Clinical review

The parents' journey: continuing a pregnancy after a diagnosis of Patau's syndrome

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Patau's syndrome (trisomy 13) is a rare condition, associated with high mortality, a range of congenital abnormalities, and severe physical and cognitive impairment. Many affected pregnancies will miscarry, and most babies born with the condition will not survive more than a few days or weeks. Less than 10% of babies with the condition will survive their first year of life.¹

Parents who discover during pregnancy that their baby has a serious chromosomal condition such as Patau's syndrome suddenly find themselves on an unexpected journey away from their hopes for a healthy baby and a normal family life towards a new and challenging situation.

Many parents in this situation will decide to end the pregnancy, but a few will decide to continue. This is the story of one couple who discovered at the 20 week scan that their baby had Patau's syndrome, and decided to go ahead with the pregnancy. Baby Benjamin was born at 35 weeks and lived for three minutes after birth. Much of the story is told in the couple's own words, recorded in an interview three months after the baby's death.

This article draws on interview data collected in 2003-4 for a wider research project on experiences of antenatal screening, now part of the DIPEX website collection (personal experiences of health and illness; see www.dipex.org/antenatalscreening for other findings and video clips from the interviews). This research included people who had routine screening experiences as well as parents of babies diagnosed with a variety of conditions, some of whom decided to end the pregnancy and some to continue.

A journey interrupted: arriving at a crossroads

The couple had two other children and had been waiting until they had moved to a bigger house to have another baby. Thus they set off on a planned and much anticipated journey of normal pregnancy and birth. They were looking forward to their 20 week scan as a chance to see the baby, although the mother felt upset on her way to hospital without really knowing why.

Mother: "I'd had my 10 week scan, my dating scan, and everything seemed to be normal. I was very anxious about my 20 week scan, and I don't know why. I remember driving there thinking I'd missed the junction off the dual carriageway and being in floods

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of tears. And there was no—as far as I was concerned—there was no reason, because it was an early morning scan. And I kept thinking, 'I've missed my scan date' because they were very busy, and I wasn't going to get my scan. But as far as I was concerned it was an opportunity to see my baby. We went in for the scan, like any of my other pregnancies, and she scanned me, and she seemed to take for ever. And she said she couldn't find my baby's lips, and his mouth. His fist was in his mouth. And there were some abnormalities with his kidneys."

Suddenly the parents found their expectations of a normal pregnancy were disrupted, and they were referred to a specialist centre for a repeat scan and further tests. In this period of uncertainty, the father's instinct was to gather information before the next scan to help them decide their next steps, whilst the mother wanted to believe everything would be all right.

Mother: "I wasn't that worried about it [the specialist scan] because I thought, 'Oh, well, you know, we'll go off, and we'll be scanned, and we'll go for lunch somewhere. . . .'"

Father: "I'd spent the weekend on the internet, so all these things that he [the consultant] was listing off as problems I'm thinking, 'Yeah, I've heard of that. Yeah, I've read that one, yeah.'"

Mother: "But I hadn't looked at the internet. I hadn't seen the amount of problems that could be and what was linked to what."

Father: "The trouble is, it's often said, 'A little knowledge is a dangerous thing.' I know in the old days, general practitioners always used to dread people going through the door with a medical encyclopaedia under their arm, and I'm sure people feel exactly the same about the internet now."

Interviewer: "But from your point of view it prepared you a bit?"

Father: "It did, yeah."

Mother: "I chose not to look at it."

Deciding which way to go

Once a diagnosis is made during pregnancy, parents immediately face a choice—do they want to continue with this unplanned journey on a new and uncharted course, or do they want to end it, by terminating the pregnancy? For some the way ahead may seem obvious, but others hesitate for some time.

The parents in this case emphasised that they did not have objections in principle to termination and supported others' right to choose that path. For them, it was a question of giving their baby a chance of surviving, however slim, and of being able to live with themselves in the years to come. They have not regretted their decision to go ahead.

