ERNDiM

Quality Assurance in Laboratory Testing for IEM

ERNDIM Participant Meeting

DPT Centre Switzerland, scheme year 2025

09.10.2025, Madrid

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Conflict of Interest

- Déborah Mathis has nothing to disclose

Contents

- Organisational aspects:
  - participants
  - samples
  - tests required in 2025
  - 2025 schedule
  - scoring and reporting
- Reports on individual sample performance
  DPT-SB-2025-A, B, C, D, E, F
- Overall performance and scores
- Conclusion, suggestion

Organisational aspects

Geographical distribution of participants

21/21 laboratories submitted results for both survey by the deadline.

Australia (3 labs), Austria (2 labs), Canada (3 labs), China (1 lab), Estonia (2 labs), Germany (3 labs), Norway (1 lab), Sweden (2 labs), Switzerland (1 lab), USA (2 labs)

Samples

The samples have been heat-treated. Four samples were from our institute, one was provided by one of our participants, and one was the common sample.

Tests required for 2025 scheme

Analyses of amino acids, organic acids, GAGs and oligosaccharides were required in 2025.

Organisational aspects

Schedule of the scheme 2025

Scheme Year:	2025	
DPT Centre:	Switzerland	
CSCQ Sample dispatch date:	05 February 2025	
	1st Submission Round	**2nd Submission Round**
Sample ID's:	DPT-SB-2025-**A** DPT-SB-2025-**B** DPT-SB-2025-**C**	DPT-SB-2025-**D** DPT-SB-2025-**E** DPT-SB-2025-**F**
Analysis start & Website submission availability*:	17 March 2025	02 June 2025
Results submission deadline*:	07 April 2025	23 June 2025

Evaluation of reports

Scoring system based on two criteria

A	Analytical performance	Correct results of the appropriate tests	2
		Partially correct or non-standard methods	1
		Unsatisfactory or misleading (in some instances will be evaluated also as a critical error), or No result submitted	0
I	Interpretative proficiency	Good (diagnosis was established and appropriate further tests were recommended)	2
		Helpful but incomplete	1
		Misleading/wrong diagnosis (will be most likely evaluated also as a critical error), or No result submitted	0

Due to the large variability in reporting results in various countries, **recommendations pertaining to treatment** are not evaluated in proficiency testing, but **recommendations for further tests** may be considered in scoring interpretation.

- The **total score** is calculated as a sum of these two criteria.
- Maximum of 4 points per sample
- Maximum of 24 points per year

DPT-SB-2025: The samples

A: Mucopolysaccharidosis type VI (MPS VI, **common sample**)

B: Barth syndrome

C: Thymidine phosphorylase deficiency (MNGIE)

D: Mucopolysaccharidosis type IIIC (MPS III)

E: Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency

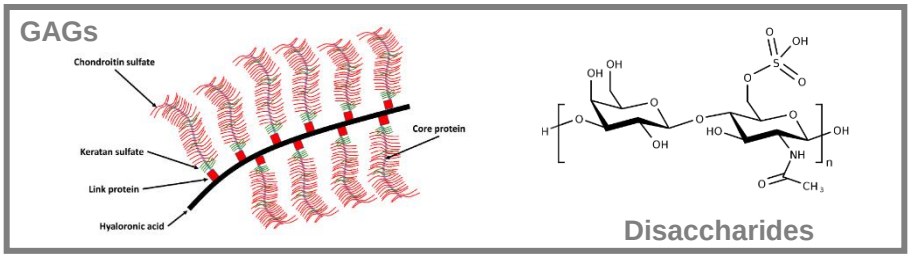
F: Multiple acyl-CoA dehydrogenase deficiency (MADD)

Total scores and proficiency

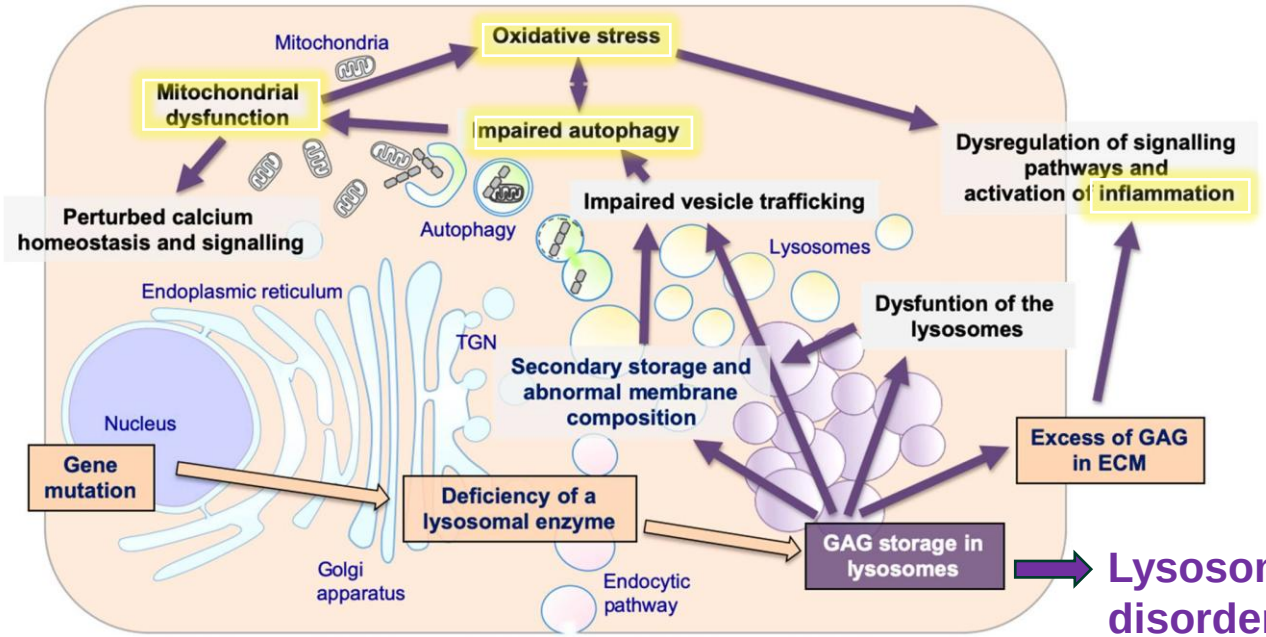
	A - MPS VI			B-Barth			C-MNGIE			D-MPS III			E-MCAD			F-MADD			Σ
	A	I	Σ	A	I	Σ	A	I	Σ	A	I	Σ	A	I	Σ	A	I	Σ	
1	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	24
2	1	1	**2**	2	2	**4**	2	1	**3**	0	0	**0**	2	2	**4**	2	2	**4**	17
3	1	1	**2**	2	2	**4**	1	1	**2**	0	0	**0**	2	2	**4**	2	2	**4**	16
4	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	24
5	1	1	**2**	2	2	**4**	1	1	**2**	2	2	**4**	2	2	**4**	2	2	**4**	20
6	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	24
7	1	2	**3**	2	2	**4**	2	2	**4**	1	0	**1**	2	2	**4**	2	2	**4**	20
8	2	2	**4**	2	2	**4**	2	2	**4**	0	1	**1**	2	2	**4**	2	2	**4**	21
9	1	1	**2**	2	2	**4**	2	2	**4**	0	0	**0**	2	2	**4**	2	2	**4**	18
10	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	24
11	1	1	**2**	2	2	**4**	2	2	**4**	0	0	**0**	2	2	**4**	2	2	**4**	18
12	2	2	**4**	2	2	**4**	2	2	**4**	0	0	**0**	2	2	**4**	2	2	**4**	20
13	1	1	**2**	2	2	**4**	1	0	**1**	2	2	**4**	2	2	**4**	2	2	**4**	19
14	2	2	**4**	2	2	**4**	2	2	**4**	0	0	**0**	2	2	**4**	2	2	**4**	20
15	2	2	**4**	2	2	**4**	2	2	**4**	0	0	**0**	2	2	**4**	2	2	**4**	20
16	1	1	**2**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	22
17	1	1	**2**	2	2	**4**	2	2	**4**	0	0	**0**	2	2	**4**	2	2	**4**	18
18	1	1	**2**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	22
19	2	2	**4**	2	2	**4**	1	1	**2**	2	1	**3**	2	2	**4**	2	2	**4**	21
20	2	2	**4**	2	2	**4**	2	2	**4**	0	0	**0**	2	2	**4**	2	2	**4**	20
21	1	1	**2**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	2	2	**4**	22
%	74	76	**75**	100	100	**100**	90	86	**88**	50	48	**49**	100	100	**100**	100	100	**100**	

