

National Metabolic Biochemistry Network

Minutes of Stakeholder meeting held on 27th June 2007 at Birmingham Children's Hospital

Present: Tim Lang (TL)
Ann Bowron (AB)
Ann Sheldrake (AS)
Jacqui Calvin (JC)
Peter Galloway (PG)
Jim Bonham (JB Chair)
Ann Marie Preece (MAP)
Helena Kemp (HK)
Mori Pourfarzam (MP)

ACTION

1. Apologies

Fiona Carragher, Lesley Tetlow, Janet Stone, Paul Newland, Tony Fensom, Mike Badminton, Stuart Moat, Philip Mayne, Jean Kirk, John Fyffe, Ying Foo, Mick Henderson, Valerie Walker

2. Minutes of Meeting held on 11.12.06

Agreed as correct.

3. Matters Arising

3.1 ACB News draft

This item remains outstanding.

JB

3.2 Amino acid analytical guidelines

A report attached (appendix A). SM is continuing to work on comparing and creating reference ranges and Rachel Carling is reviewing CSF reference range data. SM aims to create something for publication.

SM

3.3 Custom synthesis

MP reviewed availability for substrate and a table is attached (appendix B). There may be some patenting issues with the synthesis of some compounds. MP to contact JB if this is a problem; the DoH is considering patenting issues.

MP

3.4 E-learning package

No progress.

JB/MH

3.5 Non lysosomal assays

The need for alternative provision continues to be pressing particularly in the light of actual and impending retirements. Sheffield may be in a position to develop additional assays if requested but would not want to duplicate efforts elsewhere. George Gray/SK to advise.

GG/SK

	ACTION
<p>3.6 QA on the Website JC questioned the usefulness of the QA table on the website but the group agreed that it should be maintained.</p>	JC
<p>3.7 MPS scheme ERNDIM has shown some interest in providing this as a scheme. JB to clarify.</p>	JB
<p>3.8 Education MH to confirm progress with acyl carnitines appearing on the website. Additional organic acid TIC's have been supplied to Prof Pollitt and are available on the website.</p>	MH
<p>3.9 Website problems Neil Hamilton failed to replicate the problems with opening guideline documents, but speculated that this was due to the application running slowly giving the appearance of failure.</p> <p>Access to this UKGold server was believed to be a firewall problem, but Neil Hamilton requested that any error messages are reported to him in detail to let him follow them up and trace the problem if any.</p>	All
<p>3.10 Ammonia audit A summary of the findings has been distributed by YF and JB has contacted Graham Shortland BIMDG for comment/endorsement of the standards. JB to seek feedback from Graham Shortland at BIMDG meeting. SK agreed to follow up any correspondence subsequent to the issue of the audit summary.</p>	JB SK
<p>3.11 Workforce planning YF has taken over responsibility from Colin Samuell but was not present to report. MAP agreed to re-distribute templates to stakeholders for completion and attempt to collate the results. This was welcomed by the group.</p>	MAP
<p>3.12 Quality day 2006 TL had sent the presentations to Sheffield but they are not on the website. JB to check with Nickie Harries if sent.</p>	JB
<p>3.13 eGFR JB has discussed this with Paul Griffiths and Paul agreed to take this up with the new National Paediatric Biochemistry group if formed.</p>	PG

4. Reports/Liaison with Other Groups

4.1 UKGTN

JB reported that much of the recent work of this group had been around tariff setting for tests. There was concern expressed by some molecular geneticists about the proposed 3-band model, in particular that it would leave the service open to "cherry picking" by the private sector.

The group noted the extension of funding for both the molecular genetic and cytogenetic HST's and trainees. Representation has been made by JB and John Crolla on behalf of JCMG and Alastair Kent on behalf of GIG to the DoH to similarly continue funding for biochemical genetic HST's.

4.2 UKNSLN

JC reported that the first of the new style UKNSLN meetings had taken place on 8th June but that attendance had been rather poor perhaps affected by the ISNS meeting but the day had been very useful and it was hoped that attendance would be better at the next meeting scheduled for 18th September.

