

Needs Assessment and Review of IMD services in the UK

Professor Anne Green

Lead Scientist

www.metbio.net

Acknowledgements

- Jim Bonham , Sheffield
- Mick Henderson, Leeds
- Stakeholder Colleagues
- Public Health Genetics Unit
 - Hilary Burton
 - Simon Sanderson
- DH Genetics
- Mary Dowling Birmingham
- Birmingham Children's Hospital
- ACB Training Committee
- Royal College Pathologists (SAC Clin Biochem)
- British Inherited Metabolic Disorders Group (BIMDG)

- Set Scene/Background
- National Lab Network
- Review of National needs

UK Background – Genetics Agenda

- Specialised services definition
 - Includes lab biochemical genetics
- Genetics White paper monies 2001
 - huge development monies

UK Background in the Labs

- **lab services for IMD under pressure**
 - increasing workload
 - demands for new tests, screening developments
 - robustness of rare, v rare tests
- **no manpower or training plan**
- **part of general clinical biochemistry**
(retirements & succession planning, specialisation)
- **lack of awareness of issues by others/DH**

Metabolic Biochemistry Labs in UK



DH Genetics Project

- **Form a network of stakeholder laboratories across UK**
 - Communication
 - Sharing of information
 - Common practices
- **Address key issues**
 - Manpower plan
 - Training strategy
 - Testing for rare disorders (service provision)
 - Assay directory
 - Develop guidelines
 - Quality initiatives

Genetics Development Project

- **DH project**
 - Oct 2002 - Oct 2004
(extended to April 2006)
- **Hosted – Birmingham Children's Hospital**
- **Lead Scientist**
 - 0.4 wte
 - A/C support

Training Strategy

- **Paediatric Metabolic Biochemistry**
 - recognised sub modality (**HPC**)
- **Lead Trainer** (DH funded)
 - Dr Mick Henderson – June 2004
- **Local Trainers** (DH funded)
 - London
 - Sheffield
 - Birmingham
- **HSTs**
 - 8 **new** posts from DH Genetics
 - (3 posts funded WDDs)



[Home Page](#)

[About Us](#)

[Stakeholder Details](#)

[Stakeholder Resources](#)

[Links](#)

[Training and Education](#)

[Best Practice Guidelines](#)

[Quality](#)

[Metabolic Assay Directory](#)



NHS Funded Project
Copyright © MetBioNet 2004

Metabolic Biochemistry Network

WELCOME TO THE METABOLIC BIOCHEMISTRY NETWORK

The Network is a group of specialist laboratories providing tests for the diagnosis and management of patients with Inherited Metabolic Disorders across the United Kingdom. The group comprises 17 stakeholder laboratories and 5 associate laboratories.

The Network was formed in October 2002 with funding from the Department of Health and is part of the NHS Genetics Services. These services are commissioned as NHS specialised services (definition set number 20).

Activities of the Network are co-ordinated by a Lead Scientist - Dr. Anne Green at Birmingham Children's Hospital (Metbionet@aol.com) with support from our Administrator Mary Dowling (mary.dowling@bch.nhs.uk), and is hosted by Birmingham Children's Hospital NHS Trust. See 'About us' for more details about the aims, background and current activities.

The Network has an assay directory to source laboratory testing services in the UK for specialist metabolites and enzymes for inherited metabolic disorders. There is also an active training and education initiative and best practice guidelines aimed to help local non specialist laboratories and clinical teams.

Comments/suggestions about the website should be directed to the Lead Scientist (anne.green@bch.nhs.uk) or the Administrator (mary.dowling@bch.nhs.uk)

For more information, please explore the links on the left.

Thank you for visiting MetBioNet

MetBioNet News

23 February 2006 Associate Laboratories Workshop

22 November 2005 Amino Acid Disorders workshop

[News Archive](#)

[MetBioNet Home Page](#)

[Content Disclaimer](#) [Website Management](#)

View Stakeholder Details

BIRMINGHAM
West Midlands Laboratory for Inherited Metabolic Disorders and Neonatal Screening
Department of Clinical Chemistry
Birmingham Children's Hospital NHS Trust
Steelhouse Lane
Birmingham
B4 6NH

Fax No: 0121 333 9911
Web Site: <http://www.bch.org.uk/departments/imd/laboratoryservice.htm>

For Metabolic Queries Contact Duty Metabolic Biochemist	Tel No: 0121 333 9864
--	-----------------------

Senior Clinical Scientists
Dr. Anne Green
Consultant Clinical Biochemist & Head of Department
Tel No: 0121 333 9922
e-mail: anne.green@bch.nhs.uk

Mrs. Mary Anne Preece
Consultant Clinical Biochemist & Head of IMD Section
Tel No: 0121 333 9940
e-mail: maryanne.preece@bch.nhs.uk

Dr. George Gray

NHS

Met Bio Net NHS

Newcastle
Leeds
Sheffield
Cambridge
London

1 of 2 8.26 x 11.69 in


Training and Education

National Metabolic Biochemistry Network - Mozilla Firefox

File Edit View Go Bookmarks Tools Help

http://www.metbio.net/education/

Customize Links Free Hotmail Windows Marketplace Windows Media Windows



MetBioNet Training and Education

Update on Training Activities

Following the agreement to fund the trainer and trainee posts for the Metabolic Biochemistry Network following represents the current position.

Trainers

A lead trainer, Dr. Mick Henderson, Leeds was appointed in July 2004 (0.4 wte). Subsequently further trainers were appointed for the three clusters i.e. North, Midlands and South. These are as follows:-

Northern Cluster

Dr. Jim Bonham	(Sheffield Children's Hospital)	shared post
Professor Rodney Pollitt	...	(0.4wte)

Southern Cluster

Dr. Colin Samuels	(Great Ormond Street Hospital)	(0.4wte)
-------------------	--------------------------------	----------


Midlands Cluster

Dr. R.G.F. Gray	(Birmingham Children's Hospital)	shared post
Miss. K. Hall	...	(0.8wte)

The 8 HST posts were appointed to during the period November 2004 – January 2005 in the following centres:-

- Birmingham Children's Hospital
- Southmead Hospital, Bristol
- Bristol Royal Infirmary

MetBioNet Home Page
Educational Resources
Meetings
Further Information
Case study template



