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DPT Netherlands
Annual meeting 2025

Madrid, 9-10-2025

Agenda

ERNDIM
QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

1) Welcome
2) Modifications, acceptance of the agenda
3) Comments or feedback by participants on annual report 2024
4) DPT scheme: organisation, scoring
5) Discussion of the 2025 samples
6) Planning and organisation of DPT-NL 2026
7) Any other business

DPT NL scheme organisation (1)

Tests required

- Minimal panel for DPT:
  Dip stick, amino acids, organic acids, purines-pyrimidines oligosaccharides and quantitative GAG

- DPT-NL additionally recommends to perform: GAG subtyping, sialic acid, creatine-guanidinoacetate, bile acids, sugars-polyols

- Use of cluster labs is allowed, but must be routine clinical practice and must be declared with result submission

DPT NL scheme organisation (2)

CSCQ

Sample dispatch

Website reporting

- Provide quantitative data as much as possible
- Enter the key metabolites + interpretation in the tables, also if only qualitative/normal
- Please do not enter results or recommendations in “Comments” windows
- Recommendations for further investigations are scored with interpretation
- Advice for treatment is appreciated, but not scored

Interim reports, including scoring

Annual report

Sample contribution:

- 2025: 2/6 samples donated by participants
- 200-300 mL
- 20% discount DPT scheme next scheme year
- please contact scientific advisor

Patient A (common sample)

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

Diagnosis: Mucopolysaccharidosis type VI (OMIM 253200)

Common DPT sample provided by DPT-F

Participants:	18 (DPT NL)
Reports:	18
Correct diagnosis:	16 (MPS VI); 18 (MPS)
Proficiency:	96%

Results discussed by C. Vianey-Saban (Friday morning)

Patient B

Clinical info: Presented at age 1 y with vomiting and lethargy. Current age is 38 y.

Diagnosis: Beta-ketothiolase deficiency (OMIM 203750)

Sample provided by Erasmus MC Rotterdam

Diagnosis confirmed by two mutations in the ACAT1 gene

Participants:	18
Reports:	18
Correct diagnosis:	17
Proficiency:	92%

Previous survey (different sample): 2007-L; 19/19 correct diagnoses

Patient B; tests

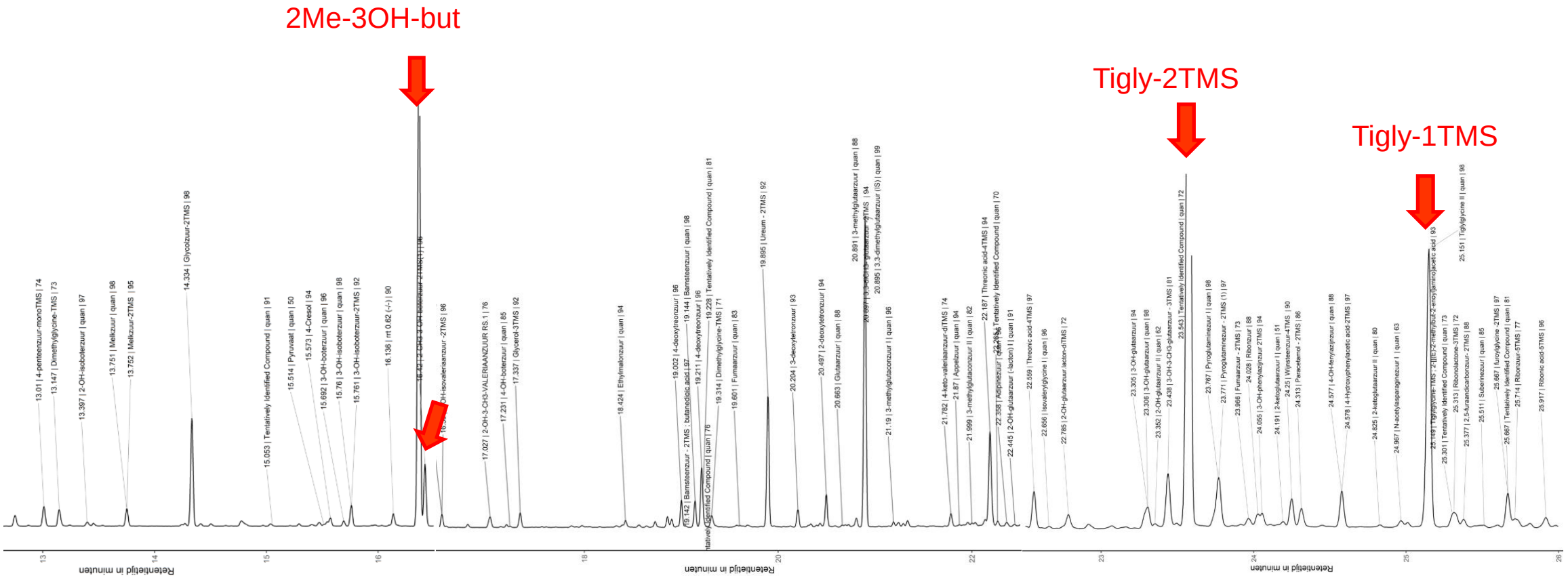
Clinical info: Presented at age 1 y with vomiting and lethargy.
Current age is 38 y.

