japan.[21] They reported on 24 cases, had a median survival of 152 days, and a 1-year survival of 25%. While some may consider this improvement, it should be noted that the 2-year survival was 4%, the same as would have been expected had no interventions been offered. The main differences are in time and morbidities with interventions. Nearly 90% required mechanical ventilation, and 70% were unable to be extubated. Only 5 patients were discharged home after lengthy hospital stays, with only 1 survivor to the end of the study. Interestingly, the surviving infant only had IUGR as a prenatal finding, was discharged at 30 days with a spontaneously closing patent ductus arteriosus as the heart defect, and required no surgical intervention.

Nelson et al[50] reported on a cross section of hospitalizations in 5 separate years in the United States and found that children with T13 and T18 receive

significant hospital care. They found that 36% of the hospitalizations were for children older than 1 year and received 2764 major procedures, although the exact number of individuals receiving these procedures is not clear as a fair number may have received multiple procedures. Importantly, the benefits and burdens are not able to be explored in this population database.

Acharya et al[42] analyzed the Pediatrix Data Warehouse, which comprises 270 neonatal intensive care units (NICUs) in order to look at major anomalies and birth weight and their influence on NICU interventions and mortality in this population. They found that a majority of infants do not have an anomaly that would need neonatal surgical repair, that these infants received a wide variety of NICU interventions, and that of those making it to an NICU, 40% survive to NICU discharge. What they did find is that there is a significantly higher mortality for those who are male, had very low birth weight (VLBW), had anomalies associated with neonatal repair and those who received more interventions. Others have demonstrated that prematurity and VLBW are poor predictors of NICU survival.[42,51,52] While there are limitations to the inquiry, it is valuable information that can help with prognosis in the prenatal consultation.

Cardiac Interventions

Much of the controversy on neonatal intervention in this population has centered around cardiac surgical intervention. A common myth was that these infants cannot survive these surgical procedures. This has been proven wrong both anecdotally and in the literature.[24,38,43-45,53] There have been studies that conclude that cardiac surgical intervention can be done successfully and increase the chance for discharge and longer survival.[43,54] Other studies show that cardiac surgery contributed to increased survival rate but not the rate of discharge alive in these patients.[38,54] Graham[53] has an interesting review of many of these studies, and as one dives deeper into them, one realizes how complex they are to interpret. Graham concludes that we should not be offering or performing these interventions on these infants as they are not proven and add burden. He argues that there is a selection bias toward healthier, bigger children with less complex heart defects who receive surgical procedures, and it is unclear if the children survived because they received surgery or because they represented "survival of the fittest." He argues that these surgical procedures cause significant morbidity in patients, including prolonged hospital stays and ventilator dependence, and have not demonstrated to improve neurologic and psychomotor disabilities and therefore should not be offered.

Population Studies With Interventions Incorporated

Population-based studies that might be more inclusive of interventions have demonstrated some improved numbers for these infants. In Ontario, 174 children with T13 and 254 children with T18 were live-born between 1991 and 2012.[22] At the end of follow-up, 24 children with T13 and 23 children with TT18 were alive. Survival did not change over time. Median survival time for children with T13 was 12.5 (interquartile range [IQR] = 2-195) days and for T18 was 9 (IQR = 2-92) days. One-year survival was 19.8% (95% confidence interval [CI], 14.2-26.1) for children with T13 and 12.6% (95% CI, 8.9-17.1) for children with T18. At 10 years, 12.9% (95% CI, 8.4-18.5 [n = 13]) of the T13 cohort was alive and 9.8% (95% CI, 6.4-14.0 [n = 16]) of the T18 cohort was alive. If an infant lived up to 30 days, then the chances of 1-year survival were 46% and 36% for T13 and T18, respectively. If an infant lived up to 6 months, the chance of 10-year survival was 50% and 60% for T13 and T18, respectively.

In the United States, Meyer et al[55] have reported 1-year survival in infants with T18 (13.4%) and T13 (11.5%) and a 5-year survival (12.3% for T18; 9.7% for T13). While these numbers do show improvement, the differences are small, reveal a high mortality, and likely demonstrate that a majority of families and physicians still do not want to necessarily provide aggressive neonatal intervention.