NHS
NHS Funded Project
Copyright © MetBioNet 2004

Done


Training and Education - Documents

National Metabolic Biochemistry Network - Mozilla Firefox

File Edit View Go Bookmarks Tools Help

http://www.metbio.net/education/


Customize Links Free Hotmail Windows Marketplace Windows Media Windows



Training and Education

[MetBioNet Home Page](#)

[Training documents](#)



NHS Funded Project
Copyright © MetBioNet 2004

Training documents

Current trainers and trainees

Higher Specialist Trainee Log Book July 2005

Paediatric Metabolic Biochemistry: A sub-modality of Clinical Biochemistry Anne Green November 2003

RC Path. Recruitment And Training Anne Green January 2003

Recommendations for A grade core training MetBioNet Training Group Spring 2005

Recruitment and Training for Clinical Scientists (Discussion Paper)

Specialist Training For Clinical Scientists Metabolic Biochemistry (Biochemical Genetics) Jim Bonham, Anne Green May 2002

Training Strategy document Anne Green September 2003

Whilst the metabolic biochemistry network has taken every effort to provide accurate and up to date information for this website the stakeholders accept no liability or responsibility for the accuracy or completeness of any information on this site.

All items may be subject to change from time to time

Website Management

Training and Education - Presentations

National Metabolic Biochemistry Network - Mozilla Firefox

File Edit View Go Bookmarks Tools Help

http://www.metbio.net/education/

Customize Links Free Hotmail Windows Marketplace Windows Media Windows



Training and Education

MetBioNet Home Page

Chromatograms

Presentations



NHS Funded Project
Copyright © MetBioNet 2004

Presentations

Amino Acid Pot Pourri Mick Henderson May 2005 Mick.Henderson@leedsth.nhs.uk

Assessing Hyperinsulinism Lesley Tetlow May 2005 lesley.tetlow@CMMC.nhs.uk

Case Presentation: An Irritable Infant with a Surprise Diagnosis Mick Henderson August 2003 Mick.Henderson@leedsth.nhs.uk

Diagnosis of CDG Enzyme Analysis and Other Investigations Viki Worthington April 2005

Fat oxidation and defects of the mitochondrial respiratory chain Simon Olpin June 2005

Galactosaemia and Immunoreactive Trypsin Dr Jacqui Calvin April 2005 jacqui.calvin@addenbrookes.nhs.uk

GSD diagnosis – can liver biopsy be avoided? Sue Alger April 2005

Management of neonatal jaundice Cath Harrison May 2005

Mitochondrial Diseases Due To Nuclear Gene Defects Garry Brown June 2005 garry.brown@bioch.ox.ac.uk

Mitochondrial DNA Disease: Clinical and histochemical features Doug Turnbull June 2005 D.M.Turnbull@newcastle.ac.uk

Mitochondrial Respiratory Chain Enzyme Analysis Simon Heales June 2005 simon.heales@uclh.nhs.uk

Neonatal Biochemistry: Investigation for Inherited Metabolic Disorders (IMDs) Anne Green May 2005 anne.green@bch.nhs.uk

Organisation of Laboratory Testing Dr JR Bonham June 2005 jim.bonham@sch.nhs.uk

Paediatric Emergencies Jean Kirk May 2005 Jean.Kirk@luht.scot.nhs.uk

Peroxisomal disorders Guy Besley July 2003 Guy.Besley@CMMC.nhs.uk

Presentation and investigation of mitochondrial disease in children Andrew Morris June 2005 andrew.morris@mmc.nhs.uk

Presentations remain the intellectual property and responsibility of the author. Any queries relating to content should be addressed to the author and not to MetBioNet. Permission to reproduce or otherwise use any of the material contained within these presentations must be sought from the author.

Website Management

Metabolic Assay Directory

National Metabolic Biochemistry Network - Mozilla Firefox

File Edit View Go Bookmarks Tools Help

http://www.metbio.net/

Customize Links Free Hotmail Windows Marketplace Windows Media Windows



[Home Page](#)

[About Us](#)

[Stakeholder Details](#)

[Stakeholder Resources](#)

[Links](#)

[Training and Education](#)

[Best Practice Guidelines](#)

[Quality](#)

[Metabolic Assay Directory](#)



NHS Funded Project
Copyright © MetBioNet 2004



Metabolic Assay Directory



[Introduction](#)

[Help](#)

Find Analytes starting with:

A	B	C	D	E	F	G	H	I	J	K	L	M
N	O	P	Q	R	S	T	U	V	W	X	Y	Z
Number												

Or use the search below:

Please select Laboratory **OR** Analyte to search

Analyte:

Laboratory:

Metabolic Assay Directory - Search

National Metabolic Biochemistry Network - Mozilla Firefox

File Edit View Go Bookmarks Tools Help

http://www.metbio.net/

Customize Links Free Hotmail Windows Marketplace Windows Media Windows



[Home Page](#)

[About Us](#)

[Stakeholder Details](#)

[Stakeholder Resources](#)

[Links](#)

[Training and Education](#)

[Best Practice Guidelines](#)

[Quality](#)

[Metabolic Assay Directory](#)



NHS Funded Project
Copyright © MetBioNet 2004



Search Results - Registered Analytes



[New Search](#)

Acyl carnitines (by tandem MS)

Laboratory name	Specimen	Sample requirements
Birmingham - Birmingham Childrens Hospital NHS Trust	Plasma & Blood Spot	
	plasma or serum	0.2ml
Cambridge - Addenbrookes Hospital	Blood Spot	
Cardiff - University Hospital of Wales	Blood	Blood spot - LiHeparin
Glasgow - Royal Hospital for Sick Children	Blood spot and Li Hep Plasma	50 ul
Leeds - St. James University Hospital	dried blood spot, plasma	
London - Great Ormond Street Hospital	Bloodspot	
London - Guy's and St. Thomas' Trust Chemical Pathology	Plasma	1ml Freshly Frozen
Manchester - Willink Biochemical Genetics Unit	Dried blood spot	or 1ml EDTA blood
Newcastle upon Tyne - Spence Biochemical Genetics Unit	Dried Blood spots or Whole blood (0.5ml EDTA)	Send by 1st class post
	Fibroblasts	Skin biopsy or cultured cells
	Urine	1ml
	Tissue	At least 30mg send frozen
Sheffield - Sheffield Childrens Hospital		