Mucopolysaccharidosis (MPS) and Glycosaminoglycans (GAGs)

- MPS are a group of genetic diseases characterized by a **deficiency of lysosomal enzymes** required for the degradation of GAGs → accumulation of certain GAGs
- GAGs are **polysaccharides** containing a repeating **disaccharide unit** → mainly located on the cell surface and in the extracellular matrix



Pathogenesis of Mucopolysaccharidoses (MPS)



→ Organ damage, skeletal abnormalities, central nervous system impairement

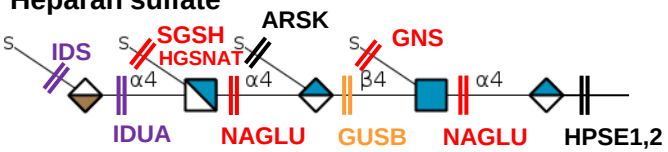
Lysosomal storage disorders

Int. J. Mol. Sci. **2020**, *21*(7), 2515; https://doi.org/10.3390/ijms21072515

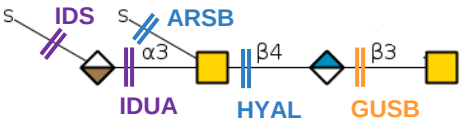
Mucopolysaccharidosis (MPS) and Glycosaminoglycans (GAGs)

Abb	Enzyme name	MPS type	Disease name	Degrada-tion of:
IDUA	L-iduronidase	I	Hurler-Scheie syndrome	HS, DS
IDS	iduronate 2-sulfatase	II	Hunter syndrome	HS, DS
SGSH	N-sulfoglucosamine sulfohydrolase	IIIA	Sanfilippo A	HS
NAGLU	alpha-N-acetylglucosaminidase	IIIB	Sanfilippo B	HS
HGSNAT	heparan-alpha-glucosaminide N-acetyltransferase	IIIC	Sanfilippo C	HS
GNS	N-acetylglucosamine-6-sulfatase	IIID	Sanfilippo D	HS, KS
ARSK	arylsulfatase K	X		HS
HPSE1,2	heparanase		Urofacial syndrome	HS
GUSB	beta-galactosidase	VII	Sly	HS, CS, DS, H
ARSB	arylsulfatase B	VI	Maroteaux-Lamy syndrome	CS, DS
HYAL	hyaluronoglucosaminidase	IX		CS, DS
GALNS	N-acetylgalactosamine-6-sulfatase	IVA	Morquio syndrome A	KS, CS
GLB1	beta-galactosidase	IVB GM1	Morquio syndrome B GM1	KS
HEXA,B	Hexosaminidase A, B	GM2	Tay-Sachs disease (def HEXA) Sandhoff disease (def HEXA+B)	KS
NAGZ		-		H

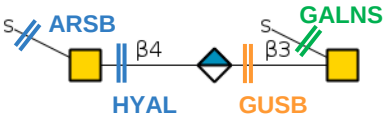
Heparan sulfate



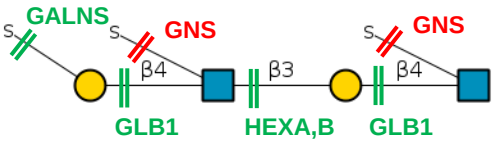
Dermatan sulfate



Chondroitin sulfate



Keratan sulfate



Hyaluronan

KEGG PATHWAY: Glycosaminoglycan degradation - Homo sapiens (human) (genome.jp)

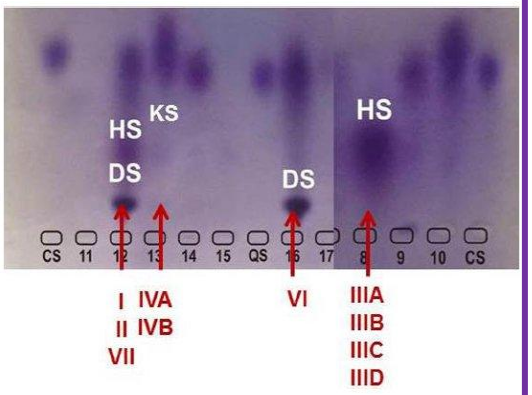
IdoA: L-iduronic acid
GlcA: D-glucuronic acid
Gal: D-galactose
GlcN: D-glucosamine
GalNAc: N-acetyl-D-galactosamine
GlcNAc: N-acetyl-D-glucosamine

