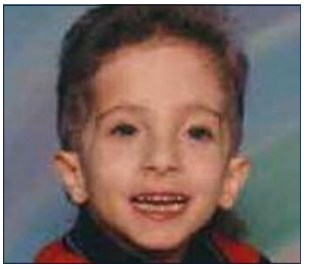
Unique

Mosaic Trisomy 16 Stories






Cameron

Cameron was born in 1999 after his mother discovered during pregnancy that he had mosaic trisomy 16. An amniocentesis was performed at 18 weeks after the triple test showed a very high risk (1: 8) of a chromosomal disorder. This revealed the mosaic trisomy and showed that 50 per cent of Cameron's cells contained an extra chromosome 16. When Cameron's own blood was tested after birth, no trisomy 16 cells were found.

Cameron was born after his mother went into spontaneous labour at 27 weeks. Medication delayed delivery until 28 weeks but Cameron was born by emergency Caesarean after the cord prolapsed and his heart stopped. At birth his Apgar scores were 1 at one minute and 5 at five minutes. He was ventilated immediately but had an intraventricular haemorrhage (a bleed into the brain) at birth. He stayed in hospital for 11 weeks and came home on oxygen. Cameron was born tiny, weighing 794 grams. By two months of age he weighed 1440 grams (3lb 3oz) and by his first birthday he weighed 6820 grams (15lb 4oz). At four years and five months, he weighed 12 kilos 700 grams (28lb). He has always been short and remains so, on or below the 0.4th centile for height and growth since birth.

Feeding has been difficult. Initially, Cameron's heart and lung problems left him too breathless to breastfeed but he took small amounts by bottle. He has had gastro-oesophageal reflux (his feeds return up the food pipe) since birth and after developing an aversion to swallowing, was fed by nasogastric tube until he was 2 years old, when a gastrostomy tube was fitted, allowing him to be fed direct into the stomach. By 2½, he started to drink and to eat by 3¾.

After a slow start, Cameron's motor development has been within the normal range. He sat at 11 months and walked at 18 months. At 4½ , he is very agile, can run, jump, play football and ride a bicycle with stabilisers. Occupational and physiotherapy input played a part in Cameron reaching his developmental milestones. However, his main problem is physical strength. Cameron has no learning difficulties. At 4½, he knows his alphabet and was starting to form letters at the age of 3. He is talkative and has a good vocabulary, using long and complex sentences and understanding at the same level. He had speech therapy to the age of 3½ when it was discontinued as unnecessary. He is determined and cheerful and makes friends easily.

Medically, Cameron was born with a small coarctation (tight narrowing) of the aorta, which resolved without treatment. He developed pulmonary hypertension and was oxygen dependent for his first year and developed chronic lung disease. His pulmonary hypertension was successfully treated and he is now healthy and has no after effects. He also has one lowered

eyelid and one lazy eye for which his 'good' eye has been patched daily. The haemorrhage at birth left no lasting effects.

Katie

Katie was born in 1998 after a triple test followed by amniocentesis at 19 weeks showed mosaic trisomy 16. Katie's mother was given an appointment for a detailed scan at 20 weeks which showed that her baby was small but appeared well. Her brain and heart seemed well formed although there was some concern over her spine. Having seen their baby on scan, Katie's parents found it impossible to even dream of ending the pregnancy. They decided that they would care for her and deal with any problems she might have.

Between 31 weeks and 36 weeks in the pregnancy, Katie did not grow. When a routine scan at 36 weeks confirmed that the blood supply from the placenta was poor and that Katie was not growing, a Caesarean section was scheduled and she was born the following morning. The theatre was packed with midwives, paediatricians and obstetricians.

Katie's parents held hands as the obstetricians delivered their daughter – a normal, healthy, if small baby weighing 1650 grams (3lb 10oz). At birth, a sample of blood was taken from the umbilical cord, and this showed no trisomy 16 cells. Katie's parents decided against taking biopsy samples from her skin.

Although Katie was well, she was reluctant to feed and she spent her first four days in special care where she was fed by nasogastric tube. After three more days on the postnatal ward with her mother, Katie was ready to go home.

