PKU: A PARENT'S PERSPECTIVE

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Having an infant diagnosed with Phenylketonuria (PKU) can be a scary and challenging experience. Louise Conlisk's daughter Caitlin was diagnosed with the condition and here she tells us how she and her husband have been helped and supported to live with PKU.

Louise has worked as an ERP consultant for over 12 years and now lives and works in Melbourne, Australia. Her interests include reading, running and time spent outdoors. Since becoming a mother, she is so 'time-poor' that all her interests are mostly baby-related!

For full article references please email info@networkhealthgroup.co.uk

My husband and I are both originally from Co Galway in Ireland and moved to Melbourne almost 10 years ago. Like a lot of other people from our side of the world, the original plan was to stay a year or two in Australia, but here we still are and have now started our little family. We welcomed our beautiful healthy daughter Caitlin into the world on Valentine's Day 2015, at the Epworth Freemason's hospital in East Melbourne.

The second day after Caitlin was born she was given a heel prick test. Blood was withdrawn from Caitlin's heel and was tested to determine if Caitlin had any genetic disorders. Approximately nine days later we were contacted and told that our daughter had Phenylketonuria (PKU). PKU is a rare genetic disorder that prevents the normal breakdown of a protein, phenylalanine (Phe). Untreated PKU can lead to intellectual disability, seizures and other serious medical problems such as brain damage.

On that first day after being told, we felt as though we had been hit by a massive blow. It was so hard to comprehend the fact that our perfect newborn baby girl had an incurable genetic illness and would never be able to eat protein freely or likely never eat meat, fish, eggs or dairy. We couldn't comprehend what Caitlin would be able to eat.

The metabolic team at the Royal Children's Hospital, Melbourne (RCH) set up an appointment for us to come to the hospital immediately after Caitlin's PKU diagnosis. In that initial meeting we were advised of a medication that could help stimulate the deficient enzyme to help allow more protein into Caitlin's diet. The medication called Kuvan,[1] we were told, did not work for all patients with PKU, and as the chance of our baby being responsive to Kuvan was one in five, the odds were against us. In order for our child to be prescribed Kuvan, there needed to be a minimum 30% drop in Phe levels after a load test. We hoped and prayed that Caitlin's levels would drop enough to be approved and given the medication.

TESTING FOR RESPONSIVENESS

In order to test Caitlin's responsiveness to this medication, the medical team inserted a tube into Caitlin's nose and down into her stomach. Caitlin being only nine days old was too young to swallow the tablet in the normal way. It was pretty difficult to watch the medical team feed a tube through her nose. Luckily, she had an excellent response to the load test.

Caitlin's levels were periodically taken over a 24-hour period and after that time, her Phe levels had dropped from 1100+ to below 400, well in excess of the required 30% drop to be granted access to the medication. This good news helped us to accept the fact that our very young baby had PKU and gave us hope that we had something that would help us live with this chronic illness.

Kuvan is not available across the whole of Australia. Currently, it is available for PKU patients in the states of Victoria and Tasmania (the states covered by the RCH in Melbourne). We thank our lucky stars every day for the fact that we live in Victoria. This was definitely the silver lining in our cloud.

In England, Sapropterin (Kuvan®) is not routinely commissioned and priority groups eligible for treatment with Sapropterin are the minority of pregnant women with PKU who are unable to establish adequate dietary control and achieve the target non-teratogenic range of Phe (100-300 μmol/L).[2] For more information on Kuvan in the UK, see 'More Information' at the end of this article.

One of our many questions about Kuvan was in relation to the cost. Initially we were informed that it would cost approximately $100 (approximately £50 or 64 EUR) a tablet with the amount of tablets per day increasing as the child grows. We worried about how we could afford this expense, but were relieved to hear that the cost of the medication was going to be covered by the hospital (with a minimal out-of-pocket charge to be covered by us).

STARTING THE MEDICATION

As advised by the RCH, Caitlin did not start taking Kuvan until a few weeks after we introduced solids into her diet, which was close to when she turned seven months old. The Kuvan dosage was calculated using Caitlin's weight and, initially, we were allowed to increase her protein by a small amount. As the weeks went by, if Caitlin's levels were low or within range, the dietitian would on occasion recommend an increase of 0.5g to her daily protein allowance. Caitlin started on 2.0g of protein a day and now, at 15 months old, Caitlin is allowed to have 9.5g of protein daily. This is significantly more daily protein than what Caitlin would be able to have without the Kuvan medication. Caitlin is still breastfed along with bottle feeds of her PKU Anamix Infant/Gel drink. I am confident that when I stop breastfeeding, her protein intake capacity should increase. Caitlin's current dosage is two tablets a day, one in the morning and one in the evening.

Caitlin is administered Kuvan by dissolving it in a small amount of water and then mixing it with her food such as jam or apple puree which is then spoon fed to her. Caitlin normally takes the medication without any problem as she is quite used to it. On occasion, if she isn't hungry, or is tired, it can take longer to get her to take the medicine. If we are eating out, we need to remember to bring the tablet and a small syringe to mix a small amount of water to it, so it is just a few more things to put in the baby bag.