Outcomes

- **Training**
- **Web site**
- **Workshops**
 - 2 – 2003/4
 - 3 – 2004/5
 - 4 – 2005/6

Outcomes

- Web site
- Training
- Workshops
- **Best Practice Guidelines**
 - 5 completed
 - 11 in preparation

Best Practice Guidelines

National Metabolic Biochemistry Network - Mozilla Firefox

File Edit View Go Bookmarks Tools Help

http://www.metbio.net/

Customize Links Free Hotmail Windows Marketplace Windows Media Windows



[Home Page](#)

[About Us](#)

[Stakeholder Details](#)

[Stakeholder Resources](#)

[Links](#)

[Training and Education](#)

[Best Practice Guidelines](#)

[Quality](#)

[Metabolic Assay Directory](#)



NHS Funded Project
Copyright © MetBioNet 2004

Best Practice Guidelines

Network members and associated colleagues are preparing best practice guidelines aimed to help the local laboratory and clinical teams. These are laboratory based guidelines which have been formulated by expert opinion. They are not evidence based guidelines. If users/readers have any comments on any aspect of these guidelines the network would very much want to hear from you. Please contact the Lead Scientist e-mail metbionet@aol.com or alternatively the Lead Author as detailed in the individual guidelines. Clearly, guidelines may become out of date before it is possible to update them, so users must bear this in mind when using them.

Contents

[Guidelines for the Biochemical Investigation of Patients with Foetal and Neonatal Hydrops](#)

[Guidelines for Investigation of Fits and Seizures \(Instruction Sheet for CSF sample collection \)](#)

[Guidelines for the Investigation of Hypoglycaemia in Infants and Children](#)

[Guidelines for the Investigation of Hyperammonaemia for Inherited Metabolic Disorders](#)

[Appendix - Notes on the measurement of ammonia in blood/plasma](#)

[Skin Biopsy - Information Sheet for parents/carers](#)

[Skin Biopsy - Consent form](#)

[Neonatal Jaundice in Inherited Metabolic Disorders](#)

Disclaimer for all Guidelines

These are laboratory guidelines reflecting current base practice in specialist metabolic laboratories the UK. They are not evidence based but reflect expert opinion. The network cannot accept any responsibility for any errors/omissions and users must take responsibility for use.

Website Management

Outcomes

- Web site
- Manpower & Training
- Workshops
- Best Practice Guidelines
- **Quality**
 - Oversee QA (annual meeting)
 - CPA recognition as Paediatric Metabolic Biochemistry
 - EQAS summary
 - Audit initiatives

Outcomes

- Web site
- Manpower & Training
- Workshops
- Best Practice Guidelines
- Quality
- **Service Provision** (Questionnaire)
 - Training/staffing – special skills
 - Equipment needs
 - Rationalisation/development rare tests
 - Out of hours needs

Service assessment

- Questionnaire
 - October – Dec 2003
 - 100% return
- Lab size
- Core test provision
- Specialist test provision
- Equipment
- Turnaround times
- Staffing
- Training

Stakeholder Laboratories(n=16)

Lab size/ Populations served

Size of Population

- 4 million or greater 5
- 3 - 3.9 million 2
- 2 - 2.9 million 6
- 1 - 1.9 million 3