Feeding got off to a slow start: Katie was breastfed with difficulty for three weeks but then took to the bottle well and moved on to solids at 4 months without problems. From the age of 18 months, her appetite has been small and she has needed prompting to eat. Although Katie has a very varied diet, she can take as long as an hour to eat a small amount. Katie's reluctance to eat has been paralleled by slow weight gain. Her parents were told that she would always be small for her age and she has been. At 6, Katie had not

grown or put on any weight for the past two years. Her growth hormone levels were normal and paediatric and genetic consultants were unable to offer any solutions. By now, Katie was aware of her height and wondered why she was smaller than all her friends. After seeking a second opinion, it was decided to start Katie on growth hormone treatment.
Katie's motor development has proved to be normal. She received physiotherapy for four months from the age of two months, but sat alone at four months and walked without problems at 14 months. She has no learning difficulties, started writing and using a computer at 4 and reading at 5. She attends a regular (mainstream) school without any learning support. She has no difficulties with speech or language and both understands and expresses herself fluently. She has a strong, outgoing personality and is very determined.
Katie has no birth defects and her only health problems have been recurrent ear and chest infections, leading to repeated drainage of the middle ear to improve her hearing. She has mild asthma which has improved since babyhood.
All in all, Katie is a happy, healthy little girl whose only problem is her small stature.

Emily

The pregnancy with Emily was a rollercoaster from the start. Emily was conceived after treatment following two years of infertility. An early episode of unexplained spotting was followed two weeks later by a scan showing an unusually small sac and the doctor warned Emily's anxious parents that there was a good chance of losing the pregnancy. But the next scan was reassuring and the first serious concerns only surfaced when two serum screening tests (Tri-screen) came back with an abnormal result. Then came the blow. The amniocentesis showed 100 per cent of trisomy 16 cells. Everyone was dumbfounded, not least the doctor who could still find a strong heartbeat.
Emily's mother was referred immediately for a level II ultrasound scan and a

meeting with a genetic counsellor who revealed that the baby was a girl. Her parents named her Emily Marie and on ultrasound, Emily appeared to wave at her parents. She seemed perfectly healthy but with 100 per cent trisomy 16 cells the medical prognoses were not positive and Emily's parents were warned that they might very likely lose their baby. They left the hospital that day with very mixed emotions but they decided not to end the pregnancy. 'If this baby was not going to live, it would not be by our hands,' Emily's mother said.

Every month, Emily's growth was monitored and every month she appeared a bit further behind in size. As the radiographers measured from head to toe, each part of her seemed to be a bit further behind than the last. More worries! But, again, nothing really seemed wrong.

Six months into her pregnancy, Emily's mother had a fetal echocardiogram (an ultrasound scan of the baby's heart) and this time the news was devastating. Three or four major things were found to be wrong. A second scan was scheduled with a specialist whose verdict was more encouraging: some narrowing in the aortic valve (the valve between the left ventricle and the aorta, the artery from the heart to the body) that would need monitoring but was not too worrying. More good news came with further results from the amniocentesis that now showed that seven per cent of the cells were normal, confirming the genetic counsellor's belief that Emily must have mosaic rather than full trisomy 16.

At 31 weeks, the waters broke, Emily's mother was given a steroid injection to mature the baby's lungs and kept in hospital on strict bed rest while Emily's progress was monitored regularly. Within days, she was moving noticeably less and the doctors decided it was time to deliver her.

At 3.36pm on May 27th 2000, two months premature and weighing 2lb 6 oz (1078g), Emily came out crying loudly. Her Apgar scores were excellent. All the same, she was transferred to neonatal intensive care and after developing a serious bowel condition (necrotising enterocolitis, to which preterm babies are vulnerable) was to spend almost three months in hospital. Meanwhile, samples taken at birth confirmed 100 per cent trisomy 16 cells in the cord blood and the placenta. Four months later, a skin sample was to show only four per cent trisomy 16 cells while Emily's own blood showed none.

Emily could not breastfeed but she took expressed breast milk for three months. Reflux (feeds returning from the stomach up the food pipe) was a major problem and she regularly scared her mother when her heart rate dipped while she was eating. Yet she slowly improved and gained weight until aged three months she was ready to come home.

