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An Infant With Trisomy 18 and a Ventricular Septal Defect

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abstract

Decisions for critically ill infants with trisomy 18 raise thorny issues about values, futility, the burdens of treatment, cost-effectiveness, and justice. We presented the case of an infant with trisomy 18 to 2 neonatologists with experience in clinical ethics, Annie Janvier and Felix Okah, and to a parent, Barbara Farlow. They do not agree about the right thing to do. *Pediatrics* 2011;127:754–759

CLINICAL ETHICS CASE REPORT: PART I

Infant Jones was born at 39 weeks' gestation with a prenatal diagnosis of trisomy 18 diagnosed by amniocentesis. At birth, he was noted to have a murmur and was subsequently diagnosed with large perimembranous ventricular septal defect (VSD). Electroencephalography revealed seizure activity. He was placed on phenobarbital. Ultrasound of his head showed prominent extra-axial fluid but no evidence of a bleed. The parents received counseling from the genetics department to inform them that most infants with trisomy 18 do not survive their first birthday. They were told that some infants survive longer but with severe cognitive impairments. The palliative care team offered support for the parents and discussed hospice care.

Should the parents be offered heart surgery to correct the VSD?

Annie Janvier

Every fetus or child with trisomy 18 is different; some die in utero, others cannot tolerate labor, others die at birth despite “maximal therapy,” and still others survive for years without intensive care. Many parents who receive a prenatal diagnosis of trisomy 18 choose to terminate their pregnancy. Parents who choose to continue the pregnancy may do so for different reasons. What were and are the parents' expectations and goals for their son? Did they expect him to die in utero? Did they oppose abortion but desire only comfort care at birth? If so, I would support their decision and not give an extensive menu of every possible intervention their son could have. If they did want to pursue life-prolonging interventions, I would explore their current understandings. What do they know about the VSD and different treatment options?

The discussion may be complex. VSD surgery does not usually need to be performed in the first weeks of life. Neonates with VSD are rarely in serious heart failure. Instead, infants with a VSD can usually be managed medically for at least a couple of weeks, if not months.

What are the risks of VSD surgery? For children without trisomy 18, the mortality rate is ~2% to 3%. The surgery, of course, requires intuba-

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ABBREVIATION

VSD—ventricular septal defect

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tion, cardiopulmonary bypass, surgery by an expert team, and admission to the ICU with gradual reduction in technologic intervention. Presumably, for a fragile child, the risks of surgery are higher, but we do not know how much higher. We need to realize—and communicate—the limits of our knowledge on this topic for children with trisomy 18. We do not really know whether VSD surgery in a stable but fragile infant with trisomy 18 actually increases or decreases his or her life expectancy and quality of life.

The alternatives to VSD surgery are medical treatment, other types of experimental closure of the VSD (such as the Amplatzer septal occluder), or comfort care. Most infants in this situation (eg, trisomy 18, large VSD, heart failure) will probably die with comfort care and purely symptomatic treatment, but even this is not certain (I have an ex-patient in this category who is now 4 years old). When heart surgery did not exist, many children without trisomy 18 with large VSDs survived, developed Eisenmenger syndrome, and eventually died of cor pulmonale in their teens, 20s, or 30s. Generally, children with trisomy 18 do not survive to be 20 to 30 years old. Thus, without surgery, this infant might well die of something other than heart failure. Surgery should be discussed with these parents, but they should be informed how uncertain we are about the benefits of surgical intervention.

Barbara Farlow

This family has had months to prepare for the birth of the infant. During that time, they have likely followed blogs or even met families with beautiful, happy, and intensely loved children with trisomy 18. From these contacts, they have also certainly heard about children with trisomy 18 who died young. Maybe they have heard about funeral services for these children

that are packed with grieving friends and community members. If so, they may have reasonably concluded that children with trisomy 18 can provide something essential and important to those who love them. The parents probably do not consider their child to be “a trisomy 18.” Instead, they probably think of him as a child with a serious medical condition for whom a personal assessment and plan of treatment should be developed. Many pediatricians think that offering VSD surgery to prolong the life of an infant with trisomy 18 is not in the best interest of the child or a prudent use of resources. But, to deny parents a choice about surgery on the basis of the doctor’s assessment that the child’s life will be short and of limited quality would be wrong.

