

Meeting minutes 20<sup>th</sup> November 2018 Moran's Red Cow Hotel

Attendees: Fergus Woodcock, Karen Willetts, Karen Lavery, Amy Meegan (Nutricia), Pauline Woodcock, Colm Galligan, Laurin Grabowsky (Mater staff), Alison Sheerin (Mater staff), James O'Byrne (Mater staff), Kelly Ann O'Connell, Joe Quinn, Aine Brodwin, Gavin O'Donnell, Bernadette Gilroy (via skype).

PKUAI Charity status: The application for PKUAI charity status has proven to be a difficult process with many challenges. We will continue in this objective and we are hoping for a positive resolution in the coming months.

Kuvan update: we remain hopeful and believe we will get some news on Kuvan following the Oireachtas Joint Committee on Health which will take place 21<sup>st</sup> Nov.

UPDATE: Michael Barry from the NCPE confirmed that Kuvan has been recommended by the NCPE on a phased basis, has been discussed by the HSE drugs group and now lies with the HSE Leadership team.

New low protein products: Access to new products, in particular GMP products, were discussed. Cambrooke and the Mater are hoping to start a tolerance test study for their products which is a requirement for an application to the HSE. This all takes a lot of time and can be very frustrating for patients. There was a discussion on whether PKUAI should challenge the HSE on their complicated and time-consuming application requirements.

Delayed blood results: Delayed blood results had been discussed at a previous PKUAI meeting. Blood results are being delayed too often. Approximately 40 delayed results over a 7-year period due to the analyser breaking down or staff shortages. A home blood monitoring kit would be very helpful but this is unlikely to be possible for a number of years. There is an extra long delay for adults as result goes from Temple St to the Mater then to the patients. This is even the case for pregnant PKU women where any delay is even more concerning. The Mater are looking to launch a text message system but this likely won't happen before March 2019. The PKUAI drafted a letter to Temple st noting our concerns and asking how the PKUAI might support the unit going forward in this regard. Fundraising for a new analyser or advocating for additional lab staff has been suggested by our members.

PKU research prioritization – UCD HRB PPI Ignite. Bernadette, on behalf of PKUAI, recently applied for funding through the UCD HRB PPI Ignite programme. We are grateful to Philip Watt of CF Ireland who acted as a co applicant to ensure that we met the eligibility criteria as we await charity status. We are delighted that our application was successful and we were awarded €1,500 to have a PKU research prioritisation workshop delivered to give our members. It will be an opportunity to voice our opinions on our needs and priorities when it comes to pku research. It is hoped that the information gathered will inform health researchers on our organisation's research priorities. Staff from the Mater who attended the meeting and Bernadette Gilroy said they would seek possible venues. We will notify our members once a facilitator has been appointed and a suitable venue confirmed.

IT Tralee Research: Recruitment for research entitled 'The self-expressed quality of life of Irish adults with early diagnosed PKU on long term dietary therapy' is taking place. Research is essential for ascertaining where resources should be spent. All PKU adults were encouraged to participate. If you are interested please see more info on [pku.ie](http://pku.ie) and/or get in touch via email to [info@pku.ie](mailto:info@pku.ie)

Rare Diseases Ireland: Rare Diseases Ireland (*formerly GRDO – the Genetic and Rare Disorders Organisation*) is a non-governmental organisation with the mission to act as a national alliance for voluntary groups representing the views and concerns of people affected by or at risk of developing genetic or other rare diseases. A key activity for Patient Organisations, including PKUAI, in the rare diseases community in Ireland is advocacy. In order for our patients to have equitable access to the best treatments and services it was agreed we must work collectively as part of RDI to ensure that we speak with a united voice on behalf of the PKU community in Ireland. PKUAI will join RDI.

IPPOSI Access to Medicines event: Karen Willetts and Bernadette Gilroy recently attended an IPPOSI Access to Medicines event on behalf of the PKUAI. Bernadette as spokesperson was invited to speak on the PKUAI's experience of HTA and reimbursement. Bernadette was asked to explain PKU as part of her introduction to the other patient organisation representatives in attendance, many of whom were unfamiliar with pku. She spoke about the challenges associated with dietary therapy the only form of treatment in Ireland highlighting current research to support this claim and the limited variety of food products available which has significant implications on adherence particularly for adolescent and adult patients. She highlighted PKUAI's frustration for medical food companies having to complete a 30-day trial for compliance and palatability compared with 7-day trials elsewhere with no reasoning behind this time period. The system lacks transparency, accountability and timelines. On a positive note, she was delighted to speak about our organisation's involvement in the clinical pathway document for Kuvan with Clinicians and the NCPE. We felt empowered, valued, respected and trusted as a patient group to be part of the process and we remain hopeful for a positive outcome on kuvan.

ESPKU Conference: PKUAI were represented this year by Karen Willetts, Bernadette Gilroy and Colm Galligan. There were 523 attendees, 140 with PKU from 38 countries. The following feedback was given;

- Lots of talks on GMP-glycomacropeptide (= 'big sugared protein')- formulations still need to contain ~30% raw amino acids. Likely to cause increase in PHE levels. Needs very careful introduction.
- LNAAs (large neutral amino acids)- in usual protein substitutes, can help block high blood levels of PHE from crossing into brain where it causes damage. Potential to take much higher amounts in tablet form to help protect brain from PHE.
- Home PHE testing- multiple devices in development. 1 of them is undergoing FDA review
- Tremor- can become apparent in people with PKU around the 10 year old mark.

- Sleep- disturbance in people with PKU, especially if poorly controlled. Large scale study to look at this.
- Long term IQ/ executive function- appeared across many studies. Bottom line is that there doesn't appear to be a significant difference between 'normal controls' and people with PKU who have had good control. However, there are limitations in terms of standardizing the types of tests used.
- Short term PHE levels vs long terms average- affects different cognitive functions, but overall normal task accuracy, but at reduced speed.
- PKU doesn't increase biomarkers associated with Alzheimer's. However, there are markers of damage in those who are poorly controlled.
- Exercise- importance of maintaining optimal nutrition levels before, during and especially after exercise in people with PKU to avoid muscle catabolism which released PHE back into the blood.
- Transition from paediatric to adult services was discussed at the Delegates meeting where it was highlighted that this can be tricky step for patients and support should be provided. A pilot programme for transitioning was taking place in Sweden and was used as an example of what could be provided. The Mater are looking at the 'Ready steady go' process by NHS and how it could be brought to Ireland.
- An education session took place on Health Technology Assessments where it was outlined what an HTA is and how they work. We then discussed the limitations of HTA's where rare diseases are concerned.

European patient's advocacy Forum: Karen gave us an update from this event which she was invited to attend by Biomarin. The session was about the power of effective data-driven storytelling. The key note speaker was Declan Noone from the European Haemophilia Consortium who demonstrated how data can be used to support messaging. The take home message is that surveys, such as the one carried out by PKUAI in 2016 to back our Kuvan submission to the NCPE, are highly effective.

AOB and next date: PKUAI will have a strategy meeting on the 2<sup>nd</sup> of 3<sup>rd</sup> week of January and the next open to all meeting will take place on the 28<sup>th</sup> February.