Diagnosis of MPS

1. Total GAGs in urine by DMB-test

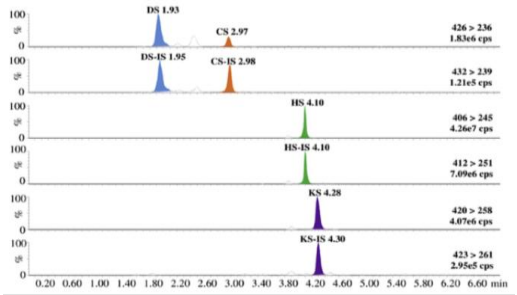
2. Differentiation of GAGs in urine

Electrophoresis of GAGs



Vairo et al. The Application of Clinical Genetics 2015:8 245-255

LC-MS/MS traces of GAGs after hydrolysis → DS, CS, HS, KS



Mathis et al. JIMD report, 2024;65 116-123.
Auray-Blais et al. Analytica Chimica Acta 2016:236 139-148

Detection of non-reducing ends of endogenous oligosaccharides with LC-MS

HNAc (1S)	HN-UA (1S)
HNAc (2S)	UA-HN-UA (1S)
HNAc-UA (1S)	$(HNAc\text{-}UA)_2$ (1S)
UA-HNAc (1S) early RT	$(HNAc\text{-}UA)_2$ (2S)
UA-HNAc (1S) late RT	$(Hex\text{-}HNAc)_2$ (2S)
	(HN-UA)2-HNAc (S)

Saville et al. Genet Med. 2019;21:753-757
Lawrence et al. Nat Chem Biol. 2012;8:197-204.

DS: Dermatan sulfate
CS: Chondroitoin sulfate
HS. Heparin/Heparan sulfate
KS: Keratan sulfate

A – MPS VI

Common sample

Mucopolysaccharidosis (MPS) and Glycosaminoglycans (GAGs)

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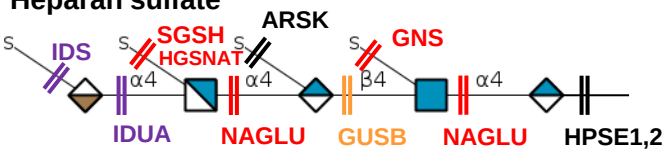
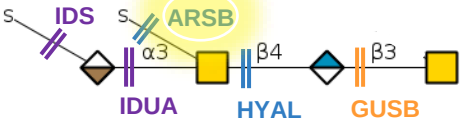
NAGZ		-		H

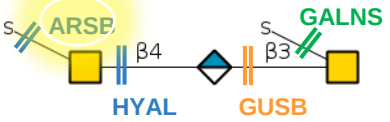
Heparan sulfate



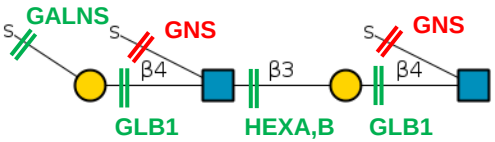
Dermatan sulfate



Chondroitin sulfate



Keratan sulfate



Hyaluronan

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GlcNAc: N-acetyl-D-glucosamine

Sample A – MPS VI

Clinical information

- 15-year-old boy. Dysmorphic features, scoliosis, size -1.5 SD, normal intellectual development. Under treatment.

Diagnosis

- Mucopolysaccharidosis type VI

Analytical

- Detection of increased dermatan sulfate or differentiation profile compatible with MPS type VI was scored two points (10/21 labs).
- Detection of increased total MPS or differentiation profile indicative of an incorrect MPS type was scored one point (11/21 labs).

Sample A – MPS VI

Interpretation

- Mucopolysaccharidosis type VI as main diagnosis was scored two points (11/21 labs).
- Other or unspecified types of mucopolysaccharidosis, or diagnosis based on clinical presentation, were scored one point (10/21 labs).

Overall impression

- Analytical and interpretative proficiencies were 74% and 76%, respectively.
- All laboratories reported MPS as the main diagnosis; however, only half correctly identified the specific MPS type. 4/21 laboratories did not perform MPS differentiation.

MPS VI in previous circulation

Swiss	2008	2008-C		LSD	MPSVI	Mucopolysaccharidosis type VI (Maroteaux-Lamy)		70	65	72
Swiss	2022	C		LSD	MPSVI	Mucopolysaccharidosis type VI		84	84	84

D – MPS III

Mucopolysaccharidosis (MPS) and Glycosaminoglycans (GAGs)

Abb	Enzyme name	MPS type	Disease name	Degrada-tion of:
IDUA	L-iduronidase	I	Hurler-Scheie syndrome	HS, DS
IDS	iduronate 2-sulfatase	II	Hunter syndrome	HS, DS
SGSH	N-sulfoglucosamine sulfohydrolase	IIIA	Sanfilippo A	HS
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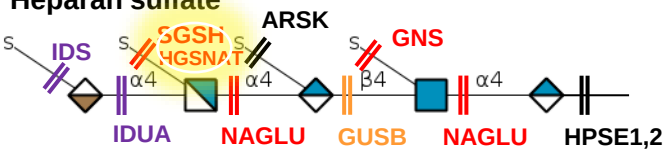
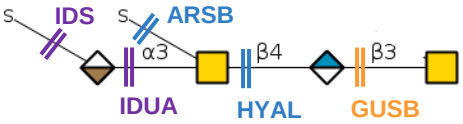
GALNS	N-acetylgalactosamine-6-sulfatase	IVA	Morquio syndrome A	KS, CS
GLB1	beta-galactosidase	IVB GM1	Morquio syndrome B GM1	KS
HEXA,B	Hexosaminidase A, B	GM2	Tay-Sachs disease (def HEXA) Sandhoff disease (def HEXA+B)	KS
NAGZ		-		H

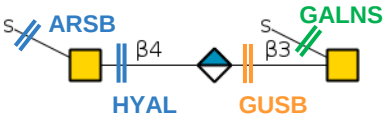
Heparan sulfate



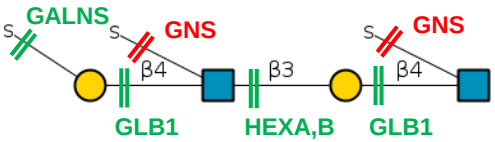
Dermatan sulfate



Chondroitin sulfate



Keratan sulfate



Hyaluronan

KEGG PATHWAY: Glycosaminoglycan degradation - Homo sapiens (human) (genome.jp)

IdoA: L-iduronic acid
GlcA: D-glucuronic acid
Gal: D-galactose
GlcN: D-glucosamine
GalNAc: N-acetyl-D-galactosamine
GlcNAc: N-acetyl-D-glucosamine

Sample D – MPS III

Clinical information

50-year-old woman with retinitis pigmentosa but otherwise in good general condition. Slight scoliosis. Genetically confirmed: compound-heterozygous for two variants in HGSNAT gene.

