

National Metabolic Biochemistry Network

Minutes of Stakeholder meeting held on 16th July 2008 at Birmingham Children's Hospital

Present: Anny Brown (AyB)
Mick Henderson (MH)
Steve Krywawych (SK)
Stuart Moat (SM)
Simon Olpin (SO)
Mary Anne Preece (MAP)
George Gray (GG)
Mark de Hora (MdH)
Jim Bonham (JB chair)
Philip Mayne (PM)

ACTION

1. Apologies

Alan Cooper, Mike Badminton, Guy Besley, Ann Bowron, Jacqui Calvin, Fiona Carragher, John Fyffe, David Isherwood, Helena Kemp, Jean Kirk, Paul Newland, Morteza Poufazam, Janet Stone, Lesley Tetlow

2. Minutes of Meeting held on 23.01.08

Accepted as correct.

3. Matters Arising

3.1 Receipt of aminoacid method questionnaire

AyB confirmed that this had been sent

3.2 Aminoacid reference ranges

Work is continuing and requests have been sent to approximately 10 laboratories to provide data together with EQA data to check for possible results bias. SM to follow-up and progress

SM

3.3 Non lysosomal assay availability

GG and SO to arrange a meeting to include AyB, Alan Cooper, GG, SO, Janet Stone, Marie Jackson to discuss the robustness of current arrangements..

GG/SO

GG also to discuss Krabbe reagent need with Jean Kirk GG

GG

3.4 MPS scheme continuity

JB confirmed that a laboratory in the Netherlands had agreed to consider taking this over and that Marie Jackson had also been contacted.

3.5 Educational presentation

MH confirmed that the acylcarnitine presentation was now available on the website.

3.6 QA Summary on the website

JC had confirmed that this was now on the website.

3.7 Website review

JB confirmed that a meeting had taken place with Neil Hamilton and an update is presented later in the minutes.

3.8 Ammonia Audit

SK confirmed that the source data had been retrieved. SK to review and prepared a report. MAP to check status of latest report and pass on to SK

SK/MAP

3.9 QA meeting report

JB confirmed that these were not on the website but had contacted MAP, SK and TL to send reports on 2005, 2007 and 2006 respectively.

MAP/SK/
TL

3.10 RCPH guidelines

JB confirmed that RCPCH planned to audit the use of the altered consciousness guidelines and to begin work within the next 2 months.

3.11 Commercial sponsorship for meetings

PN had not progressed this at this time. MdH has secured support for the annual BMS meeting.

4. Reports/Liaison with Other Groups

4.1 UKGTN

No report but work going on to designate centres with respect to specialist commissioning.

4.2 BSAG

No report but meeting planned for 22.7.08, to raise issue of date of newborn screening

4.3 JCMG

Report offering advise to the House of Lords select committee looking at genetics. Also concern expressed about the availability of funding for research from DH for genetics. SM suggested that it might be worth approaching Wellcome Trust. It was agreed that JB would liaise with BIMDG to identify likely topics for research activity and potential leads for these. PM suggested that a "Research page" on the website might be useful.

4.4 GenCAG

No report.

4.5 UKNSLN

A new chair, Paul Griffiths, has been appointed. The next meeting is planned for August.

4.6 RCPATH SAC

No report

4.7 SAGE

GG presented an update on the E learning project with an outline of the "knowledge sessions" to be available electronically - attached to the minutes.

5. Expanded Newborn Screening

JB outlined progress with this project. It was encouraging that the NSC in the person of Dr Anne Mackie have embraced this project.

There remain decisions to make about:

- The disorders to include, certainly IVA, MSUD and GA1 need to be agreed. Additional treatable conditions may be included following the completion and return of vignettes which is being co-ordinated by Dr Mackie.
- The centres to include for collaboration, this should be aimed at achieving 500k births in a reasonable time scale.
- The research questions to be answered which will include operational issues such as the reliability of the assay, the positive predictive value, an assessment of possible dysbenefit such as the effect of false positive results and a measure of clinical benefits once key outcome measures have been agreed.

These issues will be discussed at a meeting planned for 26th September to be held in Sheffield.

6. Training and Education

6.1 Update

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

A number of HSTs have now moved on to permanent posts or left the profession. It is heartening that several new HSTs have been employed. These changes are summarised in the table attached. We

are also seeking to include a record of clinical scientists in career posts, but still in training in stakeholder laboratories.

