

Newborns with Trisomy 18: To Treat or not to Treat? Have Times Changed?

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12:00 noon – 1:30 pm

Introduction of the Case: Dr. Ruth L. Fischbach

Today, we confront a particularly troubling ethical dilemma -- how best to care for a newborn with Trisomy 18, which is also known as Edwards Syndrome. You will soon hear more about this syndrome from our presenter, Dr. George Hardart. But for now, just know that Trisomy 18 occurs when a baby is born with three **Number 18** chromosomes instead of the normal two chromosomes, one from each parent. Babies with this genetic condition in the majority of cases – but importantly **NOT ALL THE CASES** – are born with life threatening abnormalities and have an extremely ominous prognosis.

The issues often rest on whether or not to provide extensive surgical procedures and invasive life-sustaining measures that may inflict pain and suffering onto the newborn, even though the newborn may have a short life expectancy. Would comfort care measures be more humane? Who makes the decision? The parents? The physicians? On what basis should the decision be made?

Today's presentation and discussion will be especially informative. You will be presented with what we know about Trisomy 18, and the historical decisions for offering care that were made. You may be surprised to learn that *The Lancet*, one of the leading medical journals, as recently as 2009, stated in an article: and I quote -- "a newborn infant with Trisomy 18 should be considered as a patient with a hopeless outlook who ought not to be subjected to invasive procedures" "unquote. {See the reference: Avoidance of emergency surgery in newborn infants with Trisomy 18. *The Lancet*, Volume 339, Issue 8798, Pages 913-915}

Yet we know of cases where individuals with Trisomy 18 are living into their 20s and beyond and appear normal.

So today, you will get to explore the medical and societal factors that shape our current approach to care of these infants. Dr. Hardart will present two infants who were born at Children's Hospital with Trisomy 18 and offer a framework for considering the ethical justifications for competing treatment strategies. And along the way, you will be invited to offer your opinions on how decisions are made.

I would like to welcome some important guests today who have come from far distances to tell us about their much loved children, born with Trisomy 18.

We are privileged to have Dr. George Hardart here to present the medical and ethical elements of the case.

Presentation of the Case: Dr. George Hardart

Dr. Hardart's event proved to be outstanding and was one of our most well received events to date - 280 people registered and attended! Because we were going to discuss two cases of Trisomy 18, our notice was picked up by several parent groups and was carried on the Internet, resulting in phone calls from out-of-state and guests coming from as far away as Minnesota. It was very important for the audience to hear the voices of the mothers and the doctors.

There are three forms of Trisomy 18: complete, Mosaic 5% (huge range of clinical features/phenotype), and partial (only a part of chromosome 18 is triplicated). Traditionally, clinical management had categorized this disorder as a lethal anomaly and only palliative care was suggested. Today, however, it is more common for families to request corrective treatment for these children at birth. With the advancement of medical care through new technologies (surgery, tracheostomies, physical therapy), these Trisomy 18 children are able to hit milestones that a one-year old might hit but at extremely delayed rates (usually around the age of three years). Some have language skills that enable them to follow simple commands and play independently. Even though the science has changed and allowed progress, Dr. Hardart questioned whether the ethics had changed. The decision-making standards, the amount of information disclosed, and the rights of persons with disabilities have all evolved. Parental autonomy is the dominant ethic of medical decision-making and parents are told the information that is regarded as the "reasonable person" standard. The Internet and support groups have allowed many families to gain more information and an empowered voice.

In the first case, Dr Hardart presented a baby boy born with Trisomy 18 at another hospital whose parents requested full treatment and surgery for their son and so was brought to CHONY. The baby required a gastrostomy with tube feeding at birth. He stayed in the NICU for two months until he was ready for discharge. He was treated with two medications for congestive heart failure. The baby required 16 hours of home nursing, and could only be fed through the gastrostomy tube. At 9 months, the baby showed severe pulmonary hypertension due to his heart disease. For the next two years the baby was hospitalized many times for pneumonia and asthma, but was never *critically ill*. Upon reaching 3 years, he still could not speak but was able to communicate his feelings in other ways. He shook his toys, rolled to move, and reached for things he wanted. In his final episode, he developed a severe cold that led to asthma, and he was brought back to the hospital. Eventually he needed to be intubated in the PICU. After 3 weeks, he experienced a cardiac arrest and was unable to be resuscitated.

In the second case, Dr. Hardart presented a couple who was given a prenatal

diagnosis at 20 weeks of Trisomy 18. The team involved discussed all the treatments available for the baby, but only recommended comfort care at birth. The family agreed to only comfort care at birth and signed a DNR order. At birth the baby was found to have a severe heart condition requiring either corrective surgery or medication (Prostaglandin E1) that would make it impossible for the baby to leave the NICU. The parents, seeing their baby, removed the DNR order the following day and requested surgery for the baby. The baby had the heart surgery at five weeks and then stayed in the NICU for two months coping with many complications from the surgery as well as infections and effusions. At three months, the baby was discharged but was only able to be at home for two weeks, before having to be readmitted to the hospital. At this point, the baby stayed in the hospital for another six weeks where he needed continued positive airway pressure (CPAP), suctioning, and oxygen. At four months of age, the baby was discharged once again. Unfortunately, the baby suffered a cardiac arrest nine days later and died.

