

National Metabolic Biochemistry Network

Minutes of Stakeholder meeting held on 9th November, 2004 at Birmingham Children's Hospital

Present:	Guy Besley	(GB)	Jean Kirk	(JK)
	Jim Bonham	(JB)	Steve Kuywaryich	(SK)
	Ann Bowron	(AB)	Mori Pourfarzam	(MP)
	Anne Brown	(ABr)	Mary Anne Preece	(MAP)
	Jacqui Calvin	(JC)	Geraldine Roberts	(GR)
	Fiona Carragher	(FC)	Lesley Tetlow	(LT)
	Anne Green	(AG)	Valerie Walker	(VW)

By invitation:

Juliet Oerton	(JO)
Pamela Phillips	(PP)

ACTION

1. Apologies

Peter Galloway, Ying Foo, Janet Stone, Helena Kemp and Mike Badminton.

2. Minutes of meeting held on 22.06.04

These had been previously circulated and were agreed as a true record.

3. Matters arising

3.1 Objectives & Action Plan 2004/5 and 2005/6

These have been finalised and previously circulated to all Stakeholders.

3.2 Department of Health Report July 2004

Formal report was completed in July 2004 and has previously been circulated to all Stakeholders.

3.3 Web Issues

The chairman reported on a very productive meeting with Kim Bartlett and Neil Hamilton on 14th September 2004 (minutes of the meeting on metbio website). There was general discussion about the website people were generally happy although felt that the appearance could be improved with more professional web design. This is something to be considered for the future. It was agreed that associate labs should retain their current level access and not have access to the Stakeholder site.

AG reported that an annual maintenance contract is now in place but is awaiting a formal definition of the contract from Kim Bartlett. AG to chase up.

AG

The chairman also reported confirmation that the software is owned by Metbionet and if an alternative web provider was required for the future we would be able to access this at a small cost.

Any further developments of the website beyond the metabolic assay directory would cost require further funding.

Future developments were discussed it was agreed that a message board would be helpful. This should be initially restricted to Stakeholders with consideration of expansion to associates in the future.

The development of the training and education site was clearly paramount with our training initiatives and Mick Henderson would be taking this forward. Further funding would be required from the training budget. AG to communicate this to Neil and Kim.

AG

It was also agreed that it would be very valuable to have a disease and test link on the Metabolic Assay Directory in due course.

Some members reported difficulty accessing the website i.e. this was very variable in some instances they were denied access to the Stakeholder site via their passwords. AG would discuss this with Neil.

AG

It was also noted that the internet instruction are actually on the Stakeholder restricted site and need to be available when access to this is blocked.

AG

3.4 Genetics Services Development Pilot Learning Event

The Department of Health meeting took place on 20.10.04 which was attended by Anne Green, Mick Henderson and Jim Bonham. A summary of our Network activities was presented and discussed at the meeting. It was very well received and we have had favourable comments about the work undertaken to date.

Note:-

A copy of the presentation has previously been circulated to all Stakeholders.

3.5 Guideline Update

The guidelines on the web now include hydrops, seizures (+CSF sampling) and hypoglycaemia.

Final draft of the hyperammonaemia guideline is ready for completion.

First or second draft of Rhabdomyolysis, Skin Biopsy Consent and Neonatal Jaundice have been prepared.

Outstanding guidelines include Developmental Delay, Sudden Unexpected Death in Infancy, Cardiomyopathy and Lactic Acidosis.

The need for the SUDI guideline was emphasised now that the RCPATH document has been published.

MAP

Future guidelines for the future were suggested:- Autism to ask Neil Dalton and Encephalopathy Jim Bonham volunteered to take this forward.

3.6 Workshops Update

Organic acid workshop will take place this week November 11th.

Non lysosomal enzymes organised by George Gray December 2nd in Birmingham.

Future dates for the diary:-

Acyl carnitines, Guy's Hospital, January 13th 2005.

Carbohydrate disorders, Cambridge, April 25th 2005.

Mitochondrial disorders ?May June 2005. Jim Bonham to confirm.

Additional workshops suggested for 2005/6 include:-

Amino acids - It was agreed that this should be scheduled for November/December 2005 in Bristol.

AB/ABr

Other possibilities for the future for further discussion at future meetings:- Purine and pyrimidines, orotic acids, hypoglycaemia, screening. Details of all workshops to be updated on the web.

