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ERNDIM
Diagnostic proficiency testing
2006
Southern Europe

ANNUAL REPORT 2006

In 2006, 19 labs participated to the Proficiency Testing Scheme Southern Europe.
Organizing Centre: Dr Christine Vianey-Saban, Dr Cécile Acquaviva-Bourdain, Service Maladies Héréditaires du Métabolisme et Dépistage Néonatal, Centre de Biologie et de Pathologie Est, Lyon.

Geographical distribution of participants

Country	Number of participants
France	7
Italy	5
Spain	4
Portugal	2
Swiss	1
TOTAL	**19**

Logistic of the scheme

- 2 surveys 2006-1 : patient P1, P2 and P3
  2006-2 : patient P4, P5 and P6

- Origin of patients : 4 out the 6 urine samples have been kindly provided by participants

- Patient P1 : Aromatic L-aminoacid decarboxylase – Hôpital Debrousse
- Patient P2 : Hyperoxaluria type I – Dr Encarnacio Riudor, Dr Jose Antonio Arranz, Hospital Maternoinfantil Vall d'Hebron, Barcelona.
- Patient P3 : Mucopolysaccharidosis type VI - Hôpital Debrousse
- Patient P4 : Hypophosphatasia – Pr Willems, SKML, Nijmegen. This sample has been sent to all labs participating to the DPT scheme in Europe
- Patient P5 : Lysinuric protein intolerance - Dr Begoña Merinero, Universidad Autonoma de Madrid
- Patient P6 : MCAD deficiency - Dr Marie-Hélène Read, CHU de Caen

- Mailing : samples were sent by DHL at room temperature.

Timetable of the schemes

- April 25, 2006: shipment of samples of Survey 1 and Survey 2 by DHL and of the forms by e-mail
- May 19, 2006: deadline for results submission (Survey 1)
- June 19, 2006: analysis of samples of the second survey
- July 10, 2006: deadline for results submission (Survey 2)
- July 17, 2006: report of Survey 1 by e-mail
- August 22, 2006: report of Survey 2 by e-mail
- October 5, 2006: meeting in Prague
- April 28, 2007 : Annual Report sent by e-mail

Date of receipt of samples

Once again, DHL has been very efficient.

	Survey 1 + 2
+ 24 hours	16
+ 48 hours	2
+ 72 hours	1

Date of reporting

All labs sent reports, but with more delay compared to last year. This will no be possible in the future, when submission will be on the website.

	Survey 1	**Survey 2**
	(3 weeks)	(3 weeks)
Receipt of results :	19 labs	19 labs
Before deadline	17	14
+ 1 day		3
+ 2 days		1
+ 5 days	1	
+ 6 days	1	
+ 28 days		1
No answer	**0**	**0**

Scoring of results

The scoring system established by the International Scientific Advisory Board of ERNDIM is still the same. Three criteria are evaluated:

A	Analytical performance	Correct results of the appropriate tests	2
		Partially correct or non-standard methods	1
		Unsatisfactory of misleading	0
I	Interpretation of results	Good, diagnosis is established	2
		Helpful but incomplete	1
		Misleading / wrong diagnosis	0
R	Recommendations for further investigations	Complete	1
		Unsatisfactory of misleading	0

Since most of the laboratories in Southern Europe don't give therapeutic advices to the attending clinician, this criterium is not evaluated.

The **total score** is calculated as the sum of these 3 criteria without weighting. The maximum that can be achieved is 5 for one sample.

Meeting of participants

It took place in Prague on October 5 from 14.15 to 16.00, during the ERNDIM / EuroGentest Meeting.

❖ Participants

Representatives from 11 labs were present: A Ribes (Hospital Clinic, Barcelona), JA Arranz (Vall d'Hebron, Barcelona), I Redonnet-Vernhet (Bordeaux), S Funghini, E Pasquini (Florence), U Caruso, M Cassanello (Genova), I Tavares de Almeida (Lisbonne), P Ruiz-Sala (Madrid), C Marsac (Paris), ML Cardoso (Porto), C Rizzo (Roma), M Rebollido (Santiago de Compostella).

❖ Informations from the Executive Board and the Scientific Advisory Board for next year

- In 2006, a fifth centre has been created in Basel (Dr Brian Fowler). There is now 5 DPT-centres in Europe.
- Certificate of participation for 2006 will be issued for participation and it will be additionally notified whether the participant has received an assistance letter. This assistance letter is sent out if the performance is less than 50%. Good performers are those whose performance is more than 75%.
- No assistance letter has been sent in 2005.
- We remind you that every year, each participant must provide to the scheme organizer at least 300 ml of urine from a patient affected with an established inborn error of metabolism or a "normal" urine, together with a short clinical report. Each urine sample must be collected from a single patient (don't send urine spiked with pathological compounds). Please don't send a pool of urines. For "normal" urine, the sample must be collected from a symptomatic patient (don't send urine from your kids !). Annex 1 gives the list of the urine samples we already sent.

