

## National Metabolic Biochemistry Network

### Minutes of Stakeholder meeting held on 23<sup>rd</sup> February, 2005 at Birmingham Children's Hospital

<b>Present:</b>	Ann Bowron	(AB)	Anne Green	(AG)
	Guy Besley	(GB)	Mick Henderson	(MH)
	Jim Bonham	(JB)	Helena Kemp	(HK)
	Jacqui Calvin	(JC)	Mori Pourfarzam	(MP)
	Fiona Carragher	(FC)	Mary Anne Preece	(MAP)
	Ying Foo	(YF)	Steve Krywawych	(SK)
	Peter Galloway	(PG)		

#### ACTION

#### 1. Apologies

Mike Badminton, Philip Mayne, Lesley Tetlow, Valerie Walker

#### Note:-

Paul Newland did not attend.

#### 2. Minutes of meeting held on 09.11.04

These were approved as an accurate record of the meeting without any amendments.

#### 3. Matters arising

##### 3.1 Workshops update

Three workshops had taken place since our last meeting - Organic Acids, Non-Lysosomal Enzymes, Acylcarnitines. These were all considered to be very valuable and the organisers were thanked.

##### Future Workshops:-

Carbohydrate Disorders, Mitochondrial Disorders and Amino Acids are planned (see list of workshops attached).

##### Carbohydrate Disorders

JC outlined the plans for the carbohydrate workshop which was in the final stages of planning. Programme to be circulated.

JC

##### Lysosomal Disorders

George Gray has produced a report raising several issues for taking forward. The content of the report was approved and it was agreed that the actions need to be taken forward. Ying Foo volunteered to undertake this with George Gray.

YF

AG to write to George Gray to inform him of the proposed way forward.

AG

Ann Bowron and Ann Brown presented some ideas for the amino acid workshop for discussion. It was agreed that a small working group should take forward the planning for data collection.

AB/ABr

It was confirmed that the mitochondrial workshop would take place on June 15<sup>th</sup>

The possibility of having a workshop for associate labs was supported.

AG

### 3.2 Guidelines Update

The up to date position on guidelines is attached.

Various ideas for new guidelines were proposed including:-

Liver Dysfunction in the older child, Fiona Carragher.

FC

Dysmorphology, Guy Besley/George Gray.

GB

Unexplained tubular function in IMD, Steve K.

SK

Unexplained neurology in Adults, Peter Galloway/George Gray.

PG

It was suggested that this latter topic should be preceded by a workshop.

### 3.3 Web Update

Members seemed happy with the general functionality of the web.

An e-mail message board is planned and will go ahead.

New items need to have date posted.

AG

As the site develops there is a need for someone to undertake website management to keep up to date items on general news/workshop updates, guideline reviews, training updates and metabolic assay directory. To date Janet Stone has undertaken the co-ordination of the assay directory, but as different functions develop e.g. guidelines there will be a need to keep these under review. It was agreed that there should be a web co-ordination group. AG to take this forward.

AG

For other developments see items 5, 6 and 8.

### 3.4 MCADD screening and notification via BPSU

Further to the discussions at the previous meeting with the MCADD collaboration group the BioSS Scheme for notification is proceeding. Members should have been circulated with information from Juliet Oerton.

### 3.5 Workforce Strategy

There is no further information at present although it is believed that a meeting is planned by the workforce group to look at genetic scientists in April.

### 3.6 LSD-NSCAG designation

AG reported that it had been confirmed from the DH that the services for lysosomal storage diseases are solely concerned with the treatment of confirmed cases by enzyme replacement therapy or similar treatments i.e. it does not include the diagnostic services.

### 3.7 Agenda for Change

Various labs are at different stages in the Agenda for Change process. There is very little information available. Sue Hill had not responded to our letter asking for clarification.

Heads of laboratories are encouraged to communicate with AG directly and keep an update on progress and outcomes.

ALL

## 4. Report/Liaison with other organisations

### 4.1 UKGTN (AG)

New gene dossiers have been approved by NSCAG. Members are reminded that if they are planning to provide molecular tests and seek approval from GTN they need to be part of GTN.

Further work has been undertaken to define the molecular tests for IMD (see item 9).

National tariffs. A Preliminary meeting took place in January where Jim Bonham represented the Network.

Pilot studies are being undertaken in certain laboratories within the UKGTN. Further work to follow. JB will keep us informed.

JB

The UKGTN website has now been launched [www.ukgtn.org](http://www.ukgtn.org) There are some preliminary issues with the search facility which are being addressed. The organisation is planning a search by disease and by gene.

