

## National Metabolic Biochemistry Network

### Minutes of Stakeholder meeting held on 23<sup>rd</sup> January 2008 at Birmingham Children's Hospital

<b>Present:</b>	Tim Lang	(TL)
	Anny Brown	(AyB)
	Ann Bowron	(AB)
	Jacqui Calvin	(JC)
	Steve Krywawych	(SK)
	Alan Cooper	(AC)
	Jean Kirk	(JK)
	Stuart Moat	(SM)
	Mary Ann Preece	(MAP)
	Lesley Tetlow	(LT)
	Paul Newland	(PN)
	Mick Henderson	(MH)
	Simon Olpin	(SO)
	George Gray	(GG)
	Jim Bonham	(JB Chair)

#### ACTION

#### 1. Apologies

Mori Poufarzam, Anne Sheldrake, Janet Stone, David Isherwood, Valerie Walker

#### 2. Minutes of Meeting held on 27.6.07

Accepted as correct.

#### 3. Matters Arising

##### 3.1 ACB Newsheet report

Now abandoned.

##### 3.2 Amino acid analytical guidelines

AB summarised progress on behalf of the amino acid working group.

- A workshop on monitoring is planned for 14<sup>th</sup> May 2007.
- A previously circulated method questionnaire circulating ERNDIM participants in the amino acid scheme may not have been received by all laboratories. AyB to check.
- Guidelines when produced to be sent PN.
- SM is continuing work on creating age related plasma reference ranges. He will contact the group asking for 50 sets of results from outpatients to help complete the data. SM to write.
- Rachel Carling is continuing work on the CSF amino acid reference ranges.

AyB

SM

### 3.3 E-learning package - MCADD

No progress has been made with the development of this educational package.

### 3.4 Non lysosomal assays

The provision of these assays needs to be kept under review. Problems with substrate availability were identified for some assays. SK felt that synthesis for some compounds might be available from UCL. SO will contact SK to discuss. SO and GG were asked to prepare a summary review of the position on assay availability for the next meeting.

SO/GG

SO said that Sheffield was expanding their repertoire of enzyme assays and molecular genetics relating to peroxisomal disorders, but were keen to avoid unnecessary duplication.

### 3.5 MPS schemes

Interest had been expressed within ERNDIM to taking on the MPS scheme from Manchester when Margaret returns next year. JB to inform AC of the name of the interested laboratory so that liaison can occur.

JB

### 3.6 Education - acylcarnitines

Roy Talbot has sent an educational power point presentation to MH for inclusion on the website. MH to pass onto Neil Hamilton for the website.

MH

It was noted that Charles Turner's presentation at the QA day on October provided useful information. The introduction of MCADD screening emphasised the need for educational material on the website relating to acylcarnitine analysis.

### 3.7 Website problems

Some problems with getting things onto website in a timely way was noted. JC mentioned the QA summary in particular. JC to resend to JB and JB to ensure that it is on the website.

JC  
JB

JB also to check that the QA day reports for 2006 and 2007 (not yet received, SK to send) are mentioned on the website.

JB

There was also discussion about the organisation of the website and it was agreed that MH, JB and GG would meeting with Neil Hamilton to review layout and contents. In particular it was felt that it would be valuable to index presentations both by subject and meeting. Key words could also be attached to existing and new presentations and then searched. The development of a site map would also be useful.

MH/JB/  
GG

### 3.8 Ammonia audit

JB had not received a response from Graham Shortland on behalf of BIMDG and would need to chase up. SK said that he would try and recover the source data collected and would review the findings and prepare a report with recommendation for publication in the ACB Newsheet and BIMDG newsletter.

JB  
SK

Areas for re-audit in the light of the recommendations would be identified.

### 3.9 Workforce planning

MAP would re-circulate the workforce questionnaire and would collate the data in time to attend the Genetic Workforce review meeting on 14<sup>th</sup> March in London. MAP to action.

MAP

### 3.10 Quality report

JB to check that the QA meeting report is on the website.

JB

## 4. Reports/Liaison with Other Groups

### 4.1 UKGTN

Commissioning of laboratory services and the creation of tariffs in Molecular Genetics and newborn screening is under active discussion. It is likely that with the inclusion of newborn screening as part of specialist commissioning that a tariff will emerge, comparison with existing centres suggests that this may be in the region of £20 for five conditions currently screened.

### 4.2 Blood Spot Advisory Group UKNSLN

This has not met since the last MetBioNet Stakeholder meeting but a meeting is scheduled for 3<sup>rd</sup> March 2008.

### 4.3 JCMG

JCMG has discussed the trainee role in Biochemical Genetics and has been able to offer valuable support to ensure that this function is re-instated.