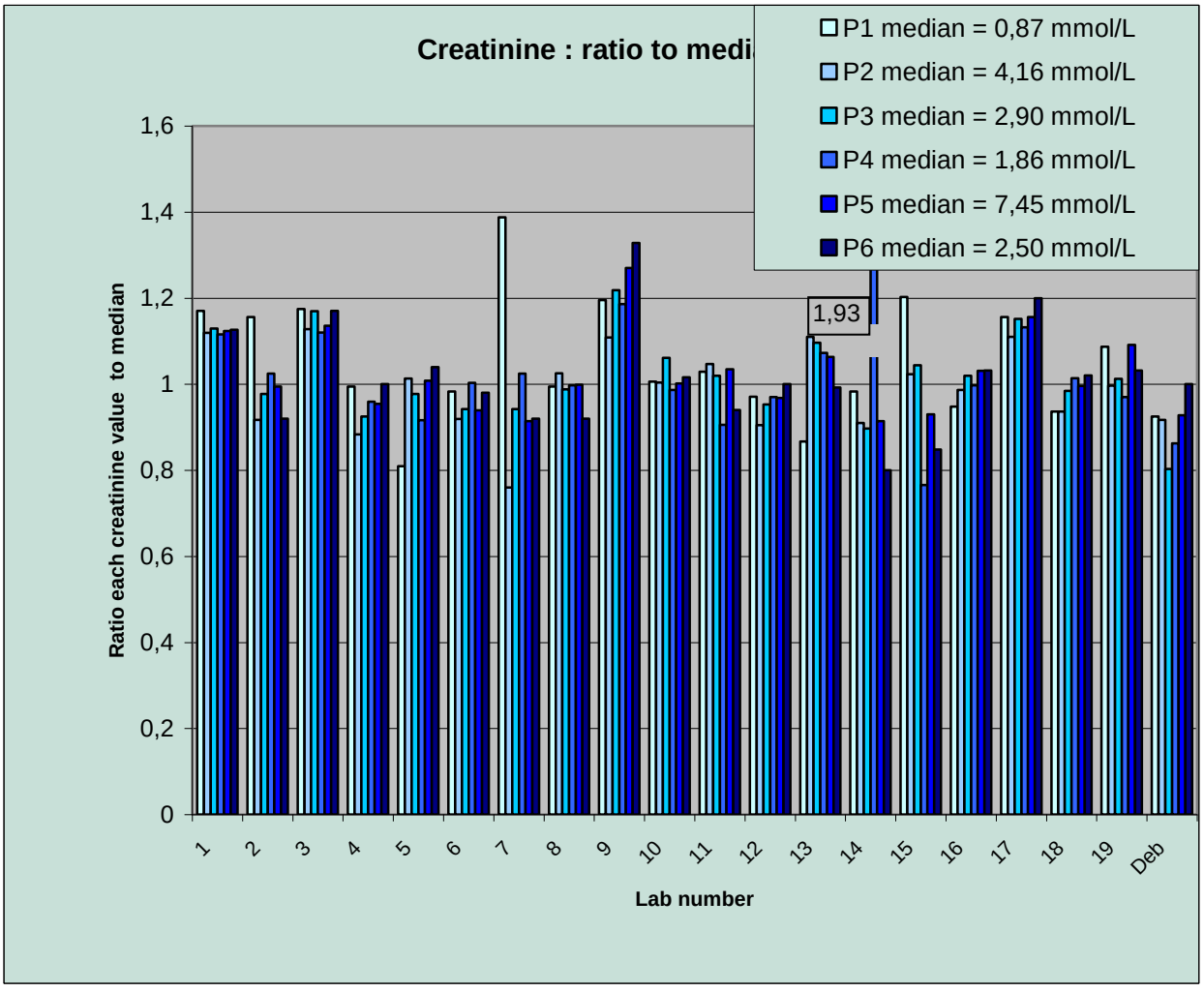
  As soon as possible after collection, the urine sample must be heated at 50 °C for 20 minutes. Make sure that this temperature is achieved in the entire urine sample, not only in the water bath. Then aliquot the sample in 10 ml plastic tubes (minimum 48 tubes), add stoppers and freeze. Be

careful to constantly homogenize the urine while aliquoting the sample. Send the aliquots on dry ice by rapid mail or express transport to: Christine Vianey-Saban, Service Maladies Héréditaires du Métabolisme et Dépistage Néonatal, 5ième étage, Centre de Biologie et de Pathologie Est, Groupement Hospitalier Est, 59, Boulevard Pinel, 69677 Bron cedex, France. Please send me an e-mail on the day you send the samples.

❖ Discussion of results

- **Creatinine measurement**

Results again significantly improved compared to last year.
If we exclude the 2 wrong values, coefficient of variation is < 11 % for all samples.



- **Patient P1 – Aromatic L-aminoacid decarboxylase (AADC) deficiency**

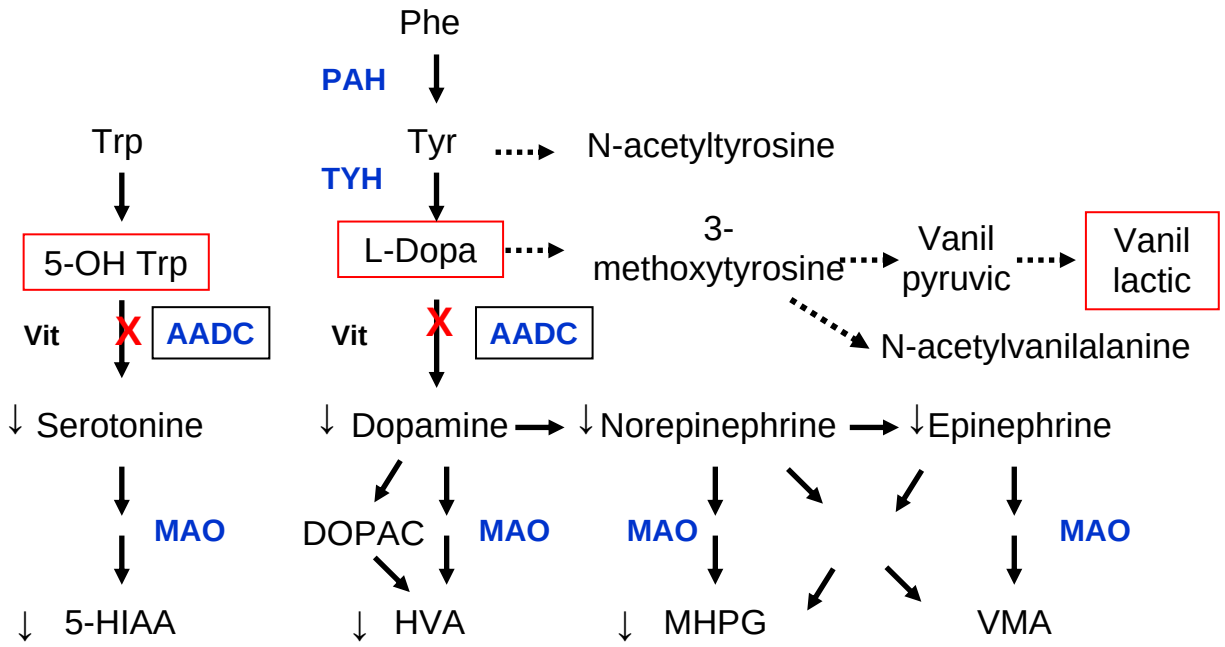
The urine sample was collected from a 4 month-old boy (he is now 11 month-old). Diagnosis was established at 2 1/2 months of age. He is the second child of non consanguineous parents. Pregnancy and delivery were uneventful. He was hospitalized at 1 month of age because of bronchiolitis. Absence of visual contact, oculogyric crises (ascribed to seizures), ptosis, tremor, and mild hypotonia were noticed. He was treated with valproate. CT scan revealed unilateral subdural hematoma. Two weeks later, he was hospitalized in intensive care unit because of respiratory syncytial virus infection. Several times per day, he needed to be resuscitated because of apneas leading to severe bradycardia. Urinary organic acids revealed an increase of vanillactic acid, and urinary amino acid

analysis, using tandem mass spectrometry, an increase of L-DOPA (13 mmol/mol creat - controls ND) and of 5-hydroxytryptophane. CSF and plasma were sent to Pr R. Wevers in Nijmegen: very low levels of HVA, MHPG and 5-HIAA were noticed, contrasting with high levels of 3-methoxytyrosine, L-DOPA and 5-hydroxytryptophane. Diagnosis was confirmed by measurement of AADC activity in serum (Pr R. Wevers, Nijmegen):