The trainer group is now established and consists of Mick Henderson, George Gray and Lesley Tetlow. We have met twice in addition to the training group meeting in February. We are establishing a work programme aimed at creating syllabi for A grade and HST clinical scientists working in metabolic laboratory medicine. These will form the basis for e-learning packages that will be mounted on the e learning for health website in collaboration with the Royal College of Pathologists. We now need to identify authors for the 12 elements of the A grade programme. George Gray is taking the lead on our participation with this project and is liaising with the ACB Education Committee to co-ordinate efforts.

7 Report from the BMS Group

The BMS training group have continued to meet (11th March and 3rd July) in London. In March we discussed the BMS training post which will be filled by Mark de Hóra who will, amongst other duties, represent the group at the stakeholder meetings.

The group have continued to work with equipment suppliers to provide affordable training courses for laboratory staff. Biochrom have already provided very successful courses and work is in development with Waters and Shimadzu.

Anne Sheldrake and Mark de Hóra met with IBMS representatives to discuss the Diploma in Expert Practice in Biomedical Science. A diploma in newborn screening is now nearing completion and will be available to IBMS members when it has been through the accreditation procedure. It is hoped the diploma will be available to members by the next IBMS congress in September 2009.

The groups' annual conference will take place on September 12th in Birmingham. Topics will include, organic acids, acylcarnitines, transferable skills and individual project reports from laboratory staff.

8. Web Review

8.1 Update

GG offered a report on the meeting with Neil Hamilton and the plans to review the website these include plans:

- To ask all Stakeholders to review their laboratory details and make any amendments.
- To create a new page for Meetings and Workshops
- To create a site map for the Home Page and the Education and Training Page
- To have a separate database containing spectra of individual metabolites linking from the Chromatograms
- To create a page for Audit
- To sort the chromatograms and presentations to enable easier location
- Update Trainer & Trainee Details

8.2 Assay directory

JS sent the following written report: "Work is progressing albeit a little slowly on the next phase of the directory - due to other work pressures. I am aiming to have a draft ready in the next 6 weeks - in effect providing an index relating disease name to the test required. I will send this to you for circulation to the stakeholders for comments and suggestions."

9. Audit

Unfortunately Tim Lang could not attend the meeting. Suggestions for future audit topics included analytical approaches to the laboratory identification of homocystinuria which AyB volunteered to co-ordinate this in partnership with Cardiff and MAP suggested an audit to determine the adequacy of clinical information available on the request form. MAP would contact other centres to undertake a survey.

AyB/
MAP

10. Workforce Planning

MAP agreed to update the existing data.

MAP

11. Quality Assurance schemes

Jacqui Calvin had updated the web entry and invited comments/Emails from the group about news of additional schemes/analytes etc which she would incorporate into the Table.

MH confirmed that six QA samples had already been collected via members of CLIMB for potential use in EQA schemes.

12. Guidelines

Paul Newlands was unable to attend the meeting but reported little progress partly attributable to the pressure of work and partly due to a lack of response from those contacted to undertake work.

13. Finance

JB reported that the finances showed little change and that while it would definitely be worth pursuing commercial sponsorship for meetings, there was no immediate crisis and MetBioNet could continue for the next two/three years at the current level of expenditure.

14. Meetings planned

- LT would be asked to confirm the date for the October MetBioNet QA day to be hosted by Manchester
- GG was progressing work for the planned prenatal diagnosis workshop to be hosted at Birmingham in November 2008, a date will need to be confirmed.
- MAP/SM confirmed their willingness to arrange a B12/propionate workshop in spring 2009
- SK offered to help with JB to arrange a workshop in London on intermediary metabolites with a possible date in April 2009, Fiona Carragher to be contacted.

15. Any Other Business

15.1

MH outlined the plans for a dried blood spot card to be used for tests other than newborn screening that required dried blood spot collection. In general this was welcomed but the following comments were made:

- It was suggested that to avoid confusion all reference to phenylalanine should be deleted indeed the feeling was that named boxes should be avoided
- It was suggested that a different colour and possibly different number of spots should be used to avoid possible confusion with newborn screening cards
- Several people reflected the view that the **NOT FOR NEWBORN SCREENING** comment should be enlarged and be more prominently placed
- There was some discussion about how these cards would reach those who wished to make a request

15.2

JB reminded the group of the Email circular issued on behalf of John Rasco requesting contact with families affected by dicarboxylic aminoaciduria or renal Fanconi syndrome.. SM indicated that they may have a family with one of these conditions.

