

slide one

hello my name is karen and im from Dublin in Ireland. I live here with my husband jon and our 2 kids, Ciara and luke . Ciara is 9 now and Luke is 5.

slide 2

Ciara is our first child so as any new parent we were delighted when she was born in 2008. For the past 50 years all babies in Ireland are tested for pku through the newborn screening program. Ciara had this done a few days after she was born. I didn't know what they were testing for, I just knew it was something that had to be done and that I should make sure her foot was warm so that the blood came out more easily. Watching her get pricked was difficult, as it hurts, so I was glad when it was finished and then promptly put it out of my head.

Everything carried on normally with feeding, nappies and showing off our new baby to our friends and family. until a few days later we got a phone call from a nurse saying Ciara had pku and we had to bring her into temple st hospital here in Dublin. Neither jon and I had ever heard of pku so we were very shocked and worried. We stayed in temple st hospital for a week where Ciara had more blood tests done and was fed a special baby formula. We learnt all about pku there from the doctors, nurses and dietitians who were amazingly supportive. We learnt that if her pku was managed Ciara could development normally. We were very worried. I think I cried for the whole week. I know its silly but I love chocolate and the thought of never sharing a slice of chocolate cake with my daughter was very difficult to accept. After the week in hospital we got to bring Ciara home but the support from the staff at temple st continued with regular blood tests, phone calls and hospital visits.

As the years went by jon and I discussed having a sibling for Ciara. We knew that any child we had would have a one in 4 chance of also having pku. In 2012 we took our chances and Luke was born. As he was high risk he had an early additional test for pku. I was convinced he wouldn't have pku but unfortunately that wasn't the case and was diagnosed pku a few days after birth. He was put on a special formula and As with Ciara I could still breastfeed him a little. So they would have carefully measured amounts of special formula and then breastmilk.

slide 3

Now Ciara and luke are older they follow a highly restricted diet, they cant have chocolate, meat fish poultry, no pasta , no diary no bread, no nuts or pulses, no lots of things. their special diet, a low protein diet, is made up of 10 g of natural protein a day, they get this from things like peas – 20g of peas have 1 g of protein, potatoes, 45g of potatoes has 1 g of protein, 2 Weetabix has 4.5 grms of protein. This natural protein is very carefully weighed and measured everyday. It is very important that they don't have too much of this as it can effect their blood levels and could lead to learning disabilities among other things.

They can eat as much protein free foods as they like. Examples of these are grapes, melon, apples. The irish health service provide specially manufactured low protein foods, these are things like pasta, flour to make bread and cakes and milk. You can also buy some special low protein products on the internet which are really pricey.

As Ciara and Luke still need all the other elements of protein apart from the one they can't break down they have to have a special drink. I don't know how the kids manage to drink these as they really taste bad and have a very strong smell. We've had many battles over the years trying to get them to drink these drinks 4 times a day. We have regular hospital visits and phone calls with the nurses and dietitians. We do regular blood tests to make sure their blood levels are within range and if the levels are high we have to reduce the amount of natural protein they can have. I think without the support from other families with PKU, the doctors, dietitians and nurses in Temple St we would have been much worse off.

slide 4

There are many challenges with PKU. It is invisible, to look at Ciara and Luke you would not know there was anything different about them. It is rare, I had certainly never met anyone with it before and it is very unknown. It is very unusual to come across someone who has heard of it and if they have heard of it they know very little about it and sometimes even incorrect information.

The highly restricted diet is very challenging, saying no to your child all the time is very necessary and very hard. Jon and I still eat normally, we have pizza and meat like most other people. Sometimes I can see in Luke's eyes that he so wants to eat off my plate but can't and sometimes that can turn me off my food.

Over time I have met many different people with PKU here in Ireland and abroad and I'm aware that they have treatment options and low protein foods abroad that we don't have here in Ireland. That makes me very frustrated. If Ciara and Luke don't manage to keep their blood levels within range they risk suffering cognitive deficits, not being able to listen and concentrate in school, or suffer from anxiety, head aches, epilepsy and tremors to name but a few symptoms. As all parents I want to make my children's management of PKU as easy as possible so it's tough to know that just a short distance away there are children with PKU who have the chance to benefit from different options. I just can't understand why all that can be done to help keep their blood levels in range and therefore avoid the symptoms, is not being done here in Ireland.

As a family we spend a massive amount of time planning meals, every meal I prepare, I prepare 2 meals, one for Jon and I and a separate low protein version for the kids. There's also the supply of special low protein foods and substitute drinks to manage. If you run out of low protein milk you cannot just pop down to the shop. It has to be ordered in through the chemist. All this takes a lot of time and energy.

A big challenge we recently went through was Ciara taking her special protein substitute drink in school. Her teacher told us that Ciara was just so tired in the afternoons at school she was unable to participate in class. So she now takes one of her drinks in school with her lunch. For her it's like an energy boost. As I said it tastes and smells pretty bad, she's having it in front of her classmates and has responsibility for taking it herself, so that was a really big step for her.

With such a restricted diet days out, social occasions and holidays all require a huge amount of planning. We cant just go out spontaneously. We have to make sure we have the protein substitute drinks with us and make sure there'll be food the kids can have or bring it with us.

As Ciara and luke grow up and start living more independently, going out with friends, going to parties its going to be really difficult for them to manage their own diets. Obviously ill do all I can to help them but what teenager wants their mum checking on them all the time. Having spoken to other pku families I understand that the transition from the parent managing the diet to the person actually doing it themselves, during the teenage years and when they're young adults, is very challenging and a lot of people really struggle with this and therefore suffer the negative impacts of high levels just at a busy time in your life when youre doing exams and making decisions that can affect the rest of your life.

Another challenging time will be when Ciara is older (hopefully quite a bit older) and wants to have children of her own, if her blood levels are not really low and not really well controlled pre-conception and during the pregnancy she risks her babies health. Unborn babies can be severely affected by high blood levels in the mother. The babies can have small brains, mental retardation and a lot of other horrible things.

slide 5

I have a pretty big wish list but I think this just shows up the amount of room for improvement in the care of pku in ireland

If there is a treatment option that helps keep Ciara and lukes blood levels in normal range I want them to have access to it. Ive seen how kuvan can help some people with pku and I want to see if it can help my kids and if so I want it to be prescribed to them.

If there is low protein food which they can eat without causing high levels I want them to have it.

I want Ciara and luke to have access to specialised nurses dietitians and doctors the whole way through their lives including into adulthood.

I want more people to learn about pku and for those with pku to have access to all information relating to their condition. I hope to help do this through the pku association of Ireland which is a patient support group here. The pku association does great work, from sharing low protein recipes to providing support to each other, to hosting the European society of pku annual conference here in Dublin last year.

I hope the espkus guidelines for the management of pku are implemented here in Ireland.

Most of all, as any other parent, I want my children, despite pku, to have happy healthy lives with the best possible long term health outcomes.