

National Metabolic Biochemistry (Biochemical Genetics) Network

UPDATE ON ACTIVITIES JUNE 2003

Infrastructure and Administration

Office space established with mailing details for stakeholders, associated members, professional bodies, national group links, and circulation lists. Budgetary arrangements established via Finance Department at Birmingham Children's Hospital as host institution.

Formation of Stakeholder Group and Agreement of Objectives

A stakeholder group of 17 members across the UK established. Objectives and work plan for 2003/4 agreed at stakeholder meeting on February 10th, 2003.

Training initiatives

Training workshops are planned for 2003/4 on the following topics:-

- Organic acids
- Acyl Carnitines
- Very long chain fatty acids
- Lysosomal enzymes

These will be specialist workshops for scientists (clinical scientists and biomedical scientists) currently working in specialist laboratories.

Quality Assurance

Overseeing of quality assurance has been handed over to the Network from the British Metabolic Disease Quality Group. Remit includes:-

- How are the stakeholder laboratories performing in specific schemes?
- Are existing schemes appropriate to our needs?
- How could/should they improve?
- Do we need new schemes?

Specific QA Initiatives

An initial meeting is planned for October 22nd 2003 for all stakeholders organised by Dr. Jim Bonham in Sheffield.

Best Practice Guidelines

A series of best practice guidelines has been commissioned by the Network to be lead by individuals from within the Network. These are:-

- Metabolic autopsy (sudden unexpected death in infancy)
- Inborn Errors of Metabolism (IEM) and hydrops Inborn Errors of Metabolism and cardiomyopathy postmortem
- Hypoglycaemia and IEM
- Hyperammonaemia and IEM
- Developmental delay and IEM
- Fits and seizures and IEM
- Prolonged neonatal jaundice and IEM
- Lactic acidosis and IEM
- Rhabdomyolysis and IEM

It is hoped that some of these will be completed during 2003 with the remainder during 2004.

Where some of these guidelines overlap with other initiatives, e.g. by the British Inherited Metabolic Disease Group, there is liaison in order to produce a jointly owned guideline.

Web Site - www.metbio.net

The Network web site has been commissioned via Professor Bartlett and Dr. Neil Hamilton, University of Newcastle-upon-Tyne. The web site has now been designed and launched with stakeholder details (April 30th, 2003). The next stage is to expand to provide additional sites for a test/assay data base, education and training site, and details of best practice guidelines.

Questionnaire on Service Provision

It has been decided to delay this by a few months to coincide with data collection for the purposes of establishing the test/assay data base on the web in order to avoid two separate questionnaires.

Training and Recruitment

The difficult position with regard to recruitment to clinical scientist posts continues with unfilled posts and no Grade B scientists in training. Discussions took place with the Royal College of Pathologies and the Association of Clinical Biochemists during January 2003, and the following agreed:-

1. To adapt the MRCPATH examination in clinical biochemistry to include more options for specialist questions on paediatric metabolic biochemistry
2. To create training HST posts (grade B clinical scientists) for paediatric metabolic biochemistry. The training provided by the specialist HSST post would match the options within the MRCPATH examination for paediatric metabolic biochemistry.
3. Create a sub-modality of clinical biochemistry for clinical registration purposes as metabolic/paediatric biochemistry. This would enable recruitment of more specialised postdoctoral scientists with a specific expertise, e.g. in enzymology, chromatography, for whom a wide based and comprehensive general biochemistry training is not essential for the job.

A Working Group (Lesley Tetlow, Mick Henderson, Jim Bonham and Anne Green) was convened to take these initiatives forward. Progress is as follows:-

1. **MRCPATH**

The modifications to the MRCPATH have been agreed with the Chief Examiner and more questions, both theoretical and practical, are to be included.

2. **Creation of HST posts**

There has been no significant progress with this need. This is largely due to the inability to progress discussions at a national level with the Workforce Development Confederations in order to produce a national strategy. Efforts to engage with the Chief Scientist are continuing via the RCPATH/ACB.

3. Competences in paediatric/metabolic biochemistry are being prepared in order to request registration as a sub-modality. These are being progressed by the Association of Clinical Biochemists for submission to the Association of Clinical Scientists.

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