AG

3.7 Electronic Learning Pilot

AG reported that has been discussed with Kim Bartlett and a contract has now been agreed for delivery of this for March 31st 2005. Kim will work with Mick Henderson and the training infrastructure to undertake this.

4. MCADD Screening and Notification

Juliet Oerton and Pamela Phillips from the UK Collaborative project for Newborn Screening for MCADD were in attendance and outlined the need for a notification system of new MCADD cases via the laboratories. A draft questionnaire was presented. Juliet confirmed that MREC approval had been received to enable this process. Initial comments on the questionnaire were received. Revised version will be circulated via the Network for final comments.

JO/AG

It was agreed that it would be important to have a system to ensure that diagnoses via the molecular labs were not missed and that a separate notification system via those few labs should be undertaken. It was suggested Juliet contact Maureen Boxer to organise this.

5. Reports Liaison with other organisations

5.1 The Genetic Testing Network

Feedback from the meeting September 9th 2004:-

1. All new DNA based tests from GTN approved laboratories need to have completed gene dossiers approved via the GTN for final agreement by GENCAG. Stakeholders were reminded that if they are submitting gene dossiers then they need to be part of the GTN.
2. Molecular tests for IMD will be taken forward via a group led by Guy Besley (date now agreed 13th December 2004). This will include Mike Badminton, Louise Brueton (Clinical Geneticist, Birmingham) and Maureen Boxer. The group will first look at the existing tests on the GTN list to assess and confirm utility and as a second part will look at the need for new tests for the future.
3. National Tariff exercise - The chairman reported that GTN is undertaking an exercise to look at setting a National Tariff for molecular tests. A pilot is being undertaken by John Old in Oxford for haematology DNA tests. Six laboratories have been selected (details not known) to proceed with this exercise. It was agreed that the National Tariff setting exercise was an important issue for biochemical tests and that the chairman should seek further information. AG to contact Jaquie Westwood.
4. It was noted that GTN now has their own website www.ukgtn.org. Tests which should be provided across the UK are now detailed on this site and their status regarding commissioning. It was noted that in some parts of the UK there are issues between availability of funding via commissioners and the service needs.

AG

5.2 JCMG Committee Medical Genetics Feedback

Meeting held on 19.10.04

Constitution in terms of reference

Constitution of the committee was debated and the terms of office of individual members. The JCMG now have a representative from the National Screening Committee as observer status. This is Alison Streetly. It was noted that AG's membership is via the Royal College of Pathologists and would be up for review in September 2005. The Biochemistry Network does not have a formal representative on the committee - this was raised by AG at the JCMG and will be considered in September 2005. It was also noted that the BIMDG do not have a representative.

Royal College of Pathologists Group

We are still awaiting the final outcome of the human tissue bill. In anticipation of this it had been agreed that the RCPATH would be establishing a group to look at specimen storage and the JCMG agreed to take this forward with appropriate representation. A Network representative is needed for this.

Note:- It is subsequently being agreed that Jim Bonham's name will be taken forward.

AG

Department of Health Report

The genetic reference labs are being funded for a further two years. NSCAG and GENCAG have now separated and are now independent organisations. The Department of Health announcement from NSCAG about enzyme replacement therapy was noted. There is some confusion about what this exactly includes. Chairman to write to Julia Stalibrass at NSCAG for confirmation.

AG

Jean Kirk reported that initiatives were proceeding in Scotland to look at service provision for lab IMD and there was some confusion about how this related to NSCAG.

Contents and Confidentiality

The JCMG document which has previously been in draft form is to be finalised and circulated.

Antenatal Screening Update

Pat Ward presented an overview of the current position re antenatal screening. The importance of the National Electronic Library was noted.

IMD Review

The JCMG have commissioned a review of IMD services under the chairmanship of Hilary Burton (Public Health Genetics at Cambridge). The first meeting of Stakeholders will take place on 10th November.

Education

The genetics education site has now been launched www.geneticseducation.nhs.uk

5.3 GENCAG and Relationship to Newborn Screening

Jim Bonham outlined the issues arising from screening being part of the genetics medical definition set and providing a very large workload of biochemical genetic testing. It was felt that as screening directors ought to have more of a role in inputting into this advisory process. It was noted that several members expressed concern at the meeting planned for Tuesday 1st February 2005. Several members were not aware of the meeting and there was concern about the programme i.e. there is no laboratory director on the programme on a meeting on Newborn Screening "current issues, future challenges". It was agreed that Jacqui Calvin would take this back to the UKNSLN.