Diagnosis

- Mucopolysaccharidosis type IIIC

Analytical

- Increased heparan sulfate or differentiation profile compatible with MPS III was scored two points (10/21 labs). Increased total MPS was scored one point (1 lab).

Sample D – MPS III

Interpretation

- Mucopolysaccharidosis type III as main diagnosis was scored two points (9/21 labs). Other types of mucopolysaccharidosis or unspecified or diagnosis according to the clinical presentation were scored one point (2 labs).

Overall impression

- Relatively poor overall proficiency with 49%. Only half of the labs detected increased heparan sulfate and concluded to MPS type III. 6 labs reported normal concentration of MPS, whereas 7 labs reported elevated MPS concentration. The other labs did not report any result for total MPS.

MPS III in previous circulation

Swiss	2008	2008-F	Common	LSD	MPSIII	Mucopolysaccharidosis type IIIA (Sanfilippo)		67	65	70
Swiss	2019	2019-F		LSD	MPSIII	Mucopolysaccharidosis type IIIA (Sanfilippo)		80	80	80

C – MNGIE

Sample C – MNGIE

Clinical information

33-year-old male with leukoencephalopathy and muscular hypotonicity.

Further: The urine was obtained from a 33 year old patient with abnormal MRI scan, leukoencephalopathy, demyelinating neuropathy, muscular hypotonicity and wasted appearance. The diagnosis was confirmed by mutation analysis.

Diagnosis

- Thymidine phosphorylase deficiency; mitochondrial neurogastrointestinal encephalopathy syndrome (MNGIE)

Analytical

- Detection of increased thymidine and/or deoxyuridine was scored 2 points (17/21 labs).

  Thymidine range: 14-50 mmol/mol creat.

  Deoxyuridine range: 35-66 mmol/mol creat.
- Detection of increased uracil and/or thymine was scored 1 point

Sample C – MNGIE

Interpretation

- Thymidine phosphorylase deficiency (MNGIE) as the main diagnosis was scored two points (16/21 labs). Diagnoses of dihydropyrimidine dehydrogenase deficiency (DPD) or dihydropyrimidinase deficiency (DHP) were scored one point (4/21 labs).

Overall impression

- Overall, a mitigated proficiency of 81% was observed. Analytical proficiency was of 76% with 6 labs failing to detect one or both specific metabolites. Interpretative proficiency was of 86%. 4/21 labs concluded incorrectly to DPD or DHP deficiencies. One lab did not identify a specific diagnosis.

MNGIE in previous circulation

Scheme	Year	Sampl	Comm	Group	Abb	Diagnosis	Educational	A	I	Total
Swiss	2010	2010-A		PP	MNGIE	MNGIE / Thymidine phosphorylase deficiency	Educational	50	50	50
Swiss	2019	2019-D		PP	MNGIE	MNGIE syndrome		88	78	83

B – Barth syndrome

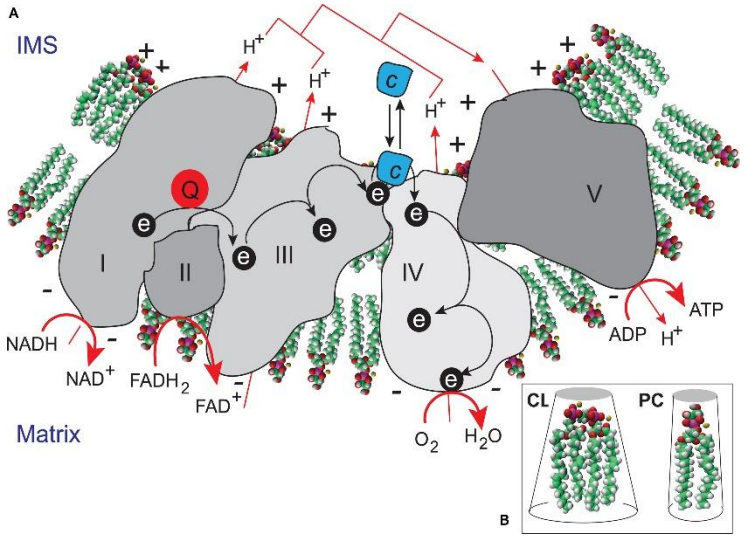
Sample B – Barth syndrome

Clinical information

41-year-old man with cardiomyopathy; neutropenia in childhood.

Diagnosis

- 3-Methylglutaconic aciduria in association with Barth Syndrome (OMIM 302060).