- With L-DOPA as substrate = 0.5 mU/L (controls : 9.0 – 56.0)
- With 5-HTP as substrate = 0.0 mU/L (controls : 1.0 – 7.1)

Mutation analysis of *AADC* gene is under investigation. The patient was treated with vitamin B6 (the cofactor of AADC) and with Selegiline, a MAO B inhibitor, which also inhibits the reuptake of catecholamines. With this treatment, he has a better contact, and an improvement of tone and of sleep disturbances. But he has persistent oculogyric crises and psychomotor retardation.

The urine sample was a mix of urine collected before and after treatment



Diagnosis

- 12 labs gave a correct diagnosis
- 2 labs concluded to a probable neurotransmitter metabolism disorder but did not identify vanillactic acid.
- 5 labs gave no diagnosis or a wrong diagnosis

Most labs who performed **aminoacids**, reported an increase of glycine (due to valproate therapy). Some reported an increase of hydroxyproline.

Eighteen labs performed **organic acids**. Eleven of them identified an increase of vanillactic acid: 28 - 75 mmol/mol creatinine – median = 52. One lab did not identify vanillactic but gave AADC deficiency as diagnosis (?). Ten labs reported an increase of homovanillic, but we could not explain why, since this patient is treated with a MAO inhibitor (see figure). Can this be due to the inhibition of the reuptake of catecholamines due to Selegiline? One lab also reported a peak of N-acetylvanilalanine. This metabolite had been reported by Abdenur et al, Mol Genet Metab.2006;87:48:53. They also reported an increase of vanilpyruvic and N-acetyltyrosine, but these 2 metabolites could not be detected in this patient.

Advice for further investigations was satisfying for those who reached a correct diagnosis.

The following **scoring scheme** was accepted:

- Analytical performance: identification of vanillactic (score 2), vanillactic not identified (score 0).
- Interpretation of results: AADC deficiency (score 2), possible neurotransmitter metabolism disorder (score 1), no or wrong diagnosis (score 0).
- Advice for further investigations: neurotransmitters in CSF or AADC activity or mutation analysis of *AADC* gene (score 1).

- **Patient P2 – Hyperoxaluria type I (alanine:glyoxalate aminotransferase deficiency)**

This 10 year-old girl, presented at 1 year of age bilateral nephrocalcinosis, with high excretion of oxalate and glycolate. Mutation analysis revealed that she is heterozygote for the common c.853T>C mutation of *AGXT* gene (inherited from her mother), but the other mutation has not been identified. Treatment with pyridoxine and citrate was instituted with excellent resolution of clinical and radiological symptoms. Now, 10 years later, she has vision problems, probably due to oxalate crystals, and renal stones, appreciated by radiological examination.

Diagnosis

Fifteen labs reached a good diagnosis, whereas 4 labs gave no diagnosis or a wrong diagnosis

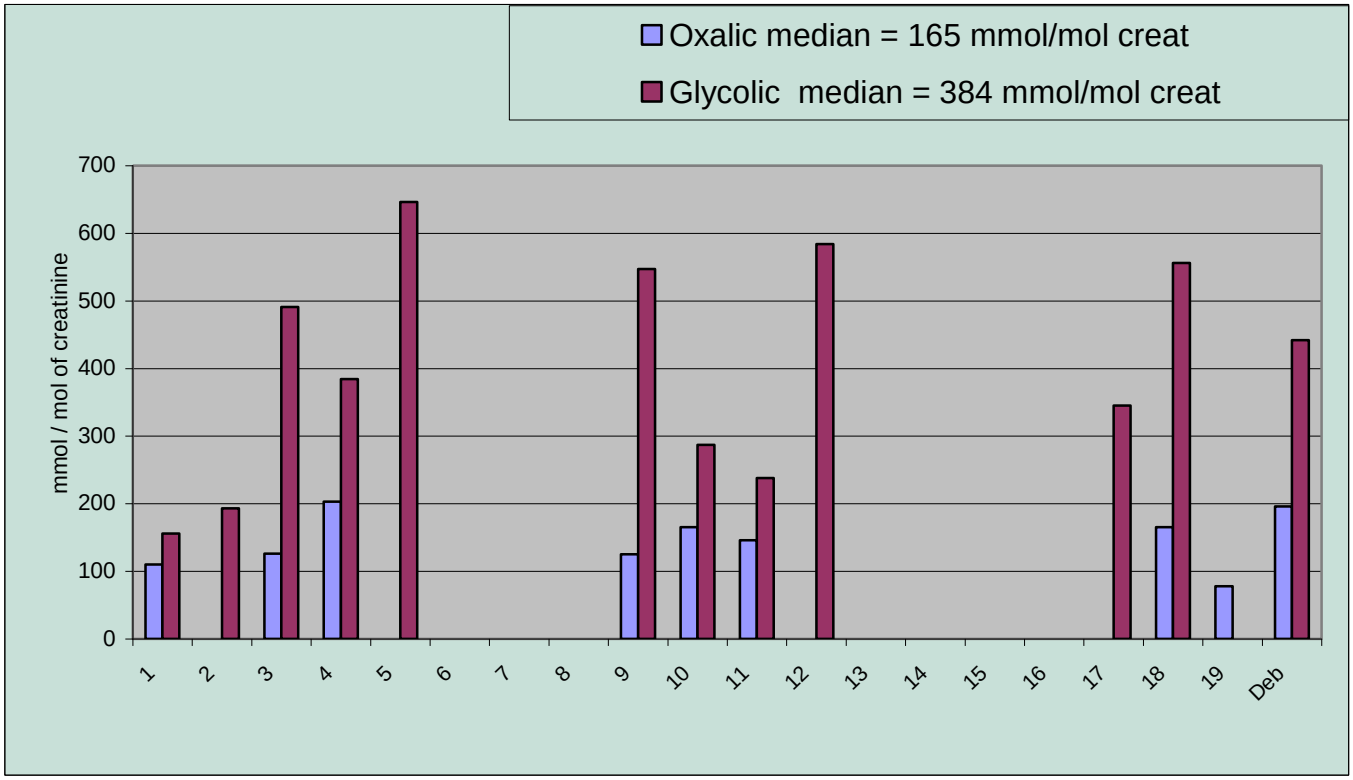
Amino acids

All labs, except 2, performed aminoacids:

