

National Metabolic Biochemistry (Biochemical Genetics) Network

Update on Current Activities July 2005

1. Training Strategy

1.1 Trainers/Trainees

The training strategy is progressing well under the leadership of Dr. Mick Henderson.

The following resources are currently in preparation:-

For specialist HSTs

Training syllabus and logbooks
Training days

For Pre-registration clinical scientists in general training

Collaboration with ACB regional tutors to plan and organise placements in specialist laboratories
Define curriculum of core training
Contribute to formulation of MRCPATH examination
Various website resources are also under production - see below

2. Workshops

Workshops on carbohydrate disorders and mitochondrial disorders took place in April and June 2005 respectively.

Further workshops organised for 2005 are:-

- Amino acid disorders (22nd November, 2005)
- Associate Labs (? March 2006)

3. Web Site - www.metbio.net

- The metabolic assay directory (MAD) was successfully **launched on May 1st** and is working well. A disorder search facility is under development.

The general functionality of the site has been improved, and current developments include:-

- ♦ An email messaging system
- ♦ Training and Education

Specific training and education resources to be created are:-

- ♦ Case based learning package
- ♦ Database of chromatograms
- ♦ Collection of clinical cases
- ♦ Collection of relevant lectures
- ♦ Educational links to other sites

4. New Guidelines commissioned are:-

- Unexplained liver dysfunction and metabolic disorders in the older child
- Dysmorphology and inherited metabolic disorders
- Unexplained tubular function and inherited metabolic disorders
- Unexplained neurology in adults with inherited metabolic disorders

5. Quality

The annual Quality Day will take place 18th October 2005.

A review of specialist QA schemes has been completed and is now on the web site

6. Audit

A national audit on ammonia measurement across all centres has been undertaken. A re-audit is planned for late 2005.

7. Service Needs

The Network has contributed to the JCMG National exercise on IMD Service Needs (Hilary Burton), with information and diagnoses across the UK. Data from the 2004 questionnaire on manpower, equipment and workload has also been summarised. These data will be available as a full report.

8. Publications

The Network presented a poster at the European Meeting on Laboratory Medicine in Glasgow May 2005.

A poster focussed on training and education will be presented at the SSIEM meeting in Paris in September 2005.

Professor Anne Green
Lead Scientist
National Metabolic Biochemistry Network
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