- **CPA Accreditation**
 - 15/16 accredited

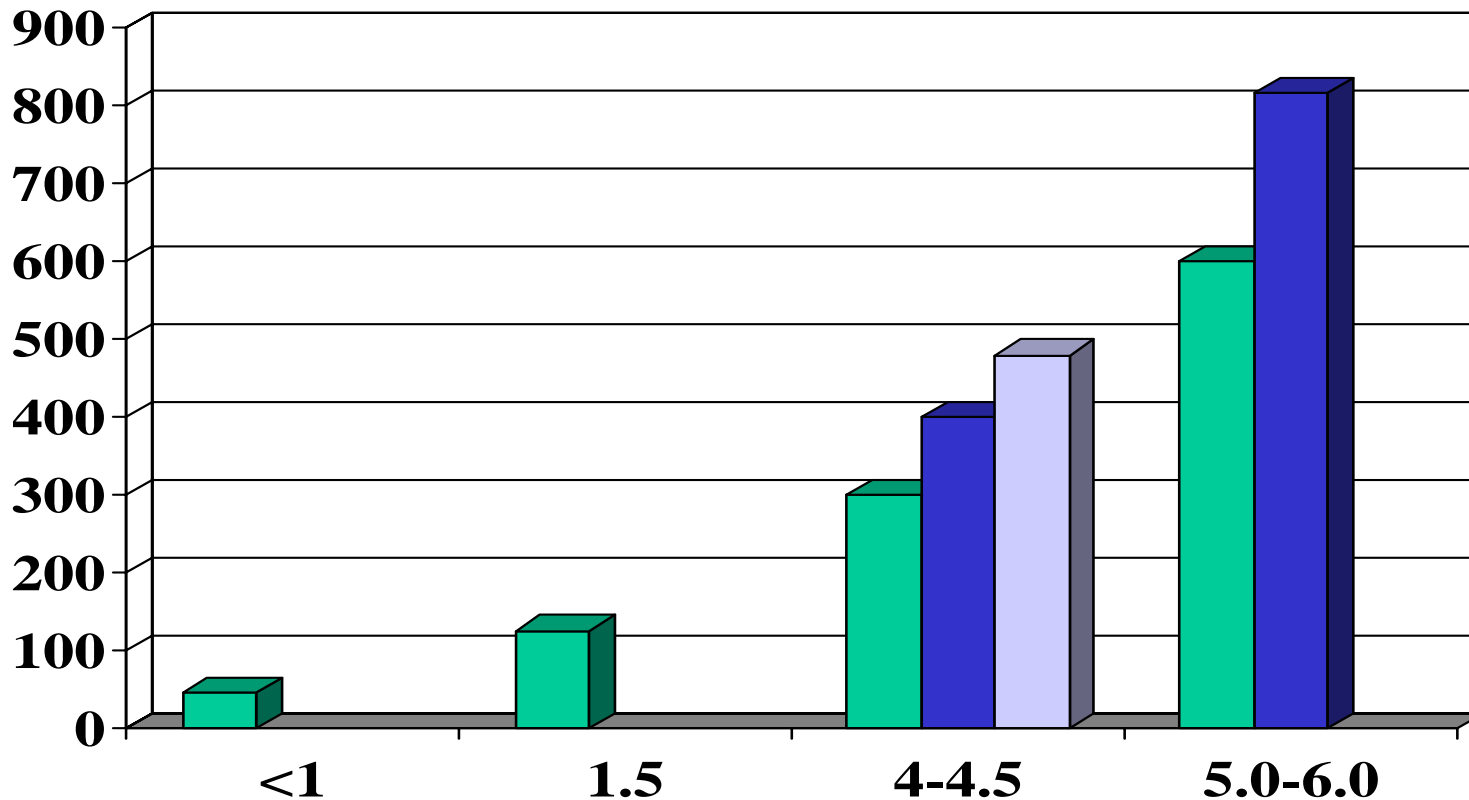
Manpower 2004/5

Clinical Scientists Grade	Number (wte)
Consultant (scientist & medical)	22 (13.6)
Principal	24 (19.85)
Senior	25 (21.7)

