ERNDIM DPT Center Prague

Institute of Inherited Metabolic Diseases
General Faculty Hospital
and
Charles University 1st Faculty of Medicine
Ke Karlovu 2, 128 08 Prague 2, Czech Republic
phone: ++420/224 967 694, 224 967 679
fax: ++420/224 967 081 or 224 967 119

Proficiency Testing Centre Prague
Annual Report 2006

1. Introduction

In 2006 proficiency testing in our centre was running as a regular ERNDIM scheme.

2. Geographical distribution of participants

Nineteen laboratories from 16 countries have participated in our Diagnostic Proficiency Testing scheme in 2006, for details see the below table:

Country	**Number of participants**
Austria	2
Croatia	1
Cyprus	1
Czech Republic	1
Denmark	1
Finland	1
France	1
Germany	2
Greece	1
Kingdom of Saudi Arabia	1
Latvia	1
Malaysia	1
Poland	1
Slovakia	2
Switzerland	1
Turkey	1
in total	**19**

3. Logistics of the scheme

- ✓ Two surveys: 2006/1 – samples A, B and C
  2006/2 – samples D, E and F
- ✓ Origin of samples: Four urines were obtained from patients with known diagnoses and one sample was obtained from a patient without any known IEM (samples were provided by the DPTC participants and by the organizers) and a common sample from DPT centre Amsterdam/Nijmegen was distributed in all five DPT schemes; all samples have been reanalyzed in our lab after heat-treatment, diagnostically relevant metabolites were detected in all six samples after 3-day incubation at RT.

- ✓ The organizers acknowledge Drs. Marita Virtanen, Darina Behulova and Wanda Gradowska for providing samples for 2006 surveys.
- ✓ Six heat-treated urines together with results protocols were shipped at once to the participants at ambient temperature using the courier service Pegasus Express. Sixteen parcels were delivered within 4 days but three parcels shipped to more distant countries were delivered within 7 – 8 days and replacement was sent by FedEx (delivery within 2 and 3 days). The deadline for the results submission was extended for these three participants. Next year Pegasus Express will not be used for transportation of samples.
- ✓ The following protocol for heat inactivation is being used: 1. Add thiomersal 100 mg/l of urine; 2. Heat urine to 56°C for one hour in water bath. Make sure that this temperature is achieved in the entire urine sample, not only in the water bath. The urinary samples have to be frozen until shipment.
- ✓ Tests required in 2006: amino acids, organic acids, mucopolysaccharides, oligosaccharides and purines/pyrimidines

4. Schedule of the scheme in 2006

Sample distribution	March 20
Start of analysis of Survey 2006/1	March 27
Survey 2006/1 – results submission	April 17 May 2 (extended deadline)
Survey 2006/1 – report	May 31
Start of analysis of Survey 2006/2	May 29
Survey 2006/2 – results submission	June 19
Survey 2006/2 – report	August 21
Annual meeting of participants	October 5
Annual report 2006	December 12

5. The receipt of samples and results

Date of receipt of samples (samples sent on March 20, 2006)

Date of receipt (reported by participants)	Number of participants	Date of receipt (reported by courier service)	Number of participants
1 day	1	2 days	11
2 days	5	3 days	3
3 days	3	4 days	2
4 days	1	7 days*	1
8 days	1	8 days*	2
21 days	1	-	-
not indicated	7	-	-

* Replacement samples were sent by another courier service to 3 participants.

Deadlines of the results submission

	2006/1	2006/2
in time	13	15
1 day delay	1	-
2 days delay	3	1
7 days delay	-	1
11 days delay	-	1
19 days delay (equipment malfunction)	1	-
no answer	1	1

6. Scoring of results

Three criteria are being evaluated: analytical performance, interpretative proficiency and recommendations for further investigations. Due to the large variability in reporting results in various countries recommendations to treatment are not evaluated in proficiency testing, however, they are still reported and summarized by the scheme organizers.

