

## MAINTAINING AND IMPROVING QUALITY IN THE DETECTION OF INBORN ERRORS OF METABOLISM

In 1966 Donabedian proposed three questions which could be asked about the quality of any health care system:

- **Structure** – *The suitability of resources staff, training, buildings, equipment etc.*

The Network is becoming pro-actively involved in many aspects of staffing by:

- The proposal to create trainer and higher specialist trainee posts
- Helping re-design a competency framework for Paediatrics and inherited metabolic disorders
- Dialogue regarding the design and appropriateness of the MRC Path examination for this area.

Discussions agreed with CPA should ensure that there is a clear understanding of the overall levels of staffing and equipment needed to provide a safe and reliable service.

- **Process** – *This includes test selection, methodology (including internal quality control and external quality assurance) and results interpretation and their use.*

The Network is approaching some of these issues by the production of “best practice guidelines” which should aid test selection and interpretation and analysis specific workshops, which will focus upon methodology. This will begin to be available later in 2003.

External quality assurance when establishing lifelong diagnosis is clearly essential. Particularly so when one considers the marked biochemical and clinical heterogeneity associated with these conditions and very often the limited amount of sample available in a crisis situation with which to secure a diagnosis. The relative rarity of the disorders has led to European and International collaboration to provide suitable schemes.

At a National level, NEQAS is involved with schemes relating to urinary amino acid analysis, mucopolysaccharidoses and orotic acid. At a European level ERNDIM offers schemes relating to: qualitative and quantitative urinary organic acid analysis, quantitative amino acid analysis, special assays (intermediary metabolic etc), purine/pyrimidine analysis, acyl carnitine analysis and white cell cystine assay. In addition ERNDIM offers participation in Regional proficiency schemes where laboratories are provided with clinical details and a sample, they are then required to choose the investigations to be undertaken and interpret the results

### ***Special Event***

On 22<sup>nd</sup> October 2003, there will be the first of an annual series of QA workshops where participants can learn from one another and discuss the findings from established schemes:

This year, the proficiency scheme for UK and Scandinavia and the National MPS scheme will be discussed. The meeting will also include a workshop to consider interpretation of quantitative amino acid analysis.

This meeting will be held in Sheffield on Wednesday, 22<sup>nd</sup> October 2003. Please contact Dr J Bonham ([jim.bonham@sch.nhs.uk](mailto:jim.bonham@sch.nhs.uk)) or fax 0114 276 6205 to reserve a place. The cost will be £15.00 and the meeting will last from 11.00am until 4.00pm.

- ***Outcome*** – *audit is the chief means of establishing outcome in health care.*

In the study of rare disorders it is particularly important that collaborative audit and research underpins practice. In future years the Network will develop suitable audit projects to inform and improve practice.