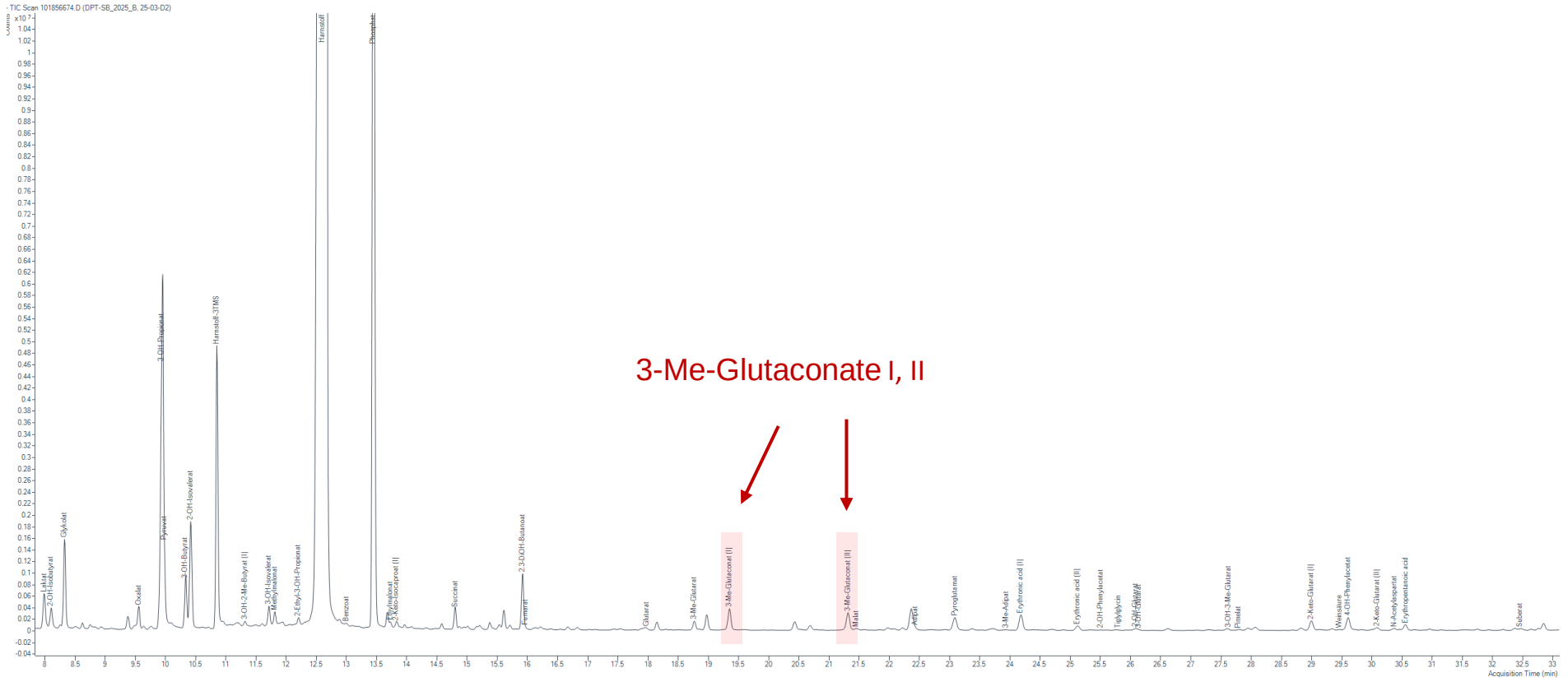
Mutations in the tafazzin gene (TAZ) cause Barth syndrome (X-linked disorder). The enzyme tafazzin act as acyltransferase for cardiolipin biosynthesis. **Cardiolipins** are complex lipid found inside the mitochondria membrane and intimately connected with the electron transport chain.

https://doi.org/10.3389/fgene.2015.00359

Sample B – Barth syndrome

Analytical

Detection of increased 3-methylglutaconic acid was scored 2 points (21/21 labs).



Sample B – Barth syndrome

Interpretation

- Barth syndrome or 3-methylglutaconic aciduria was scored two points (21/21 labs).

Overall impression

- Excellent overall proficiency of 100%.

Barth syndrome in previous circulation

Scheme	Year	Sampl	Comm	Group	Abb	Diagnosis	Educational	A	I	Total
Swiss	2013	2013-D		OS	3MGA	3-Methylglutaconic aciduria (Barth syndrom)		68	68	68
Swiss	2022	A		OS	3MGA	3-Methylglutaconic aciduria (Barth syndrom)		100	98	99

DD: 3-Methylglutaconic aciduria (simplified)

Group	Patogenic mechanism	Disease	Old type	Gene	Protein & funtion
Primary 3-MGA-uria	Organic aciduria	3-MG-CoA hydratase deficiency	I	*AUH* *HMGCL*	3-MG-CoA hydratase, Leucine catabolism HMG-CoA lyase def.
Secondary 3-MGA-uria	Defective phospholipid remodelling	TAZ defect , Barth syndrome	II	*TAZ*	Tafazzine, cardiolipin remodelling
		SERAC1 defect MEGDEL syndrome	IV	*SERAC1*	SERAC1, phosfatidil-glicerol remodelling, cardiolipin composición
	Mitochondrial membrane associated disorder	OPA3 defect, Costeff syndrome	III	*OPA3*	OPA3, respiratory chain protective function
		DNAJC19 defect, DCMA syndrome	V	*DNAJC19*	DNAJC19, mitochondrial protein import
		TMEM70 defect	IV	*TMEM70*	TMEM70, complex V assembly , mitochondrial membrane insertion
	Unknown	NOS 3-MGA-uria	IV	Unknown	Unknown

E – MCAD

Scheme	Year	Sample	Comm	Group	Abb	Diagnosis	Educational	A	I	Total
Swiss	2006	2006-C		OS	MCAD	MCAD deficiency		95	95	96