4.3 Blood Spot Advisory Group

JB reported on the last meeting held on 9th January. The meeting scheduled for the 3rd July has been cancelled. The topics covered at the January meeting included concerns about continuation of screening card supplies, the implementation of national roll-out for MCADD screening, the need to consider expanded screening, the continuation of funding for the Child Health Co-ordinators and the development of a group led by Anne Green to review the suitability of the EQA arrangements for current screening programmes.

4.4 JCMG

JB reported on the May 2007 meeting. There was discussion of the adverse effects of the NHS R&D policy on genetic research and all specialist areas, in particular the inadmissibility of laboratory based research for some funding streams. The group also supported the request for continued funding from the HST posts and trainees.

4.5 GenCAG

Discussions at the March meeting of GenCAG included the 18-week access targets, and monitoring of turnaround times.

4.6 RCPATH SAC for PPP

LT was not present.

4.7 Expanded screening

It was agreed that a meeting would be organised jointly by MetBioNet (JB) and UKNSLN (JC) to consider the possibility of expanded screening looking at varying disorders in different regions. The recommendations of that meeting will inform a wider meeting to be organised by Carol Dezateux on behalf of the Programme Centre.

JB/JC

4.8 MCADD Board

HK reported on the first meeting of the new Board on 7th June. Terms of reference were agreed and a gateway reference number allocated. Information will be provided via the Programme Centre website and the next meeting is planned for October. Patient/Professional information will be based on that used during the Pilot Scheme.

A meeting to be organised by Juliet Oerton will look at the current protocol including the use of the C8:C10 ratio and related issues.

5. Commissioning Issues

Attention was drawn to the Network Specification, Standards of Care document and Commissioning framework now available on the BIMDG and MetBioNet websites.

The possible inter relationship between emerging multidisciplinary regional networks and MetBioNet was discussed. In particular a recent meeting to inform the Carter report to Ministers scheduled for September emphasised the need to maintain a national perspective.

It was felt that the development of regional multidisciplinary networks and a national MetBioNet laboratory network were not mutually exclusive and both could be explored.

6. Training and Education

6.1 Update

MH provided a written update which is attached (appendix C).

7. Report from the BMS Group

AS summarised a recent BMS group meeting at which 40 delegates attended and 2 presented reports on their MSc projects. Future meetings planned included a practical training day, perhaps at Nottingham training centre and a Biochrom training day. AS felt that the group had raised the profile of metabolic disease within the IBMS and AS had been invited to join the IBMS clinical advisory group as a result. A session on screening is planned for next years IBMS conference with contributions from MH and HK. AS wishes to thank MH for all his encouragement and support in forming the group.

	ACTION
<p>8. Web review</p> <p>JC asked about the route of including new tests in the Directory, this is via JS. The correctness of the order of the alphabetical listing in the directory was also questioned. JS to check.</p>	JS
<p>9. Audit</p> <p>9.1 Update</p> <p>Suggestions for new audit topics included pyruvate measurement - its conduct and clinical usefulness and the take up of the recently issued unexplained encephalopathy guidelines. TL to consider both these as audit topics.</p>	TL
<p>10. Workforce planning</p> <p>MAP will circulate the current proforma for completion and collate the findings.</p>	MAP
<p>11. Quality</p> <p>11.1 QA Schemes</p> <p>JB was asked to liaise with Finlay MacKenzie and report on progress with the urinary amino acid scheme.</p>	
<p>12. Guidelines</p> <p>12.1 Update</p> <p>PN provided a report which is attached (appendix D). SK agreed to review the hyperammonaemia and hypoglycaemia guidelines and PG volunteered to review the prolonged neonatal jaundice guideline with Phillipa Goddard. MAP agreed to liaise with George Gray about the Adult Neurology guideline.</p>	PG MAP
<p>13. Network Issues</p> <p>13.1 Finance</p> <p>JB confirmed that there were sufficient funds to continue for financial year 2007/8 but asked if a subscription of approximately £1k per laboratory could be levied in the future if needed, this was agreed with a reduced rate for associate laboratories. JB noted that around £10k would be ring fenced to provide some funding relating to the HST training work if DoH money cannot be accessed.</p>	
<p>14. Meetings planned</p> <p>14.1 Future workshops</p> <ul style="list-style-type: none"> ▪ SK agreed to organise and host a Quality Day in London in early October 2007. SK to outline a programme and agree this with JB. ▪ B12 and propionate - MAP/SM to organise in Autumn 2007 ▪ Intermediary metabolites - JB/FC to organise in Spring 2008 ▪ Pre natal diagnosis - AC/GG to organise in Spring 2008 	SK MAP/SM JB/FC AC/GG

