Report of the ERNDIM National Representatives meeting

Basel, Switzerland, May 9, 2008

1. Participants

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UK	Sheffield	Jim Bonham *°	jim.bonham@sch.nhs.uk

* Chairmen during the group sessions
° Speakers

2. Programme and introduction

Programme:

9:00	Introduction	Brian Fowler	20 min
9:20	Organisation of BGT testing in 4 representative countries		
	- UK	Jim Bonham	20 min
	- Estonia	Katrin Õunap	20 min
	- Italy	Ubaldo Caruso	20 min
	- Netherlands	Marius Duran	20 min
10:40	Coffee break		20 min
11:00	Group sessions (groups 1, 2 and 3)		2 hours
13:00	Lunch time		1 hour
14:00	Report of group sessions (each group 10 min)		30 min
14:30	Develop guidelines (new groups A, B, C)		45 min
15:15	Coffee break		15 min
15:30	Final discussion and conclusion		1 hour
16:30	Close of meeting		

Introduction by Brian Fowler

see: http://www.erndim.org/Meeting_Rep/08_may_basel/menu_basel_08.htm

3. Organisation of BGT testing in 4 representative countries

- UK, Jim Bonham
- Estonia, Katrin Õunap
- Italy, Ubaldo Caruso
- Netherlands, Marius Duran

see: http://www.erndim.org /Meeting_Rep/08_may_basel/menu_basel_08.htm

4. Group work / Questionnaire 1: Recommendations to overcome deficits

This questionnaire was first presented during the first meeting in Basel, December 2, 2005 and was updated based on the feedback during the second meeting in Prague, October 2006. See also Interim Report on Biochemical Genetic Testing in Europe: deficits and needs and EQA, pages 12- 13.
Each point was subdivided into 3 questions: a) Importance or relevance today?, b) Progress made?, c) Further measures needed?

1. **Define basic standards / minimum core requirements / test repertoires in relation to size of country**

All groups stated that the recommendations from the previous meetings are still important and relevant today. It was important to define test repertoires in relation to country size. In most countries progress is reported in getting minimum core requirements and in setting-up test repertoires. However, Slovakia and Greece did not report progress but rather decline because of privatisation of laboratory services. This also means a decline in quality of service because of a lack of knowledge in private laboratories. In these countries measures are urgent to re-organise and adequately finance metabolic diagnostic services in academic laboratories. Financing of metabolic services still is a problem in many (eastern) EU countries. Further measures to be taken in general are improved staffing and structure of labs. Additionally, better data quality for xls-tables (BGT in Europe) is needed.

2. **Identification of clusters of countries / sharing where workloads are very low. Exchange visits and/or workshops**

Clustering of countries is still relevant, especially for small countries. Particular labs should specialise in particular tests.

Some progress is made in organising scientific meetings/workshops for sharing experience and knowledge. E.g. the 3 Baltic countries have started the “Baltic Metabolic Group” organising annual meetings.

Further measures are needed to develop flexible arrangements to form clusters where workloads are low and publish exchange visits. The general idea about clustering countries to join diagnostic investigations is very difficult as long as documented guidelines for equipment and minimum service packet are lacking. Economic barriers also impede such developments. Sending samples abroad may have practical difficulties for some countries, especially in the area of payment approval.

3. **Training initiatives for implementing new tests / initiatives for training in reference labs in more developed countries**
4. **National and international reference laboratories for training should be identified**

Combined answers to questions 3 and 4: There is still a need for training in reference (experienced) laboratories. Training initiatives were considered important and also demand more publicity. Training laboratories should be identified, e.g. at the ERNDIM website. Grants have been introduced by ERNDIM.

Although there is little progress made in training inexperienced colleagues, efforts to extend training facilities and financing traineeships should continue. Colleagues should be encouraged to participate in training courses. Furthermore, improved financial measures are needed.

5. **Stimulation of accreditation of laboratories by scoping present status**

We agreed that accreditation was important but that not all countries had ready access to suitable National accreditation bodies.

The directory documenting accreditation status should be a stimulation. Accreditation bodies should be aware of the existence of IEM EQA schemes, and of a candidate lab’s performance in them.