- 10 reported no significant abnormality
- 7 reported a slight increase of either Lys, Cyst, Orn, Arg or all, and 2 of them concluded to a mild pattern of cystinuria

Organic acids

Fifteen labs reported an increase of oxalic acid and glycolic acid. One lab did not report an increase of theses 2 organic acids but concluded to hyperoxaluria type I (?). Quantification of metabolites was highly variable (mainly for glycolic). In order to measure accurately these 2 metabolites, stable isotopes are available from Interchim : www.interchim.com ($^{13}C_2$ glycolic Ref CD394 or d_2 glycolic Ref CD256 - $^{13}C_2$ oxalic Ref CD255).



Advice for further investigations were correct, except that some labs advised to measure alanine:glyoxalate aminotransferase (AGT) activity in liver, which is quite invasive when mutation analysis is available.

Scoring

- Analytical : Increase of oxalic and glycolic acids (score 2), slight increase of cystine and lysine (score 1), organic acids not performed or normal profile (score 0).

- Interpretation : hyperoxaluria type I (score 2), cystinuria or no diagnosis or wrong diagnosis (score 0).

- Recommendations : repeat organic acids or blood oxalate or mutation analysis *AGXT* gene or kidney stone analysis (score 1), other investigations (score 0).

- **Patient P3 - Mucopolysaccharidosis type VI (Marroteaux-Lamy)**

This 19-year old girl, is the second child of Turkish consanguineous parents (first cousin). She was born after a normal pregnancy and delivery. She could sit at 12 months and walk at 2-years of age. She had severe statural delay. She was hospitalized for the first time in France at the age of 14 years. She had dysmorphia, gingival hypertrophy, macroglossia, disharmonious dwarfism, corneal opacity, articular stiffness, splenomegaly, aortic and mitral insufficiency, and learning difficulties. At 19-years of age, her weight is 24.8 kg, size 98.5 cm, and head circumference 57 cm

N-acetylgalactosamine-4-sulphatase (arylsulphatase B) activity in leukocytes, using a fluorimetric method after separation of ARSB from ARSA on DE52 chromatography, was 0.04 µkat/kg (simultaneous control = 3.61).

Thirteen labs gave a correct **diagnosis**, and 6 labs concluded to a lysosomal storage disease.

Twelve out of the 13 labs who performed **amino acid** analysis did not report any significant abnormality. One reported an increase of glycine.

Thirteen labs performed **organic acids**. They reported a normal profile.

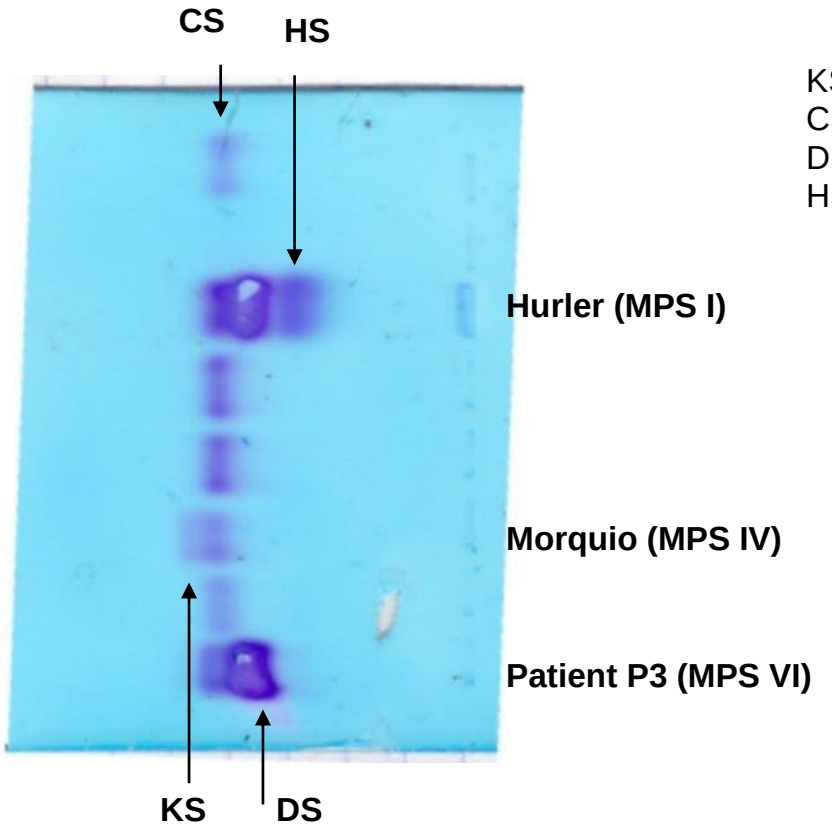
Among the 12 labs who performed **oligosaccharides**, all reported a nearly normal profile or some abnormality related to mucopolysaccharidosis.

All 16 labs who performed quantification of **mucopolysaccharides** reported an increased excretion. Two labs performed Berry test. Although they found a positive test, because GAG excretion was quite high, such test has to be abandoned because it is not enough sensitive to detect most mucopolysaccharidoses.

Sixteen labs performed identification of GAG fractions. They reported an increase of:

- dermatane sulphate n = 15
- chondroitine sulfate n = 4
- heparane sulphate n = 2
- keratane sulphate n = 1

leading to a wrong interpretation of the profile for 3 of them



KS : keratane sulphate
CS: chondroitine sulphate
DS : dermatane sulphate
HS : heparane sulphate

Advice for further investigations was OK for most labs.