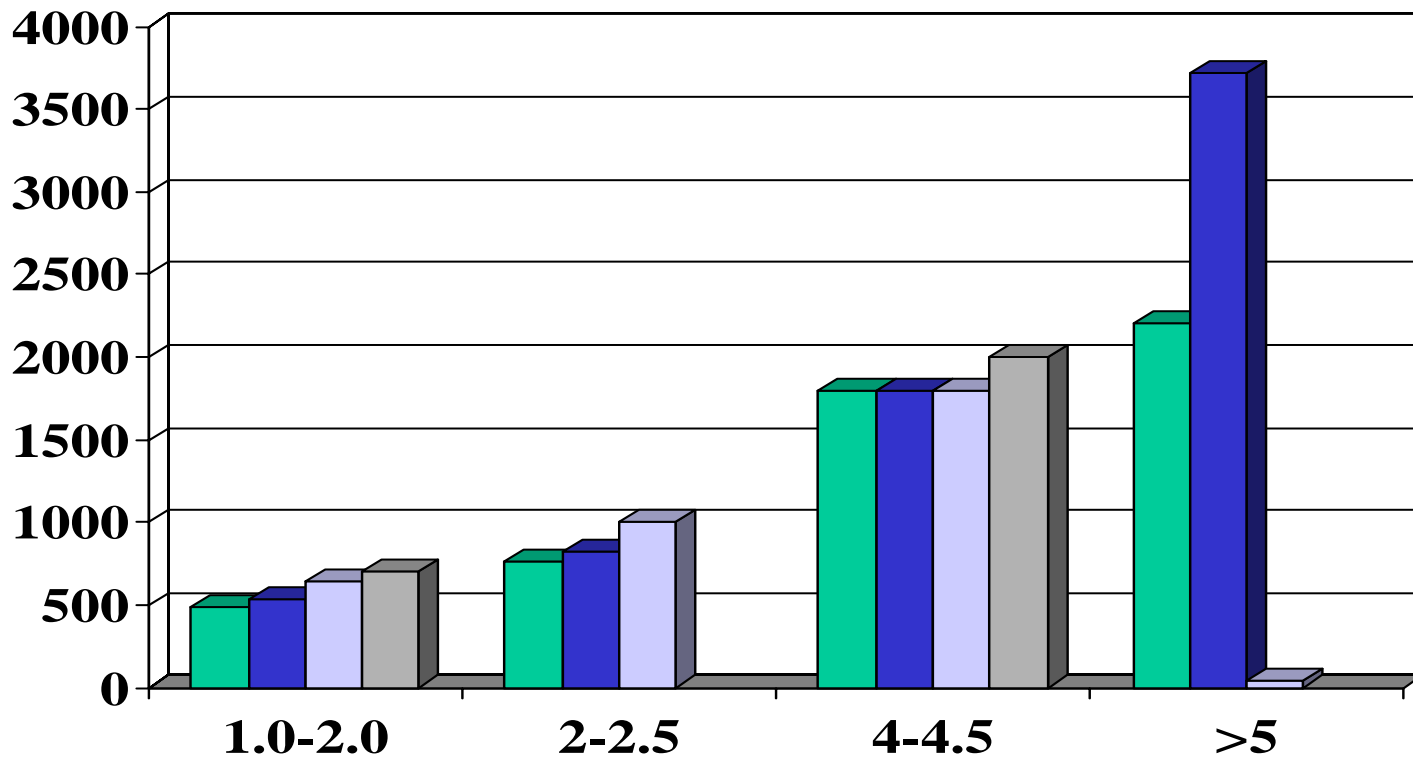
Manpower

- Manpower plan
 - Input to national workforce planning
 - Annual Q-update

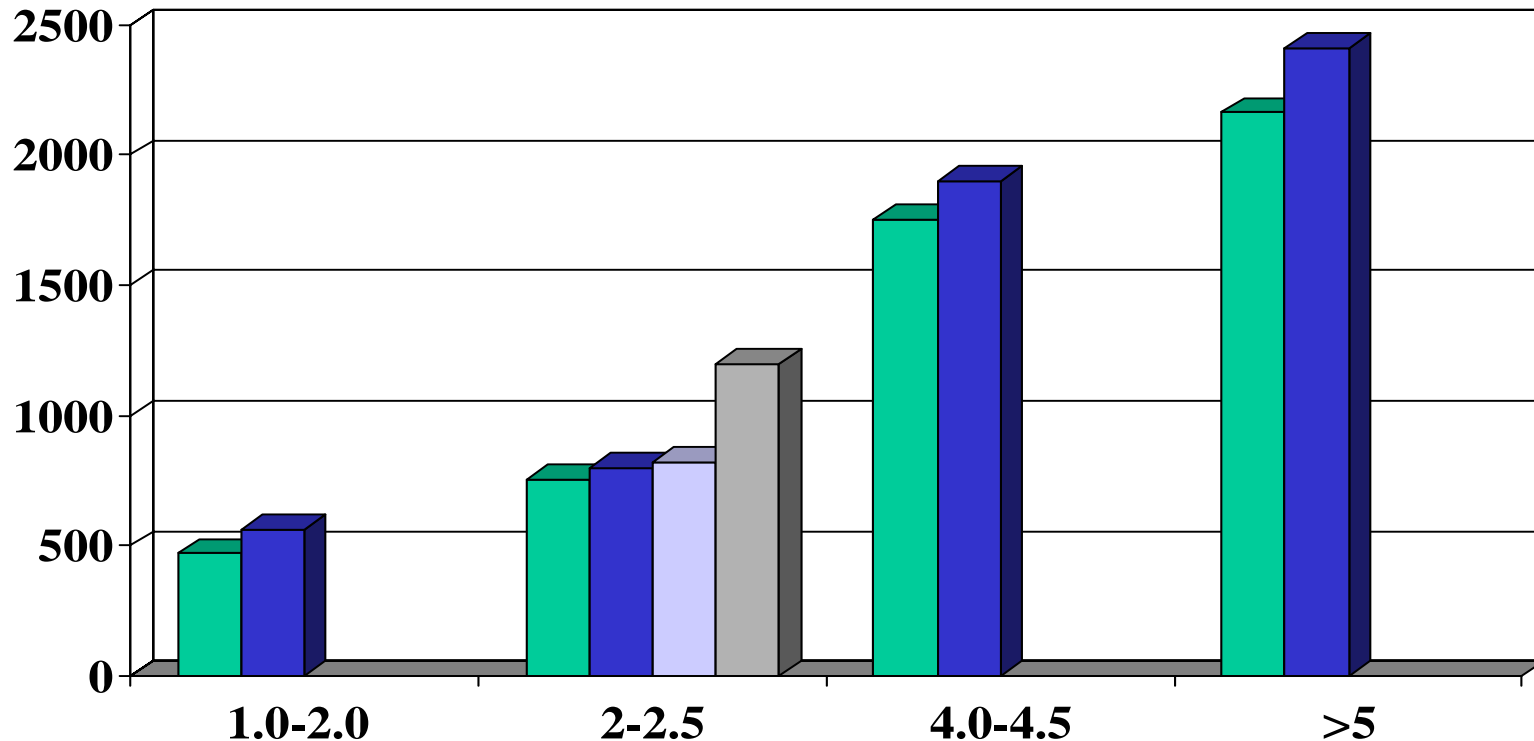
Acyl Carnitine workload (requests p.a.) vs population served(millions)



Organic Acids workload (requests pa) vs population served(millions)



Urine amino acid workload (requests pa) vs population served(millions)



Main laboratory findings

- Vulnerability of some tests
- Compromised turn around times
- No formal out of hours service
- Accommodation increasingly inadequate
- Urgent need for new and replacement equipment (AA, GCMS)
- Need recruitment of 49 new trainee clinical scientist and 46 biomedical scientists over next 5 years
- Continuing support for laboratory network



What Next

- **Dissemination of Guidelines**
- **Assay Directory**
 - Link to disorders
 - Link to molecular
- **Manpower planning & training**
- **Service issues**

Review of National Needs for IMD – whole service

- BIMDG
- Metabolic Biochemistry Network
- Joint Committee in Medical Genetics
 - Adult services
- **National review** with DH support

Main areas of investigation

- Epidemiology
- Outcomes
- Review of laboratory services
- Review of clinical services
- The nursing role
- The specialist dietitian role
- Roles and view of the voluntary sector
- Specialist commissioning



How many new cases per year?