FACTORS THAT CONFER A SURVIVAL ADVANTAGE

Table 4 shows factors that confer a survival advantage for these children. These factors should be taken into consideration when providing outcome data to families.

WHAT ARE THE OUTCOMES SHOULD THE INFANT SURVIVE TO DISCHARGE?

Understanding when and how these children who are surviving to go home might die is important to help develop the palliative plan of care for the children (see Table 5). Clearly, some medical interventions can help prevent some of the earlier deaths and lead to different management strategies. More common than needing to decide about surgical

TABLE 4. Factors That Confer a Survival Advantage for Infants With T13 or T18

Female gender
Higher birth weight
Term gestation
Absence of neonatal surgical anomalies
Medical stability (infants who do not require many interventions)

Abbreviations: T13, trisomy 13; T18, trisomy 18.

TABLE 5. Timing and Cause of Death in Children With T13 or T18

When Do Infants Die?		How Do Infants Die?
Survive the first week	32%-56%	Central apnea, airway malacia, ductal dependent cardiac lesions, withholding artificial nutrition and hydration
Survive the first month	11%-44%	Central apnea, airway malacia, aspiration, ductal dependent cardiac lesion, CHF, withholding artificial nutrition and hydration
Survive the third month	33%	Central apnea, aspiration, CHF
Survive 6 mo	3%-15%	Central apnea, aspiration, CHF
Survive the first year	2%-15%	Central apnea, aspiration, CHF, eventually pulmonary hypertension

Abbreviations: CHF, congestive heart failure; T13, trisomy 13; T18, trisomy 18.

intervention is to need to decide about feedings. Some of these infants eat fine in infancy, where others show no newborn interest or cannot orally feed. In this case, one needs to explore the withholding of artificial nutrition and hydration versus providing nasogastric or gastrostomy tube feedings.[56] An important fact is that there is no comparison trial between gastrostomy and nasogastric tube feedings showing that gastrostomy tubes are safer or better regarding aspiration or medical utilization. While they may provide parental and provider convenience, there are surgical risks, and perhaps a reasonable option should be to go home with nasogastric feedings with the option of converting to a gastrostomy tube months or even years down the line if the infant survives the other causes of death. When considering any future medical or surgical option, it is important to weigh the true benefits and risks, as these children may not get the typical benefit a child without the condition would (ie, cochlear implants or cleft palate repair for speech) and do commonly have more risk and longer recovery time with procedures. Importantly, the developmental abilities are what need to be considered in weighing the benefits and risks.

DEVELOPMENTAL ABILITIES

All families must be counseled that surviving infants with T13 or T18 will have profound neurodevelopmental disabilities.[10,57-59] These children will be dependent on their families for total care for the rest of their lives.[60] Developmental milestones achieved by these children are summarized in Table 6. There are reports of children who are able to smile, laugh, and interact with families through vocalizations and gestures, able to sit, roll, and sometimes stand or walk with assistance, and some may achieve self-feeding and toileting skills. The maximal developmental potential described in the literature approximates that of a 1-year old child. What is also described is a widening gap in milestone acquisition after 1 year of age.[57] In one study, the only 2 children who walked independently were confirmed or suspected mosaics[10]; another study reported a child with T13 who was independently walking at 9 years of age.[10,57] In the past decade, there is growing literature on the extent of interventions received by these infants and their impact on survival, but little information about quality of life of long-term survivors with the exception of parent reports and online blogs.[23] These have implicit selection biases. At the current time, we can presume that the neurodevelopmental challenges faced by these children will not change even if they survive longer than once thought and the burdens on the child and the family may in fact increase with time. Understanding the developmental outcomes of these children is paramount to counseling families that may be considering interventions for their child and what their perceived goals for the intervention are. For example, if a family is considering VSD closure for their child with T18 who is in heart failure not amenable to medical management so they can have more time with her, accepting that there are mortality and morbidity risks to the procedure, then this may be considered a reasonable option. However, if their goals are to have her heart "fixed" so she can be a "normal child," then these assumptions must be corrected before proceeding down the path to surgery.