Scoring

- Analytical performance: increase of GAG and dermatane sulphate (score 2), increase of GAG but no separation of GAG or wrong identification, separation of GAG without GAG quantification (score 1)

- Interpretation of results: MPS VI (or I H/S) (score 2), mucopolysaccharidosis (score 1), wrong mucopolysaccharidosis (score 0)

- Advice for further investigations: ARSB activity (+ other MPS enzymes) or enzymes mucopolysaccharidoses (score 1), wrong MPS enzyme (score 0)

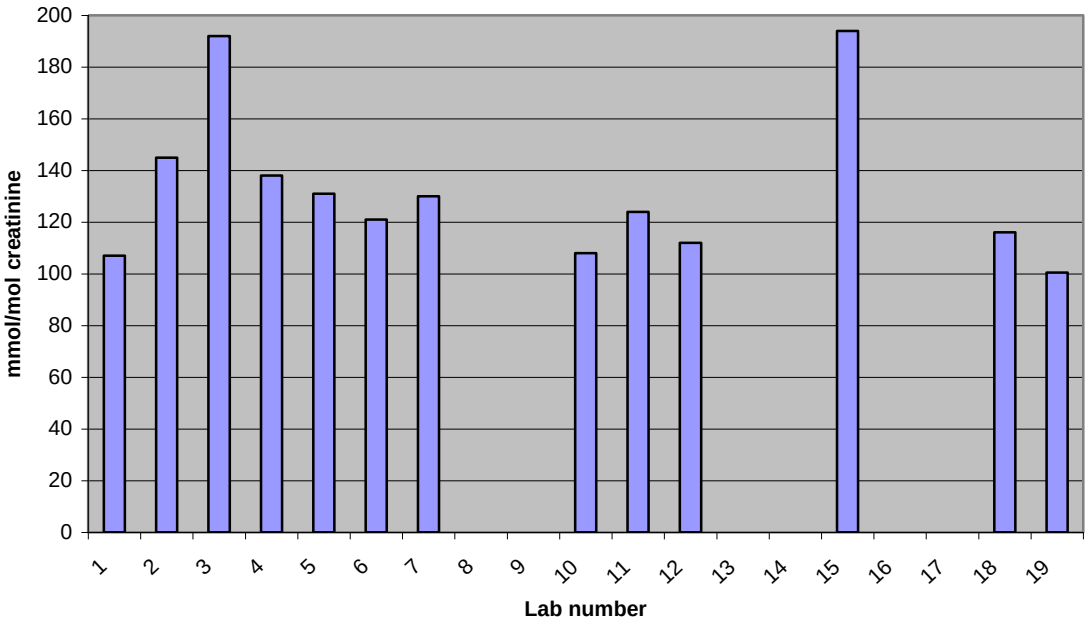
- **Patient P4 - Hypophosphatasia**

The patient, a female, presented at the age of 24 with a history of joint pains of approximately one year's duration. Relevant clinical history included delayed motor development, fatigue on exercise during childhood and premature shedding of primary teeth. The patient was of normal stature and physical examination revealed normal muscle strength and no evidence of synovitis. The forepart of her feet and her knees were painful to pressure and there were marked crepitations of the knees. There were brisk tendon reflexes without clonus and a positive Babinski response. Several joints in her hands, knees and feet had peri- and intra-articular calcifications. Diagnosis of hypophosphatasia was confirmed by measurement of alkaline phosphatase in leukocytes = 13 nmol/mg.h – control :100 – 5000). This urine sample has been sent to all DPT centres in Europe.

Diagnosis: 16 labs reached the write diagnosis, whereas 3 labs did not give diagnosis or concluded to hypophosphatemic ricket.

Aminoacid analysis was performed by all labs. Sixteen of them reported an increase of phosphoethanomine. By LC-MS/MS, phosphoethanolamine is not measurable in positive ionisation mode.

Phosphoethanolamine median = 124 mmol/mol creat - CV = 24 %



All labs performed **organic acid** profile and most of them reported a normal profile.

Interpretation and recommendations were satisfying for those who reached a correct diagnosis.

Scoring

- Analytical : increase of phosphoethanolamine (score 2), phosphoethanolamine not identified (score 0)
- Interpretation of results: hypophosphatasia / possible hypophosphatasia (score 2), no diagnosis or wrong diagnosis (score 0)
- Advice for further investigations: alkaline phosphatase activity (blood/plasma/ fibroblasts) or plasma pyridoxine-5'phosphate or mutation analysis *TNSLP* gene (score 1), other investigations (score 0)