Creatinine median value 4.3 mmol/L

→ Decompensation/intoxication

Organic acids, amino acids, purines-pyrimidines

Patient B; organic acids



	2-Me-3-OH-butyric acid	Tiglylglycine	
Median	663	366	
Ref values	11-27	<3.8	Poll-The et al Mol Genet Metab. (2004) 81:295-299
	<10	<5	(Rdam)

Patient B; OA and other investigations

Increased level of:

	nr of labs	comment
Tiglylglycine	18	
2-Me-3-OH-butyric acid	15	
2-Me-acetoacetate	3	2 labs: normal
C5:1-carnitine	3	useful?

Patient B; tiglylglycine

CLIN. CHEM. 40/10, 1879–1883 (1994) • Molecular Pathology

Tiglylglycine Excreted in Urine in Disorders of Isoleucine Metabolism and the Respiratory Chain Measured by Stable Isotope Dilution GC-MS

Michael J. Bennett,[1,4] Susan Powell,[1] Daniel J. Swartling,[2] and K. Michael Gibson[3]

Tiglylglycine (TG), an intermediate product of the catabolism of isoleucine, is increased in the urine of patients with β-ketothiolase deficiency or with disorders of propionate metabolism. It is also implicated as a useful

metabolically indistinguishable from secondary lactic acidemia due to tissue anoxia (5).

The fact that TG is not commercially available has hindered the development of an accurate quantitative assay.

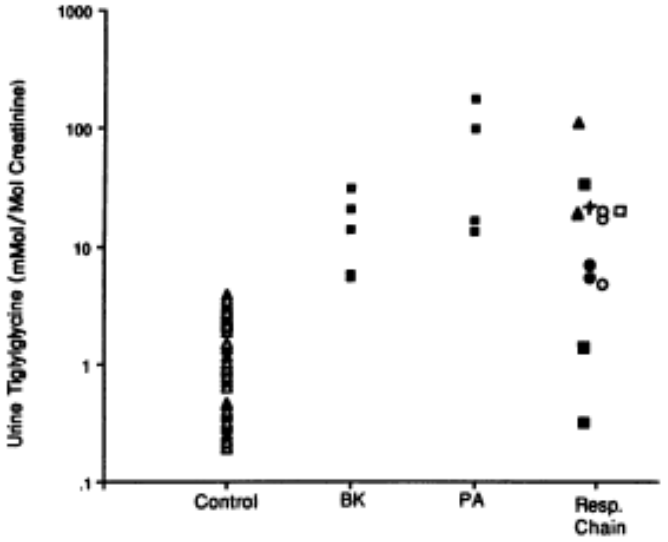


Fig. 5. TG concentrations (mmol/mol of creatinine) in urines from control subjects and patients with β-ketothiolase deficiency (BK), propionic acidemia, (PA), and disorders of the respiratory chain.

The patients with respiratory chain defects correspond to the patients in Table 1: (▲) patient 3; (+) patient 4; (□) patient 5; (■) patient 6; (○) patient 7; (●) patient 8.