Caitlin, leading as normal a life as possible

LIVING WITH PKU

Along with the medication, the other prescriptions Caitlin currently takes are PKU Anamix Infant, PKU Anamix Gel and ProZero milk. To test Caitlin's PHE levels, we do a blood test at home every week using a lancet to draw some blood from Caitlin's finger to place on a Guthrie Card. The Guthrie Cards are posted to the screening laboratory at the RCH and the results are provided to us by phone each week by a dietitian from the Metabolic team. During this call, the protein allowance for the following week is also discussed.

To manage Caitlin's protein allowance, we carefully control her food intake every day and try to spread her protein allowance throughout three main meals to hopefully keep her fuller. She has 1.5 Weetabix in the morning, which works out to be 3.0g of protein, some low protein bread/pancakes/muffins for lunch and then whatever protein allowance is left over we give her for dinner. Without Kuvan, we would not be able to give Caitlin some of the regular food we currently give her, such as regular breakfast cereal, gluten-free bread and regular pasta.

Caitlin goes to childcare four days a week since I returned to work. Due to her having PKU she cannot have the foods served by the kitchen at childcare, so we provide her with her own special lunch. We label all of her foods in her lunchbox with her name and the protein amounts contained in each food item, which we give to the childcare staff. They then put whatever food Caitlin doesn't eat into a waste lunchbox. In this way we can weigh what Caitlin hasn't eaten and accurately calculate the protein that Caitlin has consumed when not in our care.

PROS AND CONS

Kuvan has made a big impact on the foods we can give her. Having a higher protein allowance is of massive benefit in a lot of tangible and intangible ways. There are and will be psychological benefits, such as increased concentration levels and improvements to Caitlin's overall behaviour. Caitlin's quality of life is enhanced greatly, especially socially, as she can eat some 'normal' foods. In the future, there will be possibilities to actually eat a meal at a restaurant without having to bring Caitlin's own food with us. All of this will contribute to a much happier, less restrictive childhood.

Caitlin also does not have to eat as much of the low protein specialised PKU food due to taking Kuvan. If we need to give the tablet to Caitlin outside of our home, it isn't too difficult; we just need to make sure that we have a little container to dissolve the tablet in. Kuvan has made going on family holidays a possibility for us too, as we can buy more foods off the shelf instead of having to bring all specialised food with us. The fact that Caitlin can have more protein in her diet is something we are very grateful for.

The only real negative aspect of the medication is having to give a small child a tablet twice a day, which most of the time is fine ,but sometimes, as already mentioned, if we have a tired child on our hands it can be delicate.

SUPPORT

The RCH Metabolic team in Melbourne are truly amazing. We bring Caitlin to the Metabolic clinic at the RCH three to four times a year. At these clinic appointments we meet with a dietitian and a doctor and go through Caitlin's progress. We have a close relationship with the dietitians in particular and we are comfortable knowing that the management of our daughter's condition is in very good hands. The team support us with all of the changes that happen in a young child, such as starting solids, and are extremely helpful with advice. They have provided letters for airlines when we travelled home last year, armed with a suitcase full of formula and medication. The Professor of the Metabolic department is hugely experienced in his field.

THE FUTURE

We are hopeful that Caitlin's protein intake can continue to increase, given her levels are within range. As she grows older, we feel that she will be further enabled to reach her academic potential. Anything that helps my child live with this very restrictive condition and add more normality to her life is a massive bonus. We would love to consider moving back home to Ireland to be closer to our family, but Kuvan is not available there so moving back to Ireland for us is not an option.

My biggest hope for the future is that other children with PKU are given the same opportunity as Caitlin to be given this medication. I sincerely hope too, that any health professionals or decision-makers considering whether to approve this medication for use, take into account the unquantifiable benefits in helping children with PKU lead a more normal life.

More information

- NSPKU (2010). The role of Sapropterin (Kuvan®) in the management of PKU in the NHS: considerations and prospects www.nspku.org/sites/default/files/publications/Kuvan.pdf
- National Centre for Pharmacoeconomics, Ireland. http://www.ncpe.ie/wp-content/uploads/2009/06/Sapropterin-Kuvan-summary1.pdf

Comment from the Editor

At present, Kuvan is not considered a cost-effective alternative to dietary treatment and there is a limited amount of evidence for its effective use. The prospects for funding of Kuvan in the UK, however, are likely to improve if and when new evidence regarding clinical effectiveness is forthcoming. Professor Anita MacDonald has co-written research papers alongside other multinational metabolic healthcare professionals, which discuss this treatment and if used, how it should be monitored and managed. Click here . . . for references. In the future, greater scrutiny, stricter guidelines and increasing pressure to ensure cost-effectiveness will all be key factors in the decision-making before new medications are approved for NHS use. Continued support from UK clinical teams, sustained lobbying by patients/parents and carers, as well as by the NSPKU, will be crucial in ensuring that patients benefit from new developments in treatment. A pressure group by the name of Phedup has been formed, their website is www.phedup.co.uk - *Emma*