- **Patient P5 - Lysinuric protein intolerance**

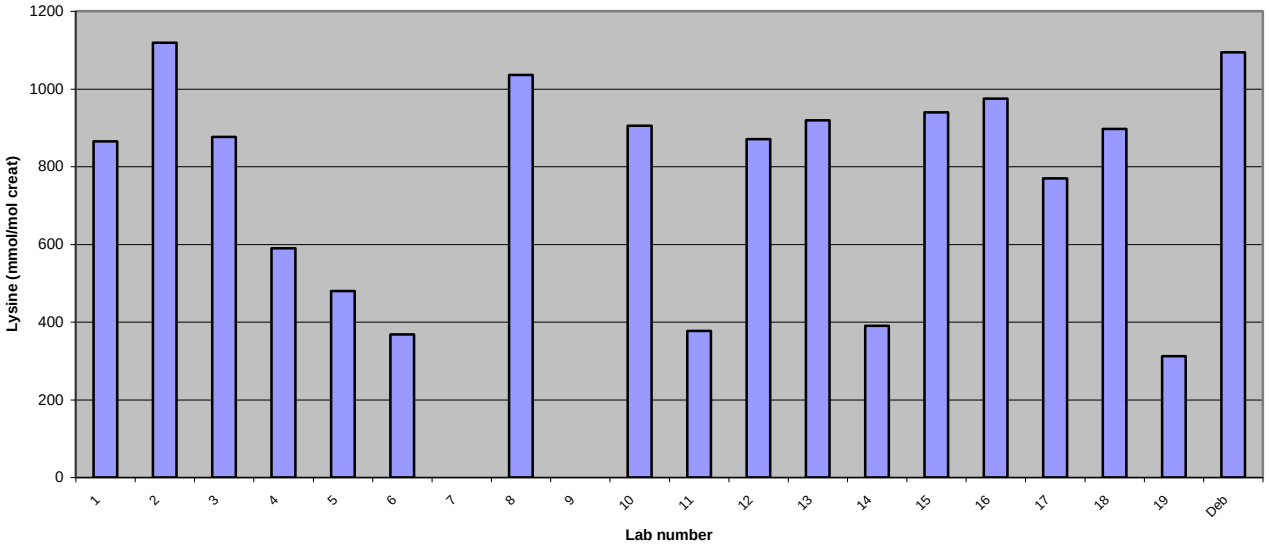
This urine sample was collected from a male who presented at 7 months of age with protein aversion, hepatosplenomegaly, psychomotor delay. At 14 years he showed osteoporosis, increased LDH, ferritine and triglycerides values, hypofibrinogenemia, bone marrow with macrophages and myeloid precursors with exclusive erythrophagocytosis. No hyperammonemia, malnutrition.
He had increased plasma glutamine (1899 µmol/L; normal 349±171) and low plasma values of lysine (62 µmol/L; normal 135-243); ornithine (19 µmol/L; normal 68±30) and arginine (55 µmol/L; normal 98±29). In urine, hyperaminoaciduria with high levels of lysine (202 mmol/mol creat; normal 15±11), arginine (76 mmol/mol creat; normal 2±1), and ornithine (23 mmol/mol creat; normal 3±3). He had hypocarnitinemia (FC 12.5 and TC 22.6 µmol/l), and normal excretion of orotic acid and organic acids.
During the last two years on hypoproteic diet with supplementation of citrulline and carnitine, the patient had increased the plasma levels of lysine and ornithine without normalization. Urine alterations had persisted.
Mutation analysis of *SLC7A7* gene has been done in the lab of Dr Virginia Nunes, Instituto Recerca Oncológica in Barcelona, showing a deletion in exon 9 in homozygous fashion, already found in other Spanish patients. The mother was carrier of the mutation. The father has not been studied.

Diagnosis

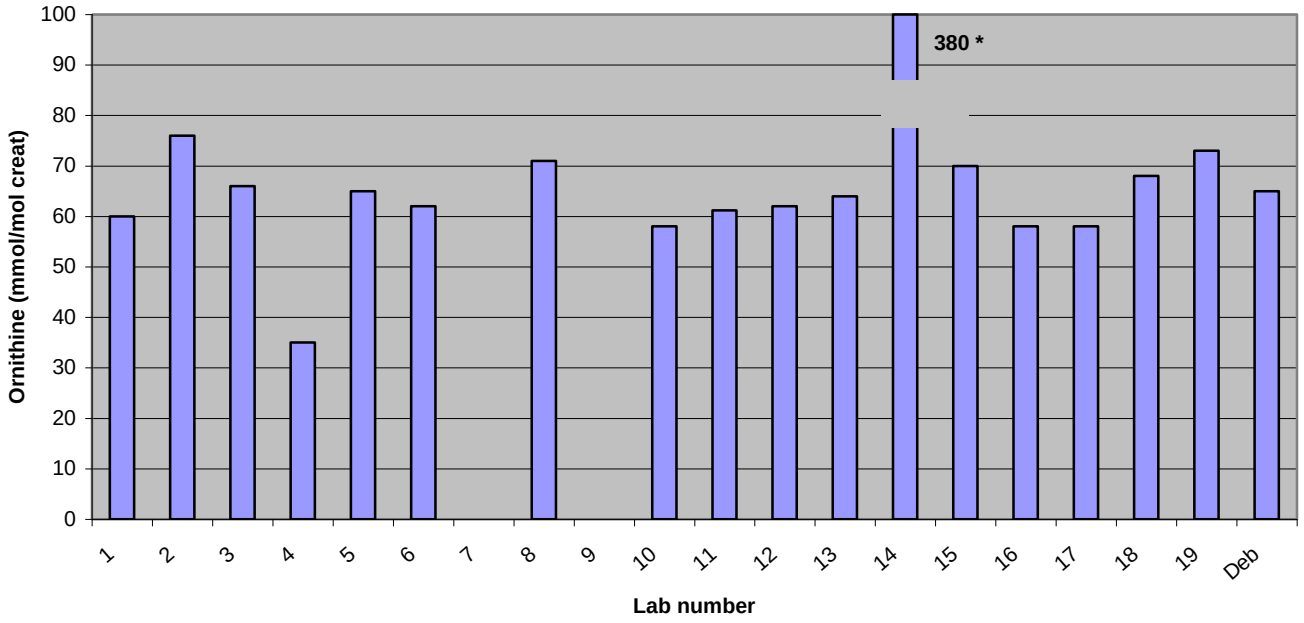
Eighteen labs concluded to lysinuric protein intolerance, whereas one lab concluded to maple syrup urine disease.

All labs performed **aminoacids** and all, except one, reported an increase of lysine. Most of them also reported an increase of arginine, ornithine (17 labs) and citrulline (10 labs). Surprisingly, there was a great variability of lysine results, but not of arginine and ornithine. Can this be due to a problem of storage (lysine can be degraded by bacteria) or is it due to a problem of linearity (the lysine values being higher than those of arginine and ornithine) ?

