



National Metabolic Biochemistry (Biochemical Genetics) Network

Annual Report 2005/6

An NHS/Department of Health funded project
Genetic Services (Specialised Services, Definition Set No. 20)

Host Trust - Birmingham Children's Hospital (BCH)

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Version: 3

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Abbreviations used

ACB	Association of Clinical Biochemists	IMD	Inherited Metabolic Disorders
ACS	Association of Clinical Scientists	JCMS	Joint Committee for Medical Genetics
BCH	Birmingham Children's Hospital NHS Trust	MAD	Metabolic Assay Directory
BIMDG	British Inherited Metabolic Disease Group	NOS	National Occupational Standards
CPA	Clinical Pathology Accreditation	QA	Quality Assurance
GenCAG	Genetics Commissioning Advisory Group	RCPATH	Royal College of Pathologists
GTN	Genetics Testing Network	UKNSLN	UK Newborn Screening Laboratory Network
HST	Higher Specialist Training/Trainee	VLCFA	Very Long Chain Fatty Acids
IEM	Inborn Errors of Metabolism	WDC	Workforce Development Confederation
INDIVIDUALS			
AG	Anne Green	JB	Jim Bonham
MD	Mary Dowling	LT	Lesley Tetlow
MH	Mick Henderson	CS	Colin Samuell
PN	Paul Newland	AS	Anne Sheldrake
GB	Guy Besley		

ADMINISTRATION AND INFRASTRUCTURE

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
<ul style="list-style-type: none"> ▪ Improve the accounting system and budgetary monitoring 	Network budget - set up accounting for 2005/6 - set up budget statements with Finance - produce end of year statements	MD Final statements, BCH Finance	Timely, accurate accounts End of year position finalised March 2006
<ul style="list-style-type: none"> ▪ To transfer admin to new Lead Scientist 	2005/6 - to transfer A/C support to Sheffield	AG/JB/MD end March 2006	All activities transferred March 2006

COMMUNICATION

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
<ul style="list-style-type: none"> Agree objectives for 2005/6 	Meeting of Stakeholders to agree objectives and priorities	AG End June 2005	Meeting 14.06.05 Objectives and work plan agreed for 2005/6
<ul style="list-style-type: none"> To communicate regularly with Stakeholders 	Newsletters	Twice yearly newsletter	Newsletter - October 2005 - March 2006
<ul style="list-style-type: none"> To communicate and liaise with related organisations (GenCag/UKGTN/ACB) - UK EuroGenTest - Europe 	Updates	Regular updates	Updates for UKGTN - October 2005 - Feb 2006 GenCAG - as required for meetings Updates for ACB Newsletter - July 2005 - November 2005 - February 2006

COMMUNICATION contd...

<ul style="list-style-type: none"> To present work about Network activities 	<p>Publications/presentations</p>	<p>Presentations</p> <p>Flyer for RCPCH members</p> <p>Plenary lecture for RCPCH - April 2006</p>	<p>Poster at EuroMedLab, Glasgow, May 2005</p> <p>Poster at SSIEM, Paris, September 2005</p> <p>Presentation to RCPATH Council, March 2006</p> <p>Circulated January 2006</p>
<ul style="list-style-type: none"> To organise a national meeting 	<p>To organise a national meeting</p>	<p>Agreed for June 2006 at Manchester (LT/GB)</p>	<p>National meeting planned for 2006/7</p>

SERVICE PROVISION

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
Service provision across Stakeholder laboratories - to progress issues	To input into National Service Review of IMD (JCMG)	AG November 2005	Summary findings and recommendations circulated to Stakeholders - June 2005 Completed Lab document to Hilary Burton - Sept. 2005
	To engage in national tariff discussions	AG/Jim Bonham	No significant progress - difficult to engage with anyone
	Engage with GenCAG/DH Commissioning on needs	AG/JB	Presentation at JCMG - Nov. 9 th 2005 IMD Review document completed (Hilary Burton) - Dec. 2005 GenCAG presentation - 23 rd Feb 2006

TRAINING and EDUCATION

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
RCPATH	To continue to liaise with the College to provide Qs input into MRCPATH examination	Mick Henderson (Lead Trainer) Ongoing	Qs provided 2005/6
ACB/ACS - Paediatric Metabolic Biochemistry submodality	To monitor registrations To provide input into the ACS process as required	AG ongoing AG to identify individuals if required	No requests notified
HST posts	Establish training syllabus and individual logbooks Organise annual assessments. Include ACB regional tutors to ensure progress towards MRCPATH	MH/LT/JB MH/local trainers	Draft completed June 2005 October/November 2005
National training strategy	Creation of web based training resources Organise supplementary one day courses Collaborate with and enhance training opportunities from other groups	MH/local trainers HSTs/local trainers Various	Web aspirations outlined with site managers April 2005 Collaboration with BIMDG July 2005 Trainee/trainer meeting Feb. 2006

TRAINING and EDUCATION contd...

