Lola

LIVING WITH PHENYLKETONURIA (PKU)

Lola's life has changed dramatically and so has her family's.

"Lola lives on only 4 grams of protein a day, living daily with dietary therapy (managing a highly restricted diet) to prevent irreversible neurological consequences."

Lola's family has been on a rollercoaster ride of emotion since their daughter's diagnosis of PKU in 2011.

Lola's mother had never heard of or understood what this condition was before Lola's birth. She recalls her feeling learning about PKU for the first time, and already with children, as beyond challenging; "We have often not been sure if the family can go out because Lola was unable to eat anything, due to the 85 % restriction on all foods which has a knock on effect on how we live our lives".

Over 800 Irish people (1: 4,500 people), live with the challenging rare condition Phenylketonuria, commonly known as PKU in Ireland today, one of the highest and most severe occurrences in Europe. This genetic lifelong disease, diagnosed at birth by the heel prick test, affects a person's metabolism - impacting their ability to breakdown protein.

Phenylketonuria requires daily treatment for the individual living with PKU, with the person caring for them managing the strict, daily dietary therapy ("diet for life") to prevent devastating neurological consequences.

10 year old Lola is a bubbly, inspiring girl living with classic PKU. Lola lives on only 4 grams of protein a day, living daily with dietary therapy (managing a highly restricted diet) to prevent irreversible neurological consequences. A typical adult (not living with PKU), consumes up to 60g or more per day.

PKU dietary therapy is extremely challenging, with up to 85% of foods restricted from the diet which means not eating – nuts, bread, meat, fish, dairy, eggs, pasta, oats, among many other items and instead eating medical foods.

Over the past number of years the PKU Association of Ireland has campaigned for the first ever drug to treat PKU – Kuvan, to be made available to families in Ireland. In 2019 after 10 years of campaigning the HSE granted approval for reimbursement of Kuvan.

In February 2021, Lola's mum received a phone call from the metabolic team in Temple Street offering a trial of Kuvan for Lola. The rest is history.

Lola and her family are overjoyed with the results. Taking the medication Kuvan has had a hugely positive impact on quality of life.

All has now changed a result of the new life changing drug Kuvan, available to Lola and members of the PKU community who are genetically compatible with Kuvan, secured through campaigning and the drug reimbursement programme.

Since Kuvan, Lola's quality of life has improved significantly and her family has felt a significant stress relief for her entire family - mum - Hazel, dad - Alan & brothers, Jamie 16, Dylan 13. Lola's new dietary protein increase (from 4g to 17g) now enables her to eat almost all key foods for body & brain development.

Lola's life has changed dramatically and so has her family.

The PKUAI & PKU community are elated with this positive stories of improved quality of life for people living with PKU .

For more information please visit - pku.ie.