

National Metabolic Biochemistry (Biochemical Genetics) Network

NEWSLETTER FOR STAKEHOLDERS

July 2003

CURRENT PRIORITIES AND PROGRESS

STOP PRESS

The Genetics White Paper: Our Inheritance, Our Future - Realising the Potential of Genetics in the NHS, was published in late June 2003 (www.doh.gov.uk/genetics/whitepaper.htm). There are plans to invest an additional £50 million in England to provide 'genetic' services.

These include:-

- | | | |
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| strengthening specialised services | - | increasing capacity of genetic laboratories |
| | - | funding additional training |
| | - | additional capital investment |
| | - | IT investment |

It is unclear at this stage what the process and timetable will be for preparing bids, but will update you when we have more information

Survey of Service Provision

The draft questionnaire is in preparation and I plan to circulate this in August 2003.

Web Site

The web site has now been launched, www.metbio.net. You should have all received a list of stakeholder IDs and passwords. If for any reason you have not got these, please contact Mary. The next stages of the web site development is to develop the stakeholders' services section to include minutes and other stakeholder documentation. We will also be including information about the planned workshops and the guidelines which are in preparation.

A major development will be the assay finder. I am pleased to report that Janet Stone has agreed to help with this. If you have any comments about the web site and any specific suggestions/requirements please contact us.

We are also hoping to develop the web site as a training resource, e.g. sharing of presentations, case histories, etc. Mick Henderson is currently giving this some thought so, if you have any ideas or would be interested to help with this, please contact Mick directly.

Education and Training

You should have all received a final version of the document 'Clinical Scientist Training Requirements for Cytogenetic Metabolic Biochemistry and Molecular Genetics Laboratories'. This is being co-ordinated by Steve Abbs at Guy's Hospital. This should be used in your discussions with the Workforce Development Confederations to help with your case in trying to get HST posts established. Please keep Anne informed of any problems.

Graham Beastall and Janet Smith have met with Sue Hill, Chief Scientific Officer, and acknowledge the difficulties we are experiencing. We are not unique - the issues are common with many other sub-specialties. There is recognition of the need for a national lead for commissioning of training posts and trainers - probably via a lead WDC - and I am optimistic that this will be taken forward.

Registration of Clinical Scientists/ Competences for Metabolic Biochemistry as a sub-modality

Draft competences for paediatric/metabolic biochemistry has now been prepared with the help of Lesley Tetlow, Mick Henderson and Jim Bonham. These have been sent to Janet Smith and Graham Beastall for comment. The intention is to finalise these and then send to the Association of Clinical Scientists (ACS) to request that paediatric/metabolic biochemistry be recognised as a sub modality of clinical biochemistry. If anyone feels that they would like to have a look at the proposals at this stage, please let me know as soon as possible. The time frame is such that I would hope that the final version would be completed before the end of summer, and submitted to the ACS by early September.

MRCPath - ? case for a specialist examination

Paediatric representation in the MRCPath examination

Training clinical scientists in our sub specialty has been highlighted as a problem. 'A' grade training is of necessity very general. The focus of those who emerge from the 'A' grade into low 'B' grade posts is very much on sitting the MRCPath part 1. The anxiety is that trainees are discouraged from specialty training until later in their careers. This limits an already small pool of people. However very constructive discussions have been taking place between the Network and the College on the best way forward to promote training in paediatrics. A separate paediatric MRCPath examination was rejected at an early stage as being impractical and probably unattractive. The College is however enthusiastic about enhancing paediatric influence by increasing the frequency of appearance of paediatric questions. There could be one paediatric choice out of five on each paper in the part 1 examination. This does put the responsibility firmly on us to

provide the material. Trevor Gray has complained about a lack of support in this respect in the past. I have begun the process by supplying him with a small variety of questions together with material for use in the practical examination. These were gratefully received. Past questions could be posted on the website with model answers. Between us we must keep this going, it is important. So beware, I shall be bullying all of you for material in the near future!

Mick Henderson

If you have any views about training, registration and the MRCPATH, please contact Anne, Mick, Lesley or Jim.

Training Workshops

Organic acids (Jim Bonham) October 2003
Acyl carnitines (Neil Dalton)
Very long chain fatty acids (Tony Reynolds)
Lysosomal enzymes (Guy Besley)

Please contact the organisers for further information.

Best Practice Guidelines

Just a reminder about who is doing what.