To promote and support a training structure for biomedical scientists	To establish a structure for biomedical scientist training	Anne Sheldrake via lead trainer	Preliminary meetings November 2005 and January 2006 Inaugural meeting planned April 2006
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SPECIFIC PROJECTS Working Groups

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
Best practice guidelines	To produce guidelines - see attached appendix	Ongoing (see Appendix 1)	Neonatal Jaundice completed Skin Biopsy completed (see Appendix 1 for those in progress) Gate Keeping transferred to PN - Nov. 2005
Web Site Development	<p>To agree gate keeping functions and leads</p> <p>To update and modernise web</p> <p>To provide a metabolic assay directory (MAD) as part of web site</p>	<p>MH/AG to agree with web managers</p> <p>MH/AG to agree with web managers</p> <p>To pilot MAD</p> <p>To launch MAD</p> <p>To link to UKGTN site</p>	<p>Agreed at meeting on 20th May 2005 (see Figure 1)</p> <p>Agreed at meeting on 20th May 2005 Web improvements completed</p> <p>Input completed early 2005</p> <p>Pilot completed April 2005 Launch of site 1st May 2005</p> <p>To consider 2006/7</p>

SPECIFIC PROJECTS

Working Groups contd...

		To link to EuroGenTest	Preliminary discussions with Brian Fowler - September 2005 - December 2005
		To develop a disorder-based enquiry function (Janet Stone)	Draft disorder-test association prepared - March 2006
		To link to RCPATH	Agreed with College - March 2006
	To monitor site and keep news updated	MD	Up-to-date details on site
	To develop an email messaging system for Stakeholders	AG with Neil Hamilton	Launched January 2006
	To develop training site	Mick Henderson - see Training and Education - page 6	
Molecular based testing	To review needs for IMD and input into Genetic Testing Network	Proposals for new tests to UKGTN on a regular basis Mike Badminton and Sarah Ball agreed as representatives	UKGTN reorganised - Jan. 2006 December 2005

SPECIFIC PROJECTS

Working Groups contd...

Workshops:- To agree topics and leads To undertake workshops	Topics for workshops 2005/6:- Mitochondrial disorders (Sheffield) Amino acid disorders (Bristol) Oxalate/neurotransmitters - Associate Lab Workshop (London)	Leads and dates advertised on web site	All 2005/6 workshops completed (Appendix 2) Report of all workshops on Web Archive Workshops planned for 2006/7 (Appendix 2)
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QUALITY STANDARDS

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
To provide a focus for quality assurance	To review performances of specific schemes as part of an Annual Quality Day	Mary Anne Preece (Birmingham) October 2005 To agree workshop venue October 2006 (AG)	Annual Quality Day - 18 th October 2005. Belfast - 4 th October, 2006
	To produce a summary of QA schemes for metabolic biochemistry and screening for web site	Jacqui Calvin June 2005	Completed - June 2005
	To develop an interactive, searchable data base quality assurance scheme developing on web	To consider commissioning 2006/7	On hold until 2006/7
Accreditation for metabolic biochemistry to be recognised by CPA	To continue dialogue and input with CPA	AG ongoing	QA Summary document sent to CPA - June 2005

QUALITY STANDARDS contd...

Audit requirements	To review audit needs	To plan and implement ammonia audit initiative - October 2005 (Mori Pourfarzam/Jim Bonham/Ying Foo) To re-audit 2006 (Ying Foo) To plan SUDI audit (AG) for 2006/7	Ammonia audit - first round completed (Ying Foo) 2005 For 2006/7 For 2006/7
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THE NATIONAL AGENDA

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
Workforce planning	<p>To liaise with professional bodies and input into national discussions</p> <p>To update Workforce Planning document</p> <p>To keep workforce data updated via training infrastructure</p>	<p>Ongoing LT/AG/JB/AG</p> <p>June 2005 (LT)</p> <p>Colin Samuelli via Lead trainer</p>	<p>Completed June 2005</p> <p>Continued dialogue with Debbie Hilder</p> <p>Annual data collection initiated September 2005</p> <p>Completed and presented to Stakeholders February 2006</p>
Agenda for Change National Occupational Standards	To continue to input into national discussions and link with UKNSLN	Ongoing Lead - LT	

THE NATIONAL AGENDA

Newborn Screening	To input into national discussions on behalf of Stakeholders	AG	First meeting December 20 th , 2005
	To work with UKNSLN on shared/complementary issues	AG	
	To represent Stakeholders on Blood Spot Advisory Group	AG	

Appendix 1

GUIDELINES

Guideline Name	Lead	Status
Autism	Neil Dalton	Draft awaited
Dysmorphology	Guy Besley and George Gray	Draft awaited
Encephalopathy	Jim Bonham	Draft awaited
IEM and cardiomyopathy postmortem	Helena Kemp	Draft awaited
Developmental delay	Peter Galloway	Second draft received
Liver dysfunction in the older child	Fiona Carragher	Draft awaited
Metabolic autopsy (SUDI)	Mary Anne Preece	Draft awaited
Prolonged neonatal jaundice	David Isherwood/Pippa Goddard	Completed 2005
Lactic acidosis	Jim Bonham	Draft awaited
Rhabdomyolysis	Jim Bonham/Simon Olphin	Third draft in preparation
Unexplained tubular function	Steve krywawych	Draft awaited