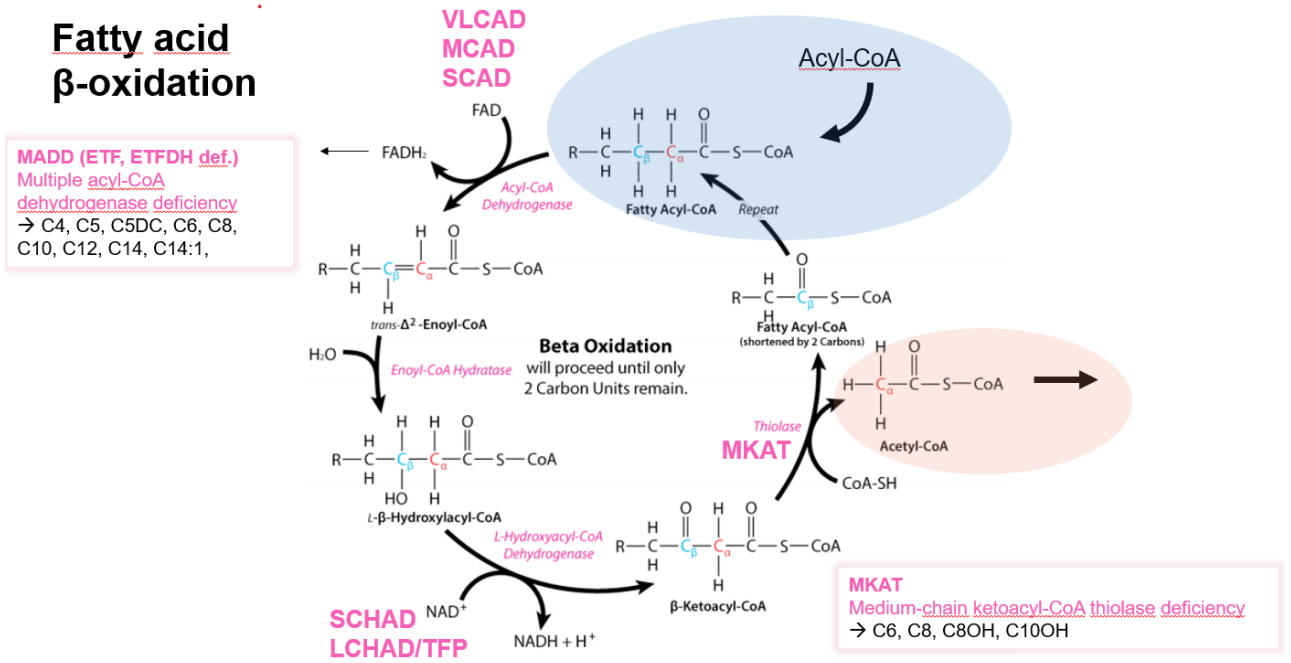
Sample E – MCAD

Clinical information

35-year-old female patient with episode of hypoglycaemia after prolonged fasting at 23 years of age. Genetically confirmed: pathogenic variants c.1102_1105delAGTT/p.(Ala396Leufs*18) and c.985A>G/p.(Lys329Glu) in ACADM gene.

Diagnosis

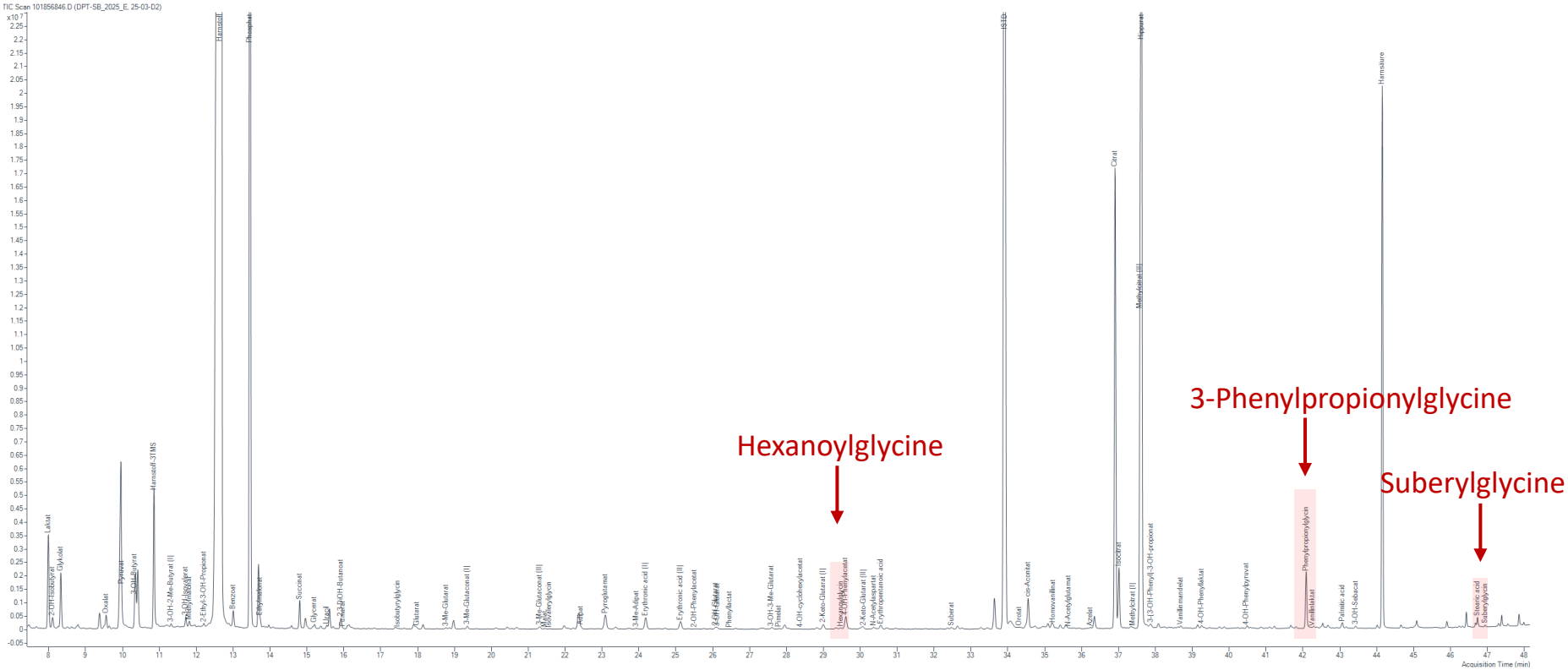
- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency



Sample E – MCAD

Analytical

- Increased concentration of at least one metabolite specific for MCAD deficiency (hexanoylglycine, suberylglycine and 3-phenylpropionylglycine) was scored two points (21/21 labs).



Hexanoylglycine: range: 2-5.5 mmol/mol creat.
Suberylglycine: range: 1-4.5 mmol/mol creat.
3-phenylpropionylglycine: range: 40-110 mmol/mol creat.

Sample E – MCAD

Interpretation

- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency as main diagnosis was scored two points (21/21 labs).

Overall impression

- Excellent overall proficiency of 100%.

