



Wednesday, 08 November 2023

Society for the Study of Inborn Errors of Metabolism (SSIEM)
South Croydon, CR2 1PN United Kingdom
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Dear Caroline and SSIEM team,

NSPKU was awarded €3,500.00 from the SSIEM for the National Conference in 2022.

Here is a report about 4 of the sessions we had in our programme of talks.

NSPKU Conference 2022

PKU Ups and Downs – Expert panel of people with PKU – D, G, C, S, H and P.

Challenges were discussed - for parents, for instance G said, “the shock of the diagnosis”. For adults with PKU: the times when blood phe levels might be in range, and clinical teams say “well, your levels are fine, so you must be ok”, as H described; or when “I can be remembering past experiences and becoming anxious” as S told us.

G said she would have given anything for someone to tell her, after the diagnosis “everything will be alright”. On the plus side, the PKU community and its kindness needs to be celebrated – and celebrating small wins like getting the right protein substitute that you or your child likes (lots of nods!).

Discussion about what can NSPKU do to help:

The NSPKU has work to do to support research in PKU. The NSPKU needs to include all of the community and support people who have been lost to follow up, who are off diet and who might want to return to diet.

R & A on PKU pregnancies

We were grateful to R and A for their talk of this rarely discussed topic that affects many people with PKU. R and A covered a wealth of suggestions and comments – here’s a 3 point summary:

1. The preconception and pregnancy experiences are intense but temporary – it will pass!
2. Be organised. For instance, get yourself a “blood spot station”, cupboard(s) stocked with low protein foods and up to date prescriptions for products you like; when eating out – PLAN and take scales!
3. Ask for help – especially from your dietitians. Get the support of all your family and friends – colleagues too, make sure they understand how important your diet and meal plans are.



Rachel Carling, Consultant Clinical Biochemist, Viapath Labs & Guys & St Thomas' NHS Foundation Trust

Rachel introduced the capitainer device - a more precise and accurate tool than a standard bloodspot card. Blood is still collected from a finger (or heel) prick but instead of letting drop of blood fall onto card, a blood drop is 'sucked up' by the device. The device collects a fixed volume of blood and forms a perfect spot. Formal evaluation of capitainer performance compared to the existing DBS card is needed and a lab-based study has already shown improved accuracy and precision, which is great news. Rachel invited the audience to help evaluate the capitainer and many people went home with devices and feedback forms with some very positive feedback.

Genetics and PKU - Rebecca Whittington, Principal Clinical Scientist, Bristol Genomics Laboratory, North Bristol NHS Trust: PKU Genotyping activity at the end of September 2022:

>900 samples received from >30 centres. The lab is funded to test 100 samples per month and at the point of the conference, Becky explained that >500 patients' genotypes have been reported with >100 different genotype combinations. For about 2% patients only 1 genetic variant of two has been identified. Becky explained the entire genotyping process and the audience valued this explanation, which helped them understand the timelines involved.

The conference was very well evaluated, and we are already planning our 2024 offer.

The SSIEM monies enabled us to ensure that we could support families and adults with PKU who have low incomes, to access our conference by giving additional subsidies or covering travel expenses.

Thank you to SSIEM for making the monies available to patient organisations and in particular awarding NSPKU €3,500.

Yours sincerely

Suzanne Ford

Dietitian for the National Society for Phenylketonuria, for an on behalf of the Society