There has also been discussion relating to the threats to research activity in Paediatrics and in particular rare disorders and laboratory work. The new DoH initiative "Best Research for Best Health" emphasises the importance of common disorders such as cancer, diabetes, stroke, heart disease etc, where RCT's can be conducted by well-established research groups. There is not a route of provision for rare conditions. The DoH has been asked for comment.

### 4.4 GenCAG

Discussions about commissioning have taken place as outlined in 4.1. Also discussion of the 18-week treatment target and its implications for genetics.

#### 4.5 UKNSLN

Concerns have been expressed about the contractual arrangements relating to the dried blood spot card and in particular the arrangements for guaranteeing continuity of supply. Anne Mackie has expressed her concern on behalf of the Blood Spot Advisory group. Kate Hall is working with the supplier on these issues.

There has also been discussion about the status codes used by IT but the lack of clarity in Child Health system procurement is delaying progress.

#### 4.6 RCPATH SAC

LT has been unable to attend the last two meetings of this group but another meeting is scheduled and she will feedback at our next meeting.

LT

#### 4.7 SAGE

GG gave a presentation (attached) to report on the work of this group. The DoH is keen to develop E-learning in conjunction with the Royal Colleges. GG is acting with others on behalf of the RCPATH. He described three types of approach:

- Modular
- Case based with branching tree structures
- Moderated Pathopedia

He also described work on a syllabus for Inherited Metabolic disease training and explained the differences between syllabus and curriculum design.

### 5. Commissioning Issues

JB outlined the current position on the network proposals. Kieran Morgan has prepared a draft report which will be submitted to Julia Stalibrass in February. It is likely that the major emphasis will be on ensuring that Inherited Metabolic Disease is included within the new areas to be part of specialist commissioning. Once this definition has been achieved and the standards of care to be achieved are recognised, then it will be up to specialist commissioners to ensure that adequate arrangements are in place for their populations. This may involve "networking" but not necessarily the formation of managed networking.

Specialist commissioners in East Midlands and the South West have already been contacted to alert them to potential needs in their areas. The specialist commissioners may designate certain centres and choose to invest in these centres if needed. In addition to this local work a National overview probably needs to be developed to assemble disease registers to assess clinical need and arrange aspects of education, training and workforce planning.

## 6. Expanded screening

The consultation period on the alternatives highlighted in the 21<sup>st</sup> September meeting is nearing completion with comments to be submitted by 31<sup>st</sup> January. This document will help form a basis for discussions scheduled to take place at a meeting to be organised by Carol Dezateux and David Elliman in April 20/21. In addition expanded screening is to feature in the ACB Focus meeting in May and at the BIMDG annual meeting in July.

JB reported that Sheffield Children's had agreed support for pre-pilot work to be undertaken and a pilot project was to be included within a South Yorkshire bid for NIHR funding under the "collaboration in applied healthcare research and care" call. If successful this would provide some funding for the 6 pilot MCADD pilot laboratories to undertake an expanded screening pilot with central co-ordination. A decision will be made in April.

## 7. Training and Education

### 7.1 Update (including trainers)

MH welcomed the continued support from the DoH for the trainers, albeit at a reduced level of 1.2wte. He proposed that the funding be divided to provide:

- A national lead 0.2wte
- An enzymology lead 0.4wte
- Additional trainer input 0.2.wte
- BMS lead 0.2wte
- Project based funding equivalent to 0.2wte

This was accepted and it was agreed that MH continue in the National lead role. GG be appointed as the enzymology lead, LT be asked to provide additional input and Anne Sheldrake's nominee be asked to fulfil the BMS lead role. The funding left could be used on a project basis as needed.

## 8. Report from the BMS group

AS could not be present at the meeting but made the following comments by email:

*"Unfortunately, I will be unable to attend Wednesday's meeting. However, we will be arranging a BMS meeting for the beginning of March where we plan to discuss the good news about the Trainer posts. In addition, Mark DeHora and myself have been invited to the IBMS on Feb 4th to discuss the Diploma in Expert Practice and how we can take it forward".*

## 9. Web review

### 9.1 Update

A meeting is to be arranged with Neil Hamilton, GG, MH and JB, see 3.7. JB to arrange.