Love, hope, and charity are esteemed qualities. The lives of these children are abundant in these qualities. Resources should be considered well spent if they prolong the life a child who lives comfortably and is loved intensely.

The option of surgery for this child should be treated like the option of surgery for any other child; that is, if there are physiologic risk factors that make surgery unlikely to be successful, that should be explained to the parents. If the risk of surgery is deemed unacceptable, surgery should not be offered. However, if there is a reasonable chance that the surgery will be successful, surgery should be offered. Parents should be counseled about the risks and benefits and be given the option of surgery or palliative care. The medical team should ensure that the parents are cognizant of the significant challenges ahead.

Felix Okah

Typically, when a diagnosis of trisomy 18 is made prenatally, as it was in this case, only a small minority (~20%) de-

cide to continue the pregnancy. Even without an abortion, two-thirds of fetuses with trisomy 18 die in utero.¹ Ninety percent of live-born infants die in the first year of life. The mortality rate is higher for boys than for girls. So, the odds are not good for this little boy.

I would need to know much more about the parents. Was the decision to continue the pregnancy based on a personal religious aversion to abortion—one that might not influence postbirth decisions about medical intervention—or was it acceptance of the fetus and infant despite the diagnosis? I would want to know the mother’s age, obstetric history, and family/social history. Do the parents have other children at home? What resources does this family have to support this child, who will have severe developmental problems if he survives beyond infancy? Infant Jones’ parents may be willing to accept limited functioning in their child if he survives. It will be important to explore how that decision will affect the financial and emotional resources of the family that will be available to their other children.

My discussion with the parents would focus on the dismal facts, particularly the fact that his male gender and the electroencephalographic evidence of seizures both suggest a particularly poor prognosis. His VSD, on the other hand, probably does not influence his chance of survival.

From an ethical perspective, I would keep in mind that treatment of Infant Jones’ heart defect will not likely improve his chances for long-term survival. In that sense, it is a burdensome and costly treatment that offers little, if any, benefit.

Therefore, I should not offer Infant Jones’ parents heart surgery to correct the VSD. I would support the recommendations for palliative and hos-

pice care and encourage the parents to go that route.

CLINICAL ETHICS CASE REPORT: PART II

When medical management of VSD was no longer adequate to control the symptoms of heart failure, Infant Jones' parents wanted to pursue surgical intervention if it would prolong and improve his quality of life. The doctors tried to talk them out of it. The parents said that they understood that their infant might not survive the surgery but that, if the doctors would not operate, they wanted to be transferred to another hospital and other doctors who would. After a multidisciplinary care conference, the medical and surgical team agreed to the surgery.

Infant Jones had a rocky postoperative course that was complicated by cardiac arrest and arrhythmia and required resuscitation and reopening of the chest. Electroencephalography performed after these events revealed generalized slowing with poor neurologic prognosis. An MRI of his brain showed marked generalized cerebral volume loss and encephalomalacia consistent with hypoxic brain injury. The patient was extubated on two occasions. Both times, he quickly went into respiratory failure and was reintubated. During this critical period, the ICU team tried to convince the parents to withdraw life support. The parents would not agree. They were constantly at his bedside, praying for his recovery.

Is further care futile? Should he be extubated over the objections of his parents?

Felix Okah

The parents wanted surgical repair of the VSD for their son if it would prolong his life. There ought to have been discussion about what "prolongation of life" means in this context and what costs are worth the risk of the defined

"prolongation of life." Survival beyond the first year of life for this child is highly unlikely. Moreover, the available evidence would suggest that "significant" prolongation of life from surgery is unlikely, because survival statistics of infants with trisomy 18 with and without congenital heart defects are similar.

Given their wish to preserve life, it surprises me that the parents were not deterred by the possibility of a fatal outcome during the surgery. This position seems *prima facie* to be at odds with the goal of prolonging life. What, then, is the underlying motivation for seeking "risky" surgery? Might guilt from bringing a child with trisomy 18 into the world be driving their decisions to "do everything possible?"