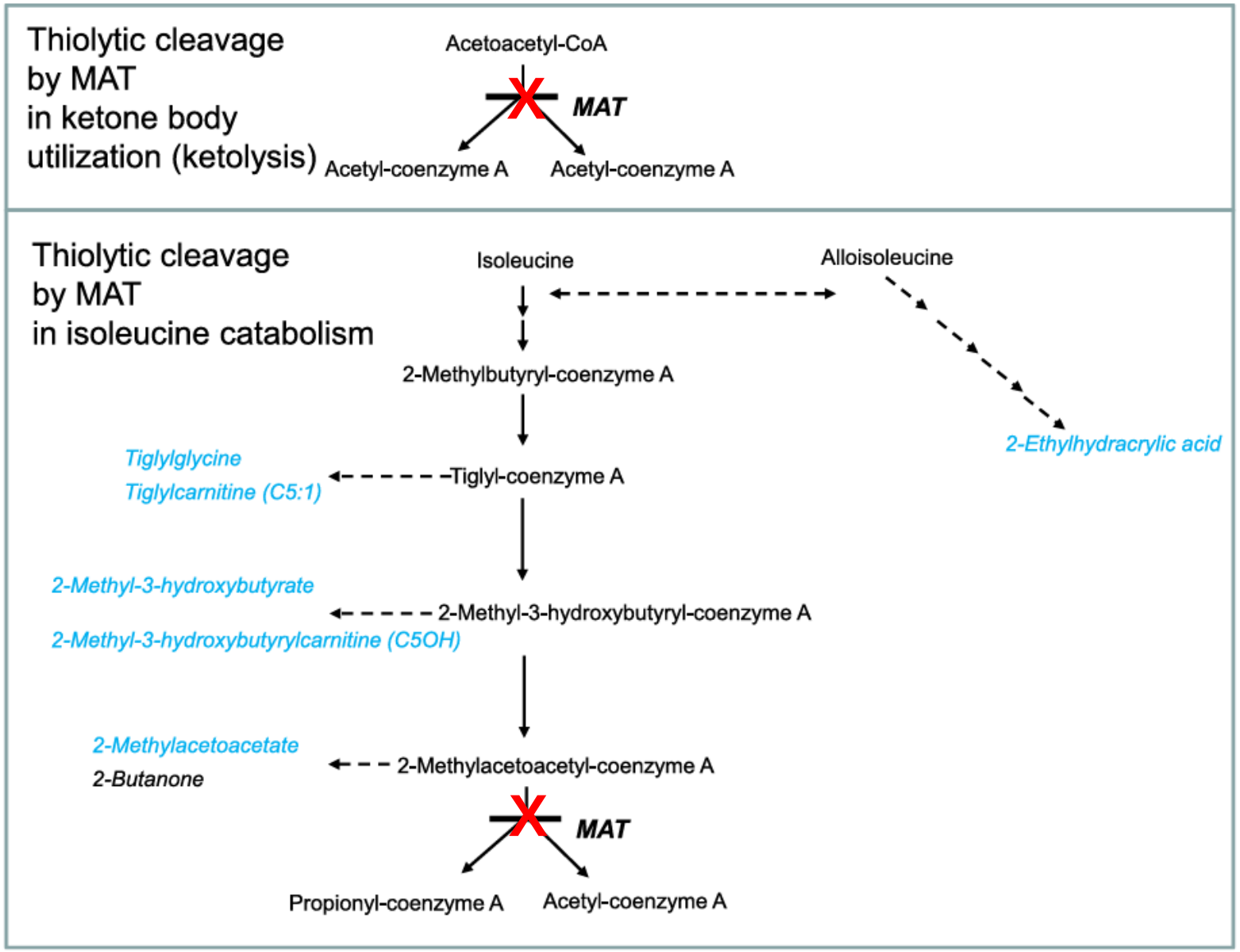
Tiglylglycine? …

Check beta-ketothiolase def
MHBD
propionic acidemia
Resp. chain defects?

Patient B; Interpretation

	Diagnosis		
	Most likely	**Alternative**	**Comment**
beta-Ketothiolase def	14	3	
MHBD def	3	9	2 labs: unlikely (symptoms)
3MCC	1	-	tigly identified as 3MCG?
MCT1 def	1	-	(in addition to BKT)
Propionic aciduria	-	1	different metabolite pattern

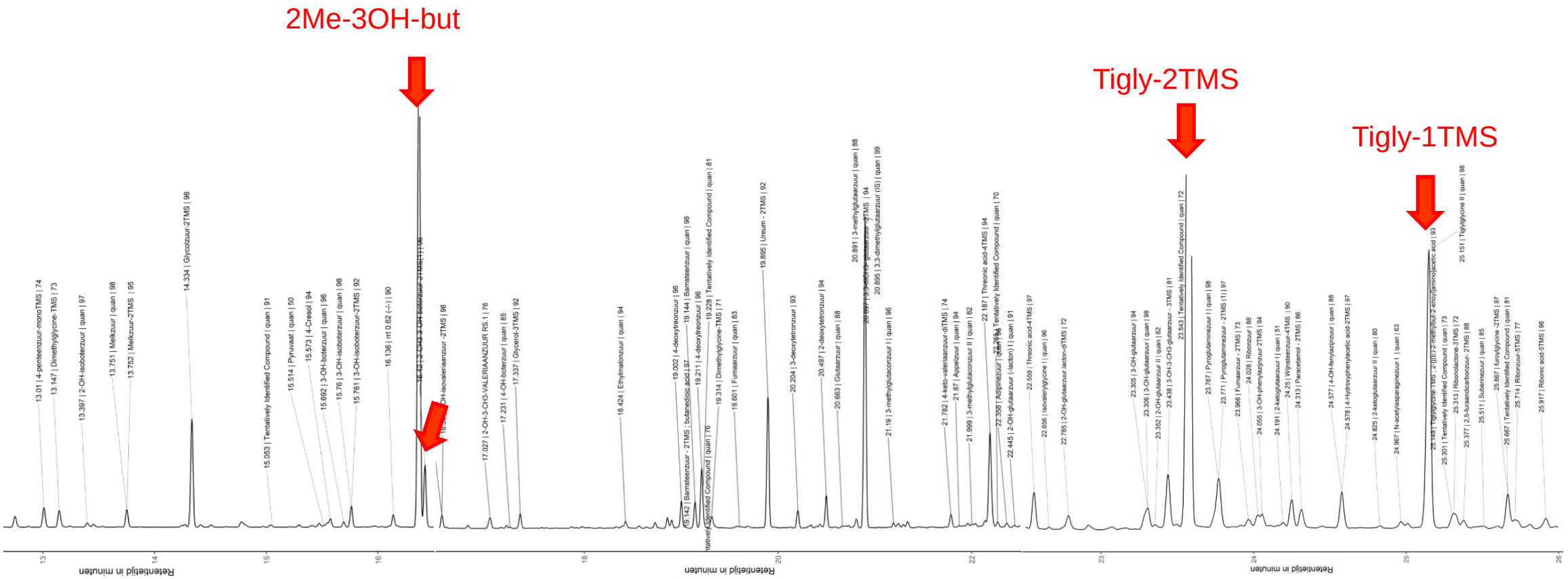
Beta-Ketothiolase def; biochemistry, clinical