15. Any Other Business

None.

16. Date and Time of Next Meeting

12th December at Birmingham at 10.30am.

The meeting closed at 3.00pm

Report from the Amino Acids Working Group

1. Monitoring of patients with amino acid disorders

BIMDG contacted to suggest a workshop. To be discussed at the BIMDG committee meeting in October.

2. Guidelines for amino acid analysis

Progress is being made (slowly).

Samples, profiles, analysis, interpretation etc

ERNDIM data to be made available for analysis.

A questionnaire has been prepared for ERNDIM participants on the methods used.

Performance can be compared with methods.

MetBioNet questionnaire to define an 'Expert opinion' on which are the most important amino acids (see separate report).

3. Reference ranges

Stuart Moat (Cardiff) collating data from defined age groups using out-patient samples and improved sample handling.

Compared with existing data – many similarities, some clear differences.

When complete will request data from other labs to add.

Working with local statistician.

CSF amino acids – Trainees group.

Trainees Group invited to join the Working Group.

Amino Acids Questionnaire

Aim to produce limited number of amino acids to use to assess performance in ERNDIM EQA scheme. An 'Expert opinion'

Questionnaire sent to all MetBioNet Stakeholder labs.

Stakeholders asked to forward to other local labs performing AA analysis.

List of all ERNDIM amino acids.

State whether always, sometimes or rarely/never useful for either screening or monitoring patients for IEMs.

26 completed questionnaires from 19 labs.

Screening

Always or sometimes useful:

Phenylalanine
Glutamine
Glycine
Isoleucine
Leucine
Valine
Citrulline
Methionine
Arginine
Tyrosine
Ornithine
Lysine
Alanine
Proline

Equivocal

Homocystine
Cystine
Glutamic Acid
Sulphocysteine
Serine
Threonine
Taurine
Histidine
Cystathionine

Rarely/never useful

Sarcosine
Histidine 1-methyl
Hydroxyproline
Asparagine
Aspartic acid
a-amino butyric acid

Monitoring

Always or sometimes useful:

Phenylalanine
Glutamine
Glycine
Isoleucine
Leucine
Valine
Citrulline
Methionine
Arginine
Tyrosine
Ornithine
Lysine
Alanine

Equivocal

Homocystine
Cystine
Glutamic Acid
Threonine

Rarely/never useful

Sarcosine
Histidine 1-methyl
Hydroxyproline
Asparagine
Aspartic acid
a-amino butyric acid
Cystathionine
Histidine
Proline
Taurine
Serine
Sulphocysteine

For discussion

? include amino acids from the equivocal range

Alternatively, use a scientific approach, i.e. those which are essential for the diagnosis of a defined group of disorders.