The decision by the physicians to perform surgery rather than transfer the patient to a different institution raises some ethical questions. They had deemed surgery to be unnecessary and dangerous, so was the decision to perform it financial or altruistic? Were the physicians concerned about the potential lost income or about lower skills of the alternative group of physicians?

It is not surprising that the postoperative course was problematic. The ensuing complications have significantly diminished the prospects of short-term survival and increased the chances of severe additional neurocognitive impairment in the unlikely event of survival.

The parents' refusal to withdraw life support is understandable in the context of wanting everything done and their earlier ability to reverse the decisions of the medical team. It is important at this point to explore the parents' motivation and reasons for their position. Do they still represent the best interests of their child, or are they protecting their own personal/emotional interests?

Should life-support be withdrawn over the objection of these parents? The answer to this question requires knowing what is in the best interest of the child and who determines it. I suspect that parents' ability to represent their child's best interests (autonomy) sometimes may be compromised emotionally. All too often, people recount the stories of those families who had wonderful and happy memories of the short lives of their children with this problem. There is no study to show how those children really felt, only subjective parental reports and equally inadequate information about the families with limited psychosocial/economic resources or those who struggled with the guilt of prolonging a life that was perceived to have endured much. Who defines pain and suffering for the sick and developmentally limited patient? Even cognitively intact persons view situations differently and exhibit significant differences in what will be tolerated for the sake of survival.

It is difficult to see how artificially supporting life under these circumstances can be in the best interest of the child or society. It is unfortunate that the child is not in a position to articulate that, and those who are making these decisions (parents and health care providers) possess biases from their own life experiences. Furthermore, such invasive and intensive support of life may be potentially harmful, because it prolongs suffering and wastes health care resources. Nonetheless, even under such difficult circumstances, keeping channels of communication open often results in collaboration between parents and the medical team and a solution that is mutually acceptable.

Barbara Farlow

Consent rights of loving and committed parents such as these should not be removed.

These parents are apparently willing to make extreme sacrifices in their lives to raise a child with severe disabilities. There is no indication that the parents are neglectful or actually want their child to suffer discomfort for no benefit. Such disagreements often reflect poor communication and lack of trust.

Many parents who have infants with trisomies lose trust because the staff treats their infant as a “trisomy 18” rather than as a unique child. Once trust is lost, it is difficult to rebuild. In parent groups, one often hears “doctors were wrong” stories.

These parents will live for the rest of their lives with the events that occur in the ensuing days or weeks. The aftermath of insensitivity can destroy families. I believe this situation needs time and compassionate communication to reach a decision that is acceptable to them all.

Annie Janvier

Futility can have multiple definitions. Were there discussions before the surgery about the expectations of the parents? If the parents wanted VSD surgery for their son to be on his high school's soccer team, the VSD surgery was futile in the first place. If the parents went on the Internet and saw (or met) other families who had faced similar challenges, with children who have survived and seem happy despite their severe disability, they might want the same for their child.

Why are the parents refusing to withdraw life support? Perhaps they do not trust physicians. They may have been told “all infants with trisomy 18 die in the first year no matter what” but then may have discovered that that is not true. They probably think continuing ongoing intensive care is in their infant's best interest. Perhaps their goal is to have the infant come home for a short time and die there rather than in

the ICU. That goal does not seem impossible to reach.

Infant Jones is not actively dying. It is difficult to predict whether he will survive with intensive care. The prognostic value of an electroencephalogram and a scan are uncertain, because there have been no studies about imaging “sick” and “less sick” infants with trisomy 18. All children with trisomy 18 have serious mental and physical disabilities. Infant Jones' problems may be more serious than usual, which may be what the team is focusing on. They probably think discontinuing the respirator is in Infant Jones' best interest and that further intensive care is “futile” because it is not “worth it.” They may think that death is a better outcome than the disabled life they predict for him. Doctors are more likely to feel this way than parents, who generally prefer life with disability over death.²

On the other hand, Infant Jones should not only serve his parents' interests. If the parents want to continue the ventilator because their son is the only element in their life keeping them together, for example, then the child's interest needs to take priority. In that case, his parents should be helped to refocus on him and his interests.