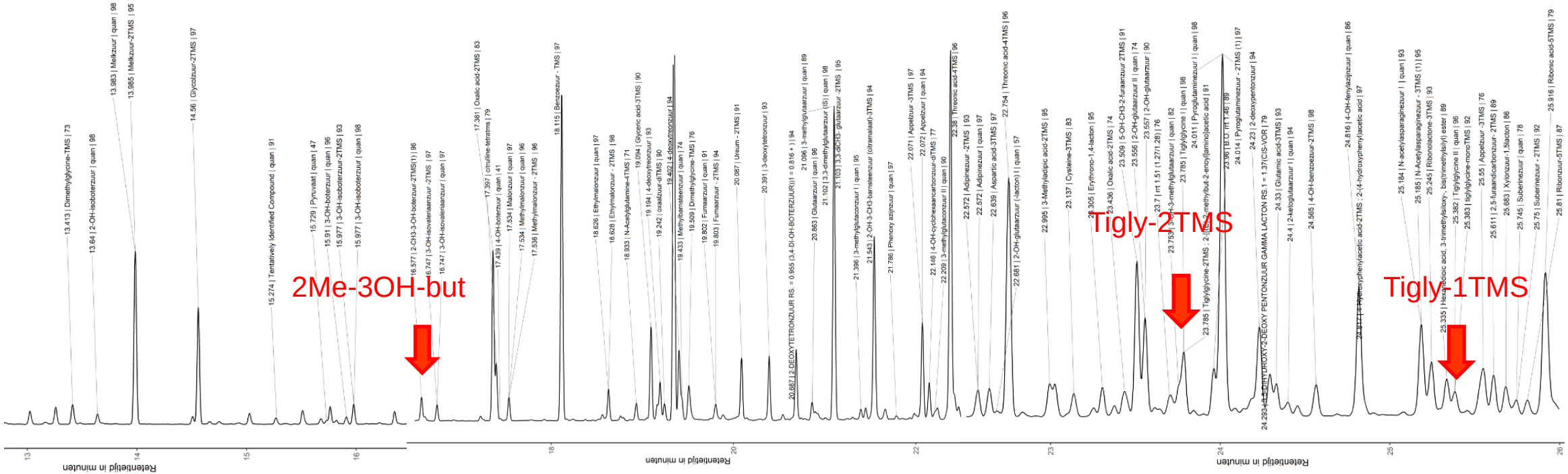
- Acute metabolic decompensation
- Infants/toddlers
- Seldom neonatally (3.4%)
- Mostly normal development (77%)

Grünert and Sass, Orphanet Journal of Rare Diseases (2020) 15:106

Patient B; organic acids



DPT 2024-B; MHBD deficiency, organic acids



How to differentiate MHBD and BKT

- Clinical symptoms
- Presence of 2-Me-Acetoacetate (not in DPT samples)
- 1 or 2 enantiomers of MHB
- Concentrations of metabolites?

Beta-Ketothiolase def; biochemistry, clinical

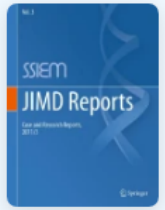
Home > JIMD Reports – Case and Research Reports, 2011/3 > Chapter

Three Japanese Patients with Beta-Ketothiolase Deficiency Who Share a Mutation, c.431A>C (H144P) in *ACAT1*

Subtle Abnormality in Urinary Organic Acid Analysis and Blood Acylcarnitine Analysis Using Tandem Mass Spectrometry

Case Report | Chapter | First Online: 01 January 2011

pp 107–115 | Cite this chapter



JIMD Reports – Case and Resear
Reports, 2011/3

Even during severe crises, C5-OH and C5:1 were within normal ranges in their blood acylcarnitine profiles and trace amounts of tiglylglycine and small amounts of 2-methyl-3-hydroxybutyrate were detected in their urinary organic acid profiles.

PEDIATRICS

Content | Authors/Reviewers | Collections | Multimedia | Blogs | Sub

CASE REPORTS | JULY 01 2011

Siblings With Mitochondrial Acetoacetyl-CoA Thiolase Deficiency Not Identified by Newborn Screening

Kyriakie Sarafoglou, MD; Dietrich Matern, MD; Krista Redlinger-Grosse, CGC; Kristi Bentler, MS, RN, PHN, CNP; Amy Gaviglio, CGC; Cary O. Harding, MD; Piero Rinaldo, MD, PhD

Address correspondence to Kyriakie Sarafoglou, MD, Department of Pediatrics, University of Minnesota Medical School, MMC 8404 13-124 PWB, 516 Delaware St SE, Minneapolis, MN 55455. E-mail: saraf010@umn.edu

FINANCIAL DISCLOSURE: The authors have indicated they have no financial relationships relevant to this article to disclose.

Pediatrics (2011) 128 (1): e246–e250.