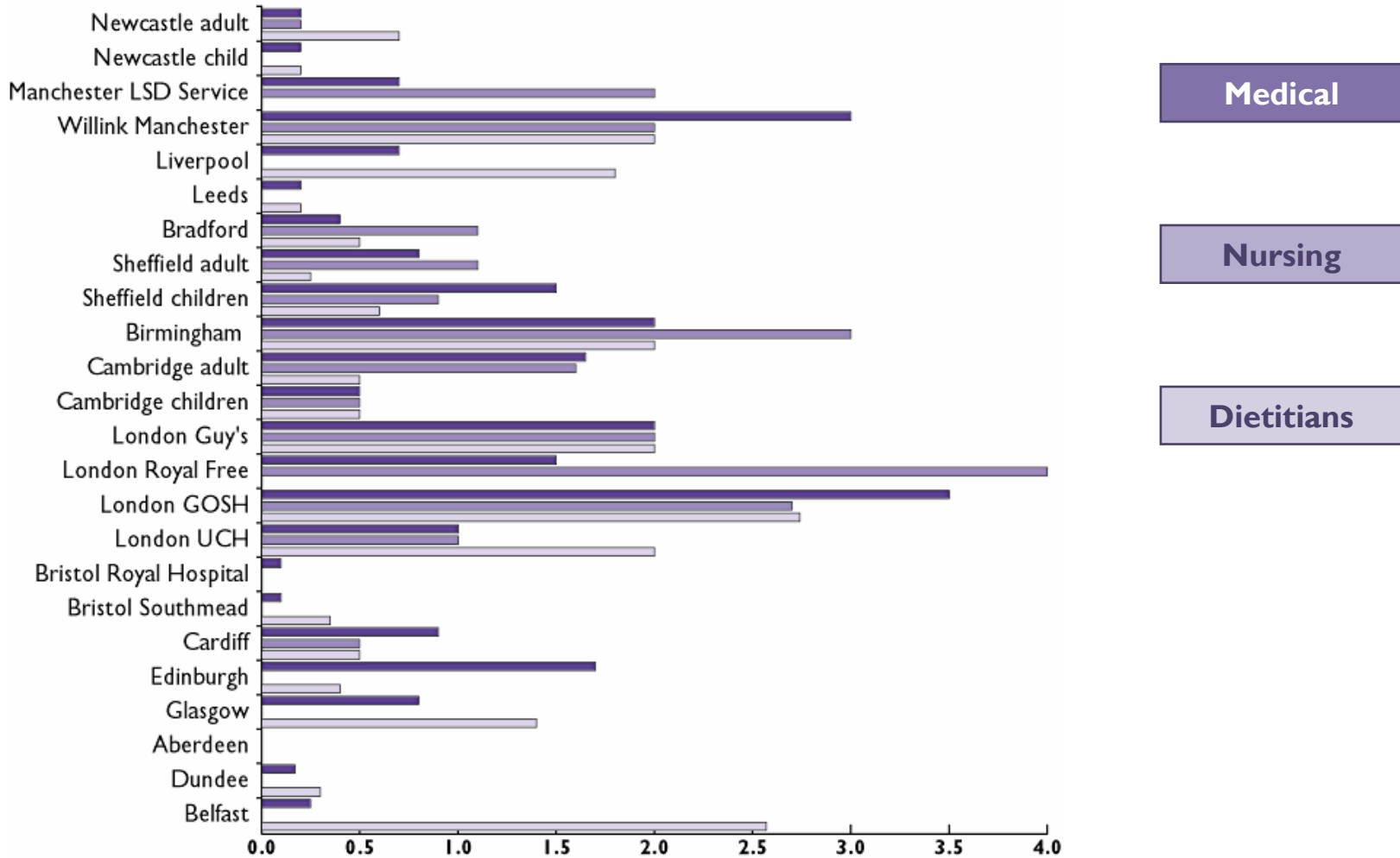
From literature

- About 900 per year

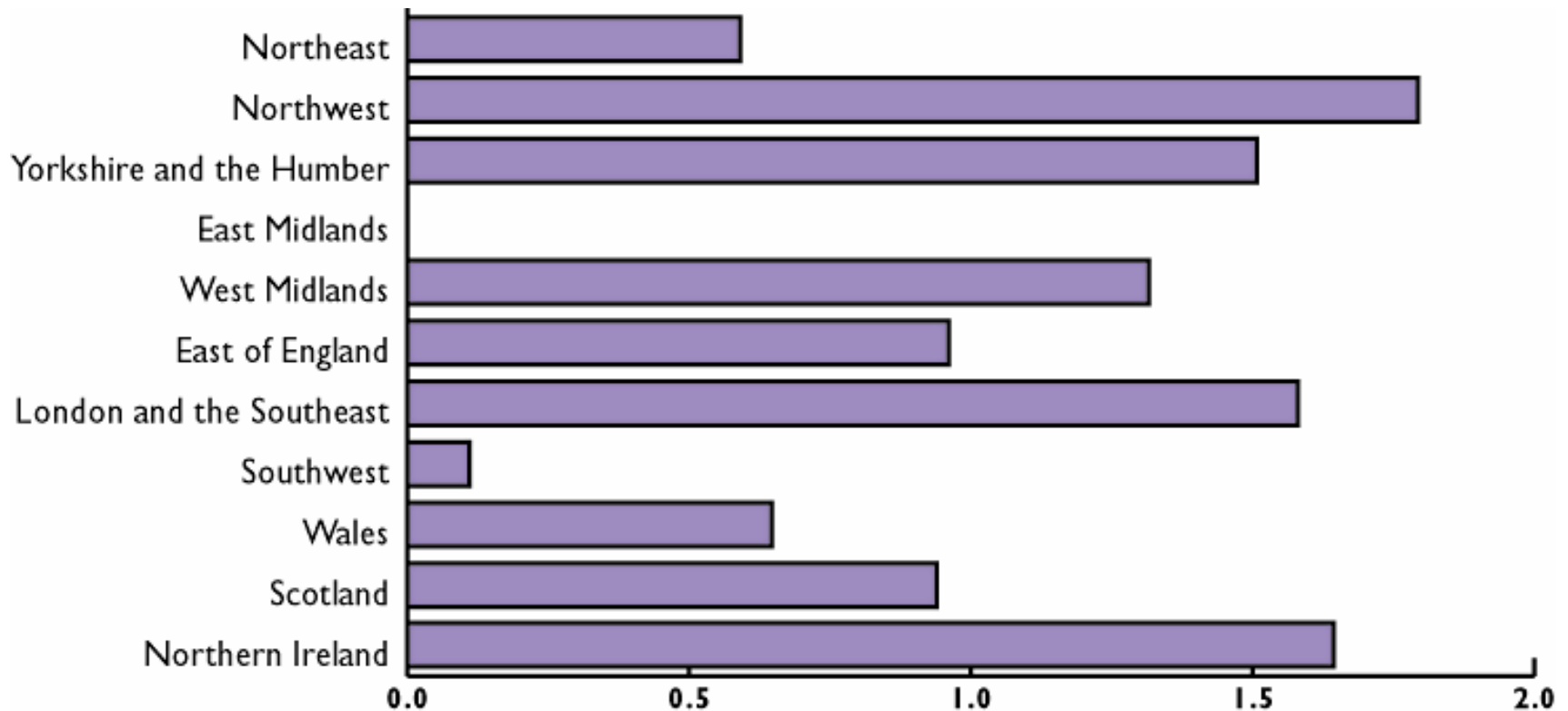
New epidemiological research

- From UK laboratory diagnoses
 - 500-600 per year
- From new diagnoses in West Midlands over a 5 year period
 - About 1 in 800 live births
 - About 800 per year in UK

