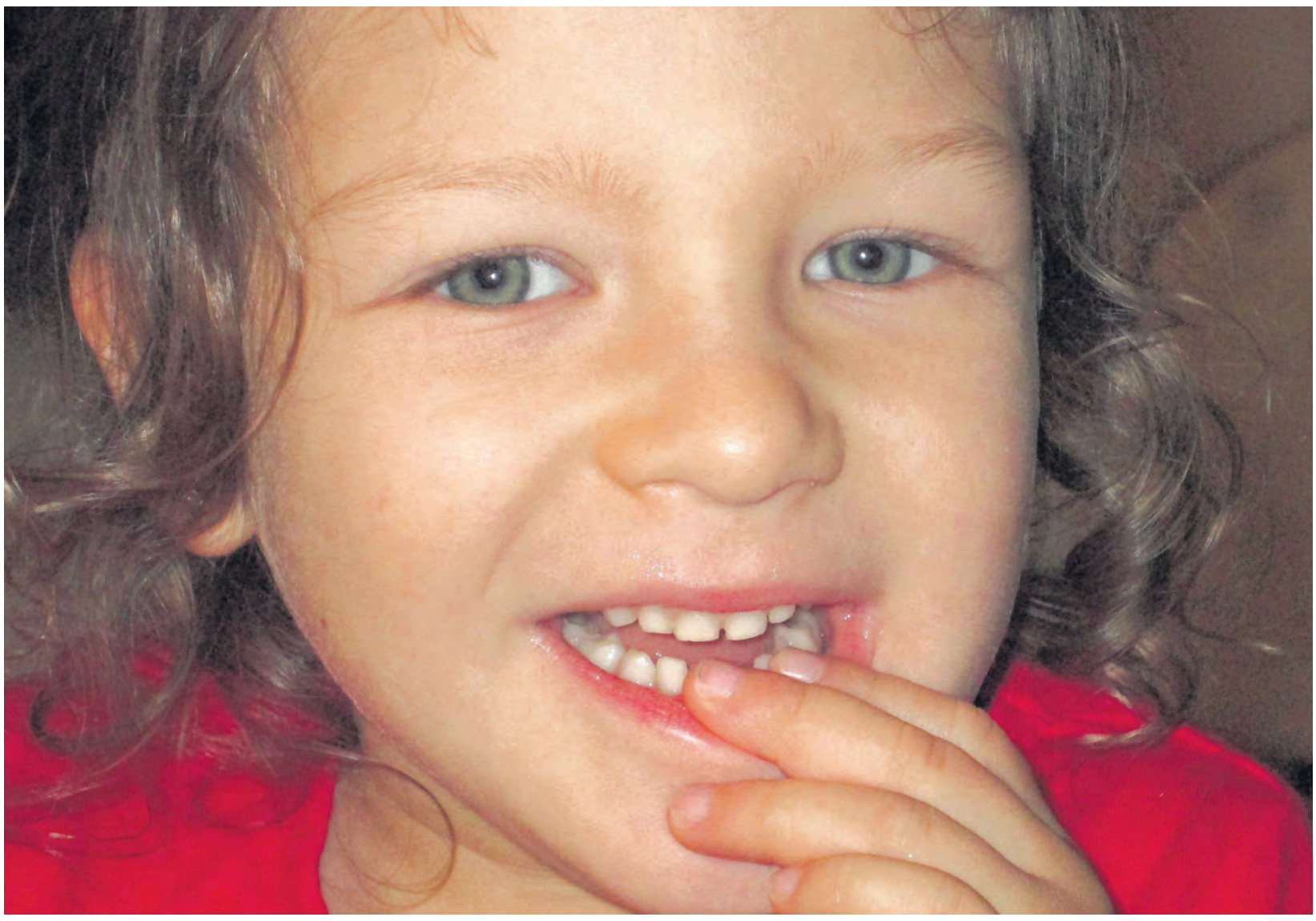
thewest.com.au The Weekend West
April 12-13, 2014

You can't stop loving them

The gift of life and the gift of a grandchild can sometimes be painful, writes **Wendy Macklin**



Rare disease: Sami Macklin suffers from Rett Syndrome, caused by a mutation in a gene on the X chromosomes which only affects one in 9000 girls.

It's marvellous being grandparents, everyone knows that. The trouble is, as your circle of loved ones grows larger, all over again you are hostages to fortune. Yes, you can give them back when you're tired and they've wrecked the house but you can't help loving them — it's in the genes.

We rushed to the hospital. Our first granddaughter had just arrived and what a thrill after so many boys throughout the family. She was gorgeous with a creamy skin and glossy brown hair — a typically exotic Australian combination of Anglo-Celt dad and Peruvian mother. "She's Samara — a gift," said Rob and Sophia. Nana and Pop were teary with joy.

Sami was a delight. She gurgled and rolled and pressed the button on her Wheels on the Bus book over and over and sang her own version. Her face lit up when she saw us and we laughed when she scooted over the polished floor on her bottom instead of crawling. She loved her Dora doll and the truckloads of toys and books from doting relatives. She could wave and say, "Bye bye."

Then, somewhere between nine months and a year, everything changed. Her words became noises and shrieks and she could no longer pull herself up and indeed, she didn't want to try. She rarely smiled.

"She's a floppy child," said a friend who ran a childcare centre." She'll take longer — but she'll get there eventually."

Sami started physio. I roamed the internet to read about other children who were slow to develop.