https://doi.org/10.1542/peds.2010-3918 **Article history**

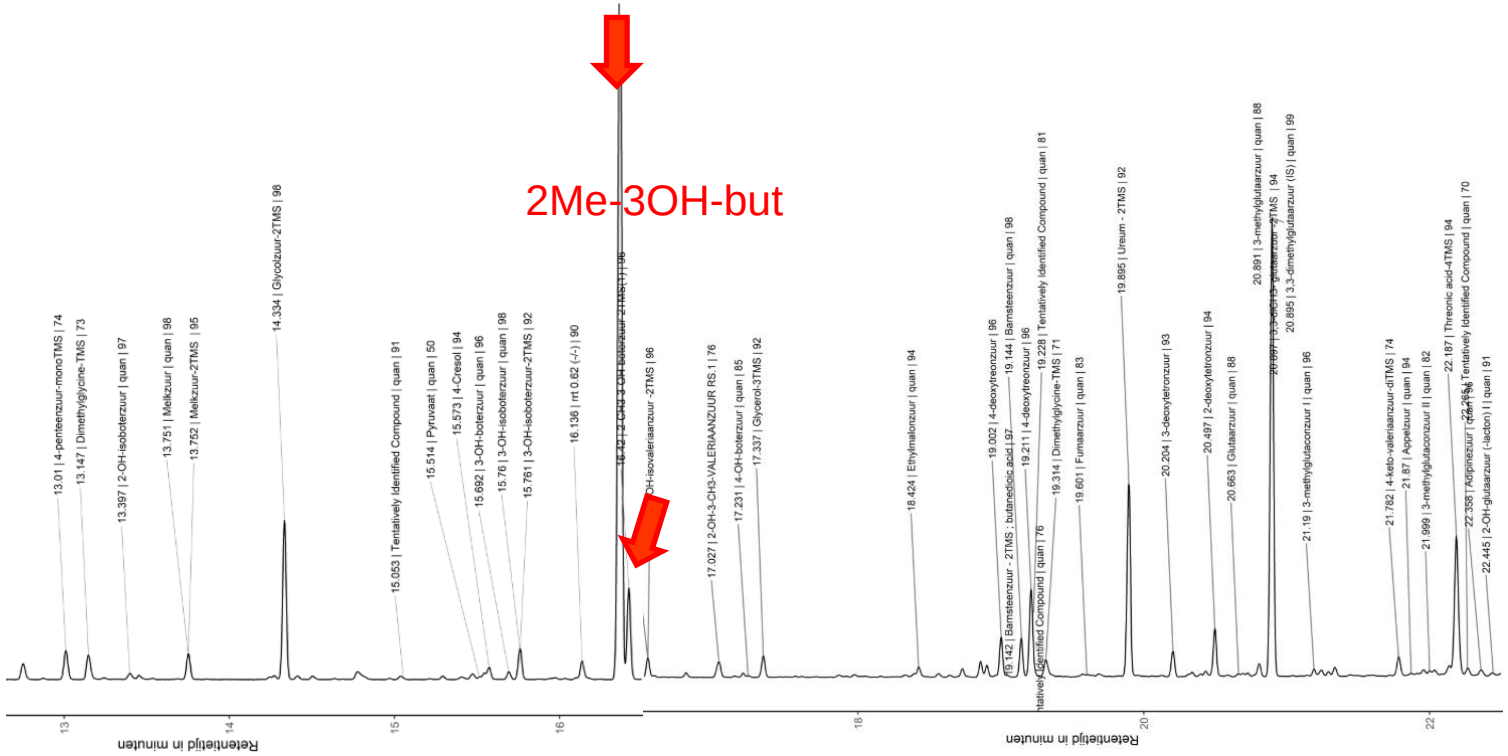
Volume 128, Issue 1

July 2011

Split-Screen | Views | PDF | Share | Tools

Screened for by all state newborn screening (NBS) programs in the United States, mitochondrial acetoacetyl-coenzyme A thiolase (T2), or β-ketothiolase, deficiency is a rare autosomal recessive disorder that causes ketoacidosis and hypoglycemia/hyperglycemia. Outcomes vary from

2 peaks of 2-Me-3-OH-butyric acid



S-2-methylacetoacetate → S-2-Me-R3-OH-butyrate

↕

Tautomerization through ‘Enol’ form

↕

R-2-methylacetoacetate → R-2-Me-R3-OH-butyrate

Patient B; recommendations

Further investigations:

Plasma/DBS acylcarnitine testing
Sequence analysis ACAT1 gene
Enzyme test in lymphocytes/fibroblasts
Repeat analysis of organic acids

Patient B; scoring

Performance item	Result	# points
Analytical	Tiglylglycine elevated 2-Me-3-OH-butyric acid elevated	1 1
Interpretation	Beta-ketothiolase def MHBD def with BKT def as alternative diagnosis	2 1
Potential critical error	Failure to report beta-ketothiolase	

Conclusions

- Clear abnormalities
- Overall proficiency 92%

Patient C

Clinical info: A 6 year old male with dysmorphic features, delayed development, abnormal behaviour and frequent infections. Current age is 29 y.

Diagnosis: Aspartylglucosaminuria (OMIM 208400)

Sample provided by VKS (Dutch patient organization)