Since then Emily has gone from strength to strength. Her forecast cardiac problems turned out to be mild aortic stenosis (narrowing), much as the cardiac specialist described from her second echocardiogram (heart scan). While she may need surgery for this later, for the time being she is just being monitored. At five months she had an inguinal hernia repaired.
In other ways you would not distinguish Emily from other girls of her age. Just after her fourth birthday, Emily is tall and skinny, like her mother at the same age. With input from occupational and physiotherapy, she sat at 9 months and walked at a year. Today she plays 'soccer', rides her bike and takes dance once a week. She has no learning difficulties and can write her name and most other letters. After initial delay in starting to speak – at 15 months she was developmentally at a 12-month level and at 22 months she was still behind - she now talks like any other child. In every other way Emily is a normal four year old.
'Emily brings a smile to everyone she meets,' says her mother. 'My mom calls her 'my golden baby' because she is more precious than gold. Emily has taught us never to give up on your dreams.'

Shayna

Shayna was born on Valentine's Day 1996, after 29 weeks of pregnancy and just eleven weeks after her shocked parents learned that she had mosaic trisomy 16. The doctors had given a gloomy prognosis (death, handicap, learning disabilities) but when two days later Shayna seemed to wave to her parents on ultrasound they decided they could not possibly terminate. At birth, Shayna was tiny (1lb 10oz; 741 grams) and had a hole between the upper chambers of her heart. That apart, she was well and after 13 weeks in neonatal care she came home.
At the age of 8, Shayna was on the small side but had otherwise defied all the dire medical prognostications. In almost all subjects at school she was performing at an above average level. She loved gymnastics and hip-hop and had just performed in her school talent show. In her mother Karen Lange's words, Shayna is 'in every way a typical little girl'.

Bobby

Bobby was born in 1991, four days before his due date. He was small, weighing just 4lb 10oz (1818 grams), and was to remain tiny all his life. However, at birth he needed no life support or medical interventions and went into neonatal care with the key aim of gaining weight. Investigations while he was there confirmed that Bobby had a heart defect called a double outlet right ventricle, which had already been identified during pregnancy, and that while he had no trisomy 16 cells in his blood, 12-13 per cent of the cells from his skin showed trisomy 16. What that meant was uncertain.

Bobby had three surgical operations in his first 14 months – to repair inguinal hernias (in the groin), to correct hypospadias of his penis (the hole was on the underside of the shaft) and to correct his heart condition. Just when his parents thought they could relax, Bobby was found to have pulmonary hypertension, a condition in which the blood enters the lungs under too much pressure. In Bobby's case, the veins that take oxygenated blood out of the lungs were found to be small and thickened and one of his lungs was also very tiny and not functioning properly.

Bobby had a hard time even fighting off colds and when he caught an RSV (respiratory syncytial virus) infection, he was on 24-hour oxygen for six weeks. By his fifth winter, Bobby was fighting off infections better but his lungs were deteriorating, the blood vessels in both lungs were thickening and narrowing and the right lung was affected as well as the left. The next winter he developed a habitual cough. Then he started to vomit, and one day he coughed up blood. Twelve days before Christmas 1996, he died in his mother's arms. He had had a pulmonary haemorrhage (a bleed in the lungs), caused by the pulmonary hypertension. Bobby was five years and three months old.

Seija

Seija was born in September 2002 after her mother's waters broke and she went into spontaneous labour at 34.5 weeks. Her parents had known she was a MT16 baby for two months, after an amniocentesis because of suspected growth delay, too little amniotic fluid and an enlarged pad at the back of the baby's neck had revealed 21% T16 cells. The geneticist warned of the risks of growth delay, abnormalities, preterm birth and the uncertainties that hang over the long term outcome for babies with MT16.

After a difficult two weeks, Seija's parents decided to go ahead and Seija was born weighing 1280 grams (2lb 13oz), with Apgar scores of 6 and 9 and a large head and fontanelle. She cried, to her parents' delight. Postnatally, Seija did well and when she left hospital aged three weeks her weight had risen to 1700 grams. She continued to gain weight steadily at over 200 grams a week, taking breast milk at first from a bottle and later from the breast. By six months she started on baby rice.