JB

### 9.2 Assay Directory

JS could not attend the meeting but made the following comments by email:

*"I am unable to attend the stakeholder meeting tomorrow but wanted to update you on the progress with the next phase of the metabolic assay directory.*

*I have made a start on linking the common/clinical name for an inborn error of metabolism with the biochemical (or in some cases genetic) test that would be needed to make a diagnosis. The table is not yet complete and I will be sending it to Neil once I have all the information there. We need to decide whether this should exist as a separate index i.e. if only the syndrome name is known then this would be the bridge between checking which test to request and then looking the test up in the original directory or whether we should try to link the information electronically so that if you look up a syndrome you will automatically be directed to the labs offering the correct test.*

*The latter may be too complicated but will discuss all possibilities with Neil.*

## 10. Audit

### 10.1 Update

TL provided a report on the recent pyruvate (attached). The recommendation contained in the report was accepted with the addition as a first sentence:

*"Many centres adopt the view that measurement of pyruvate or the calculation of lactate : pyruvate ration does not assist in the investigation of patients and should not be undertaken. If measured then certainly pyruvate should only be measured in the presence of a previously raised lactate result and only accepted if there is an accompanying lactate sample taken at the same time. The results for the pyruvate and lactate should be reported in the same units or a ratio should be calculated in the laboratory and printed on the report form".*

The group was asked to suggest other topics for audit and PN agreed to review the guidelines for any likely topics. JB agreed to check whether RCPCH had any plans to audit the "altered consciousness guideline" if this was not planned then aspects of these recommendations including the measurement of ammonia in this context could be audited. JB to action.

JB

## 11. Workforce planning

MAP agreed to re-circulate the workforce questionnaire, see 3.9.

## 12. Quality assurance schemes

### 12.1 Sample supply

MH outlined a scheme which had been agreed with CLIMB to provide patient sample for use in QA. Full consent would be obtained and the patient's consultant informed. The samples would be documented by Camilla Reed as archivist on behalf of ERNDIM and stored and distributed from a QA facility in Switzerland. The agreement could act as a model for other European countries working with parent support groups.

## 13. Guidelines

### 13.1 Update

PN updated the group on the guidelines. He confirmed that he had received the prolonged jaundice guideline and the review of the hypoglycaemia guideline. Others were outstanding and the individuals concerned had been contacted.

## 14. Finance

JB confirmed that at the current rate of usage - £2.5k pa website; £2.5k pa travel; £5k pa meeting organisation and other expenses, then sufficient funding should be available for around 5 years. PN suggested that sponsorship could be sought for meeting and agreed to co-ordinate this. This would help the funding to last for a longer period.

PN

## 15. Meetings planned

### 15.1 Future workshops

A number of IEM meetings and workshops are already planned for 2008, these include:

- A bipterin workshop, Birmingham - 5<sup>th</sup> March
- A fatty acid oxidation workshop, Manchester, BIMDG - 12<sup>th</sup> March
- Expanded screening workshop, London - 20<sup>th</sup> & 21<sup>st</sup> April
- Leeds paediatric day, Leeds St James - May
- Amino acid monitoring workshop, Cardiff - 14<sup>th</sup> May
- Screening workshop, Birmingham, Focus ACB - 22<sup>nd</sup> May
- Glutaric aciduria type 1 workshop, BIMDG - June
- BIMDG annual conference - 10<sup>th</sup> & 11<sup>th</sup> July
- SSIEM annual symposium, Lisbon, SSIEM - 2<sup>nd</sup>-5<sup>th</sup> September
- Regional meeting, Guy's Hospital, London, ACB - 25<sup>th</sup> September
- Prenatal diagnostic workshop, Birmingham, MetBioNet - Nov TBC

### 15.2 Quality assurance day

- Manchester, MetBioNet - Oct TBC

LT

It was agreed that there would be little demand for additional workshops during 2008, although days for the trainees may be indicated.

MAP/SM confirmed their willingness to organise a B12 and propionate workshop in spring 2009.

MAP/SM

JB/FC confirmed their willingness an intermediary metabolite workshop in spring 2009.

JB/FC

Dates for these workshops to be confirmed at the next Stakeholder meeting.

### 16. Any other business

MAP raised two issues

1. Genzyme's support of dried blood spot assays for Fabry disease and other lysosomal disorders. AC confirmed that Manchester was working with Genzyme in this project, but doubted whether the methodology in this form would prove reliable.
2. Laboratory reporting to the galactosaemia register. MAP expressed her concern that if laboratories did not report cases then this may be missed. JB commented that unconsented reporting of data, which included initials, DOB and condition, could not be considered anonymous and was therefore unlawful. If consent was gained then this would be ethical but this would require the clinician asking the family concerned and confirming this to the laboratory before data submission.

JB commented that the formation of a metabolic physician group was being considered in order to discuss cases and undertake research.

### 17. Date and time of next meeting

Wednesday, 16<sup>th</sup> July 2008 at Birmingham Children's Hospital.

The meeting closed at 3.00pm