Diagnosis confirmed by deficiency of AGA enzyme activity in WBC

Participants:	18
Reports:	18
Correct diagnosis:	17
Proficiency:	97%

Patient C tests

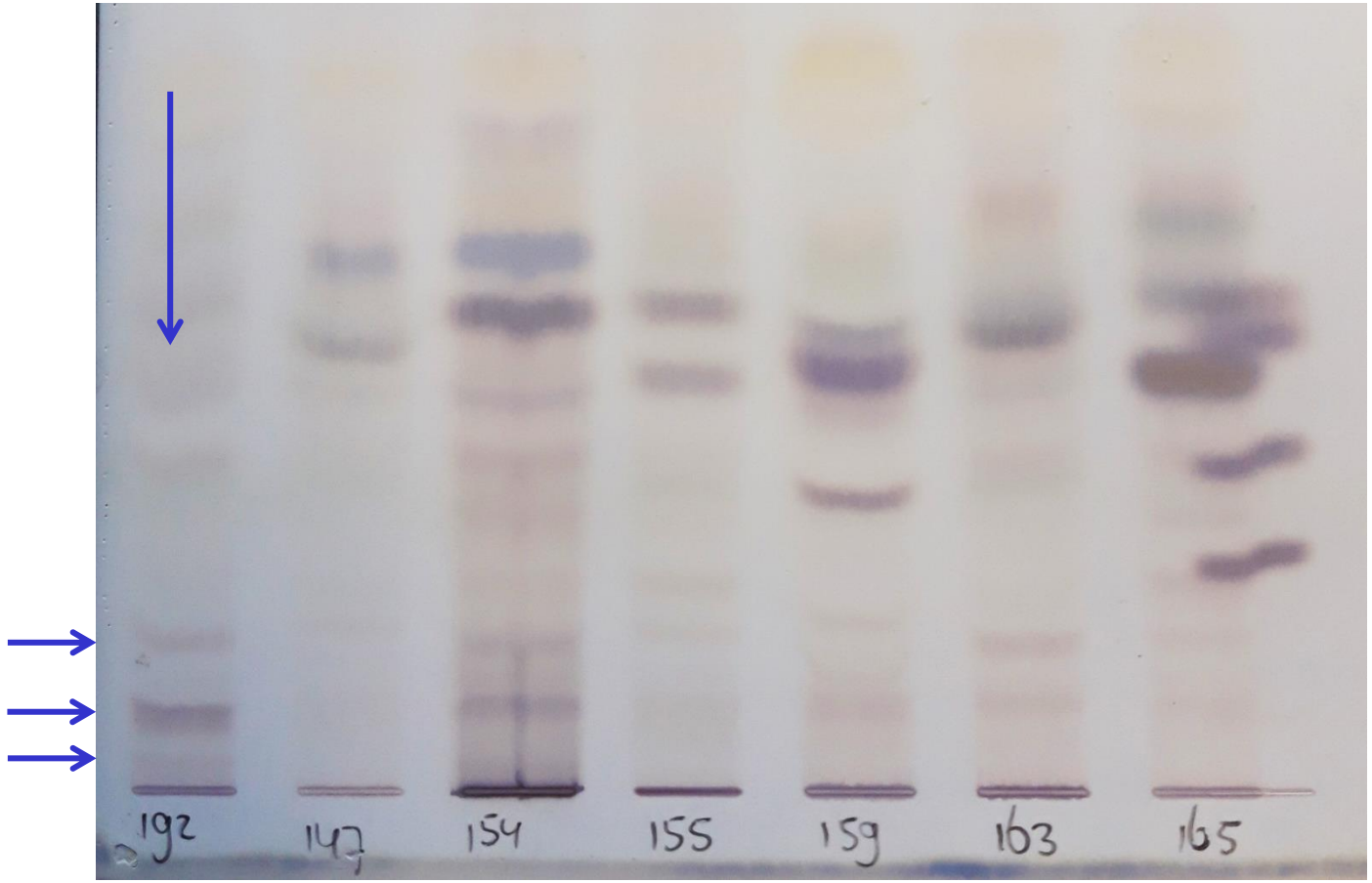
Clinical info: A 6 year old male with dysmorphic features, delayed development, abnormal behaviour and frequent infections. Current age is 29 y.

Creatinine median value 7.7 mmol/L

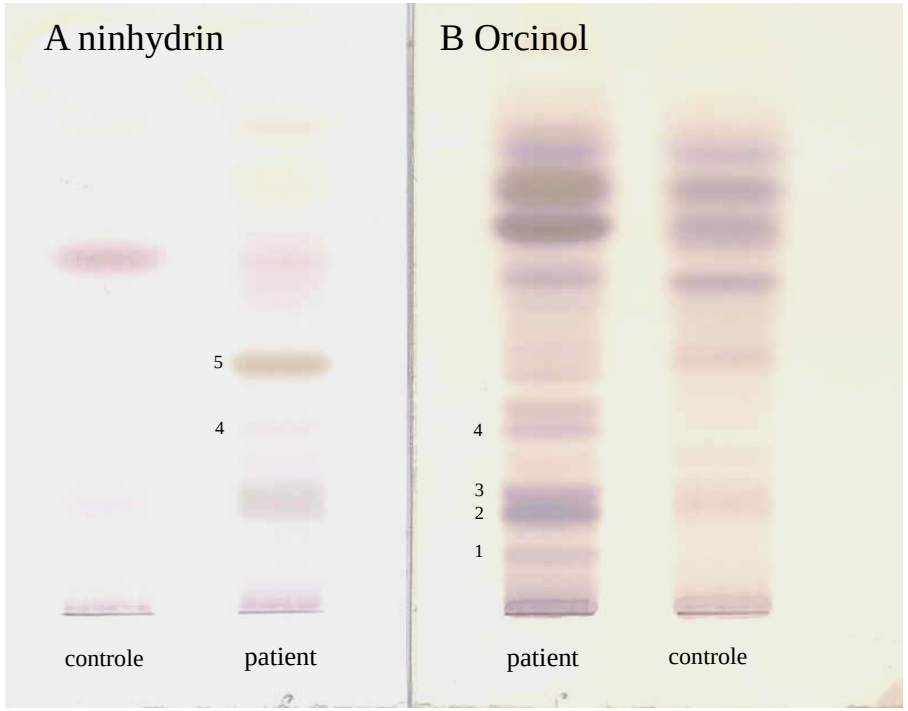
DD: IMD causing intellectual disability + dysmorphic features; storage disorder?