Lysine - median = 871 mmol/mol creat - CV = 30 %

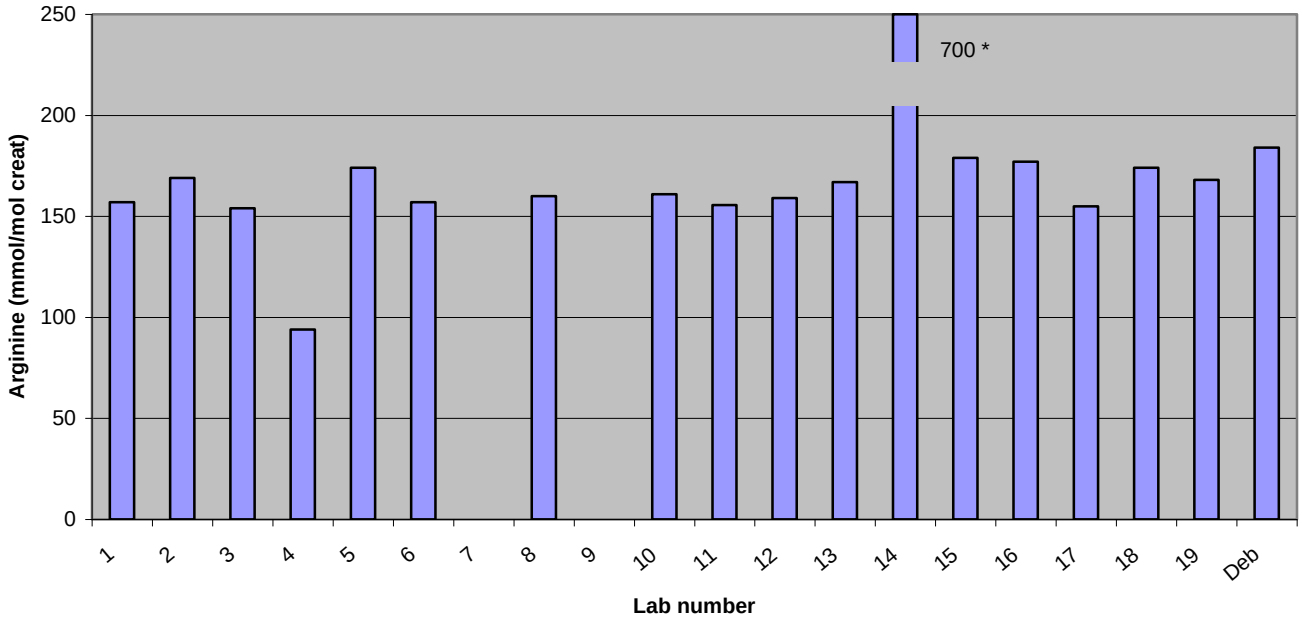


Ornithine - median = 63 mmol/mol creat - CV = 15 %



* This value was excluded for CV calculation

Arginine - median = 161 mmol/mol creat - CV = 12 %

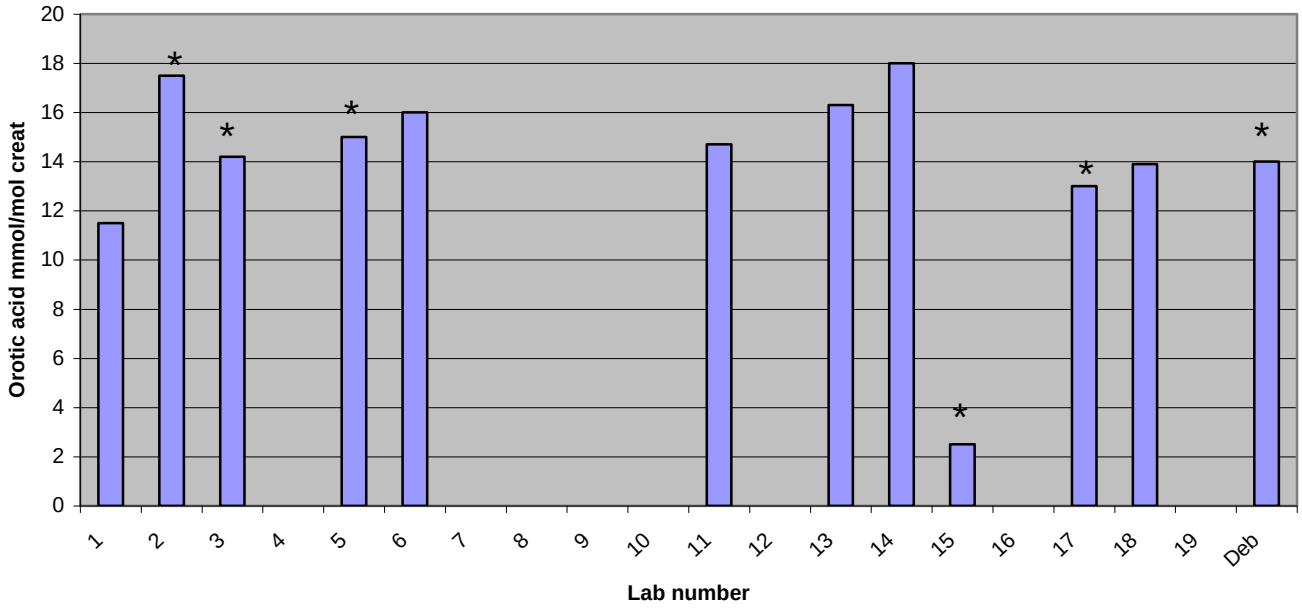


* This value was excluded for CV calculation

Among the sixteen labs who performed **organic acids**, 10 of them reported an increase of orotic acid. One lab reported an increase of 2-hydroxyisovaleric, 2-keto-3-methylvaleric and 2-keto-isocaproic acids.

Other investigations. Six labs measured orotic acid: 4 of them reported an increased value and 2 a normal value.

Orotic - median = 15 mmol/mol creat



* Other methods than GC/MS

Advice for further investigations were OK.

Scoring

- Analytical: increase of Lys (Arg, Orn, Cit) and orotic acid (score 2), increase of Lys (Arg, Orn, Cit), normal orotic acid excretion or orotic not performed (score 1), ggeneralized hyperaminoaciduria, normal orotic (score 0)

- Interpretation: lysinuric protein intolerance / probable lysinuric protein intolerance (score 2), wrong diagnosis (score 0)

- Recommendations: Plasma/blood amino acids or mutation analysis *SLC7A7* gene (score 1), enzyme study of lysinuric protein intolerance in liver or other enzyme assays (score 0)

- **Patient P6 - Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency**

This 6 year-old girl was hospitalized because of acute gastroenteritis. Metabolic investigation was required because of "ketotic vomitings". Blood acylcarnitine profile was indicative of MCAD deficiency. Her elder brother (8 year-old), who is asymptomatic, also has a blood acylcarnitine profile and an abnormal organic acid profile consistent with MCAD deficiency. Her younger sister (3 year-old) is treated with valproate because of seizures and also has a blood acylcarnitine and organic acid profile consistent with MCAD deficiency. Mutation analysis of *ACADM* gene has yet not been performed in this family.

Diagnosis

Eighteen labs concluded to MCAD deficiency or eventually to multiple acyl-CoA dehydrogenase deficiency. One lab concluded to intermittent ketotic vomitings.

The 16 labs who performed **amino acid** analysis reported no specific abnormality, except for two labs who reported a slight increase of glycine.

