

National Metabolic Biochemistry (Biochemical Genetics) Network

NEWSLETTER FOR STAKEHOLDERS

April 2004

CURRENT PRIORITIES AND UPDATE

Survey of Service Provision

The survey has been completed (October - December 2003) and initial analysis presented at February 2004 Stakeholder meeting (copy of presentation circulated with Stakeholder minutes).

A full report will be circulated to Stakeholder in May 2004.

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

A small sub group of stakeholders will be meeting before the June 2004 stakeholder meeting to make recommendations on the way forward.

Web Site

The web site is now functional, www.metbio.net, with the following sections:-

Home page

Stakeholder details

Stakeholder Resources

Links

Services

The next development under way is the assay directory. Do **please** have a look at the web site. If you have any comments and/or any specific suggestions/requirements, please contact Anne Green or Janet Stone.

The directory will have a shared test list with the ERNDIM/SSIEM assay finder. Initial population of the data base will be undertaken by Janet Stone - stakeholders will then be asked to check the input and add specimen requirement details.

We are developing the web site as a training resource, e.g. sharing of presentations, case histories, etc.

Training and Education

Our Training Strategy was completed in September 2003 (see web - Stakeholder Resources).

We have been successful in obtaining support and funding for this from the Department of Health Genetics Division. We have 3 year funding for:-

3 Trainers across England (or equivalent)

8 Higher specialist training posts

Link with workforce planning

Link with ACB

Link with trainees

Registration of Clinical Scientists/ Competences for Metabolic Biochemistry as a sub-modality

Paediatric/metabolic biochemistry has now been recognised as a sub modality of clinical biochemistry - see ACB web site (www.assclinsci.org) for a copy of the competences.

Individual planning to prepare portfolios for registration are encouraged to contact the Network Lead Scientist.

If you have any views about training, registration and the MRCPATH, please contact Anne, Mick, Lesley or Jim.

Training Workshops

Training workshops planned for 2004 are:

- Carbohydrate disorders (Jacqui Calvin)
- Organic acids (Jim Bonham)
- Acyl carnitines (Neil Dalton)
- Non-lysosomal Enzymes (George Gray)

Please contact the organisers to register your interest.

Best Practice Guidelines

Current status is as follows:-

A reminder about who is doing what.

| Guidelines | Status |
|--|----------------------------------|
| Metabolic autopsy (SUDI) <i>Mary Anne Preece</i> | |
| IEM and hydrops <i>George Gray/Mary Anne Preece</i> | On web site |
| IEM and cardiomyopathy postmortem <i>Helena Kemp</i> | |
| Hypoglycaemia <i>Anne Green</i> | First and second drafts complete |
| Hyperammonaemia <i>Anne Green</i> | First and second drafts complete |
| Developmental delay <i>Peter Galloway</i> | First and second drafts complete |
| Fits and seizures <i>Mick Henderson</i> | First and second drafts complete |
| Prolonged neonatal jaundice <i>David Isherwood</i> | |
| Lactic acidosis <i>Jim Bonham</i> | |

| | |
|--------------------------------------|----------------------------------|
| Rhabomyolysis <i>Simon Olphin</i> | First and second drafts complete |
|--------------------------------------|----------------------------------|

The directory of molecular tests is now available on the DOH Genetics Web Site (www.dh.gov.uk)

The Genetic Testing Network (GTN) is pursuing its second round of invitations to apply for GTN membership. If you provide molecular tests and have not been invited, please contact Maureen Boxer.

The Network is to set up a small working group to interface with the GTN in particular to suggest new molecular tests for IMD required for service - Guy Besley is to take the lead.

National Strategy for Inherited Metabolic Disorders Services in the UK (BIMDG)

The BIMDG produced a strategy document for the future; contact Graham Shortland. The issues for clinical IMD have been raised at the Joint Medical Genetics Committee (Chairman Heather Skirton) and we understand there are plans to undertake a needs analysis in collaboration with the Public Health Genetics at Cambridge.

GenCAG Update

The Genetics Commissioning Advisory Group

Accreditation - discussions with CPA

CPA has included paediatric/metabolic biochemistry as a sub-specialty within the CPA accreditation process. We have also provided CPA with a list of specialist QA schemes and a list of tests which defines the repertoire for paediatric/metabolic biochemistry so that these can be appropriately highlighted as part of the inspection process for any lab registering this sub-specialty. next stakeholder meeting in November. In the meantime, I have sent Cheryl Blair at CPA a list of the stakeholder laboratories.

Agenda for Change

There is concern about how metabolic biochemistry will be handled under the national occupational standards (NOS). Generic NOS have been produced for Clinical Biochemistry, but there are currently no specific ones for Metabolic Biochemistry. There is concern about lack of comparability with the other genetic specialties (i.e. cytology and molecular). A small group (Ying Foo, Jacqui Calvin and Lesley Tetlow) has been established to interact with ACB innovations.

Quality Assurance Workshop

The next workshop will be in 2004. It will be for all stakeholders and other laboratory colleagues. Just to remind you that the purpose of the meeting is as follows:-

- Overview of specialised QA schemes - how are we performing in specific schemes
- Are existing schemes appropriate to our needs - how could they improve? Do we need any new schemes?
- Specific QA projects. Which individual labs have undertaken/relevant to the National Metabolic Biochemistry (Biochemical Genetics) Network

Further details will be available from Ying Foo, fooy@gosh.nhs.uk, and on the web.

Anne Green
April 2004