



National Metabolic Biochemistry (Biochemical Genetics) Network

REPORT FOR
2002/3 and 2003/4

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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KEY OUTCOMES

- Creation of Paediatric Metabolic Biochemistry as a sub-modality
- Agreed National Training Strategy with DOH funding for trainers and trainees
- Network web site established
- Two workshops provided for Stakeholder laboratories
- Best practice guidelines now on web site

ADMINISTRATION AND INFRASTRUCTURE

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
<ul style="list-style-type: none"> Implement management infrastructure and support 	Computer and office set up (home base)	AG end November 2002	Completed November 2002
	Mailing details:- Set up lists of - stakeholder members associated members circulation lists professional bodies National Group Links	MD End November 2002	Completed November 2002
	Network Stationery: Letterheads Compliment slips	MD End November 2002	Completed November 2002
<ul style="list-style-type: none"> Implement accounting system and budgetary monitoring 	Network budget - set up budget statements - set up accounting via BCH Finance - produce end of year statements	MD End November, 2002 End of year Finance Statements, BCH Finance - April 2003 - April 2004	Completed February 2003 Completed April 2004

COMMUNICATION

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
<ul style="list-style-type: none"> ▪ Form a stakeholder group and agree objectives 	Meeting of Stakeholders to agree objectives and priorities	AG End March 2003	Meeting 10.02.03. Objectives and work plan agreed for 2003/4
<ul style="list-style-type: none"> ▪ To communicate regularly with Stakeholders 	Newsletters	Twice yearly newsletter	Newsletter (July 2003) Newsletter (March 2004) Updates - December 2002 - June 2003 - October 2003 - April 2004
<ul style="list-style-type: none"> ▪ To communicate and liaise with related organisations (GenCAG/GTN/ACB) 	Updates	Regular updates	Update for GTN - June 2003 - January 2004 - April 2004 GenCAG - December 2003 Update for ACB Newsletter - June 2003 Update on training activities (Louise Newport) - March 2004

SERVICE PROVISION

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
<ul style="list-style-type: none"> ▪ Document and risk assess the existing service and explore future back-up arrangements 	Prepare draft questionnaire for circulation	AG December 15 th 2002	Decision to delay - to coincide with work on web based assay finder
	Circulate draft for comment	AG/MD End December 2002	Completed September 2003
	Prepare final version of questionnaire	AG Mid September 2003	Completed September 2003
	Circulate final version	MD End September 2003	Circulated October 2003
	Chase up returns	MD End November 2003	Chased up end December 2003
	Analyse returns	AG January 2004	Completed January 2004 (Report available as a separate document)
	Present summary to stakeholders: <ul style="list-style-type: none"> - prepare summary - discuss outcome - agree actions 	AG Feb/March 2004 at Stakeholder meeting	Presented to Stakeholders February 25 th February, 2004 Further action agreed for 2004/5

TRAINING and EDUCATION

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
RCPath	To liaise with the College to provide Qs for MRCPPath examination	Mick Henderson (Lead) Ongoing	Increased number of questions in MRCPPath examination
ACB/ACS - Paediatric metabolic biochemistry to be a recognised submodality of clinical biochemistry	To discuss proposals with ACB officers To discuss proposals with trainers To prepare draft competencies	AG Competencies agreed and submodality available for registration October 2003	Working Group (AG/MH/JB/LT) January 2003. Discussion with trainers at Focus, May 2003). Draft competencies prepared (June 2003) Final version agreed (September 2003) with ACB Document submitted September 2003 Sub modality recognised October 2003
HST posts/WDCs	To prepare a combined document highlighting the training and recruitment issues for genetic scientists	AG Document May 2003	Document completed (JMC Genetics) May 2003

TRAINING and EDUCATION contd..