PUTTING IT ALL TOGETHER FOR THE FAMILY FACED WITH A PRENATAL DIAGNOSIS

It is important to understand the aforementioned medical literature and its limitations when counseling families with these diagnoses. The first step is to meet, listen, and understand what the family knows of the condition and is looking for in order to determine how best to approach them with an appropriate use of the literature. As our case examples show, there are families that come in with a belief and acceptance that this is not an acceptable condition for their child and they wish to terminate to the family that comes in determined, perhaps defensive about discrimination, about wanting "everything done" for their infant. While it could be argued that informed consent requires each family to hear about every option, it would be harmful to each of these families to have to hear everything about options

TABLE 6. Developmental Outcomes of Survivors With Full T13 or T18[a]

Study	Methods	Chronological Age of Survivors	Maximum Developmental Potential Achieved	Able to
Bruns[61] (n = 22)	Parent survey	13-56 mo (1-5 y)	1-12 mo[b]	Smile, vocalize, sit with and without support, walk with assistance
Baty et al[57] (n = 62)	Medical record review and parent survey	1-232 mo (0-19 y)	3-15 mo (average 8 mo)	Smile and laugh, use 1-5 words, use 1-3 sounds, sit without support, reach for objects, crawl, some independent play, some follow commands, some feeding with assistance, some toileting skills, IQ unmeasurable or in the 20s for those measured[c]
Braddock et al[58] (n = 10)	Case history and participant interviews with videotaping speech/communication	15 y (mean)	6-12 mo	Communication via hand gestures No words
Janvier et al[23] (n = 64)	Parent survey	1 to >10 y of age	3-12 mo	Smile and laugh, roll, sit up, some independent play, 1-2 words, some walk with assistance (only 2 children walked independently), few eat independently

Abbreviations: T13, trisomy 13; T18, trisomy 18.

[a]*From references.*[23,57,58,61]

[b]*With the exception of 2 cases of mosaic T13 or T18 who are not included in this review.*

[c]*Only 1 child with T13 walked unassisted at the age of 9 years in this study.*

they have no interest in. A counseling strategy might be to inform them that families in these situations choose different things and then to let them guide you in how much of each of these choices and paths they would like to hear about.

For the family in case 1, which comes in wanting to terminate, counseling should include asking the family what they have been told, correct any myths or falsities, acknowledge that yes most of these infants die at all different times during the pregnancy, and that this can make the pregnancy difficult from the physical and psychological perspectives. Instead of discussing with them a detailed option for full resuscitation, appropriate counseling would be supporting their decision. The data suggesting a survival disadvantage are all met, and it could be supportive to share these with the family. In this case, a decision to terminate might be viewed simply as choosing a time to meet their infant. For this family, perhaps even using the word "lethal" could help them in their bereavement. Importantly, the parents may choose induction, delivery, and comfort care and can receive the same palliative care support any expectant woman might receive.

For family 2, which is opposed to termination, wants complete information but does not want to cause suffering, appropriate counseling might be to explore both HLHS repair and T13 outcomes. For this family, using the language of a life-limiting or life-threatening disorder is more palatable. As an example, sharing that palliative care is considered an option for the infant with HLHS and normal genetics because of its own morbidities could help the family understand why it is not recommended to be pursued in this situation. Importantly, their goals of not wanting suffering are best achieved then with a perinatal palliative care plan. This would then include supporting the pregnancy as time for memory making, planning mode of delivery depending on goals for live birth, and supportive medical care for the infant along with the spiritual and psychological support for the family.

For family 3, the medical situation is a female fetus who is only SGA, not IUGR, and may or may not have a cardiac condition that would require neonatal intervention. The family also is not expressing any preference. Appropriate counseling then would require making sure the family understands they have options ranging from termination to full resuscitation and neonatal care. While this fetus has risks of in utero demise and death during

the birthing process, neonatal outcomes vary on the basis of whether the fetal concern of coarctation is confirmed after birth and then on whether the family would want to proceed with cardiac intervention. While all the aforementioned information is important to share, an important part of this consultation would be to elicit from the family their hopes and goals so that the literature can help guide recommendations.