Further measures: Should an IEM laboratory be defined according to a (minimum) service packet? Specification of European wide accreditation terms (norms) for IEM laboratories would be useful. It was suggested that ERNDIM could prepare a master letter for NRs to send to accreditation bodies.

6. Accreditation of the EQA schemes themselves

Accreditation of the EQA schemes remains very important. Slow but progressive process has been made.

The scheme organisers should be encouraged to speed up scheme accreditation (task for ERNDIM).

A physical seat of an ERNDIM office should be defined.

7. A survey of the scope of Metabolic Physician and Biochemist Training

All agreed that training for metabolic biochemists and physicians was important.

Progress has been made by increased dialogue and publicity and available resources (syllabus, training logbook).

The group suggested a meeting with training workshops next year in Europe. A separate extended DPT meeting with a training workshop may take place in 2009 (as there is no SSIEM conference).

One group noted that for training of physicians interested in clinical diagnostics of IEM similar problems/demands exist as for biochemists. And that there was a lack of metabolic physicians most probably because of unfamiliarity with IEM and low priority (rarity of IEM).

8. An Initiative similar to that taking place in the UK to scope the number of patients with certain IMD disorders across Europe

9. Establishment of National registers of diagnosed cases through existing national organisations / through European wide action

Combined answers to questions 8 and 9: The establishment of National and European registers of diagnosed cases of IEMs was considered desirable but difficult to impossible.

One group stated that questions 8 and 9 were difficult as it would not be clear that we were comparing like with like in terms of disease definition or to organise registers.

Improved ICD classification of disorders was needed and that reliable data depends on more specific ICD. Privacy acts often preclude the setting up of such registers. Liaison with parent groups and/or pharmaceutical companies to access their registers was suggested. We were informed that Dr. J. Walter (Manchester) is working on this for IEMs and that ERNDIM and EUGT should support it.

10. Expansion of EQA and improved availability of quality assessment materials including cell banks for biological material

All agreed that improved access to EQA samples would benefit the schemes and that parent groups may have a role to play in providing samples. Progress was made by SKML who provides reference material for certain but not all metabolites. A sample archivist should be appointed to collect specimens of interest. The UK is co-operating with a parents group (CLIMB) to encourage parents to allow provision of such samples. This approach is to be tested with an ethics committee (in Sheffield).

11. Best Practice guidelines for methodology, minimum services and QA

All welcomed best practice guidelines and stated that some progress has been made, e.g. guidelines available from ERNDIM and MetBionet.

Further measures needed are: develop more guidelines and increase publicity. It has previously been suggested that modified S.O.Ps could be used (with suitable legal disclaimer). Guidelines should be useful and not overly verbose. Documents related to methods can be mounted on the ERNDIM website. A practical guidebook edited by Dr. N. Blau was recently published (Blau N, Duran M, Gibson KM eds (2008) *Laboratory Guide to the methods in Biochemical Genetics* Springer-Verlag Berlin Heidelberg). At present, IEM procedures are less standardised than those in general clinical chemistry.

12. The advised recommendations and issues should be a Directive of the European Commission

The groups agreed that this issue was important and that progress has been made by the EC agency for rare disorders. It was noted that recognition of ERNDIM by EU as an appropriate body was desirable and that recent preliminary discussions on EU Rare Diseases initiative may be helpful. But one group stated that an EU directive in this area was unlikely.

5. Group work / Questionnaire 2: Provision of BGT services in European countries

Prior to the meeting a questionnaire was circulated to participants requesting information on the above criteria for subgroups of IEMs. During the meeting the replies for each country were discussed to attempt to reach agreement in a consensus applicable to the EU countries. Discussions led to reevaluation and redefinition of the disorder subgroups as well as the criteria. The chairs summarised the additional input. This was circulated again between all NRs and new comments were integrated into the table.