16. Date and time of next meeting

It was agreed that the next meeting would be planned for Wednesday 14th January 2009 to begin with coffee at 10.00 am in the Pan Pathology Seminar room at Birmingham Children's Hospital

The meeting closed at 14.50 pm

Grade A Syllabus in Clinical Biochemistry – IMD Component

Knowledge Sessions (Mini-Modules) (20 mins)

1 The Effects of a Metabolic Block

A description of how metabolic pathways can be disturbed by metabolic disease and the pathogenesis of the disease. The importance of cofactors and vitamins.

2 Amino Acid Disorders

A short description of the major amino acid disorders that can present at a DGH and the methods used to detect them.

Hyperglycinaemia, Maple Syrup Urine Disease, Tyrosinaemia, Homocystinuria, Cystinuria, Phenylketonuria

3 Organic Acid Disorders

A short description of the major organic acid disorders that can present at a DGH and the methods used to detect them.

Methylmalonic Aciduria, Propionic Acidemia, Glutaric Aciduria Type Biotinidase Deficiency

4 Carbohydrate Disorders

A short description of the major disorders of carbohydrate metabolism that can present at a DGH and the methods used to detect them.

Glycogen Storage Diseases, Gluconeogenic Disorders, Galactosaemia, Hereditary Fructose Intolerance. G6PDH deficiency

5 Lysosomal Storage Disorders

A short description of the major lysosomal disorders that can present at a DGH and the methods used to detect them.

Mucopolysaccharidoses, Oligosaccharidoses, Gauchers Disease, Tay - Sachs disease, Metachromatic Leucodystrophy

6 Urea Cycle Defects

A short description of the major disorders causing hyperammonaemia that can present at a DGH and the methods used to detect them.

OTC Deficiency, CPS Deficiency, Argininosuccinic Aciduria, Citrullinaemia.

7 Lactic Acidaemias

A short description of the major disorders affecting lactate metabolism that can present at a DGH and the methods used to detect them.

Respiratory Chain Disorders, PDH Deficiency

8 Fatty Acid Oxidation Defects

A short description of the major fatty acid oxidation disorders that can present at a DGH and the methods used to detect them.

MCADD, Long chain fatty acid oxidation defects, Multiple Fatty Acid Oxidation Defects, carnitine deficiency

9 Other Metabolic Disorders

A short description of the other inherited metabolic disorders that can present at a DGH and the methods used to detect them.

Peroxisomal Disorders (X-ALD, Zellwegers Syndrome), Menkes & Wilson's Disease, Lesch Nyhan Syndrome

10 Treatments and Monitoring

The current strategies available for the treatment and monitoring of inherited metabolic disease

Dietary control, Vitamin-responsive disorders, Pathway Inhibition, Chelation, Transplantation (Liver, Kidney Bone Marrow), Enzyme Replacement Therapy

11 Prenatal Diagnosis

A brief description of the techniques used for sample collection and the types of testing available for the prenatal diagnosis of inherited metabolic disease.

Foetal Sexing Chorionic Villous Biopsy, Amniocentesis, Foetal Blood Sampling. Metabolite, enzymological and molecular genetic analysis.

12 Newborn Screening

The current services and tests for Newborn Screening in the UK

PKU, Hypothyroidism, Cystic Fibrosis, Sickle Cell Anaemia, MCADD

Total time of course = 240 mins i.e. equivalent to 4 hours lectures

Pathopaedia Topics

Chitotriosidase
Cellulose acetate electrophoresis of mucopolysaccharides
Very long chain fatty acids.
Galactosaemia screening tests
Free and total homocysteine
Rhabdomyolysis
Reyes Syndrome
Leigh's Syndrome
NTBC
Cherry Red Spot
Post mortem biochemical investigations
Tandem mass spectrometry
Sulphocysteine
Metabolome
Acylcarnitines
Newborn bloodspot screening
Leukocyte cystine measurement
Fanconi syndrome

This is merely a start of a list. As the various knowledge sessions are written more instances of topics suitable for inclusion in the Pathopaedia will appear.

Case Sessions

- 1 The child with the raised ammonia
- 2 The child with lactic acidemia
- 3 The hypoglycaemic child.