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ERNDIM QA Scheme for qualitative urinary organic acid analysis

Annual Report 2002

Participation

Because of the increasing number of participants the scheme has been split between Sheffield and Heidelberg, starting from the beginning of 2001. In 2001 26 participants received their samples from Heidelberg. Since the beginning of 2002 47 participants received their samples from Heidelberg. The geographical distributions of the active participants in 2002 is 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.

Country	Number of laboratories	Country	Number of laboratories
Austria	2	Saudi Arabia	1
Canada	4	Slovakia	1
Croatia	1	Slovenija	1
Czech Republic	2	Sweden	2
Denmark	1	Switzerland	2
Finland	1	Tunesia	1
Germany	9	Turkey	1
Italy	9	United Kingdom	1
Norway	1	USA	6
Poland	1		

Table 1: Geographical distribution of participants

Samples and results

Three sets of three samples (total 9; sample number 106-114) were distributed to 47 laboratories in 2002. 41 laboratories returned results to the first circulation, 40 to the second circulation and 39 returned results to the third circulation.

Circulation	Number of returns
1. circulation	41
2. circulation	40
3. circulation	39

Table 2: Receipt of results

A few laboratories returned their results extremely late, sometimes after the circulation report. We have included such late results wherever possible, but this is not possible once the returns have been analysed and the report written. **In future, any results returned late may be disregarded**.

Instrumentation

Of the active laboratories, between 82,5% and 90% used GC-MS, between 5% and 7,5% used GC, the others did not specify their method.

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

		-2	**0**	**1**	**2**
Sample 106	Normal pattern	0	6	9	26
Sample 107	Isovaleryl-CoA dehydrogenase deficiency	0	0	0	41
Sample 108	Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type II)	3	10	5	23
Sample 109	Methylmalonic aciduria	0	0	0	40
Sample 110	Normal pattern	0	8	7	25
Sample 111	Normal pattern	0	0	0	40
Sample 112	Succinic semialdehyd dehydrogenase (SSADH)	5	0	0	33
Sample 113	Medium-chain acly-CoA dehydrogenase (MCAD)	0	0	0	39
Sample 114	Normal pattern	0	0	0	39

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 diagnosed correctly methylmalonic aciduria, isovaleryl-CoA dehydrogenase deficiency and MCAD. The sample with 4-hydroxybutyric aciduria due to SSADH deficiency was correctly identified by 85% of all participants. It is interesting to note that SSADH deficiency was also the diagnosis in a sample circulated by Heidelberg in 2001 (sample 99). At that time only 77% of all participants (26 participants) had correctly identified this sample as coming from a patient with SSADH deficiency. Two of the four laboratories who missed the diagnosis for sample 99 gave up this scheme, one missed the diagnosis again and one reported a pathological pattern and recommended further investigations.
We feel that the greatest challenge was presented by sample 108. This sample was obtained from a 10 month old boy with progressive encephalopathy, epilepsy and cardiomyopathy. Most laboratories reported a various combination of short-chain volatile acids, glutaric, adipic and suberic acids and 92% of all participants diagnosed glutaric aciduria type I or II.

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 2003. You will find your individual score for each sample in table 5. The poor performance of some laboratories scoring less than 10 this year is due to missing returns. Two laboratories only returned 1 distribution for the whole year.

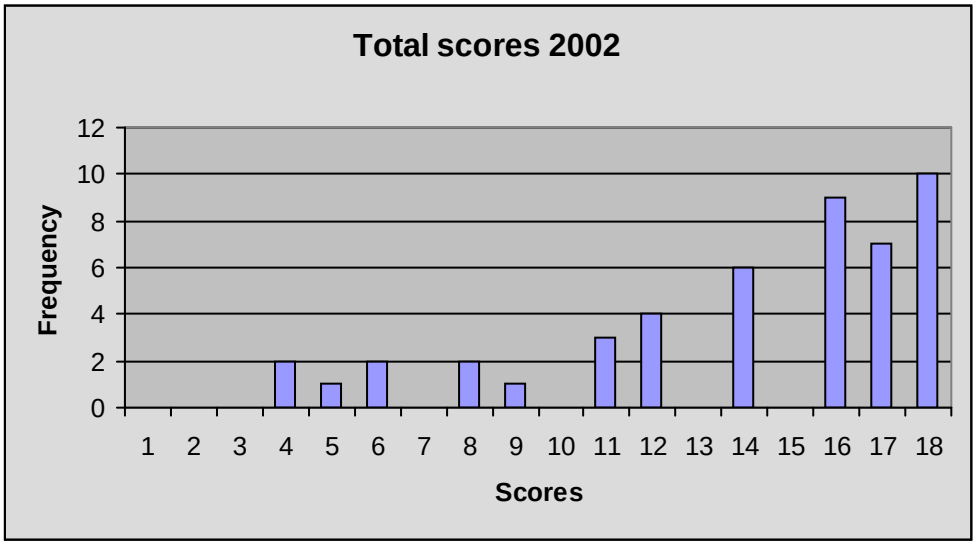

Diagram 1: Total scores 2001

Table 4: total scores 2001

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

General comments

A special thank for the laboratories who 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 120 ml urine of any interesting patients you may have. The costs will be covered by us.
Because many laboratories asked for their individual results for each circulation during the year instead of getting the results once in the annual report we will change in 2003 and give you your individual scores with each report!

Yours sincerely,

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