

Metabolic Biochemistry Network

**Survey of Service
Provision**

Oct – Dec 2003

- Share preliminary data
- What additional feedback do labs want
- Agree need for any further analysis
 - Workforce numbers project
 - ? others
- Ownership of data
- Next Stages

Contributors

- **Stakeholder Labs**
 - circulation 17
 - completed returns 17 (**100%**)
 - completed returns analysed 16*
 - * 1 lab does not provide core tests
- **Associate labs**
 - circulation 3
 - completed returns 2
- **Local Lab**
 - 1
 - * not included as a local service only (population < 500)

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

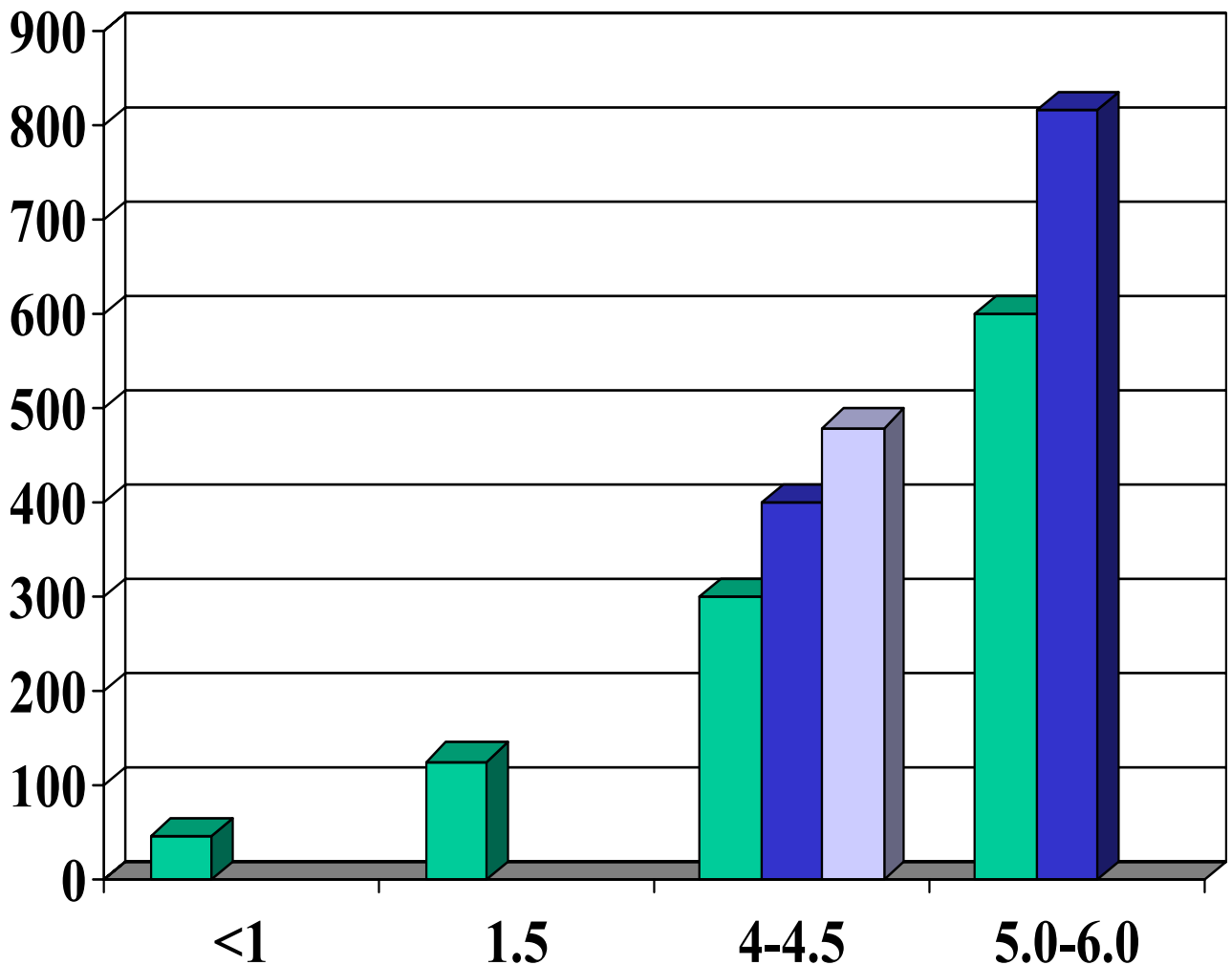
- 15/16 accredited

Core Tests and Workload

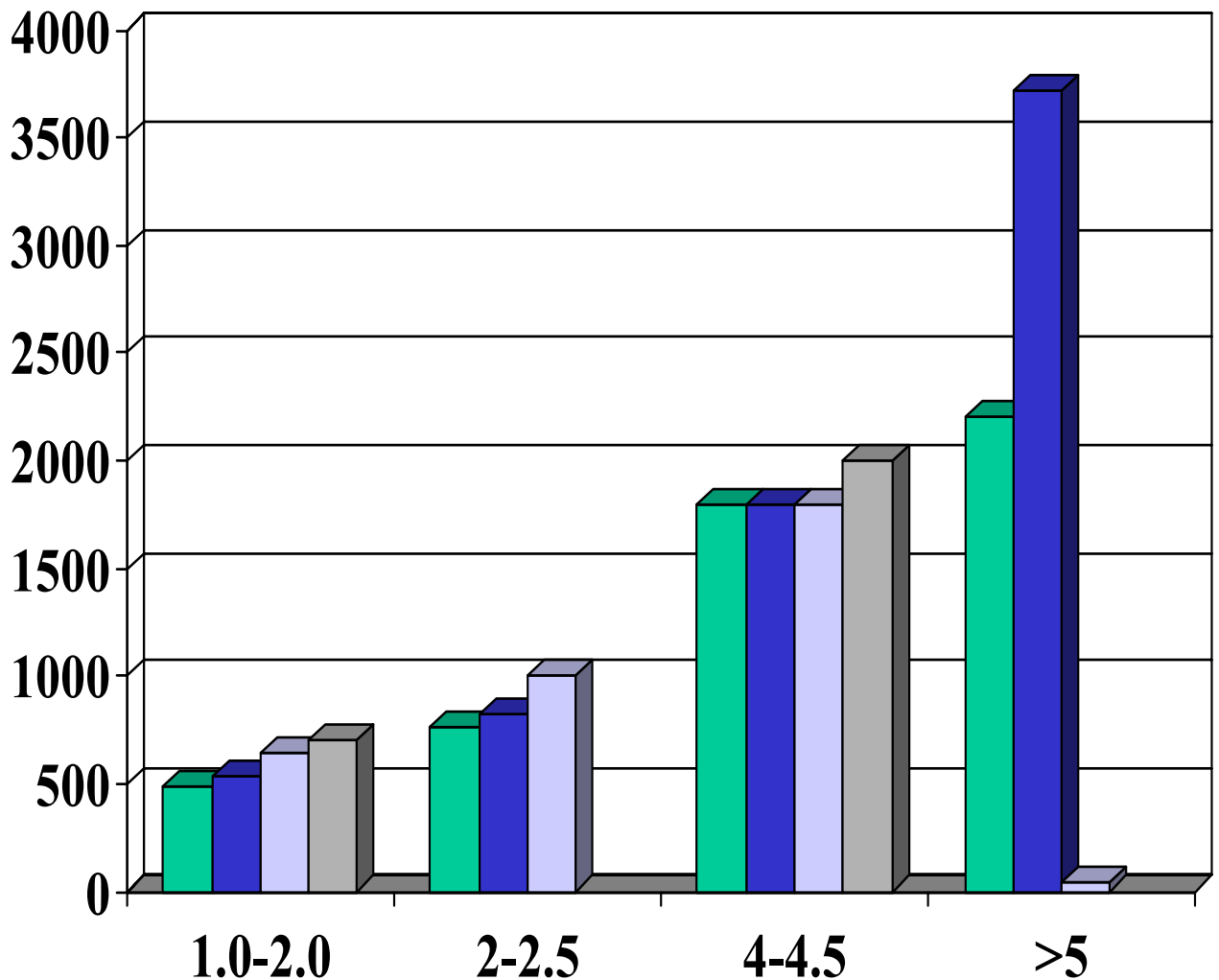
Core tests provided by Stakeholder Labs

Ammonia	15/16	*1 provided by chem path on site
Amino Acids (U)	16/16	
Amino acids(P)	16/16	
Organic Acids	16/16	
Acyl Carnitines	9/16	2 labs planning to provide
FFAs/3HB	13/16	
GAGs	14/16	
Mono/Disac	14/16	