Investigations in hospital showed that Seija was a healthy if tiny baby and follow-up showed that she was babbling by six months and was contented and communicative. By eight months she was given an all clear by cardiology, the only clinical area where there were any concerns. By 14 months, Seija was standing alone, eating normally and very vocal. Her length was on the 10th centile and her head circumference on the 25th, reinforcing the view that her slow growth before birth was caused by issues of placental function. She was found at birth to have two copies of her mother's chromosome 16 and no copy of her father's - maternal uniparental disomy 16, which is known to intensify growth delay in the womb.

Seija's mother takes up her story.

'My first reaction when I found out that I was expecting was 'I hope I can carry this one.' I had already had two miscarriages and didn't want to give up hope but was tired of failing. My obstetrician performed the first ultrasound around seven weeks and arranged for another around 12 weeks. He mentioned then that the fetus looked a little small for the gestational age but knowing that the baby was alive was enough for me. Around 18 weeks, I had another ultrasound and this showed that the baby was too small for the due date to be determined. Another scan at 21 weeks showed growth retardation. I started to have a bad feeling.

'The baby might have Down's syndrome'

'Two weeks later the obstetrician told my husband and me that the baby looked so small that he would like to investigate further. He told us that he saw some signs that a Down's syndrome baby might have, such as growth retardation and extra skin around the neck and asked us if he could perform an amniocentesis right away. I was reluctant, as I knew there was a small chance that I might miscarry, but my husband convinced me that we should. We then went to see a genetic counsellor who explained to us what they are looking for with the rapid-result FISH tests for the most common chromosome abnormalities (signals that show whether there is an extra copy of chromosomes 21, 13, 18, X and Y) and that more thorough tests would be done to look at all the chromosomes.

'The genetic counsellor sounded excited when she called with the FISH

results: we were not carrying a Down's baby. It was a relief, but it was not to last long.

Not Down's but Mosaic Trisomy 16

'The long-term results showed that the baby had Mosaic Trisomy 16. This term was new to both of us and we had to ask the GP to write it down. She said 21% of the baby's chromosomes were affected by mosaic trisomy 16, 79% were normal. She suggested seeing a genetic specialist, as it is so rare that she hadn't found much information about it. We left without knowing what to do. I started surfing the net and found little information, but everything we found was devastating. Then I found the DOC16 site at www,trisomy16.org with its stories of children who are healthy and doing well. I couldn't read all the stories there without tears in my eyes.
'Next day, we saw a genetic counsellor who had collected all the information she could find, but admitted it was limited and she had never personally heard of Mosaic Trisomy 16. We didn't know what to do when we left her.

A teddy bear called Hope

'I was scheduled for another ultrasound and told my husband that I wanted to contact Karen Lange, the founder of DOC16, to see if I could get more information. Karen sent me a lot of information together with a teddy bear whose name is Hope.
'At the next ultrasound appointment, the obstetrician spent over an hour examining the baby. In my mind, this might be our last chance to see her alive and I had tears in my eyes all the time. Afterwards, he spent another hour discussing what he had seen and what our options were. It felt as if I was facing a death sentence. Yet he told us something we didn't expect to hear: he could see the growth retardation, but all her organs seemed to be there and our daughter was at the good end of the spectrum. It was like a last minute reprieve, a glimpse of hope.

Hoping for the best, preparing for the worst

'In our hearts we both decided that there was no way we could end the pregnancy, so we agreed to carry on. 'Hoping for the best, preparing for the worst' was something we kept saying to each other for the rest of the pregnancy.
'After making the decision, I felt much better. It was still hard seeing other pregnant women and I could not face attending prenatal classes. Some days were better than others, but I was unable to enjoy being pregnant.
'Around that time we had to decide how much treatment we would like for our daughter when she was born. The words of the head paediatrician will stay with me for ever. 'If this was a normal baby, we would do whatever we

can to save her,' he said. 'But she is not. We need to know how much treatment you would like for her.' I felt as if I was carrying a non-human baby, something unwanted. Yet she was a most wanted, loved baby who just happened to have a rare disorder.

A most wanted and loved baby

'My Caesarean section was booked. Finally, I thought, I would get to see my daughter. Outwardly, I was quite calm - excited to see her but scared in case she was not born alive. We heard her cry before we saw her and then she was taken away to a paediatrician who examined her for a couple of minutes and said 'She is normal'. We couldn't believe our ears. She was put into an incubator and transferred to a special care nursery. I wasn't really aware who was in the room but my husband told me later that there were at least seven doctors, all curious to know what was going to happen.

