BMJ

BMJ 2011;343:d3625 doi: 10.1136/bmj.d3625

PRACTICE

A PATIENT'S JOURNEY

Destined to die

At 18 weeks' gestation Pauline Thiele's baby was diagnosed with trisomy 18, a usually lethal syndrome. Her problems began when she chose not to terminate the pregnancy

Pauline Thiele *patient*[1], Simon Blair *paediatrician*[2]

[1]Red Hill South, VIC 3937, Australia ; [2]53 Cranbourne Road, Frankston, VIC 3199, Australia

This is one of a series of occasional articles by patients about their experiences that offer lessons to doctors. The *BMJ* welcomes contributions to the series. Please contact Peter Lapsley (plapsley@bmj.com) for guidance.

Parked on the side of the road I listened as my obstetrician told me over the phone that my serum screen result showed an increased risk of trisomy 18. My world felt like it had been turned upside down and I listened in stunned silence as he tentatively told me that our 18 week old baby was not going to live.

I searched the internet and downloaded reams of information. The next day an emergency amniocentesis was performed. I watched my baby on the monitor and heard the fast rhythm of the heart beat. The sonologist explained that the news wasn't good, explaining that, as well as trisomy 18, the baby also had a lumbar spina bifida. Any glimmer of hope that I had been holding onto disappeared.

The weekend was tumultuous and I struggled with the realisation that the dream of raising this child would never happen. The baby moved and I welcomed the movement, and I knew that I loved this little one and would not take his or her life with a termination.

Unfortunately the communication from my family, although well intended, did not help; with each foisting their personal opinions on us, with the exception of my mother, who supported me unconditionally. Throughout my pregnancy four friends were to provide me with a great deal of emotional strength. My greatest strength, however, came from within and I was thankful for this inner strength that my life's experiences had supplied. With sadness I watched my husband struggle with the impending death of our child.

My obstetrician confirmed the diagnosis and informed us that we were to have a son, whom we named Liam. When offered the option of termination, we declined. Telling him that I had retrieved information from the internet he acknowledged that he could provide no further information. Without any referral to genetic counselling or paediatrics my decisions were based solely on information retrieved from the internet and I was left feeling abandoned by the medical profession.

Throughout my pregnancy, care was shared between the obstetrician and the general practitioner. On hearing of Liam's diagnosis my general practitioner called immediately and offered her absolute support.

One day I stopped at the hospital where I was booked for delivery and went to the midwifery ward. When my file was pulled from the shelf I was shocked to discover that the ward knew nothing of Liam's impending birth and there was very little documentation. Although I had assumed that the ward would now know, I was still to receive no professional backing.

As a registered nurse I found it reasonably easy to understand the information about trisomy 18, but my textbooks provided inadequate information about spina bifida, and suddenly at 31 weeks' gestation I felt I needed to know how to care for Liam if he was born alive. Unable to get an appointment with a specialist, I turned to the internet and sent off several emails, and soon afterwards correspondence arrived from the United States from four developmental paediatricians. Three of these wonderful men offered to make the initial contact for me with the appropriate professionals, two provided me with information about how to care for a baby with spina bifida, and one of these two doctors also told me what I could expect to see with trisomy 18. Overcome with emotion I wanted to shout for joy and sob from despair.

With new awareness I drove to the hospital to see if I could receive some support from the paediatrics department. Stopping at the midwifery ward first I was dismayed that my first visit had not been communicated to them. To me this spoke volumes about what some people thought of my son, but I loved him to my very core and his life was just as important to me as that of any other baby. Surprised by my news the midwife told me that the hospital didn't actually provide care for babies with spina bifida.

Correspondence to: P Thiele liam194@gmail.com

News came that the hospital would not allow Liam's delivery. When we visited the department of obstetrics to challenge the decision, we were told that the hospital was not prepared for the resuscitation of a baby with trisomy 18. Defiantly I explained that we had not asked for any extraordinary measures and bluntly said that I would not allow resuscitation that would cause Liam pain or distress but, rather, a peaceful death was all that we wanted. Next day I had an appointment with a doctor at the paediatrics department. Still expecting a dispute, I spoke to the paediatrician and realised that I needed to fight no more, for this wonderful man offered me his total support. Finally someone spoke to me face to face about trisomy 18, and all of a sudden I wanted to cry, not from despair but from relief. The paediatrician was blunt and challenged me when I asked for the use of antibiotics, clarifying that to do so would be to use extraordinary measures. I explained that I wanted morphine to be given to Liam for any distress even though I knew it could cause respiratory depression. He told me that he would back this choice but that some people would accuse us of murder for the use of morphine in a neonate. Incredulous, I explained that I would stand my ground if there was any confrontation about this. Softly he told me that he was happy for me to do so but that this would not be a good time in my life to have to fight.

That evening I received a phone call from a palliative care specialist, who offered his unconditional support. This doctor was known for his obstinacy, in that he would not compromise if it meant that his patient's interests were harmed in any way, and I was grateful to have him on our side.

Receiving professional backing from these departments led me to believe that the rocky road would now become smooth, but how wrong I was. The next week we were told that the hospital's legal counsel was refusing to allow Liam's delivery at the hospital for various reasons, including the possible use of morphine.