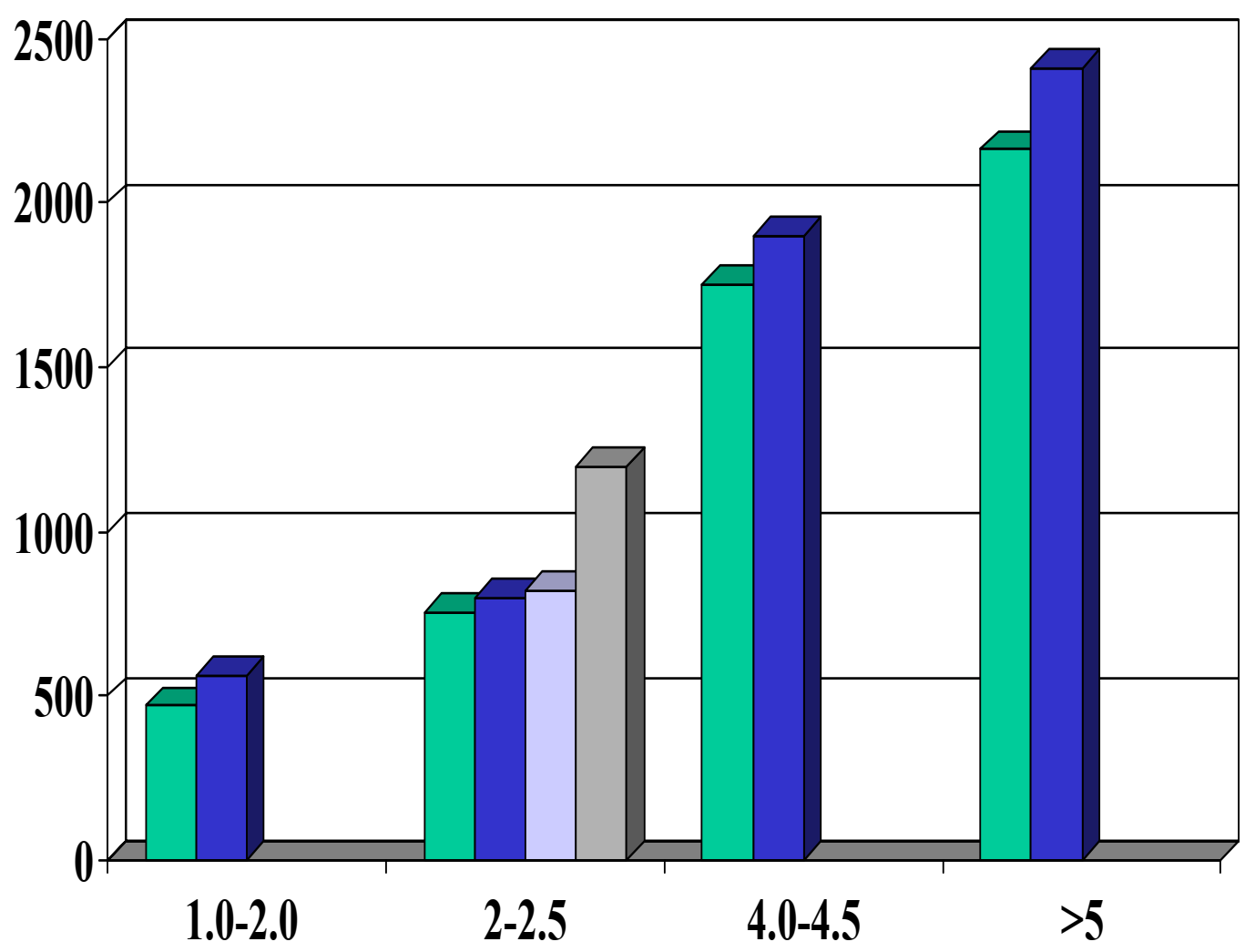
Acyl Carnitine workload (requests p.a.) vs population served(millions)



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



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



Non Core Tests

Intermediate level

Test	No labs	Requests pa
Biotinidase	10	14 - 315
Cystine (leuc)	5	65-300
7 -Dehydrochol	4	41-180
Gal 1 P	7	27-100
Gal1PUT	11	15-276
Oligosacch	6	<48-1100
Orotic acid	6	26-177
VLCFA	7	200-1547

Redundant Tests For Discussion

- Oligosaccharides
- Galactose 1 phosphate
 - Poor methods
 - Evidence of value? (for Gal monitoring)
 - Use LFTS for dietary non compliance
- Thiosulphate
 - Not reliable (false+ves and –ves)
 - Replace with sulphocysteine and urate
- Cystine uptake