Substrate / Standard	Availability	Alternative source
Biotinyl-6-Aminoquinoline	Available - Mori Pourfarzan	
[D]-Hexanoylglycine	in progress - Mori Pourfarzan	ten Brink
Glutaryl carnitine	in progress - Mori Pourfarzan	
Heptanoylglycine	Nigel Manning	ten Brink
[D]-Glutaryl carnitine		PerkinElmer Chromsystems
3-ketopalmitoyl-CoA		
2-methyl-acetoacetyl-CoA		
Fumarylacetoacetate		
3H-Galactocerebroside	New batch synthesis required in 08/09	
[D]-Globotriaosylceramide		
4MU-deoxychitobiose		
LC-Hydroxy-Acyl carnitines		

National Metabolic Biochemistry Network (Biochemical Genetics)

Report to Stakeholders June 2007

1. HST training

a) Annual Meeting of Training Group

The training group met in February, a report summarising the discussions that took place has been circulated to Stakeholders and is appended here.

b) HSTs moving on

Annette Powell was appointed to a higher grade post at the Birmingham Children's Hospital last year. Camilla Reed was successful in attaining a permanent post at the Sheffield Children's Hospital in May this year.

Carys is on maternity leave, having given birth to a healthy baby boy. We wish Carys and Tomos well.

We have had correspondence from Peter Prinsloo in Nottingham about the appointment of an HST biochemist who will concentrate on metabolic medicine. Dr Prinsloo requests that we include this appointee in the specialist HST group. George Gray has been in contact with them and will help to organise training. Mick would like the Stakeholder group to consider this. It should be born in mind that the group has not been involved in this appointment. The trainee is not based in a stakeholder laboratory and it would be difficult to provide the same level of training and metabolic experience in that environment.

c) Trainers

Ying Foo has temporarily suspended her duties as Local Trainer for the South. Helen Aitkenhead will continue in this role supported by Mick. Mick will go down to London to assist Helen with the annual reviews later in the year.

There is still uncertainty about future funding for the trainer posts. Mick is prepared to continue in the role of lead trainer, albeit at reduced time commitment, regardless of the funding. This would depend on the ratification and support of the stakeholders. There are some funds remaining and it is proposed that these should be used to support future training events.

2. HST Training Day

A successful training day, exclusively for the HST group, was held in Leeds in April. This was on the subject of amino acids. Tony Briddon and Martin Smith supported the event. The event was organised largely by Rachel Carling and Carys Lippiatt. The HSTs were asked to prepare for the event by bringing information about the service in the own labs and by answering a questionnaire. A report will follow. Future events are planned, probably at a rate of two a year

Mick Henderson

National Lead Trainer

Thursday, 21 June 2007

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Appendix D

Title	Author	Issue Date	Review Date	Volunteer reviewer	Correspondence	Additional Comments
Autism	Neil Dalton					
Developmental Delay	Peter Galloway	2nd Draft	Dec-07		Anne	Complete
Dysmorphology	George Gray and Guy Besley	Jun-07	Jun-09		George Gray	? Not yet on web site?
Encephalopathy	Jim Bonham				Jun-06	Almost complete, requires Downsizing for Network
Fits and Seizures	Mick Henderson		Sep-08			Complete
Hyperammonaemia	Helen Losty		Jan-06		Jun-06	Requested that someone else writes this
Hypoglycaemia	Helen Losty	Nov-04	Nov-06		Jun-06	Requested that someone else writes this
IEM and Cardiomyopathy PM	Helena Kemp				Jun-06	Response awaited
IEM and Hydrops	George Gray and Mary Anee preece	Nov-06	Nov-08			complete
Lactic Acidosis	Jim Bonham				Jun-06	Almost complete
Liver dysfunction in the older child	Fiona Carragher				Jun-06	Response awaited
Metabolic autopsy (SUDI)	Mary Anne Preece				Jun-06	Response awaited

Appendix D

Prolonged Neonatal Jaundice	Philippa Goddard and David Isherwood		Jan-06	Philippa	Jun-06	Awaiting Review. DMI no longer involved in this
Rhabdomyolysis	Jim Bonham and Simon Olpin	Jul-06	Jul-08		Anne	complete
Unexplained tubular function	Steve Krywawych					? Contact details
Unexplained Neurology in Adults	Guy Besley and George Gray				Jun-06	No progress to date Contacted Peter for update