As a pragmatic matter, removing the respirator against the parents' wishes is easy to write about but hard to do without feeling brutal and cruel.

When infants do not have trisomy 18 but have a prognosis similar to Infant Jones, we tend to defer to parents who do not wish to withdraw the respirator, even if we disagree with their choices. For example, for an asphyxiated infant in Sarnat stage 3, we would accept to continue intensive care and generally empathize with the parents. I have never heard anybody suggest withdrawing the respirator against parents' wishes in this case. We should do the same thing in Infant Jones's case. Parents do not grieve

less because their infant has an extra chromosome.

Infant Jones might die despite intensive care in the next few days. The most important thing now is to make sure that he is not in pain or uncomfortable.

CLINICAL ETHICS CASE REPORT: PART III

After much discussion, the parents agreed that Infant Jones should be extubated but not reintubated if respiratory failure ensued. This time, he was successfully extubated. Two weeks later, he was discharged to his home with palliative care support. He remained stable at home but continued to have occasional seizures and feeding problems. At 18 months of age, he got a gastric feeding tube and a tracheostomy and returned home. He has been gaining weight well. His parents are grateful for the care that he received.

Felix Okah

It is always a good thing when the family and medical team are on the same page. The parent's decision reflects a growing understanding of their child's condition and an acceptance of the fact that they may lose him at any time. The latter point is reflected in their decision to not reintubate and an acceptance of palliative care support on going home. It is not uncommon for patients who are supported long enough to develop independent respiratory capabilities that significantly change the psychosocial, but not clinical, circumstances. The survival of this child up to this point does not validate any one clinical or philosophical position and should not engender unrealistic expectations in the care providers.

The family should be commended for the care they have provided, given that boys rarely live that long, but helped to understand how the ongoing seizures significantly worsen the child's al-

ready limited neurodevelopmental potential. They ought also to be engaged in recurring discussions about the changing needs and challenges associated with growth and what care providers may or may not do in the event of significant changes in health status such as cardiorespiratory failure for any reason. These discussions should be entered into his medical records, and the decisions should be available to parents and other care providers in a written document.

This case captures the dilemma faced by parents and health care providers when dealing with situations that historically have a poor outcome and for which limited evidence exists for what the “best” path of care should be. Both parents and health care providers need to figure out what might be in the best interest of the child rather than that of the parents and the health care system. During these discussions, it is apparent that there is no consensus for what quality of life means for the child. If our experience with cognitively intact persons is anything to go by, quality of life will differ with each child and ostensibly could differ from the parents’ and health care providers’ understanding of an “acceptable” quality of life for the child. There is obviously no easy answer, and as with most human interactions, honest respectful communication will be the route for providing a “best” outcome with the individual case.

Annie Janvier

Most infants with trisomy 18 die. Doctors and nurses get used to it; we know the routine, the quiet moves, the memory box to fill with pieces of hair and footprints, and the ritual with the final physical examination. Parents do not get used to it. They have to live with the decisions that they make and their memories forever. So, I generally defer to the parents’ wishes unless I think that their requests will cause harm for the infant.

In this case, that was not so. It did not sound like Infant Jones’ existence was full of pain and suffering. Pain is easy to treat in an intensive care setting.

The only reservation I would have about this case is one of resource allocation. Physicians are often stuck in the conflicting role of doing the best for patients while using societies’ resources judiciously. These two can be conflicting and bring about value judgments. When one considers resource allocation, the principle of justice demands that similar patients be treated similarly. Patients who are expected to die or have severe neurodevelopmental sequelae (similar to those that children with trisomy 18 have) should all be treated in the same manner. So, to decide how to treat Infant Jones, we need to look at how we treat patients with severe Alzheimer disease, multiple strokes, asphyxia, or brain trauma. These cases are frequent. Generally, we defer to families in these cases. On the other hand, a case like Infant Jones’ is rare. It would not be just in our health care system to impose hospital policies and restrictions specifically for children with trisomy 18 when policies for other patients with similar severe limitations are nonexistent. If these policies existed for older patients, than the resource-allocation question would be a fair one to consider. Infant Jones is not likely to bankrupt our fragile health care budget, but the medical ICUs are.