Criteria:

- Frequency of diseases: high < 1:20.000 / medium 1:20.000 - 1:100.000 / low > 1:100.000
- Presentation: is acute presentation prevailing / important in this category? Acute / chronic / from neonatal screening (fill in: yes or no)
- Necessity of monitoring: frequency: yearly / more than 6 times/year / monthly etc.
- Timely monitoring: 24h-service / turnaround time (TAT) < 6h / different service options
- Equipment and instruments: handling: easy / medium / difficult.
- Price: cheap < 30000 € / expensive > 30000 €
- Expertise level needed: medium / high / very high
- Level of provision: regional = population < 5 million / national = popul. < 30 million / international = popul. > 30 million

Table 1		Tests / Disorders		
Criteria for decision		Amino acids		
		PKU	Urea cycle disorders	other (not organic acidurias)
Frequency of diseases		high	medium	medium
Presentation	Acute	no	yes	varies
	Chronic	no	no	no
	Screening	yes	no	no
Necessity of monitoring	Frequency[1]	6 per year	6 per year	6 per year
	TAT [2]	24 hours	6 hours	6 hours
Equipment, instruments	Handling	easy	difficult	difficult
	Price	cheap	expensive	expensive
Expertise level needed		medium	high	high
Organisation at present		regional	regional	regional
Reason for lack of service		Various concerns were expressed including: lack of trained manpower, lack of equipment, lack of training, lack of political will, lack of funding, may not be justified for small populations		

[1] varies with age and disease; [2] most severe / in crisis

Table 2		Tests / Disorders		
Criteria for decision		Organic acids	Carbohydrate metabolism disorders	
			Metabolite screening [3]	Mutation / enzyme assay [4]
Frequency of diseases		high	medium	medium
Presentation	Acute	yes	yes	yes
	Chronic	yes	yes	yes
	Screening	no	no	no
Necessity of monitoring	Frequency[1]	6 per year	6 per year	n. a.
	TAT [2]	6 hours	24 hours	n. a.
Equipment, instruments	Handling	difficult	easy	difficult
	Price	expensive	cheap	cheap
Expertise level needed		high	high	high
Organisation at present		regional	regional	national
Reason for lack of service		Various concerns were expressed including: lack of trained manpower, lack of equipment, lack of training, lack of political will, lack of funding, may not be justified for small populations		

[1] varies with age and disease; [2] most severe / in crisis;[3] excluding clin. chem. parameters; [4] normally performed once, except for prenatal diagnosis

Table 3		Tests / Disorders			
Criteria for decision		Creatine synthesis disorders metabolites	Respiratory chain disorders Inc resp chain enzymes [4] and mt DNA [4]	Organelle disorders inc MPS, white cell enzymes, glycosylation defects and peroxysomal disorders	
				Metabolite	Enzyme [4]
Frequency of diseases		low	high	medium	medium
Presentation	Acute	no	yes	no	no
	Chronic	yes	yes	yes	yes
	Screening	no	no	no	no
Necessity of monitoring	Frequency[1]	6 per year	1 – 12 per year [5]	1 – 4 per year	n. a.
	TAT [2]	1 week	24 hours	1 week	n. a.
Equipment, instruments	Handling	difficult	difficult	easy – difficult	difficult
	Price	expensive	cheap	cheap - expensive	Cheap, some more expensive
Expertise level needed		high	very high	medium	high – very high
Organisation at present		national	national	regional	national
Reason for lack of service		Various concerns were expressed including: lack of trained manpower, lack of equipment, lack of training, lack of political will, lack of funding, may not be justified for small populations			

[1] varies with age and disease; [2] most severe / in crisis; [4] normally performed once, except for prenatal diagnosis; [5] lactate / amino acids / organic acids

Table 4		Tests / Disorders			
Criteria for decision		Purine / Pyrimidine		Fatty acid oxidation defects and carnitine disorders	
		Metabolite	Enzyme [4]	Metabolite	Enzyme [4]
Frequency of diseases		low	low	high	high
Presentation	Acute	no	no	yes	yes
	Chronic	yes	yes	no	no
	Screening	no	no	no	no
Necessity of monitoring	Frequency[1]	1 – 6 per year (if treated)	n. a.	4 – 6 per year	n. a.
	TAT [2]	1 week	n. a.	6 hours	n. a.
Equipment, instruments	Handling	difficult	difficult	difficult	difficult
	Price	expensive	cheap - expensive	expensive	expensive
Expertise level needed		high	high	high	high
Organisation at present		national	national	regional	national
Reason for lack of service		Various concerns were expressed including: lack of trained manpower, lack of equipment, lack of training, lack of political will, lack of funding, may not be justified for small populations			

