

CCBT WORKSHOP



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Content :

- **What is PKU**
- **NSPKU/ESPKU**
- **Working relationships**
- **Difficulties**
- **The future**

What is PKU?

- IEM – recessive gene - RD – average incidence in Europe is 1:10,000 (Turkey 1:3,500/Eire 1:4,500)
- Enzyme PAH not working – high levels of amino acid PHE – carried in blood across blood/brain barrier > severe mental retardation/fitting/eczema et al
- Discovered by Folling>first patient fed LP diet by Bickel >NBS Guthrie
- Diagnosed at birth by heel prick test
- Treatment – lp **diet for life** – inc. amino acids/multi vits etc

The diet

- LP diet – lp flour /bread/pasta/pizza bases / biscuits
- Supplemented by amino acids/multi vitamins
- Red-Amber-Green system for food/shopping
- Red – meat/poultry dairy produce/fish/nuts-pulse/diet drinks-aspartame/sweets-gelatine
- Amber eg 1 banana a day / ½ an avocado.....
- Green - peppers/courgettes/carrots/tomato sauce/jelly tots

- Some protein allowed naturally – in system of exchanges
- Exchange = 50mg Phe=1gm protein (typically) eg 30 mls milk = 8 baked beans = 45 gms chips
- Exchanges dependent on tolerance – very mild/mild/classic PKU
- Tolerance is measured in blood Phe - UK 180-450 micromoles /adults up to 700 mmls
- >slide with different tolerances in different countries
- Classic PKU = low exchanges e.g 2-5
- Diet > can add up to an extra 17-19 hrs week (Prof A Macdonald study ref?) -in food preparation

NSPKU 1973

- Formation – structure – co. Ltd by Guarantee (1975) run by an elected Council of Management- with Dietitian/Administrator/Bookkeeper.
- Adherence to Charity Commission guidelines
- MAP= Medical advisory panel meets x 2 per year
- Meet regularly up to 6/7 time a year to discuss issues +organise An Conference
- Produce 4 issues of N & V
- Edale/Keswick weekends for children

ESPKU 1987

- Umbrella Organisation covering 36 +European PKU Associations
- Recently admitted Belarus , Palestine (Gaza) + Israel!!!
- Board (5) – SAC=Scientific Advisory Committee.
- An Conference – different countries
- Speakers invited from all over the World.
- 2012 Consensus report published in Orphanet News > call for Guidelines in treatment of PKU from experts
- 2015 Guidelines – ready to go – update every 18mths/2years
- (“Better than US Guidelines because more time and effort spent on them” not rushed out)
- Goal – World Guidelines – areas of commonality with US

Working relationships

- UK – clinicians & dietitians / industry / other like minded organisations such as GIG & RD UK
- ESPKU – as above – add Academics/ PKU Country Delegates/ EU administrators/ EU Health Attaches / Eurordis / other PKU Associations such as NPKUA and CanPKU

Difficulties

- The diet – administering – teenage rebellion – staying on diet in adulthood – maternal PKU
- Time – we are all volunteers
- Access to treatment
- Education
- Politicians/policy makers

UK-issues

- Differences in regions in access to care—
differences in clinicians opinions
- No Access to BH4
- Doctors refusing to prescribe lp food
- Engagement within regions
- NSPKU has been a “sleeping giant” for years
- Run by volunteers

EUROPE

- EU legislation unintelligible sometimes – too technical /European English!
- Differences in delivery of care in countries
- No NBS in some countries
- Lack of dietitians (eg Russia)
- Lack of metabolic consultants
- Engagement of Countries (wary)
- Politics/policy decision makers

The Future

- Equal access to treatment & care eg UK access to BH4
- PegPal – on the horizon 3rd phase of trials ...taking a long time (NSPKU contributed to research on this some 12 years ago)
- Other better chaperones for PAH being looked at
- University of North Texas looking at "Genetically Engineered Probiotics for the Treatment of PKU". (2012)
- Similar project going on in Australia
- Home blood tests – has been tried for > 15 years without success , but alleged breakthrough in US – youtube video –we'll see
- European Guidelines will empower Patient Organisations-but will clinicians abide by them - already mutterings by 1or 2 in UK
- Patient Organisations need to be more adversarial
- Every one know about CF (1:6000) but not PKU (1:10000)

Thank you.