Although the paediatrician reassured us that he and the palliative care doctor would be fighting for us, my mind spun and sobs threatened to explode at the thought of losing the support that I had finally found.

At 36 weeks' gestation I feared that Liam had stopped moving, and we drove to the hospital for delivery. Here a doctor performed a scan and, swinging the monitor to show me Liam's image, abruptly stated, "See? No heart beat!" Emotionally wrought and barely able to speak I was stunned not only by the news but by how I was told that my son had died and I knew that I would forever in my mind hold the parenthesis-like image of my son's still heart.

With a midwife's firm guidance Liam was born quickly. Tenderly I held my son and realised that the whole rollercoaster ride had been worth every bump, just to be able to hold my tiny son. After delivery our paediatrician entered the room and cradled my son tenderly, gently patting him on the bottom. Watching this interaction, I felt my heart swell with love for my son and in gratitude to this man for the fight that he had fought. Conversing easily, the paediatrician commented, "If a baby can know stress in utero then surely it can know love. Liam knew that he was loved." These words made the whole experience worth the heartache—just for my son to know that he was loved.

I gratefully thank Simon Blair, Brian McDonald and the neonatologist who encouraged me to write of my experience; I owe them all much for their support. Thanks also to the obstetrician for granting permission for Liam's story to be published.

Another version of this narrative appeared earlier in the *Journal of Medical Ethics*.

Competing interests: [Doctor author] has completed the Unified Competing Interest form at www.icmje.org/coi_disclosure.pdf (available on request from the corresponding author) and declare: no support from any organisation for the submitted work; no financial relationships with any organisations that might have an interest in the submitted work in the previous three years, no other relationships or activities that could appear to have influenced the submitted work. [Patient author] declares no competing interests.

Provenance and peer review: Not commissioned; not externally peer reviewed.

Cite this as: *BMJ* 2011;343:d3625

A doctor's perspective

Trisomy 18, or Edwards' syndrome, is a complex chromosomal disorder, in which there is an extra 18th chromosome. Children with this disorder have limited capacity for survival, and severe developmental delay is usual in survivors. The babies often have a small placenta, growth deficiency, and low birth weight. They are known to have very weak muscles, very poor respiratory effort, and a characteristic facial appearance, with malformed ears and a small mouth and jaw. Commonly they have malformations of the hands and feet, and they can also have severe congenital heart defects. As it is a genetic condition, almost any organ can be affected. Most children end up dying very early in life, usually of cardiac or respiratory failure.

Liam's extremely large heart indicated a structural defect, which would have severely compromised cardiac function and would have required burdensome treatment to allow Liam to survive the first months of life. Liam also had spina bifida, indicating paralysis and sensory loss in his legs and the likelihood of impaired bladder and bowel control. So even before I met Liam's parents there was a real heaviness in my heart that this was going to be difficult.

Delivering bad news is never easy, even at the best of times, but when confronted with a syndrome like this, even the most positive doctor finds it difficult to be encouraging. The children who do not die within the first few months are very disabled; only 10% survive the first year and are severely intellectually impaired.

Interestingly, though, when people talk about not wanting to be a paediatrician it is because they say that it is so hard that children die; what I tend to tell people is that this is not true and it is, in fact, a very positive field and that children mostly recover well. In my view, and with that as a background thought, the greatest challenge as a paediatrician is the delivery of bad news and the subsequent management of such children.

The rarity of trisomy 18 means that it has a severe and unexpected impact on families and is therefore a situation that we must manage well. With this in mind, we must approach the family as soon as possible to discuss very clearly and directly the diagnosis and talk about the likely options for the baby.

We hoped as Liam came to his date of delivery that he would be able to see his parents, albeit briefly. Barriers that were put in the way seemed artificial and bureaucratic but may be essential in this litigious and politically correct age. In contrast, the only real power that seemed to emanate from this family was that which was raw and basic—human love. Given the contrast, as a paediatrician I only ever had one course of action and that was, if possible, to place Liam into his mother's arms in hospital with the best available health care.

Simon Blair sblair7@bigpond.com

Further reading

For patients

Australia: http://livingwithtrisomy13.org/MemoriesofDanielS.htm; http://members.optushome.com.au/karens/

UK and Ireland: www.soft.org.uk; www.softireland.com

Japan: www.18trisomy.com

Reflections on my experience

- Bad news should be delivered face to face by an empathetic individual
- The role of tests for chromosomal disorders and their accuracy should be discussed before such tests are performed
- Patients should early on be offered a specialist referral—to a genetic counsellor, geneticist, perinatologist, or paediatrician—even if they don't want to access that at this stage
- No professional can know everything about every possible condition. Jointly exploring areas of uncertainty can help the relationship between doctor and patient
- Doctors shouldn't assume they know what patients are thinking. They need to ask them.
- Pregnancies complicated by a fetal abnormality should, if possible, be managed by a multidisciplinary team
- A management plan for such pregnancies should be agreed upon with the parents as soon as possible and should be shared with all relevant parties. Potential problems with such plans should be discussed ahead of time
- It's no consolation to be told by healthcare professionals that we have one healthy child or could have more; it only invalidates my unborn child.