

Sarah

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHOSE CHILD HAS
3-METHYLCROTONYL-COA CARBOXYLASE DEFICIENCY (3MCC)

Sarah Mackintosh entered the world on the 25th July 1996, two minutes after her twin brother. The twin's birth followed a pretty awful pregnancy that had seen me vomiting constantly and bedridden for a big chunk of time. I was delivered by caesarian section at 37 weeks as Alex, the presenting twin was breech, and sister Sarah transverse (sideways) across the top of him. I was so high on the morphine from the spinal block ("Oh but we don't give you enough to affect your brain" HA!) all I can remember is the obstetrician's comment when Alex was born "oh, he's a good size, look at that!" (6lb 5oz). Then Sarah was born and the doctor exclaimed, "Christ! Look at the size of this one!!" (7lb 6 oz) A fat little face appeared over the green screen and quickly contorted into a loud scream. And there they were. Our first children.



The twins and I came home after 9 days in hospital - the midwives at the Mater Hospital were determined to get me breastfeeding them both and their support and commitment saw me "twin feeding" (both at once!) till they were 9 months of age. Sarah had no medical problems, and really no indication that anything was seriously wrong. But she did have a few idiosyncrasies.

She was a hungry baby. I thought it was just my milk supply playing up until she lost weight again at her 2 week check up. From then she breast fed with her brother and had a full bottle of formula afterwards. By 6 weeks of age I got sick of this and started her on rice cereal. Instead of the usual "spit out" response, she gulped it down. By 3 months she was having cereal with her morning feed, stewed fruit with her lunch feed, and mashed veggies with dinner. Her twin brother was not the slightest bit interested in solids until 7 months. And there was nothing wrong with my milk supply or her sucking mechanism - she still craved solids even after a full bottle of expressed milk or formula.

At around two months of age, I couldn't wake her up one morning. She was a bit floppy, but still breastfed at her normal times - she just seemed to stay asleep. I decided to take her to the doctor in the afternoon if she was still sleeping. At around 1pm, after her lunchtime milk, she opened her eyes and woke up. She seemed a bit grumpy, but that was all. I told the child health nurse who said it was "odd", but since she was fine we never followed it up. What we now know is that she was probably hypoglycemic.

Sarah was an anxious baby - from the age of eight weeks. She vehemently responded to change, hated going out and refused anyone wanting to hold her besides me. She had some skin problems, cradle cap, non-specific dermatitis and the most appalling nappy rash I had ever seen.

Rob and I were pretty army-like in our approach to managing the twins. They were sleeping through the night, in their own cots, by 3 months of age. They fed together at regimented feed times. All of a sudden, at six months of age Sarah started screaming overnight. She sounded like she was in pain - we couldn't figure it out. We started controlled-crying techniques which were useless. The only thing that did seem to help (although not always) was paracetamol. We took her to the child health nurse and doctor. After a period of time she was diagnosed as having "night terrors". She would scream for up to four hours, but oddly, did not seem awake or even lucid. It really was like she was in a trance. We accepted the diagnosis of night terrors and followed the doctors advice - it did nothing. In hindsight - 6 months was when we introduced meat into her diet.

Sarah developed into a serious, surly toddler. We nicknamed her "serious Sarah" and "stropky pants" for her moodiness. Her anxiety increased and she seemed to have panic attacks if we took her to a new house, and she was terrified of animals! Baby sitting was out of the question. We just accepted this as her personality and hoped she would "grow out of it". Her twin brother could not have been more different. He was (and still is) the happiest of all our children and a real live-wire. He went to complete strangers without concern and reveled in the novelty of any situation!

In September 1997 myself and the twins contracted some sort of gastroenteritis. I was 30 weeks pregnant at the time. Sarah began to vomit Wednesday afternoon, she'd had gastro once before at 10 months of age, so we kept her fluids up and supplemented her with "gastrolyte" - a mild sugar mix. I was very ill as well, and we were all exhausted. Upon waking the following Friday morning Sarah made a strange "cat-like" cry from her room. Rob leapt out of bed to see what was wrong, she had vomited blood and appeared to be semi-conscious. Rob took her to our GP. The GP immediately recommended hospitalization on the suspicion of severe food poisoning and phoned the Mater hospital to advise of our impending arrival. Rob returned home to collect myself and our twin son as he figured if food poisoning was the correct diagnosis - we would all need to be admitted. By the time we got to the hospital, (45 minutes later) Sarah was fighting for her life.

I cannot speak highly enough of the hospital's response. Upon arrival, a young nurse immediately recognized Sarah was unconscious and asked me to deal with administrative details while she took her to see the Registrar. When I was led into the emergency room, all I can remember is seeing Sarah hooked up to several machines and the doctors and nurses looking extremely worried and muttering "she's very flat, she's very flat". The emergency registrar started questioning me about our intake of food etc in the belief it was food poisoning. Within half an hour it seemed the entire children's hospital had shut down to deal with Sarah. All Pediatric registrars and the Consultant had arrived to see Sarah, Surgeons from the adult Hospital had been consulted, x rays had been taken, an ultrasound had been performed and blood products were being flown in. In short, the response was both efficient and extraordinary. (To this day on I am still recognized by staff at the Mater by "Oh, you're Sarah's Mum? I remember that day!")