[1] varies with age and disease; [2] most severe / in crisis; [4] normally performed once, except for prenatal diagnosis

6. BGT in Europe

BGT in Europe GENERAL ASPECTS

old/new EC	Country	Pop. in Mio	Birth/Y	NBS No. Disorders	NBS No. Centres	No. BGT labs	EQA	Accred.	Training Biochemists	Training Physicians	Sucession	No. Med Centres	IEM of unusual incidence
o	**Austria**	8.2	76000	24	1	7	y	50/50	n	n	n	4	4
o	**Belgium**	10.4	103000	Fr 18/Nl 11	6	10	y	1Belac/10	n	n	variable	11	0
n	**Croatia**	4.4	43000	2	1	1	most	n	n	n	n	4	1
n	**Cyprus**	0.78	9828	2	1	1	y	in prep	n	n	n	1	2
n	**Czech Republic**	10.2	91800	3	4	7	4/7	1/7	n	n	4/7	4	0
o	**Denmark**	5.5	65000	18	1	1	y	n	n	n	y	1	6
n	**Estonia**	1.34	15741	2	2	2	y	some tests	n	n	y	2	1
o	**Finland**	5.2	54080	1	9	5	2/5	test	n	n	n	5	5
o	**France**	61	725900	5	21	28 *	y	n	y	y	n	14 **	0
o	**Germany**	82	672400	12	12	50	most	some	y	y	n	22-32	0
o	**Greece**	10.7	102720	4	1	4	y	n	n	n	n	4	1
n	**Hungary**	10	97000	26	2	6	1	n	n	n	n	8	3
o	**Ireland**	4.24	68000	5 #	1	1	y	conditional	n	n	n	1	5 ##
o	**Italy**	58	493000	3	22	25	y	n.a.	n	n	limited	26	0
n	**Latvia**	2.3	21620	2	1	1	y	n	n	n	n	1	0
n	**Lithuania**	3.6	32040	2	1	1	y	n	n	n	n	1	n.a.
o	**Luxembourg**	0.48	5660	4	1	0	y	in prep	n	n	n	in prep	0
n	**Malta**	0.4	4120	2	2	1	n	n	unstruct.	unstruct.	n	1	1
o	**Netherlands**	16.5	176550	16	5	8	y	6/8	y	y	y	8	n.a.
n	**Poland**	38	376200	2	8	5	not all	n	n	n	n	9	3
o	**Portugal**	10.6	112360	15	1	3	y	n	n	n	n.a.	2	0
n	**Slovakia**	5.4	54424	3	1	3	2/3	n	n	n	n	4	3
n	**Slovenia**	2	18000	2	1	1	1	n	n	n	n	1	1
o	**Spain**	40.4	404000	6	21	6	some	n	n	n	not always	12	n
o	**Sweden**	9	91800	5	1	2	y	y	n	n	limited	2	n.a.
X	**Switzerland**	7.5	72750	7	1	4	y	some	n	n	ad hoc	4	0
o	**UK**	60.8	650560	5	18	16	y	16/16 CPA	y	y	variable	29	n.a.
total		468.94	4637553		146	171						150	

Birth rates from: http://www.infoplease.com/ipa/A0004395.html

Populations from: http://www.infoplease.com/ipa/A0004379.html

France: *only 15 of them are providing most BGT tests, **only 8 of them are accredited by French ministry of health

Ireland: # currently (toxoplasmosis dropped in 2007, but MCADD and GA1 to begin in 2009), ## PKU, galactosaemia, GA1, resp chain defects, MPS 1 & 2.

