

National Metabolic Biochemistry (Biochemical Genetics) Network

Update on Current Activities April 2004

1. Training Strategy

Approval has been agreed

- i. To create 10 supernumerary training HST posts (grade B clinical Scientists) for paediatric metabolic biochemistry (8 additional posts - 2 already funded)
- ii. To create a lead trainer post and local trainer posts in 3 training centres across England. The lead trainer role will be advertised in the ACB News Sheet (April)

Training Workshops

A successful workshop, organised by Guy Besley, took place in February 2004

- lysosomal enzymes

Others planned for 2004 are

- organic acids
- acyl carnitines
- carbohydrate disorders
- non-lysosomal enzymes

2. Questionnaire on Service Provision - Current & Future

The questionnaire has been completed with 100% return. Initial analysis has been undertaken and discussed with Stakeholders. Key findings are:-

Core Tests

- Workload relates to population
- Acyl carnitine deficiency for some labs

Turnaround times

- Routine core tests - 30% compromised by Clinical Scientist time

Accommodation for next 5 years

- 50% have inadequate lab accommodation
- 75% have inadequate office accommodation

Equipment

- Urgent replacement for 30% amino acid analyser
- 3 year replacement for 50% of gas chromatography mass spectrometer

Training

- Inadequate training capacity-trainer time a limiting factor

Staffing

- Major issue to recruit Clinical Scientist posts

Future work - possible initiatives

- Workload units
- Disorder register

3. Web Site - www.metbio.net

The quality, best practice guidelines and education and training sites are now functional

The metabolic assay directory (MAD) is under development. A current assessment of tests provided by each centre has been completed, and it is anticipated that the MAD will be ready for launch by August 2004

4. Best Practice Guidelines

The first of the best-practice guidelines commissioned by the Network (hydrops and IEM) is now available on the web (www.metbio.net).

Others are in final draft form:-

- hypoglycaemia and IEM
- hyperammonaemia and IEM
- rhabdomyolysis and IEM
- seizures and inborn errors of metabolism
- developmental delay and inborn errors of metabolism

5. Quality Assurance

A second meeting is planned for October 2004 - details to follow

6. IT

There are plans for a major IT investment in laboratory genetics (White Paper monies). A consultation to identify needs and proposed situations is being undertaken via the Manchester Genetics Reference Laboratory. It is not clear at this stage how this might effect metabolic biochemistry (i.e. opportunity for funding). I have had some preliminary input into discussions and will keep you posted.

7. Future of the Network/Funding

Funding to continue the Network has been granted for 2004/5 and 2005/6. Key objectives are:-

- i. To continue support of the infrastructure for the Stakeholder Laboratories

- ii. To support an educational and training infrastructure across three lead training centres
- iii. To provide on-going support and development of the WEB (in collaboration with the GTN WEB), in particular a Metabolic Assay Directory
- iv. To continue to support specific projects, i.e. best practice guidelines, workshops
- v. To work with the commissioners to help plan service delivery

An Annual Report is in preparation and will be available via the Lead Scientist later this month.

Dr. Anne Green
Lead Scientist
National Metabolic Biochemistry Network
April 2004