

**National Metabolic Biochemistry Network  
(Biochemical Genetics)**

**REPORT PREPARED FOR THE  
DEPARTMENT OF HEALTH**

**Network Progress Report  
October 2002 – June 2004**

**June 2004  
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## **SUMMARY – HIGHLIGHTS**

- Network infrastructure and communication links established
- National overview of service provision completed
- Plans for National Strategy for Training for Clinical Scientists, with funding for trainer posts and higher specialist trainees secured, Lead Trainer appointed (June 2004)
- Paediatric Metabolic Biochemistry now recognised as a sub modality for registration of clinical scientists with the Health Professionals Council
- Comprehensive and up to date lists of metabolic biochemistry (biochemical genetic) tests provided by UK laboratories completed

## **BACKGROUND**

In April 2001 the Secretary of State for Health announced a commitment to place genetic services in the NHS at the leading edge of international development. The detail of this statement included an undertaking to increase the number of consultants, scientific and technical staff and genetic counsellors, and a doubling of test numbers by 2005. In July of that year, the laboratory services for Inherited Metabolic Disorders (Metabolic Biochemistry) were included within the Medical Genetics

Definition set 20, and consequently became part of the specialised services genetics commissioning arrangements overseen by the National Genetics Commissioning Advisory Group (GenCAG).

At the same time there was a growing recognition that the laboratory services for metabolic diseases were under increasing pressure, the chief concerns being:

- The increasing workload and repertoire of metabolic biochemistry tests, and the increased expectation/demands for service including availability outside normal laboratory working hours
- Poor recruitment of clinical scientists
- Lack of a national training plan for clinical scientists in this speciality
- Difficulties in maintaining a robust service in some centres due to inadequate trained staff and/or equipment needs

These concerns were exacerbated by the lack of a robust manpower plan; despite the anticipated retirement of several senior scientists within the next five years few plans appeared to be in place to recruit and train additional staff for succession planning. An application for funding to the Department of Health (DOH) to form a Network of laboratories providing these services was submitted. The plan in outline was to form a National Stakeholders group of laboratories whose major function was the provision of these services. The stakeholder group would be co-ordinated by a "lead scientist" and would address the key issues.

The DOH provided funds for the network for an initial for a two year period (2002/3 and 2003/4) and have subsequently extended funding for 2004/5 and 2005/6.

### **SYNOPSIS OF THE PROJECT - AIMS**

- Provide a baseline assessment of current services including their scope, capacity, staffing and equipment
- Perform a risk assessment of the robustness of current services with a view to developing risk sharing and back-up arrangements
- Assessment of equity of access on a National basis
- Undertake manpower planning for the specialty
- Assess national training needs in collaboration with workforce development confederations
- Provide better information and advice to liaise with accreditation bodies
- Plan testing for the very rare disorders to ensure National availability
- Monitor the adequacy of existing EQA schemes and develop promote development of new schemes as needed
- Promote best practice guidelines for investigation
- Promote clinical audit
- Promote teaching and education
- Promote and co-ordinate research and developments

### **STAKEHOLDER GROUP**

There are 17 specialist laboratories (12 in England) providing services for metabolic biochemistry in the UK and Ireland.

The group comprises: Belfast, Birmingham, Bristol, Cambridge, Cardiff, Dublin, Edinburgh, Glasgow, Leeds, Liverpool, London (Great Ormond Street and Guy's and St. Thomas's), Manchester (2), Newcastle, Sheffield, Southampton. Other laboratories are associate members. Stakeholders meet together two or three times annually and conduct most of their business by electronic mail.

## ACHIEVEMENTS/COMPLETED WORK (June 2004)

### 1. 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 with effect from October 1<sup>st</sup> 2002 .

### 2. Formation of Stakeholder Group and Agreement of Objectives

Stakeholder group of 17 members across the UK established. Objectives and work plan for 2003/4 agreed at stakeholder meeting on February 10<sup>th</sup>, 2003. Subsequent work plan for 2004/5 and 2005/6 agreed at Stakeholder meeting on June 22<sup>nd</sup>, 2004.

### 3. Workshops for Specialist Scientific Staff

Workshops undertaken 2003/4

- ① Very long chain fatty acids
- ① Lysosomal enzymes

Workshops planned for 2004/5 on the following topics:-

- ① Organic acids
- ① Acyl Carnitines
- ① Non-lysosomal enzymes

### 4. Training

Training is one of the key issues for the network.

Discussions took place with the Royal College of Pathologies and the Association of Clinical Biochemists during January 2003, and the following agreed:-

- i. To adapt the MRCPATH examination in clinical biochemistry to include more options for specialist questions on paediatric metabolic biochemistry
- ii. To create 10 supernumerary training HST posts (grade B clinical scientists) for paediatric metabolic biochemistry. The training provided by the specialist HST post would match the options within the MRCPATH examination for paediatric metabolic biochemistry

- iii. 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 in Jan 2003 to take these initiatives forward. Progress is as follows:-

- i **MRCPath**

The modifications to the MRCPath have been agreed with the Chief Examiner and more questions on paediatric/metabolic biochemistry, both theoretical and practical, are to be included. The issue as to whether this should progress to a sub specialty examination requires further discussion.