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
National training strategy	<p>To prepare a strategy document for training</p> <p>To obtain funding for trainers and trainees</p>	<p>AG August 2003</p> <p>AG December 2003</p>	<p>Strategy document agreed with Stakeholders - August 2003</p> <p>Strategy document submitted to DOH - September 2003 Funding for trainers (3) and trainees (HST posts) agreed December 2003 Job description for Lead Trainer post agreed March 2004</p>

SPECIFIC PROJECTS

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
Best practice guidelines	<p>To agree topics (see separate list, minutes of Stakeholders Group, February 10th, 2003)</p> <p>To produce guidelines</p>	AG	<p>List of topics agreed and commissioned (February 2003)</p> <p>Hydrops Guideline on web (Jan. 04)</p> <p>3 draft guidelines prepared (Jan. 04) (see appendix)</p>
Web Site	<p>To agree web site supplier</p> <p>To agree web contract/specification</p> <p>To commission site (see separate specification):-</p> <ul style="list-style-type: none"> - Phase 1 - Phase 2 <p>To provide a metabolic assay directory (MAD) as part of web site</p>	<p>AG April 2003</p> <p>July 2003</p> <p>November 2003</p> <p>AG - November 2003</p> <p>Identify Working Group</p>	<p>Web site launched with Stakeholder details (30th April, 2003)</p> <p>Phase 1 completed (June 2003)</p> <p>Phase 2 completed December 2003</p> <p>Outline agreed and list of tests identified (as part of service questionnaire)</p> <p>Working Group established to develop MAD (Lead Janet Stone - Bristol) during 2004</p>

SPECIFIC PROJECTS

Molecular based testing	To review needs for IMD and input into Genetic Testing Network	AG - to undertake review and needs for new tests September 2003	Review complete October 2003 and inputted to GTN On-going need identified - to establish sub-group for 2004/5 (Lead Guy Besley - Manchester)
Workshops:- To agree topics and leads To undertake workshops	To agree topics for workshops (see separate list, minutes of Stakeholder Group February 10 th 2003)	AG	List of topics agreed and commissioned (February 2003) Dates advertised on web site (September 2003) Workshops undertaken - VLCFA, December 2003 - Lysosomal Enzymes, February 2004 See appendix

QUALITY STANDARDS

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
To provide a focus for quality assurance	To review existing schemes and need for new schemes To review performances of specific schemes as part of an Annual Quality Day	Jim Bonham (Sheffield) October 2003	Annual Quality Day - 22 nd October 2003
Accreditation:- Needs for metabolic biochemistry to be recognised by CPA	To engage with CPA To consider specialty specific standards	AG	Agreed with CPA that metabolic biochemistry be a recognised sub specialty (April 2003) Agreed a list of specialised QA schemes - December 2003
Audit requirements	To review audit needs	To discuss at Annual Quality Day	Outstanding for 2004/5

THE NATIONAL AGENDA

OBJECTIVE	ACTIONS	LEAD/PROCESS AND TARGET DATE(S)	OUTCOME
Workforce planning	To liaise with professional bodies and input into national discussions	Ongoing Lead Lesley Tetlow (Manchester)	Initial meeting - February 2004 Network Working Group formed - March 2004 (Lesley Tetlow, Jacqui Calvin and Ann Sheldrake)
Agenda for Change National Occupational Standards	To input into national discussions To appoint a small working group	Ongoing	Attended DOH Workshop - December 2003 Working Group formed (Jacqui Calvin, Ying Foo and Lesley Tetlow)

Appendix

WORKSHOPS

(To provide training for existing or new staff)

Workshop	Organiser	Date
Quality Assurance	Jim Bonham	22 nd October, 2003
Very long Chain Fatty Acids	Ying Foo	11 th December, 2003
Lysosomal Enzymes	Guy Besley	16 th February, 2004
Urine Organic Acids	Jim Bonham	2004/5
Acyl Carnitines	Neil Dalton	2004/5
Carbohydrate Disorders	Jacqui Calvin	2004/5
Non-lysosomal Enzymes	George Gray	2004/5
Quality Assurance	Ying Foo	22 nd October, 2004
Mitochondrial Disorders	Jim Bonham	2005/6

Appendix

GUIDELINES

Guideline Name	Lead	Status
Metabolic autopsy (SUDI)	Mary Anne Preece	
IEM and hydrops	George Gray/Mary Anne Preece	Completed - on Web January 2004
IEM and cardiomyopathy postmortem	Helena Kemp	
Hypoglycaemia	Helen Losty/Anne Green	Second draft completed
Hyperammonaemia	Helen Losty/Anne Green	Second draft completed
Developmental delay	Peter Galloway	First draft received
Fits and seizures	Mick Henderson	Second draft received
Prolonged neonatal jaundice	David Isherwood	
Lactic acidosis	Jim Bonham	
Rhabdomyolysis	Jim Bonham/Simon Olphin	First draft received