Father: "The whole thing about it has been giving him absolutely the best chance that we could. It's also for our sake. We were given our diagnosis in September, and the baby was due around Christmas time. Now with the two older boys as well, we sat and we thought and we talked about this, and it's, 'Well, you know, is it going to be fair on the boys? Are they always going to associate Christmas with losing their brother or having a very profoundly disabled brother? How are we going to cope over the next four months?' And very, very early on we made a decision, or we formed the opinion that the decision we made wasn't what was best for the next four months. It wasn't what was best for the next four years. It wasn't, to an extent, what was going to be best for our two older boys. We knew that we were going to have to look ourselves in the eye in 10, 20, 30, 40 years' time and be able to say to each other, 'We gave our son the absolute best chance that we could.' And that's really why I don't think we'd do anything differently. It's been hard. . . . I went off and read up and looked on the internet and all the various websites. We knew very, very early on after diagnosis that there wasn't going to be a happy ending here. What there could be was a slightly less bad ending. We had to try to salvage something from what was a very, very bad situation."

The parents explained the relief of arriving at a decision—or, as they sometimes felt, deciding not to make a decision.

Mother: "And we sat down and we talked, and we lay up in bed for hours, trying to get our heads round the whole thing. And I think we'd basically both come to the same conclusion: that we wanted to carry on. And I think that was the first time I'd slept then for like a couple of weeks. And we woke up in the morning, I felt as if a big weight had been lifted off my head. I felt it was all gone and——"

Father: "Just because I think we knew the direction we wanted to go in."

Mother: "I felt very mixed up and very confused about what to do for the best, and once we'd made up our minds, that was it. 'I've made up my mind' and I was going to carry on. . . . I do feel that the decision we made at the time was very easy. I felt sometimes as if we never really made a decision, we just waited for nature to take its course."

Father: "Decided we'd let fate sort it out for us; abdicate the decision."

Mother: "Where I think I would have always felt, 'Oh, but what if? What if they were wrong?' or 'What sort of quality of life?' or 'He could have lived, he could have had this, he could have. . . .' And I would never, ever have known, and I needed to know."

Adapting to the new route

Having decided to continue with the pregnancy, parents have to come to terms with this new course. They face perhaps weeks or months of worrying what the outcome will be for their baby, adjusting to how their lives may change, and living with the disruption to their experience of pregnancy. The father describes their changed expectations for a different future:

Father: "It's quite vicious because you've got two stages of grieving. We grieved for our baby when he was born and died so quickly, but also in September, when we received the diagnosis, we had to grieve for the baby that we thought we were going to have. We had three months of having to get used to the idea that he wasn't going to see his first day at school, you know, I wasn't going to take him to see football matches or motor races or whatever. And you have to allow yourself to grieve for that as well . . . and it is very, very difficult."

A tough road

The mother described her determination to do the best for her baby by eating well and taking lots of rest. She remained hopeful that the baby would do better than expected, but "Towards the end it was becoming really quite hard work. . . . It was a tough road, but I don't regret it." The companionship of other women could be difficult—for example, continuing to attend a toddlers' group with her younger son.

Mother: "It's tough seeing other babies. You know, I go to a mums' and toddlers' group, and it's a very difficult position to be in, because parents of other babies or parents who are pregnant don't realise how many problems there can be, and how many different problems there can be that result in the same thing. It's not their fault; it's not your fault; but sometimes you've got to—— you know, I continued and went to the mums' and tots' group. It's very easy to hide yourself away. It's very easy for you to say, 'Well, I'm not going to go out anywhere I can meet parents or children, because I can't face to talk about this.' Or 'Oh, when's your baby due? Oh, I bet you're looking forward to having your baby. Is it a boy or is it a girl?' Nobody ever says, 'Is it okay?' or, 'Is everything all right?' Because they just assume everything is going to be okay. That was hard."

Additional educational resources

Literature

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Support organisations and web resources

Antenatal Results & Choices (ARC, www.arc-uk.org)—ARC offers information and support to parents who are making decisions during the antenatal testing process and offers a range of leaflets to help not just parents but also grandparents and other children in the family.