Clinical teams are very patchy

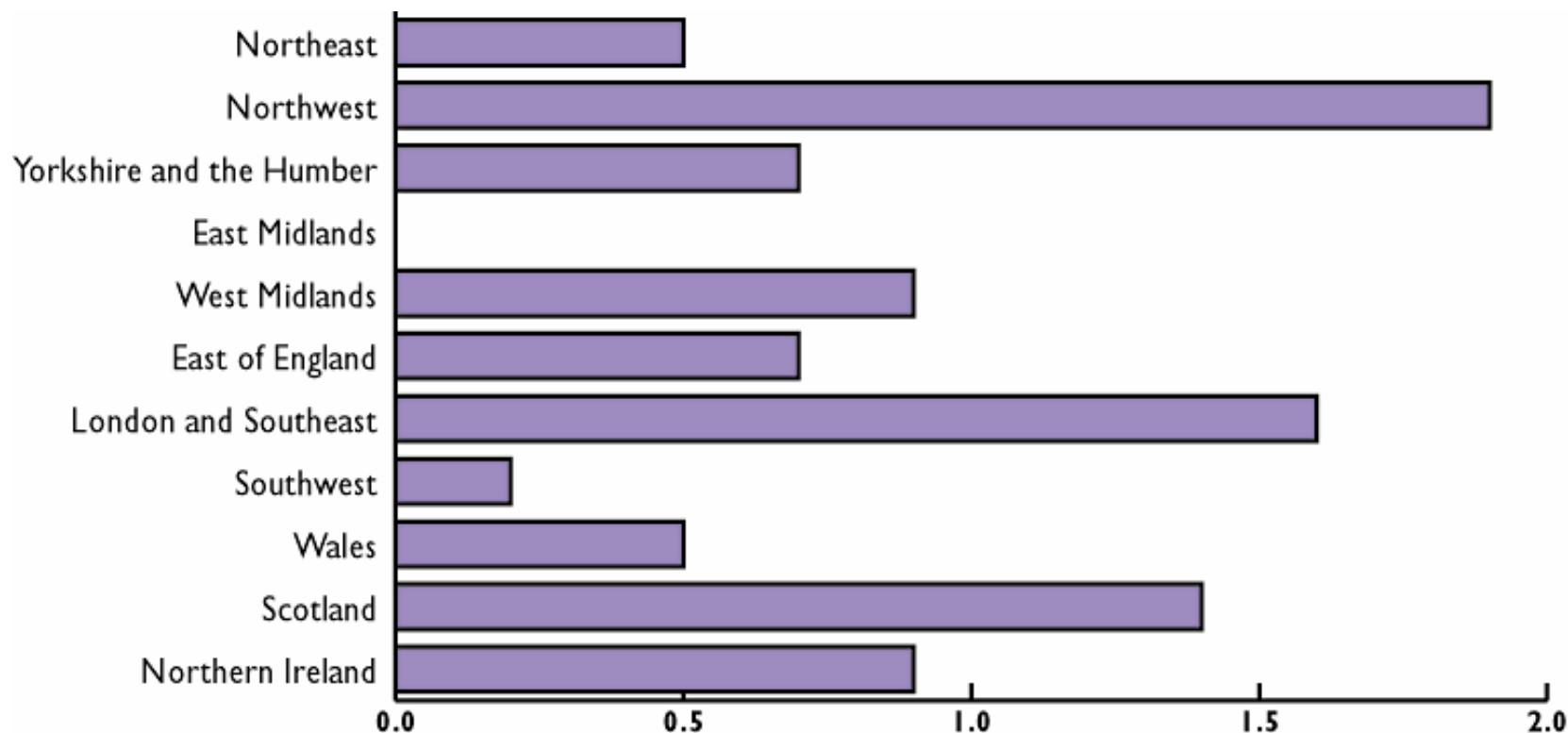


Ten-fold regional variation in total clinical workforce per million population



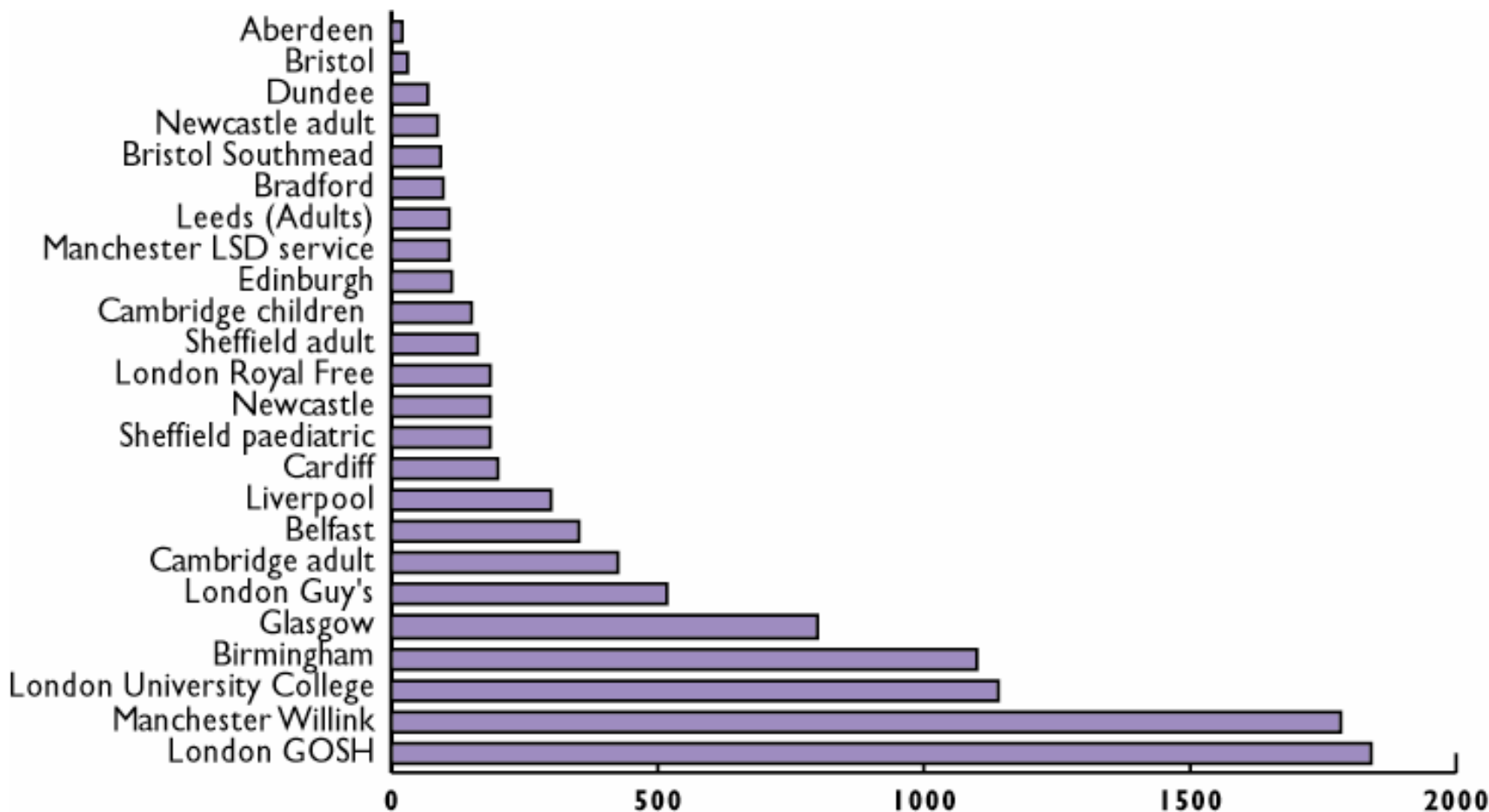
Total clinical workforce (wte) per million population