The two reference laboratories have now been made accountable to the UKGTN.

### Micro arrays

Charles Shaw Smith from Cambridge presented some work on the use of micro arrays to detect abnormalities in patients with learning difficulties. This is an expensive technology £700 per chip. The hit rate for patients with learning disabilities was as high as 10-20% but there were no controls. A group is to be set up under the Cambridge Public Health Genetics Unit, Dr. Ron Zimmersr to assess this technology.

### Information

Various initiatives are underway including tracing and tracking and a review of information systems for use in genetics. Issues relating to NPfIT are of major concern.

### Pathology Modernisation

Ian Barnes gave a brief overview of the current status of pathology modernisation with the key drivers of capacity, plurality, 13 week access, primary care diagnostics. Strategic health authorities had been charged with developing a plan to establish a diagnostic strategy board. Watch this space! There was no further information about PBRs and national tariffs. The aim is that 15% of services will be from the private sector within the next three years.

### Identity of the Network/Future Strategy

There was considerable discussion about the future for genetics. Particular issues include:- the relationship with cytogenetic techniques which are bordering those/overlap molecular techniques.

- The relationship to the national genetics reference laboratories.
- How to ensure equity of access. Have we got any evidence?
- Concerns about the slow IT development particularly the web, database and tracking.
- Is there any evidence that commissioners are listening i.e. is there any extra funding been achieved.
- Need to input more clinical information into decision making.

Other concerns are the impact of PBRs/National tariffs, Foundation Trusts and the need to engage Trust Chief Executives with the genetics agenda.

#### 4.2 UKNSLN (JC)

Jacqui Calvin updated members on UKNSLN activities. The importance and need for UKNSLN and MetBioNet to work together was emphasised and supported. There are obviously issues which concern both organisations and a real need to ensure robust and co-ordinated approach to responses to other organisations in particular the National Programme Centre and the various National screening initiatives.

Jim Bonham outlined the current thinking at the National Screening Committee levels for a new structure. There will be a need for UKNSLN and MetBioNet to link into this in due course.

#### 4.3 JCMG (AG)

An overview of the human tissue bill was presented - the human tissue authority is anticipated to come into existence later during 2005. The consent and confidentiality draft document is now in its final form prior to publication.

There is considerable progress with the review of inherited metabolic disorders see item 10.1. The National Genetics reference laboratories will be hosting a Stakeholder meeting on June 21<sup>st</sup> 2005. AG will be representing the Biochemistry Network.

There was no further progress on the Royal College of Pathologists working group on specimen/tissue storage. We understand that this is in limbo pending the establishment of the Human Tissue Authority.

### 5. Training Update (MH)

#### 5.1 Electronic learning pilot

It is unclear how much progress has been made with this. A presentation was received from Kim yesterday but unfortunately will not work on the current PC. MH to pursue.

MH

#### 5.2 Plans

MH presented a summary of progress with the training strategy. All trainees and trainers are now appointed. Mick reported on the successful meeting between trainers and trainees which had taken place on 1<sup>st</sup> February. See website for copy of minutes. The next

priorities will be to work on the training material for both Grade A's and HST's.

The Grade A training syllabus is now available and details the minimum training requirements. This document is to be circulated to all Stakeholders.

Other items discussed included the role of regional tutors, the need for ACB links and links to the molecular services.

The website will develop a training facility to include items such as chromatogram collection case reports, reading lists, lectures etc.

MH

It was agreed that a Grade A reading list would be useful. Mick to ask trainers to do this.

A purine/pyrimidine study day has been organised.

The issue of copyright for lectures was discussed. MH to take this forward.

MH

With regard to the website development for training AG and MH will be meeting with Neil and Kim in the near future to take this forward.

MH

A poster has been accepted for the Euro Med Lab meeting and one is to be prepared for the SSIEM.

MH/AG

## 6. Metabolic Assay Directory

The modifications requested following discussions at the previous Stakeholder meeting have been implemented.

AG

To request from Neil whether it is possible to have a specific page link for the Assay Directory rather than going through the home page of the website.

All Stakeholder labs were urged to check their entries with a planned go live date for 1<sup>st</sup> April. \*This was subsequently delayed until 1<sup>st</sup> May 2005 because of a service failure during March.

ALL

Various changes to layout have been completed i.e. specimen types and sample details completed. Ultimately there are plans to link to the genetic testing network directory.

Janet Stone is pursuing setting up a small group to work on service functionality of the Assay Directory in order to access tests via a disorder link.

