



Introduction

- Karen Willetts from Dublin, Ireland
- Married to Jon, 2 kids who both have PKU
- Ciara aged 9 and Luke aged 5





Finding out about PKU

- Ciara is our first child
- Heel prick test – Happy Birthday Bob Guthrie & Horst Bickel!
- The phone call that changed our lives
- Hospital stay
- Support from Temple street hospital
- Whether or not to have more children
- Luke's arrival and diagnosis





Managing PKU

Ciara and Luke's PKU is managed through a low protein diet with synthetic protein substitute drinks.

Low protein diet is made up of:

- very small quantities of natural protein. Allowed 10g per day eg 20g peas = 1g protein, 45g potatoes = 1g protein, cereals – 2 Weetabix = 4g. All closely monitored and weighed
- Protein free foods eg melon, grapes, apples
- Specially manufactured low protein products eg special pasta, bread, milk. These products are expensive, some of these are paid for by the HSE, others you can buy on line.
- Synthetic protein substitute drink 4 times a day

Regular blood test

Regular hospital appointments

Regular phone contact with Dietitians and specialized nurses





Challenges

PKU is invisible, rare and unknown

Highly restricted diet

Not having access to the best treatments and low protein foods

MASSIVE amount of planning – 15 to 20 hrs per week

Performance in school

Days out, Social gatherings, Holidays

Gaining independence

Maternal PKU





Hopes for the future

Access to all available treatment options in Ireland:
I want to find out if my children will benefit from kuvan, I've seen how it benefits other people and if so I want it to be prescribed to them

More awareness of PKU – PKU Association of Ireland.

ESPKU conference

Learn from experiences of other people with PKU

Implementation of ESPKU Guidelines

= Best possible long-term health outcomes