A	Analytical performance	Correct results of the appropriate tests	2
		Partially correct or non-standard methods	1
		Unsatisfactory or misleading	0
I	Interpretative proficiency	Good (diagnosis was established)	2
		Helpful but incomplete	1
		Misleading/wrong diagnosis	0
R	Recommendations	Helpful	1
		Unsatisfactory or misleading	0

The total score was calculated as a sum of these three criteria. The maximum that can be achieved is 5 points per sample, i.e. 15 points per survey and 30 points per year.

7. Score of participants for individual samples

Survey 2006/1

Lab no	Sample A Aspartylglucosaminuria				Sample B Ketosis				Sample C Tyrosinemia type I			
	A	**I**	**R**	**T**	**A**	**I**	**R**	**T**	**A**	**I**	**R**	**T**
1	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
2	2	2	1	**5**	2	2	1	**5**	1	2	1	**4**
3	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
4	2	2	1	**5**	2	1	1	**4**	2	2	1	**5**
5	0	0	0	**0**	2	2	1	**5**	2	2	1	**5**
6	0	0	1	**1**	2	0	0	**2**	2	2	1	**5**
7	2	2	1	**5**	2	0	0	**2**	2	2	1	**5**
8	0	0	0	**0**	2	0	0	**2**	2	2	1	**5**
9	2	2	1	**5**	2	2	0	**4**	2	2	1	**5**
10	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
11	0	0	0	**0**	2	0	0	**2**	2	2	1	**5**
12	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
13	1	0	1	**2**	2	2	1	**5**	2	2	1	**5**
14	0	0	0	**0**	2	0	0	**2**	2	2	1	**5**
15	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
16	2	2	1	**5**	2	2	0	**4**	2	2	1	**5**
17	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
18	0	0	0	**0**	2	2	1	**5**	1	2	1	**4**
19	0	0	0	**0**	0	0	0	**0**	0	0	0	**0**

Survey 2006/2

Lab no	Sample D Molybdenum cofactor def.				Sample E D-2-Hydroxyglutaric ac.				Sample F Hypophosphatasia			
	A	**I**	**R**	**T**	**A**	**I**	**R**	**T**	**A**	**I**	**R**	**T**
1	1	1	0	**2**	2	2	1	**5**	0	0	0	**0**
2	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
3	2	2	1	**5**	2	1	1	**4**	2	2	1	**5**
4	2	2	1	**5**	2	1	1	**4**	0	0	0	**0**
5	0	0	0	**0**	2	2	1	**5**	2	2	1	**5**
6	0	0	0	**0**	2	2	1	**5**	2	2	1	**5**
7	0	0	0	**0**	2	2	1	**5**	0	2	1	**3**
8	1	2	1	**4**	2	2	1	**5**	2	2	1	**5**
9	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
10	1	0	0	**1**	2	2	1	**5**	2	2	1	**5**
11	0	0	0	**0**	2	2	1	**5**	0	0	0	**0**
12	0	0	0	**0**	2	2	1	**5**	2	2	1	**5**
13	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
14	0	0	0	**0**	2	2	0	**4**	0	0	0	**0**
15	2	2	1	**5**	2	2	1	**5**	0	2	1	**3**
16	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
17	1	2	1	**4**	2	2	1	**5**	2	2	1	**5**
18	0	0	0	**0**	0	0	0	**0**	2	2	1	**5**
19	0	0	0	**0**	0	0	0	**0**	0	0	0	**0**

A – Analytical score, I – Interpretative score, R – Recommendations, T – Total score

8. Total score of participants for individual surveys and their performance in 2006

Lab no	Survey 2006/1 [points]	Survey 2006/2 [points]	Total point 2006
1	15	7	**22**
2	14	15	**29**
3	15	14	**29**
4	14	9	**23**
5	10	10	**20**
6	8	10	**18**
7	12	8	**20**
8	7	14	**21**
9	14	15	**29**
10	15	11	**26**
11	7	5	**12**
12	15	10	**25**
13	12	15	**27**
14	7	4	**11**
15	15	13	**28**
16	14	15	**29**
17	15	14	**29**
18	9	5	**14**
19	0	0	**0**