"Don't worry," wrote mothers round the world. "All children are different. My boy didn't walk 'til he was three and now you can't stop him!" And, "Don't worry! One day she will surprise you." On and on like that — always so encouraging and reassuring.

But the worries grew and the tests began. Sami was a happy and affectionate child but the months went by and she wasn't walking or talking or showing the slightest interest in any toys. The Dora doll was put away. The special shoes made little difference to her unsteady gait. At the petting zoo she didn't even look at the cuddly animals while all around, other children aged two were squealing with delight. She couldn't hold her spoon and solid food had to be in very small bits; she was grinding her teeth and her arms were always held out in front like a teddy bear. Two small cousins blew out the candles on her third birthday cake and I thought my heart would break.

More blood tests, MRIs and specialists and it wasn't autism, or cerebral palsy or . . . there were so many dreadful things children could have and they were ruled out one by one but you felt so helpless not knowing.

Life was becoming expensive because there is no government financial help if your child doesn't have a label. Rob and Sophia both had to work for the expenses and the mortgage and day care was becoming difficult because Sami was neither a baby nor a toddler and needed more careful supervision than the others. One highly qualified carer said, "I love Sami, she is a beautiful child but there is something wrong and I'm sorry but I can't keep her here with five others to mind."

> It's such a rare disease. Only about 15 girls are diagnosed each year in Australia.

It was the same everywhere and finally the family moved to Sydney where, like a miracle, there was a church-subsidised special needs centre for children under five. They would take her and there would be continuity at last. Finding St Anthony's has been the high point of the past two years; the ratio is one staff member to two or three children and Sami loves it. The painful and futile probing continued and Sami was nearly four when she went for genetic blood testing at Royal Prince Alfred Hospital.

I answered the phone and was horrified to hear my son Rob, almost incoherent with anguish. "It's terrible, terrible," he sobbed. "It's called Rett Syndrome." He dropped the phone, I dropped the phone and cried out for my husband and we raced to the computer. We only read for a few minutes and couldn't go on. In all our charmed family life, there had never been more shocking news.

This strange, rare disease is caused by a mutation in a gene on the X chromosomes and only one in 9000 girls is afflicted. Chromosomes are found in the cells and chromosomes hold genes which tell the body how to grow and develop. Boys sometimes get it but generally don't live long. The little girls will be perfectly normal for between eight and eighteen months and then begin to regress. There will be progress and regression from then on.

Most children will have trouble walking and few will talk. They often have eating problems and fail to thrive. Was there ever a more cruel diagnosis? I tried hard not to dwell on Sami's future but thoughts would force their way into my head without warning; in a mall book shop, I realised that she would never read or love words and started crying all over again. I knew that my writer husband would be going through a similar misery. The wonderful Sydney grandmother and I cried over the phone.

Both sides of the family went into shock and mourning and then were angry for a few weeks but gradually we began to calm down and think in practical terms. There would now be government assistance with physiotherapy which was vital to prevent later spine problems and hydrotherapy. Speech therapy at this stage was mostly to encourage Sami's parents to interpret what Sami was communicating with her eyes and signs and sounds. It is frustrating if you can't say what the matter is. At these times — about once a day — when she cries miserably we have learnt to use singing and music and especially High Five DVDs — on TV, mobiles and iPads and this singing group have a wondrous calming effect. I am a music person and am going to experiment with Mozart and Debussy. It was a dreadful experience for Rob and Sophia when Sami had the first epileptic fit which most Rett girls get in varying degrees of severity; thank goodness for modern medicine which is keeping this in check.

Not many have heard of Rett Syndrome which was "discovered" in Europe in the 1950s but had no international recognition until 1983.

We have some leading experts in Australia including Professor Helen Leonard and Dr Jenny Downs whom I met recently at the Telethon Kids Institute in Perth.

"It's such a rare disease that it's hard to get funding," they said. Only about 15 girls are diagnosed each year in Australia. These women want greater awareness in the medical profession and quicker diagnosis .They are also investigating the huge variation in symptoms associated with the gene's malfunction. Unfortunately, their funding runs out at the end of 2014. Our granddaughter is already registered with these dedicated researchers who track 'their Rett girls' all round Australia. There may be hope there for future treatments. The ultimate aim with ongoing research worldwide is to identify these debilitating genetic diseases in utero and to tweak the mutations into normality.

But we try not to look ahead and instead, concentrate on small but hopeful signs of progress as they happen. Sami, we now know, is luckier than some. She sleeps well and can feed herself with her hands. She loves to be with other children and they love her. "Sami is my best friend," says Allegra who is nearly three and is always hugging her. There is a big network of caring relatives and respite is essential for the exhausted mum and dad. And she is blessed with the most patient and resilient of parents. Our son and his partner have risen to this challenge in a way that makes us humble. And so proud.

Last week, we went to the beach. Sami loves water in every form and always smiles when her bare toes touch the sand. She seems to relish the texture and perhaps the feeling of security it evokes? She began to stumble towards the waves and then suddenly crashed over on her back. "No, wait," said Sophia putting out a restraining arm. We watched in silence.

Sami lay there for a few seconds and then slowly, with great difficulty, pulled herself up until she was standing proudly in front of us with sand pouring out of the glossy curls. We clapped and cheered and she laughed with joy.

Grandchildren are marvellous. They might take giant steps or small steps. It doesn't matter. You can't help loving them. It's in the genes.

For further information: aussierett@telethonkids.org.au

To donate online go to : rett.telethonkids.org.au

Grandma knows best: Wendy Macklin is slowly coming to terms with her granddaughter Sami's debilitating genetic illness.