Antenatal Screening Web Resource (ANSWER, www.antenataltesting.info/)—Aims to provide information so that you can to make the best decision for you and your family, whether you decide to test, or not to test. Site includes accounts of personal experiences (including one using a journey metaphor: having a baby with a disability is like booking a trip to Italy and then finding you have been diverted to Holland).

Contact-a-Family (www.cafamily.org.uk/index.html, tel 020 7608 8700 or 0808 808 3555)—Charity that provides support and advice to parents of children with a disability or special needs. Can put families in touch with each other.

The Compassionate Friends (www.tcf.org.uk, helpline 01179 539639)—National self help group of bereaved parents offering support and friendship to those similarly bereaved.

Stillbirth and Neonatal Death Society (SANDS, www.uk-sands.org, helpline 020 7436 5881)—Support for parents and families whose baby is stillborn or dies soon after birth.

Condition specific support groups

SOFT (UK) (Trisomies Support Group; www.soft.org.uk, helpline 0121 351 3122)

SOFT (US) (Trisomies Support Group; www.trisomy.org)

Unique: rare chromosome disorder support group (www.rarechromo.org)

Syndromes without a name (SWAN, www.undiagnosed.org.uk)

Staff support

As the parents themselves noted, Patau's syndrome is relatively rare, and deciding to continue with an affected pregnancy is unusual.

Father: "We were told at our local hospital we were the first people they'd had diagnosed for about a year, and we were the first people for 10 who'd decided to carry on."

Despite the inexperience of local staff in accompanying parents on this particular journey, the couple felt extremely well supported. Apart from one difficult moment in a waiting room with other pregnant women, they praised staff for going out of their way to offer home visits and arrange sensitive and very personalised care.

Father: "We went for an appointment, didn't we, a couple of days after the diagnosis. . . . And it was there in the scanning department, where you go for your

scan. And of course we'd just had the news that our baby isn't going to live very long, and we're going to be plonked into a waiting room full of expectant mothers who are going for their first scans. So that was very, very difficult. After that we didn't have to go there any more. We mentioned that to the fetal health coordinator, and it was, 'Right, private room, other side of the hospital.' . . . And it's little things like that that you do appreciate."

Mother: "Another thing that was very good for me was, through the last few weeks of your pregnancy you usually get offered the relaxation classes . . . and I couldn't face all that. Even though I'd had two children . . . it's still quite an emotional time, to have a baby, and I'd still got to go through a whole labour."

Father: "Yeah, we knew there was going to be a full labour, with all the breathing exercises and the when to push and the when not to push."

Mother: "And they organised me to have some private massage. . . ."

Father: "Again, just so we weren't in a room with 30 other expectant couples watching videos about breast feeding that really weren't going to be relevant to us. The people at the local hospital and the fetal health unit were without exception absolutely fantastic."

Journey's end?

The time of birth is an important transition point for parents who know their baby has a serious condition. For Benjamin's parents, having an antenatal diagnosis enabled them to retain control and make his birth and death as peaceful and non-interventionist as possible. Staff were able to concentrate on their needs as parents, remaining alongside them on the final stage. "There were an awful lot of tears in the delivery suite, and not all of them were ours," as the father remarked.

Mother: "I didn't want him to suffer. I just wanted to give him what I possibly could, without too much interference, and without too much upset and hurt for him."

Father: "One of the fortunate things to come out of the pre-screening and the diagnosis is, you know, everybody, they tried to make a birth plan. Hospitals are quite keen that everybody goes in now with a birth plan, and we knew we were going to have a very special birth, and so we could tailor not only the birth plan, but we could also tailor the aftercare plan as well. Because we knew that it was possible that we'd only have a few minutes with Benjamin. It was possible he'd be stillborn. As it turns out we did only have a few minutes. What we really didn't want—if that's all the time that he had, we wanted him to retain some dignity. As it turns out, taking him from us, rushing him away, and putting him onto a ventilator would have made absolutely no difference at all. So during those few minutes that he had——"

Mother: "He stayed completely with us."

Father: "Yeah, he was with us in the room all the time. They just gave him a little whiff of oxygen, a little bit of chest massage, and then it became obvious that, you know, his breathing was just dying away, and his heartbeat was just fading away."