MCAD in previous circulation

Scheme	Year	Sampl	Comm	Group	Abb	Diagnosis	Educational	A	I	Total
Swiss	2006	2006-C		OS	MCAD	MCAD deficiency		95	95	96

F – MADD

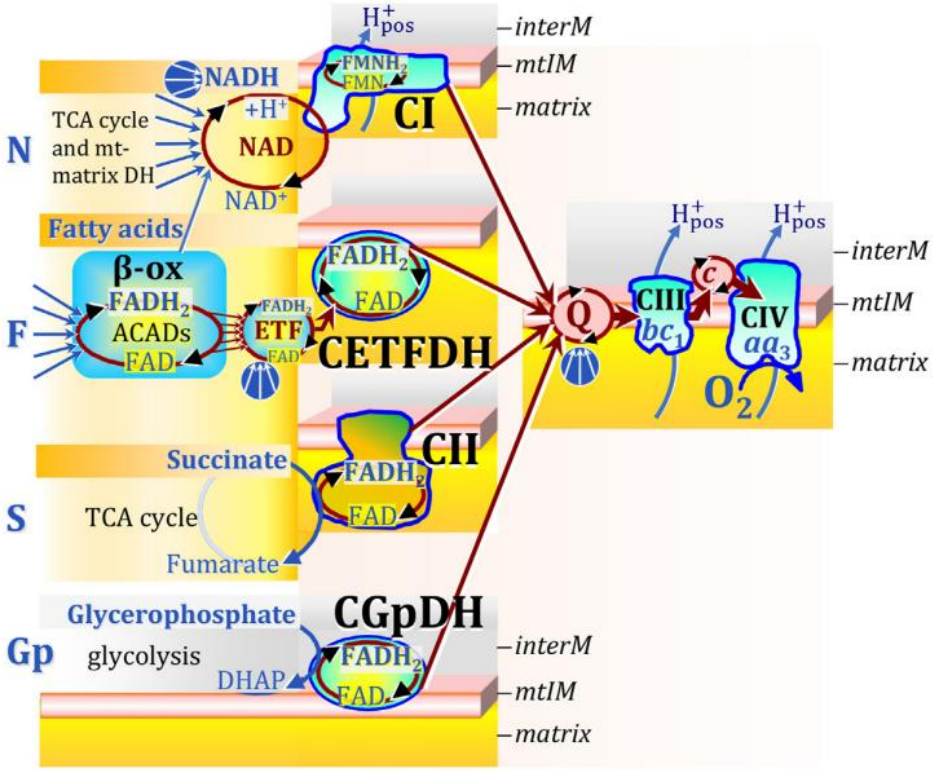
Sample F – MADD

Clinical information

34-year-old female patient presented initially with Rey like syndrome. Currently under therapy, presenting mild ataxia but normal cognition. The therapy consist of fat-reduced diet combined with L-carnitine and riboflavin supplementation.

Diagnosis

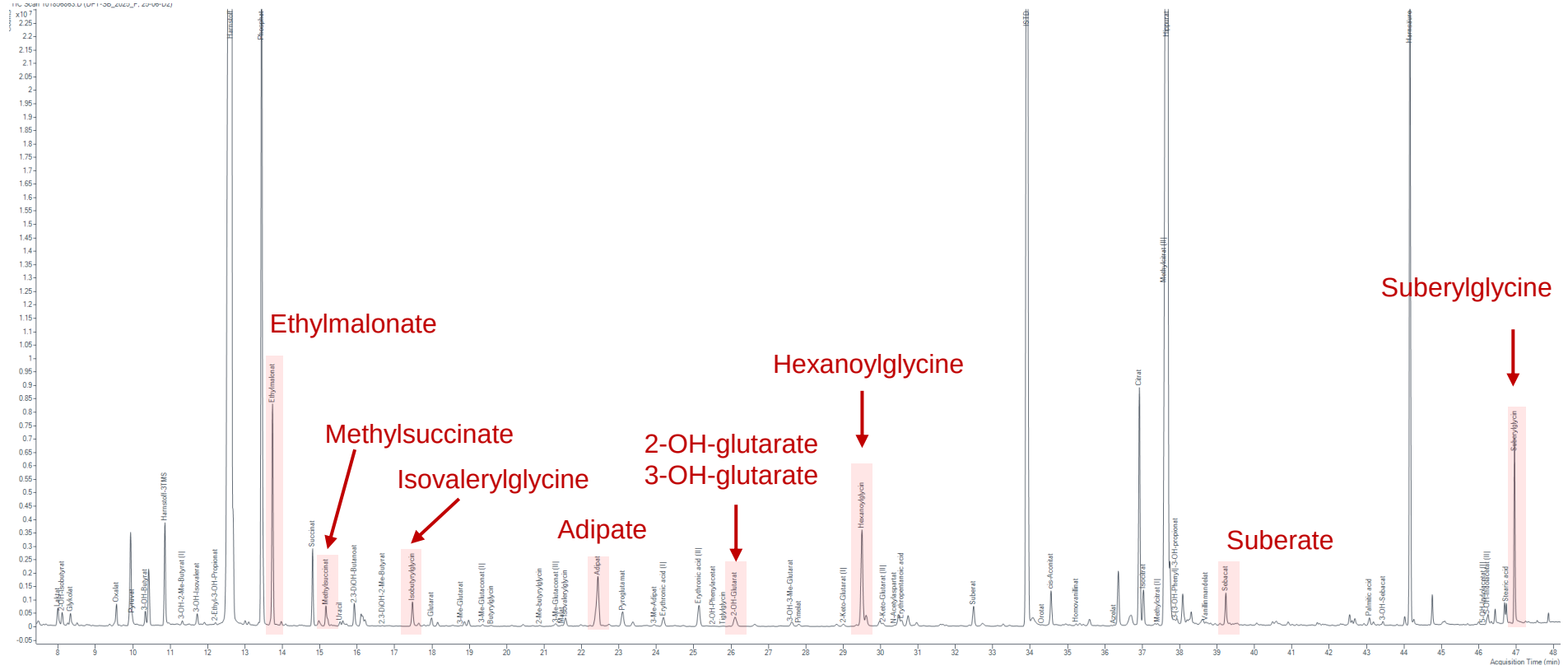
- Multiple acyl-CoA dehydrogenase deficiency (MADD). Electron transfer flavoprotein-ubiquinone oxidoreducatse (ETF-QO) deficiency.



Sample F – MADD

Analytical

- Increased concentrations of at least three metabolites specific for MADD (ethylmalonic acid, glycine-conjugates, dicarboxylic acids) were scored two points (21/21 labs).



Ethylmalonate: range: 59-153 mmol/mol creat.
Hexanoylglycine: range: 30-156 mmol/mol creat.

Sample F – MADD

Interpretation

- Multiple acyl-CoA dehydrogenase deficiency (MADD) as main diagnosis was scored two points (21/21 labs).

Overall impression

- Excellent overall proficiency of 100%.