JC

It was also agreed that the chairman would write on behalf of the Stakeholders to Carol Dezateaux expressing disappointment.

AG

6. Training

MH gave an update on the progress with the Trainer post and Trainee posts (copy of presentation attached).

In summary all HSTs are appointed to and all three sites (i.e. Southern, Midlands and Northern clusters) have appointed Trainers. A priority for the Trainers is to work on the MCADD web based case study with Kim Bartlett (reference item 3.7). An informal group has been established to review developments including MH. Colin Samuel, Local Trainer for Southern cluster, HST and a Grade A Trainee.

MH also reported on an update with regard to discussions about the RCPATH examination. There are no firm plans to change at the moment although discussions are leading towards the possibilities of a more specialist part II examination - this would require the availability of sub-speciality examiners.

7. Metabolic Assay Directory

The current position is that all Stakeholder and Associate Laboratories have been asked to complete the list of analytes and specimen detail and also provide details of accreditation status, QA schemes and provide some laboratory details. At the moment the entries are incomplete. All labs are urged to update as soon as possible.

ALL

Issues raised at the discussion with Kim Bartlett and Neil Hamilton (September 14th 2004) included whether we needed access to the assay directory via an e-mail address or other identifiers. This would enable monitoring. The view from the Stakeholders was that this was not necessary.

The next stage is to complete the entries. AG and Janet Stone have access to a trial version of the directory and are currently providing feedback to Neil and Kim. It is hoped that the pilot version will be available for all to try out during late December/January 2005. Once everyone is happy with the version it will be able to go live.

There are plans to link to the Genetic Testing Network Directory which will detail the availability of Molecular tests. There are also longer term plans to link tests to the disorders. Janet Stone has agreed to take this forward and to establish a small group to work on this.

JS

A brief overview of how the Assay Directory looked was presented. Initial comments were that it looked good but it would be helpful to have all labs listed initially by City and to ensure that all tests are in A-Z order.

8. Audit

AG thanked all Stakeholders who had contributed to the review of audit projects and presented a brief overview of completed, ongoing and planned projects (see attached summary).

The amount of Audit activity was very considerable and very encouraging. The main topics for Audit included Newborn Screening, Test Repertoire, Requesting Patterns, Sweat Tests and Protocols. It was agreed that we should undertake a prospective audit and after discussion it was agreed that the availability of ammonia investigations for neonates and children across the UK would be valuable. The North East (Newcastle) have undertaken such an audit and it was agreed that Mori Pourforzam would ask Hillary Wastell whether she would be happy to undertake this audit on behalf of the network.

MP

9. Agenda for Change

There was considerable discussion on the state of progress in individual laboratories. A document about the position of training clinical scientists in Agenda for Change had been obtained via Lesley Tetlow. The advice in this document was that Trainees should not be job matched via local processes and they should not be pressurised for agreed job descriptions at this stage. The document to be circulated to all.

AG

FC reported on the matching at the pilot site. There was still no definitive outcome. Job descriptions were not available but will be able to be circulated at a later date. It was agreed that LT and FC should get together to look at job descriptions and with the aim of producing some generic job descriptions that could be made available to everyone.

LT/FC

There was considerable concern expressed about the whole Agenda for Change process and the appropriateness of AFC for consultant level posts. After much discussion it was agreed that we should possibly send a letter to Sue Hill about this. AG to review after further discussion.

AG

10. Service Questionnaire

There was no time to further discuss this. However, it is possible that this could be incorporated in the National IMD Review.

AG

Workforce Strategy

A recent e-mail from Debbie Hilder has provided some modelling data for the workforce for Paediatric Metabolic Biochemistry. AG to discuss this with Debbie Hilder.

AG

Quality

Steve K reported back from the very successful Quality Day which had been held in London on October 29th 2004. There had been a good response to the day and some valuable actions to take forward. This will be reported on the web.

There were no other items of business and the meeting closed at 16.45 hrs.

Date of next meeting

February 23rd, Birmingham at 11.30am

