



# “Eating protein could damage my daughter”

A newborn screening program revealed Bridie's baby has a rare metabolic condition



Bridie Melham, 36, from NSW, and husband Paul, 39, are parents to Eden, 3, and Pippa, 1 year

After 10 years of marriage, my husband Paul and I had our first daughter, Eden, with the help of IVF. Eighteen months later we were delighted by a positive pregnancy test: I had conceived naturally.

I felt more sick than I did during my first pregnancy, but everything was going to plan with our second until the 20-week morphology scan. This showed that the left ventricle of our baby's heart was bigger than the right, which could indicate a heart murmur (or ventricular septal defect – VSD). We were upset but hoped for the best.

After two visits to the paediatric cardiology department at The Children's Hospital at Westmead, doctors said they were 90% sure our baby did not have a VSD, however

a paediatrician could not be completely sure until after the birth.

Ten days after my due date I went into labour. Although my waters hadn't broken, I had period-like pains every five minutes, so I rang the hospital and was advised to come in. We waited for my brother-in-law to collect Eden then drove to the hospital, by which time the contractions were three minutes apart but not too painful.

I was lucky that the midwife on duty knew my doctor and my history, so she rang the doctor as soon as

I walked into the labour ward. She examined me, said I was 7cm dilated and that I should have my baby by the end of the day. But by then I couldn't even lie on the bed. I stood up as the pain intensified and the contractions became more frequent. Two contractions later my waters broke and, after five minutes, Pippa was born. From when we drove into the hospital car park to the birth was just 50 minutes. The news was good, too: the paediatrician said there were no issues with Pippa's heart.

Three days later a nurse came in with a pamphlet, advising she would be taking some blood from Pippa. This heel prick test, also called the Newborn Screening Test, indicates whether babies have one of a number of rare inherited conditions, including Phenylketonuria (pronounced fenel-key-tone-uria or PKU) and other amino acid metabolism disorders.

For some reason I kept the pamphlet, and it was at the front of Pippa's health booklet when I received a phone call at home two days later with news that Pippa had tested positive for PKU. The booklet explained that people with this condition are missing an enzyme that breaks down phenylalanine, an amino acid in protein, which builds up in the

bloodstream and becomes toxic. Dangerous levels of it can cause mental retardation, seizures and other brain disorders.

Paul and I were devastated. I was also gripped with guilt that I had unknowingly handed down this problem to Pippa (see box). But we also counted our blessings that the newborn screening program was in place to pick up the condition.

### SPECIAL CARE

We discovered there is no cure for PKU; the only treatment is a daily diet of special formula supplemented with a miniscule amount of natural protein. I wasn't comfortable not knowing how much protein Pippa received through my breastmilk, so chose to stop breastfeeding. I felt that establishing a systematic approach of identifying how much phenylalanine her body could process each day was more important to her health.

Pippa had to be fed two types of formula: a small amount of commercial baby formula, until we worked out how much protein her body could tolerate, followed by the special PKU formula.

Tests showed that Pippa has an extremely low tolerance for natural protein – only 4g per day (one slice of bread has about 3g of protein). This tolerance will not change, even as she grows up, and has to be closely monitored each week through a heel prick test to ensure her brain is not affected. She will be on a specialised type of supplement for the rest of her life to replace the vitamins, minerals and amino acids that she cannot consume naturally.

Pippa has just turned one, and we are now transitioning to solids. We feed her 2.5g of protein by way of food, supplemented with commercial baby formula (1.5g protein) and 500mls of PKU baby formula. The foods she's eating

## PKU explained

Babies who have PKU are unable to produce an enzyme called phenylalanine hydroxylase, which breaks down the amino acid phenylalanine present in food containing protein.

There is no cure for PKU, but it is treated with a restricted, low-protein diet. Sufferers cannot eat meat, fish, poultry, nuts, cheese, bread, pasta, eggs, ice-cream, soy, lentils, chocolate, beans, gelatin or dairy products.

The PKU diet includes carefully measured amounts of fruits and vegetables, plus specially prepared protein substitutes.

A child inherits the condition when both parents pass on the defective gene. However, there is still a one in four chance of inheriting PKU. This is why in Bridie and Paul's case, Eden doesn't have PKU and Pippa does.

The PKU Association of NSW is raising funds for research being carried out by Professor John Christodoulou, head of the PKU clinic at The Children's Hospital at Westmead. Visit [www.pkunsw.org.au](http://www.pkunsw.org.au) to find out more.

now include rice cereal, apple, pear, blueberries, carrot, zucchini, pumpkin, corn, low-protein pasta and low-protein crackers.

She's just started walking. An 'old soul', she's relaxed around people and at times is very cheeky and stubborn.

I still feel guilty that Pippa will have a severely restrictive diet forever. However, we are reassured by the PKU community that the diet becomes a 'new type of normal' in the family; and we count our blessings that we have two lovely children. **R&B**

