

National Metabolic Biochemistry (Biochemical Genetics) Network

Update on Current Activities January 2005

1. Training Strategy

Trainers/Trainees

- i. The 8 additional supernumerary Clinical Scientist training posts for paediatric metabolic biochemistry have been appointed and all individuals are now in post.
- ii. The lead trainer post and the local trainer posts (North, South & Midlands) in 3 training centres across England have been appointed and are all in post.

The training group will meet on February 1st to agree priorities and objectives for 2005/6.

Training Workshops

Workshops completed in 2004 were

- urine organic acids
- lysosomal storage disorders
- non-lysosomal enzymes

Reports of this are on the website

Workshops organised for 2005 are

- acylcarnitines (Jan 2005)
- carbohydrate disorders (March 2005)
- mitochondrial disorders (May/June 2005)
- amino acid disorders (Nov/Dec 2005)

2. Web Site - www.metbio.net

The metabolic assay directory (MAD) is in the final stages before launch. Plans to link with the UKGTN website are under discussion.

An electronic learning package on MCADD has been commissioned as a possible model for other subject matter.

3. Best Practice Guidelines

Best-practice guidelines commissioned by the Network now available on the web (www.metbio.net) are:-

Hydrops and Inborn Errors of Metabolism (IEM)

Seizures and IEM

Hypoglycaemia and IEM

Hyperammonaemia and IEM

Others in final draft form are:-

- rhabdomyolysis and IEM
- Skin biopsy information sheet and consent
- Developmental Delay and IEM
- Neonatal jaundice and IEM

4. Quality Assurance

The second annual meeting took place on 29th October 2004. This was well attended and has stimulated several new initiatives including provision of information about schemes to be made available via the website.

5. Audit

A review of audit activities across all labs had been completed. A national audit across all centres on the availability of certain tests is being planned and available as a summary for 2005.

6. Service Needs

- i. The Network is contributing to the JCMG National exercise on IMD Service Needs (Hilary Burton).
- ii. Neonatal Screening.
The impact of neonatal screening on diagnostic metabolic services (staffing and other resources) is a significant developing issue.

7. Other Issues

- i. National Tarriffs - The Network is working with the specialist commissioners as part of a genetic lab initiative.
- ii. Agenda for Change (A for C) - The impact of A for C on posts/services is to be monitored.

An Annual Report (2004/5) for the Network is available via the Network Administrator, Mary Dowling (mary.dowling@bch.nhs.uk).

Dr. Anne Green
Lead Scientist
National Metabolic Biochemistry Network
January 2005