Appendix 2

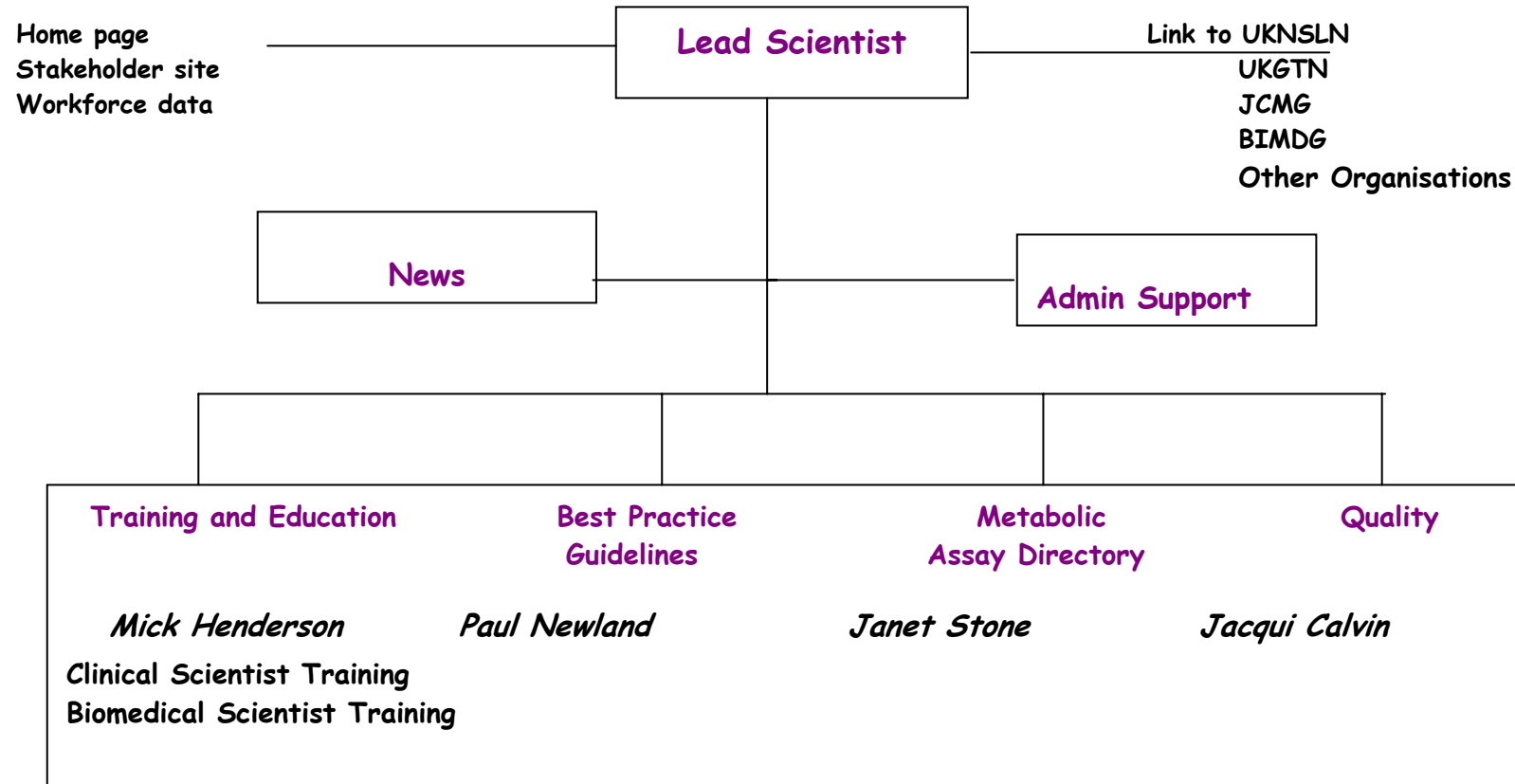
WORKSHOPS 2005/6 (To provide training for existing or new staff)

Workshop	Organiser	Date
Mitochondrial Disorders	Jim Bonham	15 th June, 2005
Amino Acids	Ann Bowran	22 nd November, 2005
Oxalate and inborn errors of dopamine and serotonin metabolism.	Gill Rumsby	23 rd February, 2006
Purines and porphyrins	Fiona Carragher/Mike Badminton	27 th June, 2006
Vitamins, including B ₁₂ and folate	Mary Anne Preece	October/November 2006
Intermediary metabolism	Ying Foo/Jim Bonham	2007

Figure 1

National Metabolic Biochemistry Network (Biochemical Genetics)

Web Gatekeepers/Network Roles



A. Green
March 2006