By this stage, I was so busy trying to accurately answer the emergency Registrar's questions, I do not specifically remember the arrival of Dr Frank Bowling, the Mater's only Metabolic consultant, who examined Sarah and suspected a metabolic disorder may be the problem. What I do remember is ALL of the other attending Doctors, whilst extremely competent, were completely dumbfounded as to the cause of her problem. Hypotheses ranging from food poisoning to telescopic bowel were being discussed - and despite the input from consultant Pediatricians, it was clear that they had no clue as to what was wrong. When Sarah was stabilized she was taken to intensive care, still in a coma. We had no explanation as to the problem and the Doctors were clearly struggling to establish one.

Later that evening both Pediatric Registrars came to tell me that Sarah had been diagnosed (thanks to Dr Bowling's intervention) with a rare metabolic disorder. They said the Mater had never seen a case such as this and the only people in Queensland who "knew anything at all" about the disorder were Dr Frank Bowling or Dr Jim McGill (who was on holidays). Dr McGill interrupted his holiday and came to see us that evening. We

know now that, had Dr Bowling not been available when Sarah was admitted to the emergency department, diagnosis would have taken days and she would have almost certainly suffered permanent brain damage.

I guess our story from this point is no different to any other parent who is told their child has a "metabolic disorder" (what the?). Jim McGill arrived at the hospital with a photocopied article from a medical journal to give to us, and his first question was "so, are you and your husband related?". (NO - we are not by the way!) We quickly realized there were only a handful of reported cases around the world and that Sarah was the only person in Australia with this condition. We realized what danger she had been in when she awoke from her coma, two days later, sat up and pointed to all the machinery stating "NO!". The doctor on duty exclaimed "oh look, she's got great motor control!" Clearly they were expecting her to have suffered major brain damage. She did not, and we are eternally grateful for all the staff at the Mater whose response no doubt made this happy ending possible. Sarah commenced low protein diet and carnitine therapy at 14 months and guess what?! No more problems sleeping overnight, no more panic attacks and our photo album started to fill with pictures of smiles and giggles. The only thing to persist was the anxiety which went from over-the-top to moderate to (now) mild.

Her doctors took her off carnitine at the age of 2 for a "trial". Guess what again? Screaming overnight reappeared within 3 days, misery and tears by end of week one. By the end of the third week (the trial was supposed to last four) I contacted Jim and said we couldn't stand her distress any longer. If he wouldn't give us the carnitine, we'd go to a health food shop and buy it ourselves! Jim managed to placate me and promised carnitine as soon as she had a blood and urine test to see what was going on. The physiological tests proved beyond doubt that Sarah "needs" her carnitine. We were assured that no-one would challenge her access to carnitine with "test results like these". Although it seems obvious in hindsight - we had not suspected that carnitine did anything other than perform some sort of medical function. We had thought the lo-pro diet was the key to her change in temperament post-diagnosis.

We have the occasional ups and downs like any family with an IEM kid. But apart from the odd hospital visit for hypoglycemia (which she hates), and the odd bad response to a "theft" of high pro food or other disruptions, Sarah is thriving. She's in grade one at school and quite frankly, we often forget she has an IEM she is so well. She has no delays, physical or intellectual and is pretty in tune with how she's traveling. She will tell us if she's not well, and we usually find a good reason (constipation because of the diet, stole some meat from her brother last night!). She is occasionally sensitive to illnesses and has been known to respond badly to protein and/or carnitine fluctuations. She can rapidly develop a "cluster" of symptoms that we now recognize as "metabolic", to the point where we now base our decision on whether or not medical intervention is necessary based on whether she's "acting metabolic". So I hold great hope that she will be the best judge of whether she needs medical help as she gets older (currently we know we need to worry when she says "Mummy, I think I need to go to the hospital")

I think Sarah's story is a perfect example of why it's so important to have all babies screened for these disorders properly at birth. She, like many with a metabolic disorder, will lead a perfectly healthy life if diet and treatment are maintained. We are the lucky ones. Because a metabolic consultant happened to be "on shift" when she arrived at the Mater, we do not have a brain damaged child. There are many others with metabolic disorders like Sarah's, who have not been so lucky. A statewide total of around five "Sarah's" present each year in Queensland hospitals, many in a worse state, and many suffer permanent and irreparable damage. Of course this damage is completely preventable, and the heartache caused to those families totally unnecessary. At the time of writing this article (May 2002) Queensland is the only state in Australia that does not protect families from this fate with Tandem Mass Spectroscopy newborn screening. [Ed. correction - Western Australia does not use MS/MS and only screens for the same four disorders as Queensland. Let's hope the "Smart State" catches up with the rest of the country soon.]

February 1, 2006, UPDATE: Re-reading Sarah's story makes me realise just how far we've come since then. She's doing incredibly well--grade 5 in school, an above average student, plays the piano beautifully, and we haven't needed a hospital admission in over 2 years. She's still managed by the metabolic clinic at the Mater (pops in twice a year with a urine sample and says "Hi!") and continues well with her low protein diet and carnitine which she pretty much manages on her own these days. We were lucky, but no-one should have their child's life riding on luck when the technology exists to protect them.

Australia has made gains in NBS since I wrote about Sarah in 2002. Our states are using tandem mass spectrometry for expanded screening, but the number of disorders that are screened varies. Efforts are underway to make NBS uniform throughout Australia.

Sincerely,

Rachael Sharman &
Rob Mackintosh
Written by Rachael Sharman
Mom to Sarah - May 2002