- ii. **Creation of HST training posts and trainer time**

The training needs for metabolic biochemistry were incorporated in a document commissioned by the Joint Medical Genetics Committee identifying the needs for all scientists within genetics. The Department of Health has agreed funding for trainers and trainees across England.

- iii Competences in paediatric/metabolic biochemistry were prepared and submitted to the Association of Clinical Scientists and resulted in obtaining recognition of paediatric metabolic biochemistry as a sub-modality of clinical biochemistry.

## 5. **Quality/Audit**

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

- ① How are the stakeholder laboratories performing in specific schemes?
- ① Are existing schemes appropriate to our needs?
- ① How could/should schemes improve?
- ① Do we need new schemes?

The first annual meeting took place on October 22<sup>nd</sup> 2003 for all stakeholders, organised by Dr. Jim Bonham in Sheffield. The second meeting is planned for October 29<sup>th</sup>, 2004 (organised by Dr. Ying Foo, Great Ormond Street Hospital, London).

Discussions with Clinical Pathology Accreditation (CPA) have taken place and Metabolic Biochemistry is now recognised as a sub speciality.

The Network plans to undertake an audit project to investigate equity of service during 2005/6.

## 6. **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 and IEM(sudden unexpected death in infancy)
- Hydrops and IEM \*

- Cardiomyopathy and IEM
- 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

\* Two of these guidelines are now completed and on the web site

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 for the UK.

#### 7. Web Site – [www.metbio.net](http://www.metbio.net)

The Network web site has been commissioned via Professor Bartlett and Dr. Neil Hamilton, Newcastle-upon-Tyne. The web site was launched with stakeholder details (April 30<sup>th</sup>, 2003). Work is in progress to provide a metabolic assay directory with links to the Genetic Testing Network and a European assay finder.

Web site services for health professionals have been established to include:-

- ① Details of Stakeholders laboratories
- ① Best Practice Guidelines for health professionals
- ① Quality initiatives
- ① Education and training site (including workshops)

#### 8. Questionnaire on Service Provision-Current & Future

A questionnaire has been completed with recommendations requiring action. There is a need to engage with the commissioning process and link with plans for newborn screening.

### NEXT STEPS, 2004-2006

#### i. Further development of the WEB site

- a. Complete development of the metabolic assay directory repertoire for biochemical genetic tests to complement the Genetic Testing Network (GTN) data base.
- b. Provide improved access to information for health professionals e.g. best practice guidelines for investigation, availability of rare tests.
- c. Develop electronic learning packages.

#### ii. Service Provision Overview

- a. Identify needs from the survey of service provision
- b. Progress the recommendations about service delivery to minimise risk to services and ensure equity of access
- c. Work with the commissioners to identify specific needs/change as part of a national strategy for service delivery linking with newborn screening

### iii. Education & Training

- a. Training workshops
- b. To plan and undertake an electronic learning project as part of the core syllabus for the MRCPATH.
- c. To establish lead training centres (3 for England) and appoint trainers for each centre, and plan the delivery of training
- d. To appoint to Higher Specialist Trainee posts (8 posts)

### iv. Quality

- a. To produce best practice guidelines
- b. Quality workshop planned for October 2004
- c. To work with CPA on the specific needs for the sub specialty
- d. To undertake an audit plan on equity of service

### THE 'HIGHS'/SUCCESSSES

- ♣ Support from colleagues – shared problems/'spirit' of co-operation very rewarding and motivating
  - willingness to change
  - conformity/removal of variables now possible
- ♣ Training Strategy supported with investment for trainers and trainees
- ♣ Paediatric Metabolic Biochemistry Sub Modality for clinical scientist registration
- ♣ Closer working with other genetic professionals
- ♣ Increased understanding of our role/needs by the Department of Health – at least we hope so!
- ♣ Web site development
- ♣ Best Practice Guidelines development
- ♣ Workshops for specialist staff

### DIFFICULTIES – THE 'LOWS'

- We are responding to a problem too late
  - very tight timescale to achieve training (3 years too late!)
  - burden falls on the same few people
- Is our service/needs really understood and supported?
  - Is metabolic biochemistry really recognised as part of genetics – feel 'poor relation/Cinderella' at times
  - Continued need to 'lobby' and raise awareness because of lack of understanding about some issues
  - Apparent lack of interest by the commissioners/how do we engage with commissioners?
- NHS Structure or lack of!
  - Lack of a coherent national structure for Workforce Development
  - Confederations has hampered the development of a national training strategy
  - Need to have a more formal link at national level
  - The future – how do we ensure continuation and build on what we have achieved?
  - How do we progress issues?
- Accounting/finance agreements
  - finance agreements have been hard to obtain in writing
  - carry over of costs to next financial year difficult
- Peoples' time – difficult in making progress in some areas (e.g. guidelines)

### LESSONS LEARNT

- Do not expect others to understand what we do!
  - keep repeating the same messages

- be patient
- don't expect too much, too soon
- Department of Health staff are very busy
  - may need to feel a nuisance at times!
  - Need to hassle
- ♣ Know what you want to achieve and keep focussed