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Universitätsklinik für Kinder- und Jugendmedizin

ERNDIM QA Scheme for qualitative urinary organic acid analysis

Annual Report 2006

Participation

The geographical distributions of the active participants in 2006 are shown in Table 1. Sheffield and Heidelberg participate in each other´s scheme and the two centres work closely together under the auspices of the ERNDIM Scientific Advisory Committee.

Table 1: Geographical distribution of participants

Country	Number of laboratories	Country	Number of laboratories
Austria	3	Philippines	1
Belgium	1	Poland	1
Canada	5	Saudi Arabia	2
Croatia	1	Slovakia	1
Cypres	1	Slovenia	1
Czech Republic	2	Spain	2
Denmark	1	Sweden	2
Germany	10	Switzerland	3
Hungary	1	The Netherlands	8
Italy	10	Turkey	1
Latria	1	United Kingdom	1
Norway	1	USA	10

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Samples and results

Three sets of three samples (total 9; sample number 142 --150) were distributed to 71 laboratories.

Four participants did not answer to any of the three circulations.

Table 2: Receipt of results

Circulation	Number of returns	Late returns
1. circulation	64 (90%)	9 (14%)
2. circulation	64 (90%)	5 (8%)
3. circulation	65 (92%)	4 (6%)

Shipment of the samples

As the years before we sent out the samples for all three circulations together. This is only for organisation reasons and to keep the costs for participating in this scheme as low as possible. Some laboratories reported all three circulations together. **The idea of the scheme is to measure the samples evenly spread over the year and report the results near the closing date!**

Table 3: Distribution of scores for individual samples (laboratories making returns)

		-2	0	1	2
Sample 142	Isovaleric aciduria	0	0	0	64
Sample 143	Normal pattern	4	0	1	59
Sample 144	classical phenylketonuria	0	0	0	64
Sample 145	Alkaptonuria	1	0	0	63
Sample 146	Canavan disease	2	0	0	62
Sample 147	Normal pattern	0	0	0	61
Sample 148	Normal pattern	2	0	1	62
Sample 149	aromatic L-amino acid decarboxylase (AADC deficiency under L-DOPA therapy)	5	24	8	28
Sample 150	Normal pattern	7	4	9	45

Scoring scheme

Individual returns for each sample were scored on the scale

2	Correct/satisfactory
1	helpful but incomplete
0	unhelpful
-2	misleading

All active laboratories gave the correct diagnoses for most pathological samples. Also the control urines were mostly considered as normal. Only one control showed increased levels of lactic acid and some participants diagnosed lactic aciduria or suspicion of mitochondrial disorders (3. circulation). The increase of lactic acid was most likely caused by bacterial contamination.

All participants identified **isovaleric aciduria** (sample 142) and **"classical" phenylketonuria** (sample 144) correctly. Informations like the results of BH_4 loading tests and determinations of dihydropteridine reductase activity would be required for further diagnostic differentiation of PKU.

In case of **alkaptonuria** the correct diagnosis was given by nearly all (98%) participants. Analytical detection of homogentisic acid was no problem for all of the participating laboratories with the exception of one. Similarly, and due to an elevation of *N-acetylaspartic acid* **Canavan disease** was correctly diagnosed by most (97%) participants.

Obviously most problems were caused by sample 149 in the third circulation. Although the relevant metabolites *homovanillic acid*, *vanillylmandelic acid* or *vanillactic acid* were identified by nearly all laboratories interpretation of the analytical findings varied. The correct diagnosis of **aromatic L-amino acid decarboxylase (AADC) deficiency** or any neurotransmitter defect were given only by part of the participants. Neuroblastoma was often diagnosed. Interpretation was greatly complicated by drug metabolites which are present in this urine sample because the urine was collected during a period of L-DOPA medication.

Comments on performance

The participants cumulative scores are shown in diagram 1 and in table 4. Cumulative scores are the scores for the whole year 2006. This year sixteen participants (23%) got full marks!
The poor performance of some laboratories scoring less than 10 is mainly due to missing returns.
Four laboratories returned no results at all, six laboratories send only results for two of the third circulations.
One participant returned only one circulation.

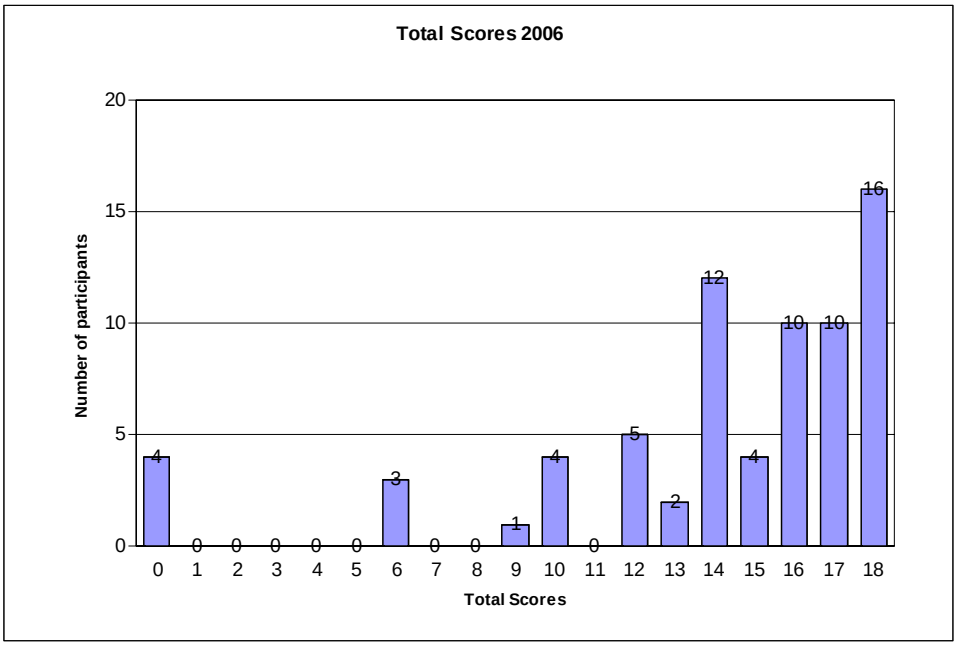


Diagram 1: Total scores 2006

Table 4: total scores 2006

Cumulative scores	Number of laboratories	Cumulative scores	Number of laboratories
18	16	**8**	0
17	10	**7**	0
16	10	**6**	3
15	4	**5**	0
14	12	**4**	0
13	2	**3**	0
12	5	**2**	0
11	0	**1**	0
10	4	**0**	4
9	1		

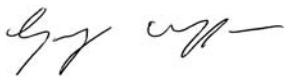
Your total score 2006

Your total score for 2006 was:

General comments

Special thank for the laboratories that supported us last year with samples. This is critical for the success of the program and will keep the scheme interesting. **It is most appreciated that you will continue to support us with urine from patients. Please send us at least 250 ml urine of any interesting patients you may have. We will cover the costs.**

Yours sincerely,



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