Dr. Hardart posed many questions for the audience. He asked in these cases whether Prostaglandin E1 should have been recommended for the first baby, a treatment that can prolong life, but not ultimately correct the problems. He discussed whether corrective surgery and full life-sustaining support should be performed on these babies, and how one can justify these decisions. Should the doctors discuss all available treatment options with the parents, or can non-disclosure be justified? Although a treatment may be disclosed, does a doctor need to offer it?

There are justifications that support a doctor not disclosing certain medical treatments available to the baby. The doctors can err on the side of paternalism towards the babies and their family by not disclosing. The babies may be able to benefit from this decision because they are not forced to cope with the burdens of invasive treatment. Parents are often not able to see that these surgeries are very risky and difficult for the baby because they are most concerned with saving their baby from dying, despite the cost. Parents can benefit from non-disclosure because they then do not have to deal with making an extremely difficult decision of prolonging life for their child at a dangerous and often painful cost, versus allowing their baby to die more peacefully but sooner.

These reasons for non-disclosure raise many troubling issues. When doctors do not disclose all available treatment, they are violating the rights of the parents to decide, which diminishes the parents' autonomy. The parents are being stripped of their right to free choice. If the parents do find out about an available treatment that the doctors have not disclosed, the relationship between the doctors and the parents are seriously affected as the doctor then becomes an untrustworthy source of information. This makes decision-making for the family much harder because they feel they cannot trust the doctor.

Many believe that it is in the best interest of the baby to not offer treatment to the baby who is born with Trisomy 18. As a doctor has pledged to uphold his/her duty and to avoid nonmaleficence, it is considered only right to NOT treat the baby. The burdens of

the surgery seem surely to outweigh any benefits from treatment. The babies most often cannot live past a year of life, even with aggressive life sustaining treatment, due to heart, lung, and gastrointestinal complications. Therefore, any treatment seems ultimately futile. The treatments available only seem to harm the baby and do not necessarily improve the quality of life, but cause the baby to suffer. While it is important to listen to the family and provide care, what matters most is the child and the burden placed on him, which is often very great and unpredictable. It is important to think about long-term consequences where parents may not necessarily have the financial capacity of caring for all the baby's medical needs, and the burdens associated with ICU care. Arguments are raised that hospital resources could be better allocated than in providing expensive and complex treatment for these babies. The hospital could use the money to provide more benefit elsewhere. Additionally, the babies are taking beds in the hospital that could be better used by another sick patient, with a better chance of survival.

On the other hand, Dr. Hardart also explained the reasons behind treating a patient diagnosed with Trisomy 18. While there are many doctors who may feel that treatment goes against their duty to avoid nonmaleficence, many doctors feel that withholding treatment goes against their duty to uphold beneficence. Treatment can be in the best interests of the baby as well as future babies to come who are diagnosed with this condition. Many of the complications present with Trisomy 18 are treatable in healthy babies. Although doctors may feel that these babies and children have a low quality of life, data have shown that doctors in general often rate quality of life of patients living with disabilities much lower than the patients and their families do.

Several mothers in the audience came from out-of-state to testify that they had children born with Trisomy 18, who were now 10+ years old, so it is wrong to write these children off. One mother explained that no one can predict what will happen to the babies. Whereas the parents have to live with their decision, doctors can move on. As the parents have found, many cases do not turn out the way the doctors predict, and some babies live well beyond their expected lifetime. Therefore, it is essential that doctors treat each case individually.

Parents are thus best at determining what is in the best interest of their baby, and their autonomy, as parents, needs to be recognized. An out-of-state pediatrician stated strongly that efforts should be made to follow what the parents want (palliative care or more likely, all-out aggressive care including surgery if required). He stated that we need to ensure that the parents are NOT filled with guilt if the baby dies – did I do everything, did I do enough?

Dr. Hardart concluded his presentation by explaining there are many lessons to be learned from these cases. Any policies that are absolute and deny the possibility that some patients with Trisomy 18 can benefit from cardiac surgery should be rejected. Each case should be individualized based on the available and appropriate options considering the benefits and burdens that may arise from treatments. At the same time, any policy that denies that certain treatments for sustaining life for the patients will be

too burdensome should also be rejected. Hospitals should have policies that address requests for inappropriate medical treatment. Therefore, hospitals should not be controlled by either of the extremes - for or against treatment. Every treatment plan must be individualized for each patient. It is important for doctors to fully disclose available treatments to the family; otherwise people are not able to make informed decisions. It is important that doctors make sure that they are always empowered to fully advocate for the patient.

One neonatologist ended by adding a word of caution. While it is important to support the parents so they are not filled with guilt should their baby die, he feels strongly that as a pediatrician, his duties and obligations always lie with treating the child with compassion, to make sure that care is given that does not lead to additional suffering, especially with babies who often have such a poor prognosis.

Both of these cases raised complex ethical dilemmas. In each case, compassionate care can always be provided.