updates based on NR meeting, May 9, 2008 in Basel

BGT in Europe TESTS AND CASES

old/new EC	country	AA		Org Ac		CHO Metab		FA ox		Resp chain		MPS		Sph-Lip		Pur/Pyr		Perox		Creat Synth		CDGly	
		Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y
o	**Austria**	2700	20	2500	30			1000	15	113	16	287	10	794	10	0	0	857	20	150	2	100	15
o	**Belgium**																						
n	**Croatia**	2000	12	750	3	25	2	400	2	14	1	100	1	70	1	10	0	240	1	0	0	40	0-1
n	**Cyprus**	350	1-2	250	1-2	30	1	50	0	40	2	100	0.2	30	0.3	5	0.1	50	0.1	0	0	20	0
n	**Czech Republic**	6300*	23*	4000	9	2500	15	350	7	700	25	1900	7	1700	14	950	4	700	2	30	0	400	3
o	**Denmark**	2500	10	1500	6	250	1-2	50	10	60	5	1500	2	150	5	250	1-2	200	2	10	0-1	100	1
n	**Estonia**	450	3	450	0.3	500	1	40	0.2	5	1	500	0.5	10	1	2	0	25	0	100	1	50	0
o	**Finland**	1500	5	1000	5	20	0	400		150	50	500	0-1	20	1-2	5	1-2	200	1	5	0-1	10	0-1
o	**France**	28000	110	15000	90	1900	70	4000	40	300	20	4000	50	1000	35	2000	5	3500	20	3200	20	600	6
o	**Germany**	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.
o	**Greece**	6000	8	n.a.	n.a.	30	1.6	n.a.	n.a.	n.a.	n.a.	390	6	2000	9	650	2	500	3.1	n.a.	n.a.	455	2
n	**Hungary**	98000	19	780	7	98000	10	98000	14		1	50	4		2				2	0	0	50	2
o	**Ireland**	2500	16	3000	11	20	0 ^^	2000	3	60	8	63	1	52	1	30	1	60	0	10	1	62	0
o	**Italy**	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.
n	**Latvia**	580	6	80	3	100	2	25	2	15	1	140	2	5	1	10	1	5	1	8	1	28	1
n	**Lithuania**	176	5	n.a.	n.a.	56	1	n.a.	2	n.a.	n.a.	36	6	n.a.	n.a.	n.a.	1	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.
o	**Luxembourg**	150	n.a.	150	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.
n	**Malta**	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.
o	**Netherlands**	n.a.	175	n.a.	100	n.a.	?	n.a.	30	n.a.	n.a.	n.a.	56	n.a.	?	n.a.	?	n.a.	75	n.a.	?	n.a.	28
n	**Poland**	1000	15	3500	25	500	10-15	700	10	100	10	350	15	150	5	50	1	250	5	0	0	800	3
o	**Portugal**	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.
n	**Slovakia**	3100	15	1600	4	340	5	530	4		4		4		5	410	0-1	450	2	n.a.	n.a.	n.a.	n.a.
n	**Slovenia**	600	1-2	600	2-3	20	1	50	0-1	10	0-1	40	0-1	10	0-1	2	0	10	0-1	0	0	20	0-1
o	**Spain**		185		87		92		20		28		71				16		15		5		14
o	**Sweden**	3000	10	3000	8	10	3	150	6	200	20	400	2	300	15	800	2	200	2	n.a.	<1	300	2
x	**Switzerland**																						
o	**UK**	110075	155	24197	89		103		106			108	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.

Cyprus: Test/y for Pur/Pyr, Perox and CDGly refer to samples that are sent abroad
Czech Republic: see questionnaire - test/y: 6300 + 100 000 Phe cases/y 23 + 16 HPA
France: estimates from 10 labs representing approximately 80% of total activity
Hungary: MS scr. started: 1. Oct. 07, screening data included
Ireland: 3 cases of galactosaemia per year from newborn screening
Spain: MPS/Sphingo-Lip taken together, year 2004
UK: Urine Plasma taken together

Comments:
c/Y =cases/year (only your own country)
Test/Y: newborn screening should be excluded
What is a test? What is requested and reported. Can be a profile of aa or a single analyte.
What is a BGT lab? Not only NBS lab. For the purposes of evaluating the present state, all labs should be included whether a single/few or complete test array. For the future (ideal) DPT can be used.
Updates based on NR meeting May 9, 2008 in Basel

7. Development and extension of BGT services in Vilnius, Lithuania

Meeting with Prof. Aleksandras Laucevičius, general director of Vilnius University Hospital Santariškių Klinikos, March 26, 2008, 09.00 am.