→ Screening incl GAG, oligosaccharides, organic acids, amino acids, purines-pyrimidines

Patient C; oligosaccharides - TLC



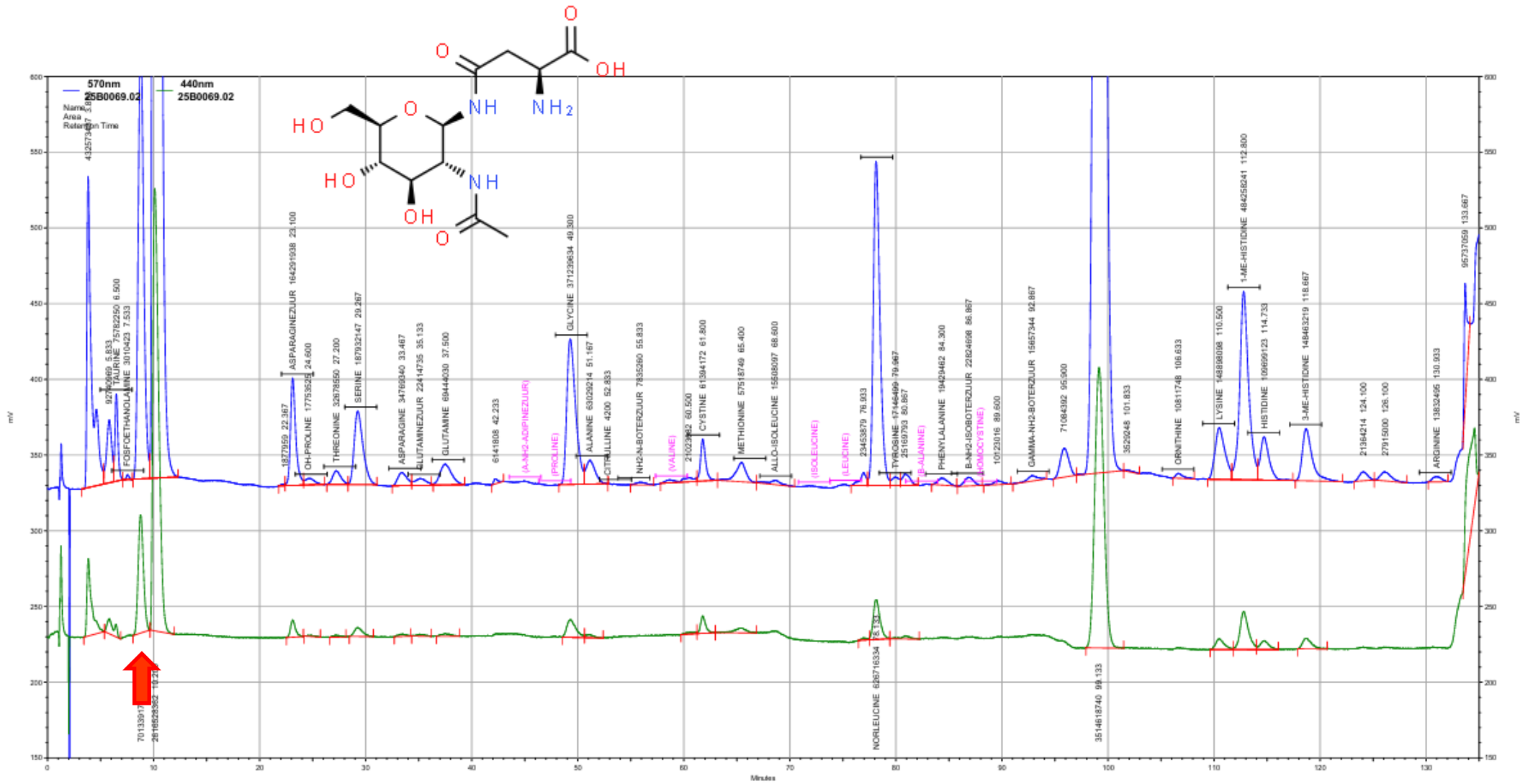
Ninhydrin staining of oligosaccharide TLC



STAINING:	**a.Ninhydrine**	**b.Orcinol**
5. GlcNAc-Asn	positive	
4. (GalGlcNAc)-Asn	positive	positive
3. abnormale band		positive
2. abnormale band		positive
1. abnormale band		positive

