

## National Metabolic Biochemistry (Biochemical Genetics) Network

### Update on Current Activities April 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

A successful workshop on non-lysosomal storage disorders took place in December 2004. It is clear that there is scope to rationalise some services and a need to develop others to make services more robust and equitable across the UK. Dr. Ying Foo (Great Ormond Street) will take a lead on this together with Dr. George Gray (Birmingham)

Workshops organised for 2005 are:-

- Carbohydrate disorders (25<sup>th</sup> April 2005)
- Mitochondrial disorders (15<sup>th</sup> June, 2005)
- Cystic Fibrosis (21<sup>st</sup> June, 2005)
- Amino acid disorders (22<sup>nd</sup> November, 2005)

#### 3. Web Site - [www.metbio.net](http://www.metbio.net)

- The metabolic assay directory (MAD) is to be **launched on May 1st**. The next stage will be to develop a disorder search facility
- An email messaging system has been commissioned for Stakeholder and Associate laboratories
- Plans to develop the web site further for training purposes are under discussion/preparation

### Website resources to be created

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

### 4. Best Practice Guidelines

Best-practice guidelines commissioned by the Network now available on the web:-  
Hydrops and Inborn Errors of Metabolism (IEM)

Seizures and IEM

Hypoglycaemia and IEM

Hyperammonaemia and IEM

Skin biopsy information sheet and consent

Others in final draft form are:-

- rhabdomyolysis and IEM
- Developmental Delay and IEM
- Neonatal jaundice and IEM

New Guidelines under production 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 in October 2005.

A review of specialist QA schemes has been completed for inclusion on the web.

### 6. Audit

A national audit across all centres on the availability of certain tests is being planned.

### 7. Service Needs

The Network is contributing to the JCMG National exercise on IMD Service Needs (Hilary Burton) and has completed questionnaires on workload and IMD diagnoses.

### 8. Publications

The Network has had a poster presentation accepted for display at the forthcoming European Meeting on Laboratory Medicine in Glasgow May 2005.

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