Barbara Farlow

I believe the parents have revealed that they are rational and realistic by agreeing to palliative care support, given the expected life span of a child with trisomy 18.

It is noteworthy that after spending more than a year with a complex child, the parents sought treatment to better nourish their son and help him to breathe. The medical system accom-

modated this request, which reveals that the parents were realistic about their son’s limitations from the start, because they have clearly satisfied their commitment to care for him and even taken active steps that will make him stronger and likely live longer. Therefore, one must question the quality of the experience of the child’s life and conclude it to be much more favorable than the medical literature might suggest. It is also noteworthy that despite being off to a rocky start, it seems that Infant Jones has been quite healthy and has not spent a lot of time in the hospital.

This child had everything going against him. He seemed to be a “bad case” of trisomy 18, and the situation looked grim after the cardiac surgery. Yet, he survived, is doing reasonably well, and is clearly loved by his family. He has not been an excessive burden to the health care system. It is interesting that the doctors turned out to be wrong about his chance of survival and the parents turned out to be right, which gives credence to the often-heard suggestion that futility assessments might be a self-fulfilling prophecy. Perhaps doctors should reconsider their long-held belief that trisomy 18 is synonymous with medical futility.

EDITOR’S COMMENTS

Cases of trisomy 13 or 18 highlight an area of deep disagreement. Most parents would not want an infant with these conditions. Many of them are grateful for the prenatal diagnosis that allows them to terminate an affected pregnancy. Others choose a different course and either forego prenatal diagnosis or, as in this case, use the information to make decisions about obstetric and neonatal care. Doctors are similarly deeply divided; some feel that aggressive treatment is futile and should not be offered, and

others defer to parents. Drs Okah and Janvier reflect this professional disagreement. Ms Farlow speaks for the parents who come down on the side of treatment. These cases raise the most fundamental questions about the

value of life, the meaning of personhood, and the limits of parental and professional authority. Deference to parents is generally the right course unless the infant is clearly suffering from ongoing treatment that is un-

likely to be of benefit. The doctors in this case did the right thing: they worked to find common ground. As often happens, the infant surprised everybody.

—John Lantos, Section Editor

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SMARTPHONES AND PRIVACY: *Whenever I log onto a website, download a new application for my smartphone, or get a statement in the mail from my bank, I am irked by the claims that the companies are busy protecting my “privacy”. Almost everyone claims strong privacy protection for the consumer. However, it would appear the strong protection means different things to different people or organizations. Take for instance applications for smartphones. As reported in The Wall Street Journal (December 17, 2010: Tech), downloaded applications are sharing a wealth of personal information about the user with the outside world. To learn what information was shared, investigators from The Wall Street Journal intercepted and recorded the data transmitted from 50 commonly used iPhone applications and 50 popular applications using Google’s Android operating system. More than half of all applications transmitted the phone’s unique device ID number, which cannot be changed or blocked, to other companies. Almost half transmitted the phone’s location. Fewer sent age, gender, and other personal information to outsiders. All this was without users’ awareness or consent. One popular text messaging application for the iPhone sent the phone’s unique ID number to eight advertising companies and the phone’s zip code, along with the user’s age and gender, to two. A popular music application common to both user platforms sent age, gender, location and phone identifiers to various advertising networks. Unfortunately, while there are ways to avoid tracking from a personal computer, opting out of tracking from a smartphone is almost impossible. Consumers don’t have many protections. Almost half the applications don’t even have a written privacy statement on their website or within their application. The companies getting information bluntly state they monitor smartphone user data as much as possible. The companies want to know which applications are downloaded and how much time is spent on an application, and the depth to which the application is explored. As the two most popular platforms for smartphones, the iPhone and Android, also generate the most money from ad revenue, setting rules for transmission of personal data is likely to be inherently challenging. What can consumers do? Alas, not much at this time. Just know that while you may be storing lots of private information on your smartphone, your phone is not a silent partner.*

Noted by WVR, MD

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