Mother: "But he did not suffer."

Father: "And then they passed him back to us, and from then on he was our baby. And he wasn't poked, prodded; there was no invasive testing done on him. All he ever knew was love and dignity, and that was very important to us. . . ."

Mother: "And I've kept all the clothes that he wore. He had a lovely smell, that lovely newborn baby smell, which is still here with us. Then he was dressed separately to rest him. But I've got all his clothes, all the things that I made for him, because if he'd have gone into the special care baby unit I didn't want blankets and that that they'd provided. I wanted all my own stuff that I could bring home, which was important. They're your keepsakes. They're your baby. Although I haven't got my baby, I do have the clothes that he wore, and that's the best thing I'll ever get, so treasure them. Mine haven't been washed. They've still got his scent on, and they're not going anywhere, they're at home with me."

Moving on together

Although Benjamin's short journey was over, his parents' own journey continued through grief, adjustment, and the stress of the nagging question, "Why me?"

Mother: "You know, there's times we've just sobbed and cried, and it's unfair."

Father: "It is; it's unfair, it's unjust. You find yourself saying, 'What have we done to deserve this? What could we have done differently? How could we have?' You know, you get to a point where it's at the back of your mind and you're thinking, 'Well, there's an extra chromosome carried. Well, so was it mine or was it my wife's?' And there's absolutely nothing you can do about that. You don't choose which egg you release at a time or which sperm is going to win the race, do you? You have to talk; you have to tell each other how you're feeling. You have to be willing to be a punchbag every now and then, for each other. . . ."

"It can weaken your relationship, or it can make your relationship stronger. I think if it weakens your relationship it's possibly because you're not telling each other what you're thinking. You have to talk to each other. This whole thing has been tragic enough as it is. It would have been even more tragic if it had pushed the couple apart as well. And that would be a real tragedy, because it's a shared thing that you've been through. And when you're both old bones, sat around talking about how your life's gone, you've got to look back on the bad times in your life and say, 'We did the right thing.' And I do think we could do that It's been hard; it's been a journey. It's been a bloody difficult journey sometimes."

Mother: "I was glad we did what we did for him."

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1 National Electronic Library for Health. Specialist Library Clinical Genetics. Palace Syndrome <http://libraries.nelh.nhs.uk/genepool/viewResource.asp?uri=http://libraries.nelh.nhs.uk/common/resources/?id=93804#Find%20out%20more>

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A shocking affair

Many years ago, when I was an anaesthetic registrar, one of my duties was to visit a psychiatric hospital for the weekly outpatient list for electroconvulsive therapy. It was all fairly straightforward. The patients were mainly women in their 50s or 60s, and there were usually six or seven at a session. The patients all sat together on benches, usually with a relative to escort them home afterwards. There were two curtained cubicles, each with a couch, facilities for ventilating the patient with oxygen, and, of course, the ECT apparatus. On the other side of the waiting area there were some curtained recovery cubicles, each with its own couch.

It was a bit of a production line system. A patient would be brought in to a cubicle, I would anaesthetise her with thiopental and suxamethonium, the psychiatrist would administer the shock, and I would ventilate the patient with oxygen until she woke up. She would then be transferred to a trolley and thence to the couch in a recovery cubicle. The nurse was then asked to bring in the next patient. And so it went on.

On one occasion, after the second or third patient, a nurse from the waiting area came in to ask if anyone knew where the

previous patient's relative had got to. She had, apparently, "disappeared." It took a few minutes for the dreadful truth to become apparent. Instead of bringing in the next patient, the nurse had brought in the previous patient's relative, who had said nothing and duly undergone electroconvulsive therapy. We were all appalled and unsure what to do.

I went over to the now recovering "missing" relative to ask how she was and apologise for the error. I received a glowing response. She said she was fine, just fine; in fact, she felt much better than when she had arrived with her friend. Honesty compelled me to explain what had happened. Fortunately, those were far less litigious days, and the elderly woman was quite charming said she fully understood and insisted that it was all her own fault for not speaking up. Indeed, she added, she now felt so much better than when she had arrived at the clinic that perhaps she should have regular treatments.

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