For family 4, the most important part of consultation is to try and build trust with the family that comes with assumptions and in a seemingly battle mode. Some of these families come in having gained trust in other parents through social media that may make it more difficult for the medical providers to gain rapport. Gaining their trust can only happen through genuine care of them by assessing their goals for their individual infant. There is no need to discuss termination as an option, for example. Because the data support that this fetus has more advantages for survival (female, only mild growth restriction, no neonatal surgical), we should certainly be able to support their desire to support their infant. Within this framework, they should still receive the information as a life-limiting or life-threatening disease with risks of fetal demise and stillbirth, causes of neonatal death, the risks of cardiac surgery when indicated, and, most importantly, how the genetic diagnosis cannot be ignored as it has implications on survival from causes of death other than cardiac, survival and recovery from surgery, and long-term neurodevelopmental outcomes. While some families may already know this information, others do come with a focus for cardiac surgery without really understanding all the other information. This can depend on the advocate they have met and what their goals and agenda might be.

In conclusion, we believe the following pearls are important to keep in mind:

- Families with a prenatal diagnosis of T13 or T18 have many choices depending on the stage of pregnancy—many families choose termination, but some decide to continue their pregnancy.
- All families and children deserve respectful communication and avoidance of language that devalues them or their infants irrespective of the choices they make about the pregnancy.
- When a pregnancy is continued, there is not only the chance of stillbirth or in utero demise but also the chance that they will meet their infant alive. Factors that may pose greater risk for in utero demise are male gender (for T18), significant growth restriction, and presence of multiple anomalies.

Summary of Recommendations for Practice and Research

What we know:	• When a pregnancy is continued, there is the chance of in utero demise at any time during the pregnancy, some risk of stillbirth yet chance at live birth, and a chance for discharge and longer term even when comfort care is chosen. • Factors that confer a survival advantage include female gender, higher birth weight, term gestation, absence of neonatal surgical anomalies, and medical stability (infants who do not require many interventions). • Children who receive surgical intervention can survive, but they have significant postoperative morbidity and mortality compared with infants without these diagnoses, and long-term survival may not change significantly. • Surviving infants will have severe to profound neurodevelopmental disabilities, and currently there are no data that show or suggest that receipt of interventions and longer survival changes these outcomes.
What needs to be studied:	• Studies that include parental voices all come from the smaller group of parents who desire more intervention, so hearing the voices of those who have chosen termination or comfort care and what language they appreciate need further study. • Studies on long-term parental mental health regarding choices made could provide information for prospective parents making decisions. • Studies on long-term family dynamics and coping could provide information for prospective parents making decisions. • Studies on healthcare utilization and cost.
What can we do today:	• Practitioners should aim to be knowledgeable and truthful about the obstetrical and neonatal outcomes as they enter into consultation. • Practitioners should provide respectful communication and avoid language that devalues parents or their infants. • Palliative care services should be provided for families that choose to pursue the path of comfort care, to assist with a focus on bonding with their child and minimizing suffering, and to help support the infants who survive to discharge home, perhaps even for weeks or months. • Palliative care services should be provided for families that choose to pursue the path of intervention to assist in supporting bonding, comfort, and decision making throughout the life span.

- For live-born infants, families can choose to pursue the path of comfort care, with a focus on bonding with their child, and minimizing suffering but not necessarily prolonging life. Even for families that choose comfort care, many infants can survive to discharge home, perhaps even for weeks or months depending on their gender, birth weight, and coexisting anomalies.
- For families that choose interventions, chances of survival to hospital discharge and later may be increased depending on individual risk factors.
- Children who receive surgical intervention can survive, but they have significant postoperative morbidity and mortality compared with infants without these diagnoses, and long-term survival may not change significantly.
- Surviving infants will have severe to profound neurodevelopmental disabilities, and currently there are no data that show or suggest that receipt of interventions and longer survival changes these outcomes; families' request for interventions must be balanced against their perceived expectations.

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