Metabolic autopsy (SUDI - Mary Anne Preece)
IEM and hydrops (George Gray/Mary Anne Preece)
IEM and cardiomyopathy postmortem (Helena Kemp)
Hypoglycaemia (Helen Losty)
Hyperammonaemia (Helen Losty)
Developmental delay (Peter Galloway)
Fits and seizures (Mick Henderson)
Prolonged neonatal jaundice (David Isherwood)
Lactic acidosis (Jim Bonham)
Rhabdomyolysis (Jim Bonham/Simon Olpin)

If anyone needs any funding and/or help, please let me know.

Genetic Reference Laboratories Update

The genetic reference laboratories now have a web site, www.ngrl.org.uk. This is listed as a useful link on our metabolic biochemistry web site.

GenCAG Update

The Genetics Commissioning Advisory Group

This group is made up of representatives from the Department of Health, Commissioners and professionals from the genetics disciplines, public health and pathology. It was established to offer advice on commissioning and the shape of the service Nationally in the light of the monies available for investment in Genetics (£30m announced in April 2002 and a further £50m outlined in the recent White Paper, "Our Inheritance, Our Future" published in June 2003). Of course, as inborn errors of metabolism and newborn screening we now firmly part of "Medical Definition Set 23 - Genetics".

July 31st will see the first meeting of GenCAG in 2003, this was delayed to follow the publication of the White Paper in June. These are exciting time and a real opportunity to develop our service alongside the expansion in Genetics and the development of the Newborn Screening program nationally. To make the most of these opportunities it is important that:

1. We have a well co-ordinated and evidence based National vision - this is very much the role of our network
2. That we clearly identify the resources (both equipment and staff) that we need to fulfil this planning.
3. That we maintain good links locally with the Workforce Development Confederation and Regional Specialist Commissioning leads and clearly spell out our needs ensuring that we are included in any relevant bidding rounds (the next to be completed by December 2003).
4. That we co-ordinate our activities well at a National level via GenCAG, Joint Medical planning group and the Genetic Testing Network.

Can I suggest that you take time to read the latest White Paper (it is available on the DoH website) and to ensure that you are involved with any local response.

Jim Bonham

Accreditation - discussions with CPA

CPA is very happy to include paediatric/metabolic biochemistry as a sub-specialty within the CPA accreditation process. At this stage it will allow us to provide CPA with a list of tests which we deem to be within the repertoire for paediatric/metabolic biochemistry so that these can be appropriately highlighted as part of the inspection process for any lab registering this sub-specialty. This is something important for us

to discuss at our next stakeholder meeting in November. In the meantime, I have sent Cheryl Blair at CPA a list of the stakeholder laboratories.

Genetic Testing Network (GTN) Update

The GTN has organised a series of educational programmes for commissioners. The first of these is planned for the South West of England in October 2003. This will then be repeated at other sites around England later in 2003/4. It is really important that members engage with these to ensure that metabolic biochemistry is appropriately represented in the presentations and discussions with Commissioners. Helena has further details about the South West programme should you require further information at this stage.

National Strategy for Inherited Metabolic Disorders Services in the UK (BIMDG)

Since the BIMDG Workshop in October 2002, the BIMDG are producing a paper to summarise their strategy and vision for the future; contact Graham Shortland. The issues for clinical IMD have been raised by the Joint Medical Genetics Committee (Chairman Heather Skirton). A brief paper summarising the position was prepared by Ruth Newbury-Ecob, Graham Shortland and myself for discussions between Professor Black (Chairman of the Royal College of Physicians) and Professor Sir Liam Donaldson and Sir Nigel Crisp in May 2003. We await to see whether there is any further progress following these discussions. The recent publication of the Genetics White Paper should be an opportunity to progress these issues.

Agenda for Change

We have no further information about job evaluations in specialist areas. If anyone has any further information, could they please contact Anne.

Quality Assurance Workshop

Jim is organising this in Sheffield on October 22nd 2003. It will be for all stakeholders and other laboratory colleagues. Just to remind you that the purpose of the meeting is as follows:-

- Overview of specialised QA schemes - how are we performing in specific schemes
- Are existing schemes appropriate to our needs - how could they improve? Do we need any new schemes?
- Specific QA projects. Which individual labs have undertaken/relevant to the National Metabolic Biochemistry (Biochemical Genetics) Network

Further details will be available from Jim and on the web.

Anne Green
July 2003