'I was transferred to a recovery room and asked the nurses to make sure that my placenta was ready to be sent to Vancouver. I had talked to Dr. Wendy Robinson a couple of times before the delivery and arranged that I would send my placenta for her research. *(Note)* The next day passed like a dream. I went to see my daughter that night and had the chance to hold her for the first time. She was incredibly tiny but quite as beautiful as I had ever imagined.

The smallest baby ever to go home

'Seija surprised everyone. Next day she was able to manage without the intravenous line to keep her sugar level up. After three days she did not need constant heart monitoring any more. A week after her birth day she was out of the incubator. She was small, but doing what she was supposed to do. At less then three weeks old, she was discharged. Her hospital paediatrician said that she was the smallest baby he had ever sent home. She weighed just over 1700 grams.

'Getting everything ready for her was quite a challenge for us. We had bought nothing, as I didn't want to come home from the hospital after losing a baby and be surrounded by a house full of baby things. The only thing I managed to buy for her before her arrival was a little hat, and even that was too big for her!

'Since then, Seija has been growing nicely. She had a hard time latching on because she was too small and too weak at first. At first I rented a breast pump to collect milk for her and I must say that was the hardest part of being a mother, as I would give her a bottle and then express, then put the bottles in the fridge or freezer and boil all the accessories from the pump. Imagine standing in front of a big pot of boiling water for 20 minutes at three in the morning! Luckily, Seija started to latch on around the last week of

November and was breastfed until she was 16 months old. I wanted to keep breastfeeding her as long as she wanted, but I got pregnant again and my milk production dropped drastically.

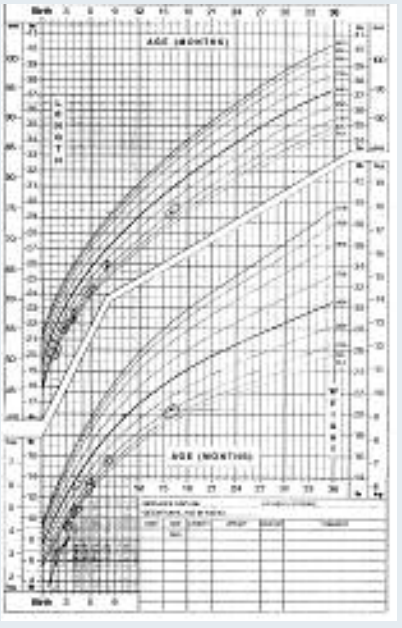
'Every month or two we have had visitors from the government's Child Development Program to see how Seija is developing. She has passed all her milestones at the normal rate. She is a little bit late with her first words but I am sure this will come shortly. At 19 months without correcting for her prematurity, her weight was 10.6 kg. She is still small but not far off her peers.

Yes, she is really here

'It took me almost a year to realise that she is here and will be staying. The turning point was our final routine visit to the geneticist. 'Normal,' he said. 'She is normal.' He has seen lots of disorders and hearing 'normal' from his mouth was quite something.

'I cannot imagine what would have happened if we had decided to terminate the pregnancy. I am glad that both my husband and I agreed that we should continue. Seija is our living proof that even being diagnosed with a rare disorder, the outcome is not always a sad story.'

Note: See link to website on back cover



The circled dots show Seija's catch-up growth

Unique recommends that you read these personal stories of families' experiences with Mosaic Trisomy 16 in conjunction with its information booklet **Mosaic Trisomy 16**, available from *Unique*.

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Disorders of Chromosome 16 Foundation
www.trisomy16.org

If you have an MT16 pregnancy and wish to contribute to research, log onto the mosaicism website at the University of British Columbia
www.medgen.ubc.ca/wrobinson/mosaic/index.htm

Unique mentions other organisations' message boards and websites to help families looking for information. This does not imply that we endorse their content or have any responsibility for it.

This leaflet is not a substitute for personal medical or genetic advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication and the medical content has been verified by Dr Wendy Robinson, Associate Professor of Medical Genetics, University of British Columbia and Monica Pearson BSc 2004.

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