Comparisons of regional rates of out-patient provision



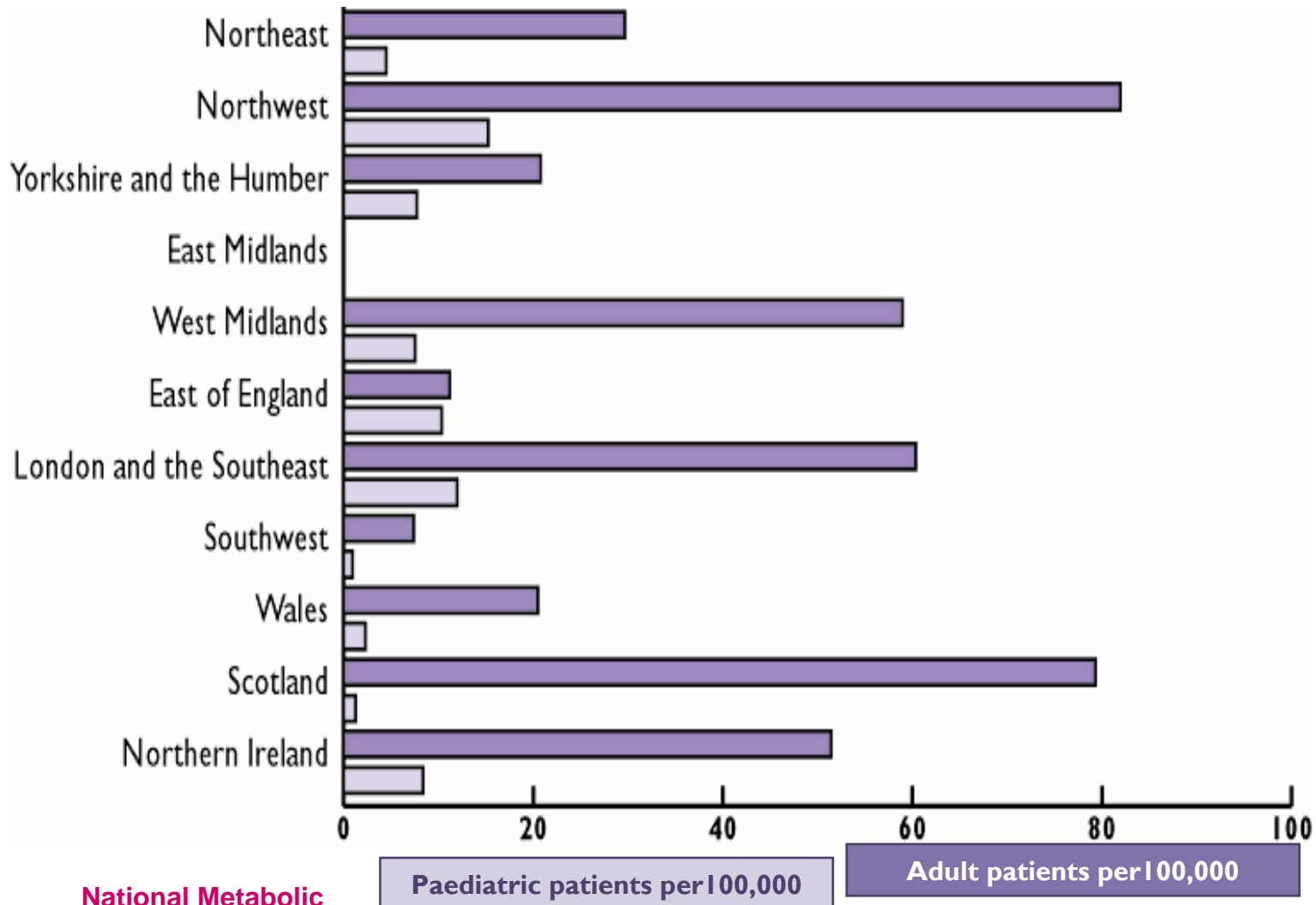
Average weekly out-patient sessions per million population

Critical mass: implications for clinical governance



Number of patients attending the service

10-fold geographic variation in patients per 100,000 population



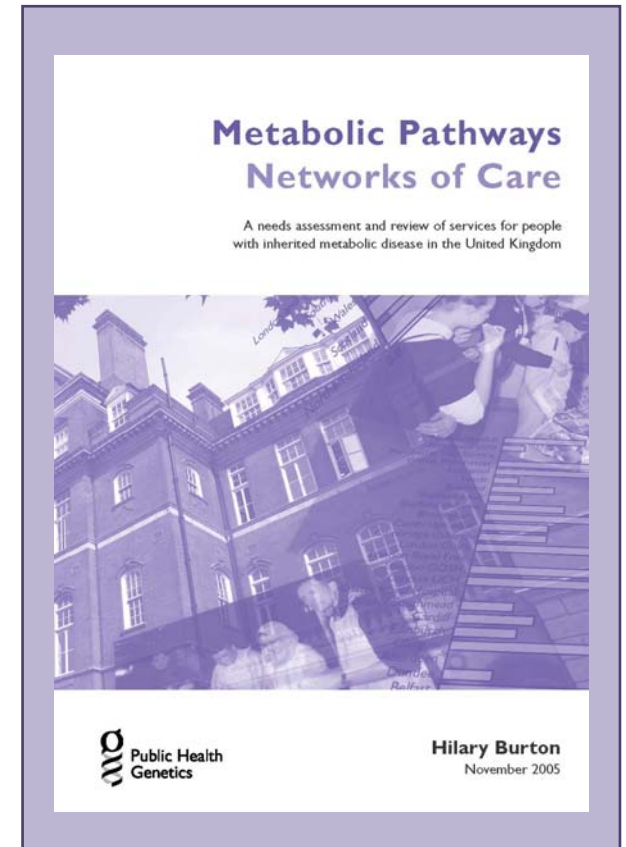
Estimates of shortfall for patients in specialist care

	Children	Adults
Estimated number for UK*	12103	6827
Totals reported	6547	3499
Shortfall	5556	3328

***Based on Northwest rates**

Summary of recommendations

- Strategic advisory group
- Explicit commissioning
- Clinical networks
- Strengthen laboratory and clinical services
- Developing shared care arrangements
- Manpower planning and education
- Close work with voluntary groups



Laboratory recommendations

- Continue and develop as integral part of MDT
- Increase workforce resources
- Capital investment
- Investment in education and training
- Review of accommodation
- Develop database for IMD diagnoses
- Detailed planning for very specialised tests

The End