



PROFILE

Killian Fitzgerald

LIVING WITH PHENYLKETONURIA (PKU)

“85% of real food is off limits”

Killian Fitzgerald from Limerick has Phenylketonuria (PKU). As an adult Killian finds weight management to be a very challenging aspect of the metabolic disorder as 85% of real food is off limits and the replacement medical and low protein foods are highly processed. Killian also found the transition from child to adult services difficult as the level of contact with the medical team is significantly reduced.

Killian regularly attends PKU Association of Ireland (PKUAI) meetings and receives tremendous support from those within the PKU online community. As a child he could never have imagined having friends that live with this rare metabolic disorder. It was not until he was fifteen years old that he first met another person with PKU and takes great comfort in knowing that he is not the only one living with the challenging metabolic disorder PKU,

‘We need more awareness. In my whole life I would say that five people knew what PKU was when I explained to them that I had it. I would love to see this change.’

RARE DISEASE

About Phenylketonuria (PKU):

PKU is a rare genetic disease that affects a person’s ability to breakdown protein resulting in ‘restricted low protein dietary therapy’ for life. It is diagnosed through newborn blood-spot screening. If unmanaged PKU can lead to profound irreversible intellectual and physical disability. There is currently no cure for PKU. It is managed in Ireland through restrictive dietary therapy since the introduction of newborn screening in 1966. Research indicates that 60% of adults and 70% of adolescents do not strictly adhere to dietary therapy and it still leads to suboptimal outcomes signifying the real need for additional medical supports. Kuvan® a medicine benefiting a subgroup of PKU patients was recently approved for reimbursement in Ireland, ten years after EMA authorization, however the lack of HSE resources means it cannot be prescribed for eligible patients. Ireland has one of the highest incidences of PKU in the world at 1:4,500 compared with 1:10,000 worldwide.