Organic acids: it was a difficult sample, not collected during an acute episode, with a normal excretion of dicarboxylic acids and of ketone bodies. All labs, except one, identified hexanoylglycine, and nearly all of them identified suberylglycine and phenylpropionylglycine. There was a great variability of quantitative results, but the problem is that no standards are available from trade companies.

One lab performed a urinary **acylcarnitine profile** and reported an increase of C8 and C10:1. We obtained the same profile and this can be useful if acylglycines excretion is low and plasma not available.

Advice for further investigations was correct.

Scoring

- Analytical: Increase of hexanoylglycine and suberylglycine and/or phenylpropionylglycine (score 2), increase of hexanoylglycine, but suberylglycine and phenylpropionylglycine not detected (score 1), normal organic acid profile (score 0)

- Interpretation: MCAD deficiency (eventually MADD) or MADD deficiency (MCAD cannot be excluded) (score 2), no diagnosis (score 0)

- Recommendations: plasma/blood acylcarnitines or mutation analysis of *ACADM* gene (or c.985A>G mutation) or MCAD activity (fibroblasts, lymphocytes) (score 1)

Scores of participants

- **Survey 2006-1**

Lab n°	Patient P1 AADC deficiency				Patient P2 Hyperoxaluria type I				Patient P3 MPS VI			
	A	**I**	**R**	**Total**	**A**	**I**	**R**	**Total**	**A**	**I**	**R**	**Total**
1	0	0	0	**0**	2	2	0	**4**	2	2	1	**5**
2	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
3	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
4	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
5	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
6	0	0	0	**0**	0	0	0	**0**	1	1	1	**3**
7	0	2	1	**3**	0	2	1	**3**	1	1	0	**2**
8	2	2	1	**5**	2	2	1	**5**	2	1	1	**4**
9	0	1	1	**2**	2	2	1	**5**	1	1	1	**3**
10	0	0	0	**0**	2	2	5	**5**	2	2	1	**5**
11	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
12	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
13	0	0	0	**0**	2	0	0	**2**	1	1	1	**3**
14	2	2	1	**5**	2	2	1	**5**	1	2	1	**4**
15	0	0	0	**0**	2	2	1	**5**	2	2	1	**5**
16	2	2	1	**5**	1	0	0	**1**	1	0	0	**1**
17	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
18	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
19	0	1	1	**2**	1	0	0	**1**	2	2	1	**5**

❖ **Survey 2006-2**

Lab n°	Patient P4 Hypophosphatasia				Patient P5 LPI				Patient P6 MCAD deficiency			
	A	**I**	**R**	**Total**	**A**	**I**	**R**	**Total**	**A**	**I**	**R**	**Total**
1	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
2	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
3	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
4	2	2	1	**5**	1	2	1	**4**	2	2	1	**5**
5	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
6	2	2	0	**4**	2	2	1	**5**	2	2	1	**5**
7	2	2	1	**5**	0	0	0	**0**	2	2	1	**5**
8	0	0	0	**0**	2	2	1	**5**	1	2	1	**4**
9	0	0	0	**0**	1	2	1	**4**	1	2	1	**4**
10	2	2	1	**5**	1	2	1	**4**	2	2	1	**5**
11	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
12	2	2	1	**5**	1	2	1	**4**	2	2	1	**5**
13	0	0	0	**0**	2	2	1	**5**	2	2	1	**5**
14	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
15	2	2	1	**5**	1	2	1	**4**	0	0	1	**1**
16	2	2	1	**5**	1	2	1	**4**	2	2	1	**5**
17	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
18	2	2	1	**5**	2	2	1	**5**	2	2	1	**5**
19	2	2	1	**5**	1	2	1	**4**	1	2	1	**4**

❖ **Total scores**

Lab number	Survey 2006-1	Survey 2006-2	Cumulative score	Cumulative score (%)
1	9	15	**24**	**80 %**
2	15	15	**30**	**100 %**
3	15	15	**30**	**100 %**
4	15	14	**29**	**97 %**
5	15	15	**30**	**100 %**
6	3	14	**17**	**57 %**
7	8	10	**18**	**60 %**
8	14	9	**23**	**77 %**
9	10	8	**18**	**60 %**
10	10	14	**24**	**80 %**
11	15	15	**30**	**100 %**
12	15	14	**29**	**97 %**
13	5	10	**15**	**50 %**
14	14	15	**29**	**97 %**
15	10	10	**20**	**67 %**
16	7	14	**21**	**70 %**
17	15	15	**30**	**100 %**
18	15	15	**30**	**100 %**
19	8	13	**21**	**70 %**

Performance

	Number of labs	% total labs
Excellent performers (100 % of good responses)	**6**	**32 %**
Good performers (> 75 % good responses)	**13**	**68 %**
Poor performers (< 50 % good responses)	**0**	**0 %**

❖ **Summary of scores**

We excluded from this table, the labs who did not send results. The percentages given are the scores obtained from labs who sent a report.

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Recommen-dations (%)	Total (%)
Patient P1	AADC def.	58	68	74	**65**
Patient P2	Hyperoxaluria	84	79	74	**80**
Patient P3	MPS VI	84	82	89	**84**
Patient P4	Hypophosph.	84	84	79	**83**
Patient P5	LPI	76	95	95	**87**
Patient P6	MCAD def.	87	95	100	**93**

DPT-scheme in 2007

Same “rules” as in 2006:

- Two surveys of 3 urines, including “normal” patients
- Results have to be sent within 3 weeks
- Scoring will be analyzed for all centres
- Poor performers: those who don’t reply to both surveys or those who received an assistance letter (score < 50 %)
- Good performers: those who reached a score > 75 %

We remind you that to participate to the DPT-scheme, you must perform at least:

- Amino acids
- Organic acids
- Oligosaccharides
- Mucopolysaccharides

If you are not performing one of these assays, you can send the samples to another lab (cluster lab) but you are responsible for the results.

Please send quantitative data for amino acids and, if possible, for organic acids.

Meeting in 2007

The next meeting for the DPT-scheme Southern Europe will take place during the 43nd Symposium of SSIEM in Hamburg, on Tuesday September 4th from 9.30 to 11.00. Further information will be sent as soon as we get more details.

We remind you that attending this meeting is an important part of the proficiency testing. The goal of the program is to **improve** the competence of the participating laboratories which includes the critical review of all results with a discussion about improvements.