

A DIFFERENT VIEW

More than a diagnosis

Pauline Thiele (liam194@gmail.com.au)¹, Siri Fuglem Berg², Barbara Farlow³

1.School of Nursing and Midwifery, Peninsula Campus, Monash University (gap year), Frankston, Victoria, Australia

2.Medical Doctor (sabbatical), Gjøvik, Norway

3.School of Health Policy and Management, York University, Toronto, ON, Canada

Correspondence

Pauline Thiele, 126 Barkers Road, Main Ridge, Victoria 3928, Australia.

Tel: +61359896420 |
E-mail: liam194@gmail.com.au

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Three women speak of and analyse their abandonment by healthcare professionals in the past five years following their foetus' diagnosis of a genetic disorder during pregnancy.

As educated women, we were bewildered by our medical care, or lack thereof. Following prenatal diagnostics of trisomy 13 or 18, we chose to continue our pregnancies. We understood that our babies would most likely die in utero or soon after birth.

To enable ourselves to make informed choices, we sought information from the internet. Here, it was discovered that some babies with these trisomies can live contented, comfortable lives for many years. We considered this information as well as the anomalies of our unborn and planned accordingly.

We did not want our babies to receive nonbeneficial, burdensome interventions, nor did we want them to suffer. Our hopes were not for a miracle but were much simpler: to meet and hold our babies and to be a family. Although our children, choices and outcomes are unique, we all hoped for time with our babies. We also all wished to be able to make informed decisions that respected the dignity of the fragile lives we carried.

Articles in the series A Different View are edited by William Meadow (wlm1@uchicago.edu). We encourage you to offer your own different view either in response to A Different View you do not fully agree with, or on an unrelated topic. Send your article to Dr. Meadow (wlm1@uchicago.edu).

IN UTERO

Pauline, mother of Liam (trisomy 18)

'Your baby isn't going to live', my obstetrician commented as I sat behind the steering wheel. Although I felt comfortable with him, he supplied me with no information about trisomy 18.

At my second ultrasound I found myself panicking over my lack of specialist support. Liam had numerous medical complications and no one had spoken to me about his care. Sadly, it was my obstetrician's lack of referral that left me feeling professionally abandoned.

Using my initiative I eventually (at 33 weeks gestation) gained the support of a paediatrician and palliative care physician. However, the hospital where they worked informed us that Liam could not be born there because of the possible use of morphine. Before finally determining where our son would be born, Liam died in utero. Cruelties at the hospital continued though when an obstetric resident performed a scan to detect a heart-beat and heartlessly pointed to the monitor and exclaimed, "See?! No heart beat!"

I am grateful to have been supported by two physicians willing to fight for us and our son's right to be born. How fortunate I feel to have been able to cradle my son, whisper words of love to him, kiss him and say goodbye.

THE NEONATAL PERIOD

Siri, mother of Evy (trisomy 18)

As an anaesthesiologist, I know about the risks of C-sections but elected for one in case of fetal distress. My request for this and fetal monitoring were denied. Evy had trisomy 18 and was born limp and blue. Ignoring the disparaging advice of midwives, my husband (also an anaesthesiologist) bagged Evy back to life.

We held and kissed Evy, so gorgeous with pitch black hair and a sweet face. The neonatologist regarded her as she lay asleep and content in her brother's arms, making sucking movements with her tongue. He urged us to remove the CPAP, saying it was futile. Futile? Her oxygen saturations were excellent; the CPAP was effective. Every minute with Evy was a gift.

No echo, blood tests or other examinations were performed. Evy was given no chance. We even had to fight for palliative care. The hospital welcomed the death of such infants sooner rather than later.

Evy lived for three days, the best days of our family's life. Could Evy have lived longer with simple interventions? Our grief is deepened by the pain of this uncertainty.

Evy's life was short, but it was valuable and purposeful – and certainly not futile.

THE INFANT PERIOD

Barbara, mother of Annie (trisomy 13)

We knew that eventually we would need to choose between palliative care or interventions, just as for any child with a serious diagnosis. Annie had trisomy 13 but without the common brain and heart anomalies. She thrived at home; smiling and needing only a small volume of oxygen. Respiratory distress led to an urgent trip to the children's hospital at 80 days of age.

Before a diagnosis was made, we were asked twice to consent to a DNR order. We responded that we needed to understand the problem and to make an informed decision. Did treatment exist that would prolong Annie's life and allow her to live comfortably? We didn't want her to live on machines. The doctor seemed flummoxed by our position.