9. Score summary in 2006

Sample	Diagnosis	Analytical [%]	Interpretative [%]	Recommen-dations [%]	Total [%]
A	***Aspartylglucosaminuria***	64	61	72	64
B	***Ketosis***	100	69	61	80
C	***Tyrosinemia type I***	94	100	100	98
D	***Molybdenum cofactor deficiency***	50	53	50	51
E	***D-2-Hydroxyglutaric aciduria***	94	89	89	91
F	***Hypophosphatasia***	67	78	78	73

"Easy" and "difficult" urinary samples were included in the surveys. The analytical and interpretative performance was very good for tyrosinemia type I and 2-hydroxyglutaric aciduria while it was poor for aspartylglucosaminuria and molybdenum cofactor deficiency. The performance for the latter two diseases demonstrates a continuous problem with diagnosing lysosomal storage disorders and disturbances of purines and pyrimidines metabolism.

10. Good performers

The participants who obtained 15 points and more (>50%) within the calendar year are assumed to be good performers. One participant did not return any results, of the remaining 18 labs fifteen satisfied criteria for good performance in 2006.

11. Annual meeting of the participants

The annual meeting of participants of the Proficiency Testing Centre Prague took place during ERNDIM/Eurogenetest meeting in Prague on 5th October 2006, twelve laboratories were represented. The following items were discussed during the annual meeting of our DPT centre (discussion minuted thanks to Dr. Jeannette Klein):

1. Information: - ERNDIM will provide quality control material
   - will be available 2007
   - not to be used as calibrators!

2. Information: - budget from ERNDIM for
   - training of biochemists (up to 6 grants per year)
   - funds for labs not being able to pay for QA scheme

3. Idea of biobanking of interesting samples
   - What is interesting?

4. Information: - Web submission of DTP results in preparation

5. Discussion of results of samples A-F
   - consensus on scoring
   - three poor performing labs will get a helping letter

6. Consensus on sample difficulties (one difficult sample per survey)
   However: - Availability of samples is problematic
   - How to assure comparability between centres?

7. Discussion: - Why don't labs perform thiosulfate test routinely?
- Experiences with urease treatment or chloroformate derivatization?
- Idea of comparative methodological study (initiated from Prague)

12. Changes planned in 2007

- ✓ Submission and evaluation of results and reporting via web: the system is now being developed by B. Fowler, P. Litynski and V. Kozich; testing of this system will be possible by help of participants from our centre – participants will be notified in due course.

- ✓ Evza Pospisilova will step down by December 31, 2006 and Petr Chrastina M.Sc. will take over as a scheme organizer on January 1, 2007 (tel.: ++420/224967647, e-mail: petr.chrastina@lf1.cuni.cz)

13. Tentative schedule of DPT scheme and fee in 2007

Sample distribution	March 12, Monday
Start of analysis of Survey 2007/1	March 19, Monday
Survey 2007/1 – results submission	April 10, Tuesday
Survey 2007/1 – report	May 9, Wednesday
Start of analysis of Survey 2007/2	May 28, Monday
Survey 2007/2 – results submission	June 18, Monday
Survey 2007/2 – report	August 6, Monday
Annual meeting of participants	September 4, Tuesday
Annual report 2007	December

Next annual meeting of participants will take place on September 4th during the 43rd SSIEM annual symposium in Hamburg.

The Executive Board of ERNDIM determined the fee for 2007 in the amount of 284 €.

14. Certificate of participation in Proficiency Testing for 2006

The certificate of participation will be provided by the ERNDIM to all participants, who returned the results of both surveys.

Prague, December 12, 2006

Viktor Kožich, MD, PhD
Scientific Advisor to the Scheme
vkozich@lf1.cuni.cz

Evženie Pospíšilová, M.Sc.
Scheme Organizer
eposp@lf1.cuni.cz