Rare & Very rare tests

- Is repertoire appropriate
- Efficiency
- Robustness

Test not easily available/required

- Creatine & Guanidino acetate
- Cobalamin defects/B12 studies/enzymes
- 5 methyl THF defects
- CDG enzymes
- Serine biosynthesis disorders(enzymes)
- Hypophosphatasia
 - ? ALP
 - pyrophosphate
- 2 methyl 3 hydroxy butyryl CoA dehydrogenase
- Sorbitol dehydrogenase
- 3 methyl glutaconyl hydratase
- Succinic semialdehyde dehydrogenase
- Glut 1 transporter
- Biopterin studies
- Respiratory chain defects service/advice

Test not easily available/required

- **Molecular tests**

- CNS disorders
- Lysosomal storage disorders
- Tyr II
- GSD
- GAI
- VLCAD
- SLO- carrier status
- MLD pseudodeficiency

The Way forward?

- Molecular tests
 - ? Molecular sub group
- White cell/tissue enzymes/Co₂ release assays
 - ?sub group

Turnaround Time/Robustness (Routine)

Amino acids	16	11/16 (68%)	Batch size (2) Equipment (2) CS time (2)
Organic acids	16	10/14	Equipment (1) CS time (2)
Acyl Carnitines	9	5/8* * no info	CS(1) Way of working / Need (2)

Turnaround Time/Robustness (Emergency)

Amino acids	16	14/15	Equipment (1)
Organic acids	16	12/14	Equipment (1) CS time (2)
Acyl Carnitines	9	6*/7 * no info	

Summary

Turnaround Times

- Instrument failures /shortage
 - GC/MS (1)
 - AAA (3)
- Staff shortages/availability
 - OAs (3)
 - Acyl carnitines (1)
 - Amino acids (2)
- Batch size too small
 - AAs (2)

Training - Q responses

(16 responses)

- Adequate Capacity
 - 6/16
 - Insufficient trainer time (7)
 - Space (2)

Out of Hours Service

	Analytical (14)	Advisory (13)
Formal service	Ammonia Lactate Organic acids & Amino acids (1)	Formal DB (4)* *with adhoc access to metabolic specialist Consultant (7)* * 1:1, 1:2
Adhoc service	Can be organised by negotiation	Ad hoc access* (2) *metabolic specialist

Accomodation

14/16 responses

- Current
 - Lab 12/14 adequate
 - Office 8/14 adequate
- Future (next 5 years)
 - Lab 6/14 & 1 with plans
 - Office 3/16 & 1 with plans

Equipment

- AAA
 - 1 instrument 14/16
 - 2 instruments 2/16
- MSMS
 - 1 instrument 5/10
 - 2 instruments 5/10
- GCMS
 - 1 instrument 10/15
 - 2 instruments 3 /15
 - 3 instruments 2 /15

Organic Acid workload and GCMS instruments

Number of Instruments	OA Workload pa
1	266 492 540 648 702 820 1000 1800 2000 2209
2	765 1250 1800
3	1800 3714* plus other non IMD assays

Equipment needs next 3 years

- AAA
 - 5 instruments required urgently (30%) –rigor mortis!!
 - 10 required in next 3 years
- TMS
 - 3 /4 required in next 3 years
- GC/MS
 - 9 required in next 10 years

Staff Needs in next 5 years

- **Clinical Scientists**
C - 8
B - 34
- **Biomedical Scientists**
3 - 6
2 - 14

Needs refining

Possible Network Initiatives

- Workload Units 12/16
 - Disease Register 11/16

 - Std Methods/SOPs 6(+3)* /16
- * (CE marking)
-
- Std reports 5/16
 - Common request from 5/16

Summary

- **Core Tests**
 - Workload relates to population
 - Acyl carnitine deficiency for some labs
- **Turnaround times**
 - Routine core tests – 30% compromised by CS time
- **Accommodation for next 5 years**
 - 50% have inadequate lab accommodation
 - 75% have inadequate office accommodation
- **Equipment**
 - Urgent replacement for 30% AAA
 - 3 year replacement for 50% AA & GCMS
- **Training**
 - Inadequate training capacity-trainer time a limiting factor
- **Staffing**
 - Major issue for CS posts
- **Future work**
 - Workload units
 - Disorder register

Issues

- **Do we need a more formal emergency service ?**
 - weekends/bank holidays
 - (national tox advice service)
- **Formalised back up between labs**
 - Routine
 - Emergency
- **Accommodation (mainly office)**
 - ?local solutions
 - ?joint initiatives
- **Introduction of new tests/rationalisation**
 - Network strategy?
- **Manpower/workforce numbers**
 - Refine data/include specialist labs
 - Screening extra needs
 - Succession planning
 - Training of other health professionals