



## 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

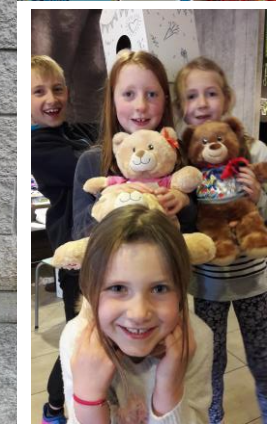
MASSIVE amount of planning – 15 to 20 hrs per week

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