JS

## 7. Audit

### National audit ammonia proposals (MP)

Mori Pourfarzam presented the proposals for a possible national audit based on the one that had taken place in the North East. There was support to undertake a national initiative and a small group comprising of Jim Bonham, Ying Foo and Mouri were asked to take this forward.

JB/YF/MP

### Summary

It was agreed that a major focus for the next Stakeholder meeting would be audit including a fuller discussion on this proposed project.

## 8. Quality

### 8.1 QA Schemes for the web (JC)

Jacqui has undertaken an excellent piece of work to summarise all the QA schemes. This will be put on the website initially as a word document but eventually available as a searchable document.

It was agreed that the search facility should enable the key in a specific assay.

It was agreed that the listings should put the national schemes first followed by an A-Z list of the more minor schemes.

Final comments on the detail should be sent to Jacqui by the end of March.

ALL

Jacqui will consult with all scheme organisers to ensure they are happy with information being made available on our website.

JC

The document will ultimately be available to CPA.

AG

### 8.2 Proposals for a quantitative amino acid interpretive scheme (JB)

Jim Bonham, Steve K and Finlay McKenzie presented a proposal for a quantitative amino acid scheme. Stakeholders were supportive of this initially as a pilot for plasma and CSF as three samples four times per year. The possibility of a marking system was also discussed.

It was agreed this should be taken forward as a pilot, but that a partnership with NEQAS was the preferred option. Jim Bonham and Steve K to take this forward with the aim of first

circulation by May 2005 and two circulations by November 2005.

JB/SK

## 9. DNA testing/links with GTN (GB)

Guy Besley reported on the meeting which had taken place with Maureen Boxer on 13<sup>th</sup> December 2004.

An agreed list of molecular tests for inherited metabolic disorders was now finalised. It may be necessary to fill in some gene dossier documents retrospectively for some of these tests. Maureen Boxer will be taking this forward with the wider GTN group. Guy was thanked for his work on this. The importance of keeping a continued link with the molecular testing gene dossier work of GTN was stressed and Guy agreed to continue to undertake this function for the Network.

## 10. Miscellaneous Items

### 10.1 National IMD Review

An initial meeting had taken place with Hilary Burton and other colleagues to map out the information required for this National piece of work. Members were thanked for returning questionnaires detailing work on diagnoses. AG to pull together a document for finalising and inputting into the National initiative. All members were urged to complete data sets as far as possible.

ALL

### 10.2 Medicine for Children's Research Network

AG reported that William Van Hoff had confirmed that unfortunately the bid for this from the London hospitals had not been successful. We understand the successful applicant is Liverpool.

### 10.3 Associate Labs

It was agreed that we should suggest to associate labs that a useful activity could be to undertake a workshop in early 2006. AG to suggest this.

AG

## Date of Next Meeting

June 14<sup>th</sup> 2005, Birmingham at 11.30am

## National Metabolic Biochemistry (Biochemical Genetics) Network

### Workshops Review 23<sup>rd</sup> February 2005

#### Workshops completed 2004/5:-

Organic Acids, Sheffield, November 11<sup>th</sup> 2004 , Dr. J. Bonham

Non-Lysosomal Enzymes, Birmingham, December 2<sup>nd</sup> 2004, Dr. R.G.F. Gray

Acylcarnitines, Fiona Carragher/Neil Dalton, January 13<sup>th</sup> 2005

#### Workshops planned 2005:-

Carbohydrate Disorders, Cambridge, April 25<sup>th</sup>, Dr. J. Calvin

Mitochondrial Disorders - (subsequently confirmed 15<sup>th</sup> June 2005), Dr. J. Bonham

Amino Acids, Bristol, ?Nov/Dec 2005 (subsequently confirmed), Ann Bowron/Ann Brown

Jan - March 2006 ?Associate Labs.

Guidelines	Lead	Target Date
Metabolic autopsy (SUDI)	Mary Anne Preece	
IEM and hydrops	George Gray/Mary Anne Preece	Completed - on web site
IEM and cardiomyopathy postmortem	Helena Kemp	
Hypoglycaemia	Helen Losty	Completed - on web site
Hyperammonaemia	Helen Losty	Completed
Developmental delay	Peter Galloway	First draft
Fits and seizures	Mick Henderson	Completed - on web site
Prolonged neonatal jaundice	David Isherwood	
Lactic acidosis	Jim Bonham	
Rhabdomyolysis	Jim Bonham/Simon Olphin	