MADD in previous circulation

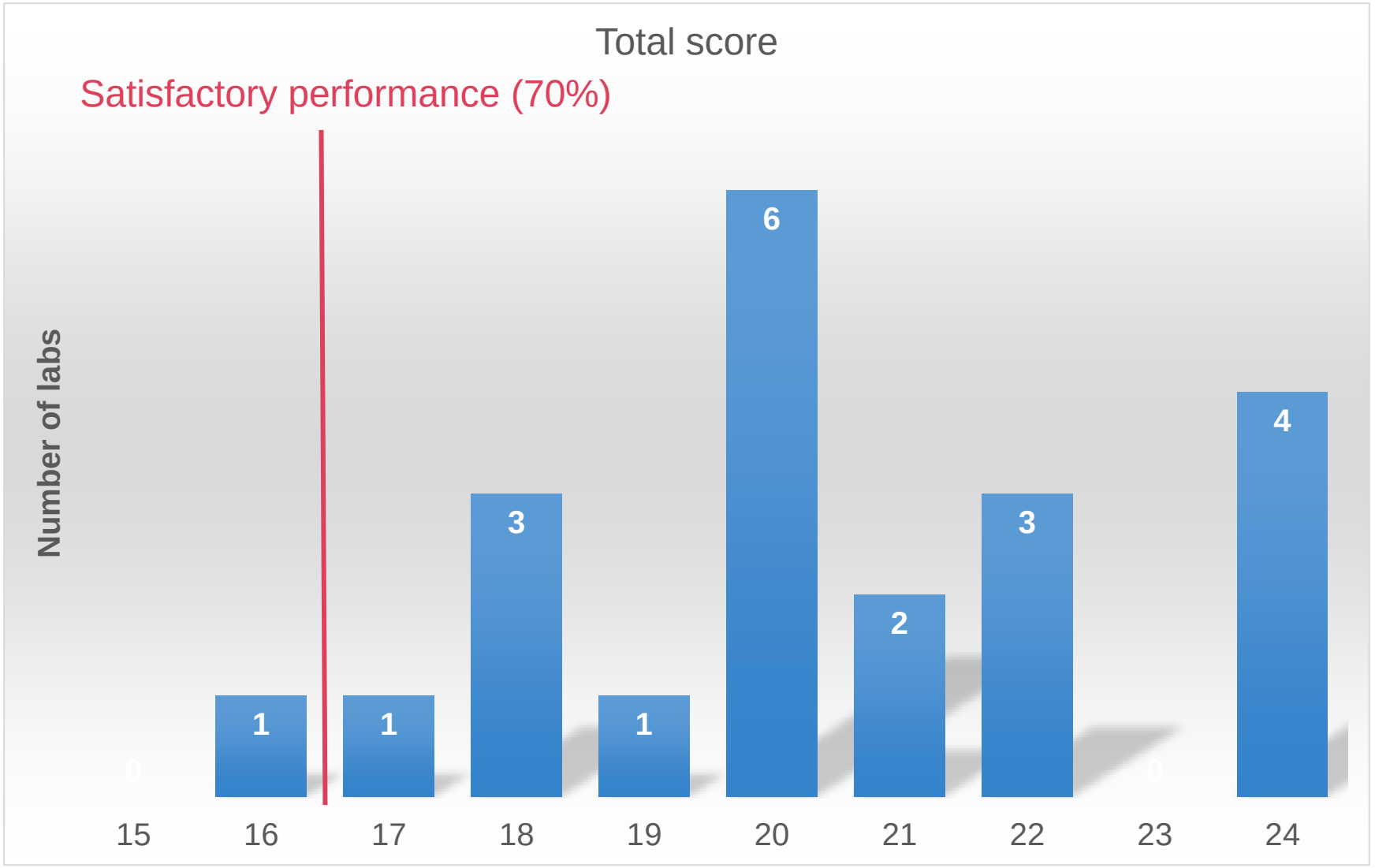
Swiss	2023	F		OS	MADD	Multiple acyl-CoA dehydrogenase deficiency (MADD)		71	71	71

Total scores and proficiency

	A - MPS VI			B-Barth			C-MNGIE			D-MPS III			E-MCAD			F-MADD			Σ
	A	I	Σ	A	I	Σ	A	I	Σ	A	I	Σ	A	I	Σ	A	I	Σ	
1	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	24
2	1	1	2	2	2	4	2	1	3	0	0	0	2	2	4	2	2	4	17
3	1	1	2	2	2	4	1	1	2	0	0	0	2	2	4	2	2	4	16
4	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	24
5	1	1	2	2	2	4	1	1	2	2	2	4	2	2	4	2	2	4	20
6	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	24
7	1	2	3	2	2	4	2	2	4	1	0	1	2	2	4	2	2	4	20
8	2	2	4	2	2	4	2	2	4	0	1	1	2	2	4	2	2	4	21
9	1	1	2	2	2	4	2	2	4	0	0	0	2	2	4	2	2	4	18
10	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	24
11	1	1	2	2	2	4	2	2	4	0	0	0	2	2	4	2	2	4	18
12	2	2	4	2	2	4	2	2	4	0	0	0	2	2	4	2	2	4	20
13	1	1	2	2	2	4	1	0	1	2	2	4	2	2	4	2	2	4	19
14	2	2	4	2	2	4	2	2	4	0	0	0	2	2	4	2	2	4	20
15	2	2	4	2	2	4	2	2	4	0	0	0	2	2	4	2	2	4	20
16	1	1	2	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	22
17	1	1	2	2	2	4	2	2	4	0	0	0	2	2	4	2	2	4	18
18	1	1	2	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	22
19	2	2	4	2	2	4	1	1	2	2	1	3	2	2	4	2	2	4	21
20	2	2	4	2	2	4	2	2	4	0	0	0	2	2	4	2	2	4	20
21	1	1	2	2	2	4	2	2	4	2	2	4	2	2	4	2	2	4	22
%	74	76	**75**	100	100	**100**	90	86	**88**	50	48	**49**	100	100	**100**	100	100	**100**	

Critical errors? To be discussed at the scientific advisory board (SAB) meeting in November

Total scores



Conclusions and future

- Good performance this year
- New SAB deputy: Kathrin Freiburghaus from Bern, Switzerland
- Please try to provide samples !

Thank you !

ERNDiM

Quality Assurance in Laboratory Testing for IEM

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