The next day, we were told that Annie needed a tracheoplasty, surgery that she would not likely survive. Soon after, she declined rapidly and unexpectedly, forcing us to make decisions quickly. Based on what were told, the choice was clear; we declined intubation and Annie died.

We were devastated to later discover that a DNR had been ordered before we had consented. The Coroner's Paediatric Committee declared the care provided was "not appropriate" and the surgery described to us was "by no means certain." To this day, we do not know if Annie should have died that day.

We sought support and information to make the best decisions for our babies. Regrettably our carefully made, reasonable goals were met with opposition or disrespected, compounding our grief and diminishing the joy of the brief time we had with our children. The medical care we requested was not selfish, nor without benefit to our children. Unfortunately, we discovered that, for some healthcare professionals, the standards of 'respect' and 'shared decision-making' are trumped by a 'one-size-fits-all' approach for trisomy babies. Providers who were empathetic became part of the precious memories we hold of our children's lives.

The medical literature provides guidelines and reviewed articles, some of which recommends withholding care to children with trisomy 13 and 18. Interestingly, the only basis for such a recommendation is an assumption that disability relates to a poor quality of life (1). It is disturbing to note that some providers declare that children with trisomy 18 have an absent 'achievement of human and social good' (2). In fact, recent literature based on parental perspectives and personal observation of physicians reveals that surviving children interact with and enrich their families lives (1,3). Such inconsistencies raise important questions about the development of policies and guidelines based on unilateral judgements about QOL. Medical literature offers conflicting advice – from withholding/withdrawing treatment, to only palliative care or to active care in some situations. Hence, it is no wonder that care for such babies and their families varies so greatly; from high quality to substandard care.

We acknowledge that physicians are trained to cure patients and to alleviate suffering. There is, of course, no cure for trisomy 13 or 18. One might ask if death is easier for the families than caring for a severely disabled child. It may be surprising but publications reveal a different truth, with parents of these children overwhelmingly describing their lives as deeply meaningful, positive and transformative. Regarding severely disabled neonates, research has revealed that neonatal providers rate quality of life lower than do parents, practitioners often with a preference for death over life with disability.

A thorough reading of the reviewed literature reveals some very strongly paternalistic views. It is encouraging to note, however, that other critiqued medical literature provides excellent care measures in the management of trisomy 13 and 18:

- 1 Prenatal diagnosis should include discussion of all options/outcomes including termination, live birth with palliation and live birth with active care.

- 2 Avoid inappropriate language and language that assumes outcome, such as 'vegetable', 'lethal' and 'incompatible with life'. With survival exceeding 8%, assumed universal lethality is wrong.
- 3 If you feel you are unable to support such families, find another healthcare professional who can.
- 4 Provide adequate information so parents are able to make an informed choice.
- 5 Each child is unique, decision-making must be individualized.
- 6 Treat parents with respect and empathy. Respect their choice, whether it be comfort care or interventions (3).
- 7 Care should be focused on the symptoms, not the diagnosis.
- 8 Do not rush into withdrawing treatment simply because of the diagnosis, allow the neonate time to 'declare' himself or herself.
- 9 Allowing surgery may make the baby more comfortable and easier to look after, perhaps giving the parents much valued time at home with their baby.

A prenatal diagnosis of trisomy 13 or 18 is a life-changing event for parents, regardless of the decisions made. This

Editor's Comments

I write this commentary alone, although I have many colleagues who would gladly share my views. I find the article by Thiele P et al. one of the most powerful and appalling we have ever published. I would have thought that the behaviour of the healthcare professionals was commonplace thirty years ago and nonexistent now. It appears I am wrong.

The authors have listed many specific ways in which doctors (paediatricians and obstetricians) and nurses (both neo and ob) can and should behave when confronted with a woman carrying an infant with T13 or T18 (and by extension a large number of other analogous genetic traits). They are terrific suggestions.

But more simply, these women are our patients, mothers or mothers to be experiencing the awful pain of an expected

difficult journey can be improved and enriched when providers try to see with our eyes and feel with our hearts. We, as any parent, love our children unreservedly. Providers need to understand that our children are unique and their lives are cherished; they are so much more than a diagnosis.

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pregnancy gone terribly awry. They need nothing more – nor less – than our help, our compassion, our assistance, our attendance in any decisions they choose to make about sustaining or comforting the short (or not so short) life of their child.

Anything less is unconscionable.

William Meadow (wlm1@uchicago.edu)
Pediatrics University of Chicago Chicago IL USA