Present: Prof. Aleksandras Laucevičius, Prof. Vaidutis Kučinskas, Dr. Jurgita Songailienė, Dr. Leo Spaapen.

Items discussed:

1. Development and extension of the Laboratory for Neonatal Screening and Diagnostics of Inherited Metabolic Disorders (laboratory NSDIEM)
2. Housing of the laboratory in the (near) future
3. Training of dr. Jurgita Songailienė in all aspects of laboratory diagnostics and treatment monitoring with regards to Inherited Metabolic Disorders
4. Urgent need to recruit clinicians (neuropaediatricians, paediatricians, neurologists, internists, other specialists) for specialization in clinical diagnostics of IMD
5. Financial cover of diagnostic testing and treatment (health insurance?)
6. Quality assurance in the mentioned laboratory; participation in external quality control schemes (ERNDIM)

Conclusions and appointments:

1. Present status:
    - Reliable thin-layer chromatographic (TLC) and electrophoresis methods are in use for diagnostics of lysosomal storage diseases.
    - Significant progress in set-up and development of gas-chromatographic mass spectrometric analysis (GCMS) of metabolites of inborn aminoacidopathies and mitochondriopathies is made.
    - GCMS validation procedures are in preparation (awaiting delivery of standards); expected time for validation: at least one year for one full-time technician. Qualitative interpretation of organic acids for diagnostics: soon available.
    - Amino acid analysis which is only performed with TLC (urines) and HPLC (plasma) awaits validation but will be only partially available in reliable quantitative numbers. An amino acid analyzer is indispensable.

    Prof. Aleksandras Laucevičius, Prof. Vaidutis Kučinskas agreed to continue with the promising development.
2. In the planned new building the Laboratory NSDIEM will have new accommodation at its disposal.
3. Prof. Aleksandras Laucevičius and Prof. Vaidutis Kučinskas agreed to financially support further training of dr. Jurgita Songailiene.
4. With the heads of the concerned departments, Prof. Aleksandras Laucevičius will discuss the implication of paediatricians, neuropaediatricians, neurologists, internists and other specialists

to participate in the clinical diagnostics and treatment of Inherited Metabolic Diseases. Possibilities for special training of clinicians will be made available.

5. For the time being financial cover of Biochemical Genetics investigations will stay unchanged.
6. Long-lasting validation procedures are started and participation in the external ERNDIM quality control schemes are well appreciated by Prof. Aleksandras Laucevičius and Prof. Vaidutis Kučinskas and will be supported financially.

Final conclusion:
The development of the Laboratory for Neonatal Screening and Diagnostics of Inherited Metabolic Disorders is progressing and can count on the broad support of the Vilnius University Hospital Santariškių Klinikos towards becoming a fully certified professional diagnostic unit.

8. Conclusions

The organisation of biochemical genetic services throughout Europe is very diverse, mainly for historical reasons. The overall incidence of inherited metabolic diseases in Europe is approximately 1:1000 births.
The provision of BGT services was discussed and 2 contradictory approaches arose: local and small versus centralised and large. Which services are available for a given population may need to be tailored to the incidence of the various disorders in that population. It was noted that a specialised biochemical genetic lab with a relatively small workload may be a better performer than a large general clinical chemistry lab performing the same number of metabolic tests.
With regard to monitoring and equipment there was discussion about services from the Metabolic Laboratory (e.g. amino acid analysis) and those from the Clinical Chemistry department (e.g. determination of blood ammonia, lactate, etc.). People who are dealing with the selective screening in an IEM laboratory cannot also provide routine clinical chemistry analyses.
For East-European (small) countries documented guidelines for establishment of laboratories with a well defined minimum service profile and minimal equipment (dependent on the number of inhabitants) should be made available by ERNDIM/EUGT. Also, additional official documents with guidelines are urgently needed to obtain governmental permission and financial support to send patient's samples to specialised metabolic laboratories abroad for assays that are not included in the minimum service profile/package.