Report on Participation meeting
European Society for Phenylketonuria allied disorders treated as Phenylketonuria
Liverpool , October 19- 20, 2012

Liesbeth Siderius
Paediatrician
Representative of Dutch Shwachman Diamond Syndrome patient organisation
EAP, rare disease working group

Phenylketonuria is one of the rare disorders which can be screened in the neonatal period with blood taken by heel prick test, as practiced by many countries for many decades. The experiences of patients and physicians as issued at the European Society for Phenylketonuria allied disorders treated as Phenylketonuria (E.S.PKU) meeting are of interest for others involved in rare diseases.

The board of the E.S.PKU has requested the scientific board to produce international **Guidelines on the Management** of PKU. Then first meeting was held at the October Conference in Liverpool.

Different groups are formed by professionals that both have experience and are internationally known as being a PKU expert based on their publications on PKU. Aspects such as Neurocognitive development and imaging, Psychosocial aspects, Nutrician / diet and drug treatment/ prevention, Adult and maternal care, as well as the care of late diagnosed patients will be addressed.

Persons with PKU are treated with a diet to prevent developmental delay and behavioral problems. The treatment exists on a reduction of natural protein diet with supplementation of an mixture of all amino acids except phenylalanine. To achieve this the use of special protein substitutes (also called amino acid mixtures) are necessary as products free or very low in natural protein are required. The costs are estimated at minimum of about €10.000 / year in the UK. There are considerable differences in reimbursement and availability of the dietary products in Europe as well as the US. The E.S.PKU has informed members of the European parliament about the **inequalities in treatment** to prevent mental disability in Europe.

Management is evaluated by **regular blood tests** of Phenylalanine. Persons in the UK send a blood sample self collected at home and get the result by phone or email. Other blood tests such as follow up on minerals and vitamins may be necessary. Discussions on the necessity of standard brain MRI or the need of specific neurcognitive testing at specific ages are ongoing.

E.S.PKU also represents persons with other -more rare- metabolic disorders such as Type 1 Tyrosinemia (1:100.000). Information and experience on these disorders is scarce, parents become experts in management.

For Urea Cycle defects a guideline has been developed. The method of the Scottish Inter-colleague Guideline Network was used. The issue of evaluation of international guidelines was discussed.
For Urea Cycle defects the emphasis on early recognition is on educating neonatologist on blood ammonia test in cases of neonatal sepsis not responding on antibiotics.

What can be learned from this patient – professional PKU meeting?
- The process of making international guidelines can be **harmonized** : in all elements of (early) diagnosis, preventive collaborative management.
- Patient/ families can play an major role as **co - managers** and evaluating the care provided.
- Some of the test used in to **monitoring** in PKU (such as vitamins and minerals) are also applicable to Shwachman Diamond Syndrome.
- Electronic / **digital data collection can support communication and data registration** at the same time, and so a process of quality improvement.
- This all fits in the chronic care model / **medical home concept** endorsed by the European Academy of Paediatrics.

LS November 10, 12