Figure kindly provided by L Kluijtmans & M Huigen, Nijmegen NL

Patient C; amino acids, Biochrom 30



Patient C; Results

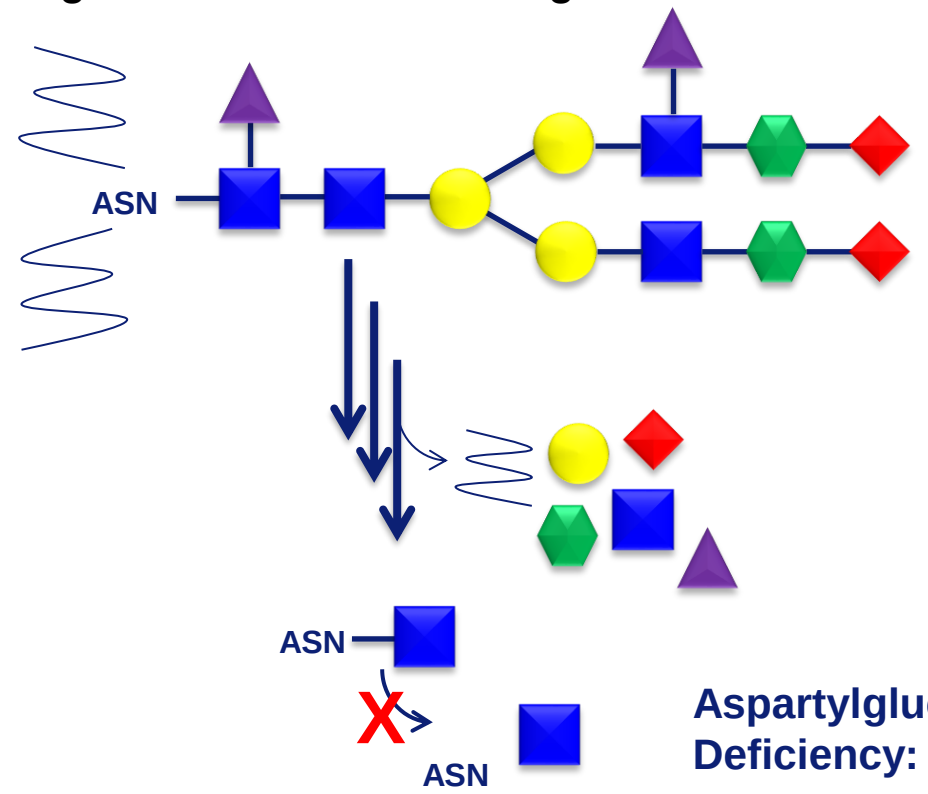
Elevated aspartylglucosamine (AA) Median value: 172 mmol/mol		11
Abnormal oligosaccharide pattern		18
GAG screening	normal	12
	elevated	2
GAG electrophoresis	normal	5 (1 borderline)

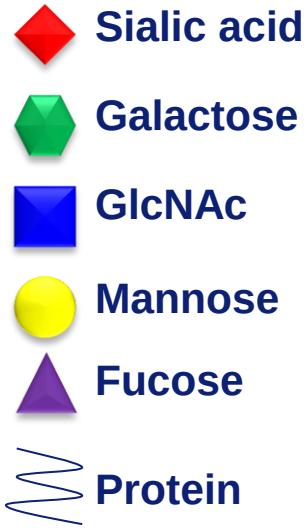
Patient C; interpretation

	Most likely diagnosis	Other possible	Comment
Aspartylglucosaminuria	17	-	
Sialidosis	1	3	
Other LSD	-	3	(GAG borderline; probably secondary)
NGLY1	-	6	usually weaker pattern; only by MS?

Patient C; degradation of oligosaccharides

Degradation of N-linked oligosaccharides





Aspartylglucosaminuria
Deficiency: aspartylglucosaminidase

Frequent in Finnish population (C163S)

No curative treatment

Oligosaccharides by MS

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Bonesso *et al. Orphanet Journal of Rare Diseases* 2014, **9**:19
http://www.ojrd.com/content/9/1/19

ORPHANET JOURNAL OF RARE DISEASES

RESEARCH Ope

Fast urinary screening of oligosaccharidoses MALDI-TOF/TOF mass spectrometry

Laurent Bonesso[1], Monique Piraud[5], Céline Caruba[1], Emmanuel Van Obbergben[1,2,3,4], Raymond Meng and Charlotte

Abstract

Backgroun
disorders du
glycoprotein
accumulatio
disorders are

Research Article

Received: 21 December 2016 Revised: 22 March 2017 Accepted: 25 March 2017 Published online

Rapid Commun. Mass Spectrom. **2017**, *31*, 951–963
(wileyonlinelibrary.com) DOI: 10.1002/rcm.7860

Development of a new tandem mass spectrometry meth urine and amniotic fluid screening of oligosaccharidos

Monique Piraud[1]*, Magali Pettazzoni[1], Louise Menegaut[1,2], Catherine Cail Yann Nadjar[4], Christine Vianey-Saban[1,5] and Roseline Froissart[1,6]

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[4]Département de Neurologie, Hôpital Pitié-Salpêtriére, Assistance Publique des Hôpitaux de Paris, France
[5]Unité INSERM U1060, Laboratoire CarMeN, Université Lyon-1, Lyon, France
[6]UMR 5305 CNRS/UCBL, Lyon, France

RATIONALE: The first step in the diagnosis of oligosaccharidoses is to evidence abnormal oligosaccharides excreted in urine, usually performed by the poorly sensitive but efficient thin layer chromatography (TLC) method. Developing a tandem mass spectrometry (MS/MS) technique could be of great interest to replace TLC.
METHODS: Abnormal underivatized oligosaccharides have been recently studied using matrix-assisted laser desorption/ionization time-of-flight mass spectrometry, allowing the unambiguous identification of oligosaccharidoses. Based on this previous work, we developed an advantageous and efficient liquid chromatography (LC)/MS/MS method using a more common triple quadrupole tandem mass spectrometer for oligosaccharides analysis.

Received: 16 November 2022 | Revised: 20 January 2023 | Accepted: 6 February 2023
DOI: 10.1002/jimd.12597

ORIGINAL ARTICLE

JIMD SSIEM WILEY

Analysis of urinary oligosaccharide excretion patterns by UHPLC/HRAM mass spectrometry for screening of lysosomal storage disorders

Marne C. Hagemeijer[1] | Jeroen C. van den Bosch[1] | Michiel Bongaerts[1] | Edwin H. Jacobs[1] | Johanna M. P. van den Hout[2] | Esmee Oussoren[2] | George J. G. Ruijter[1]

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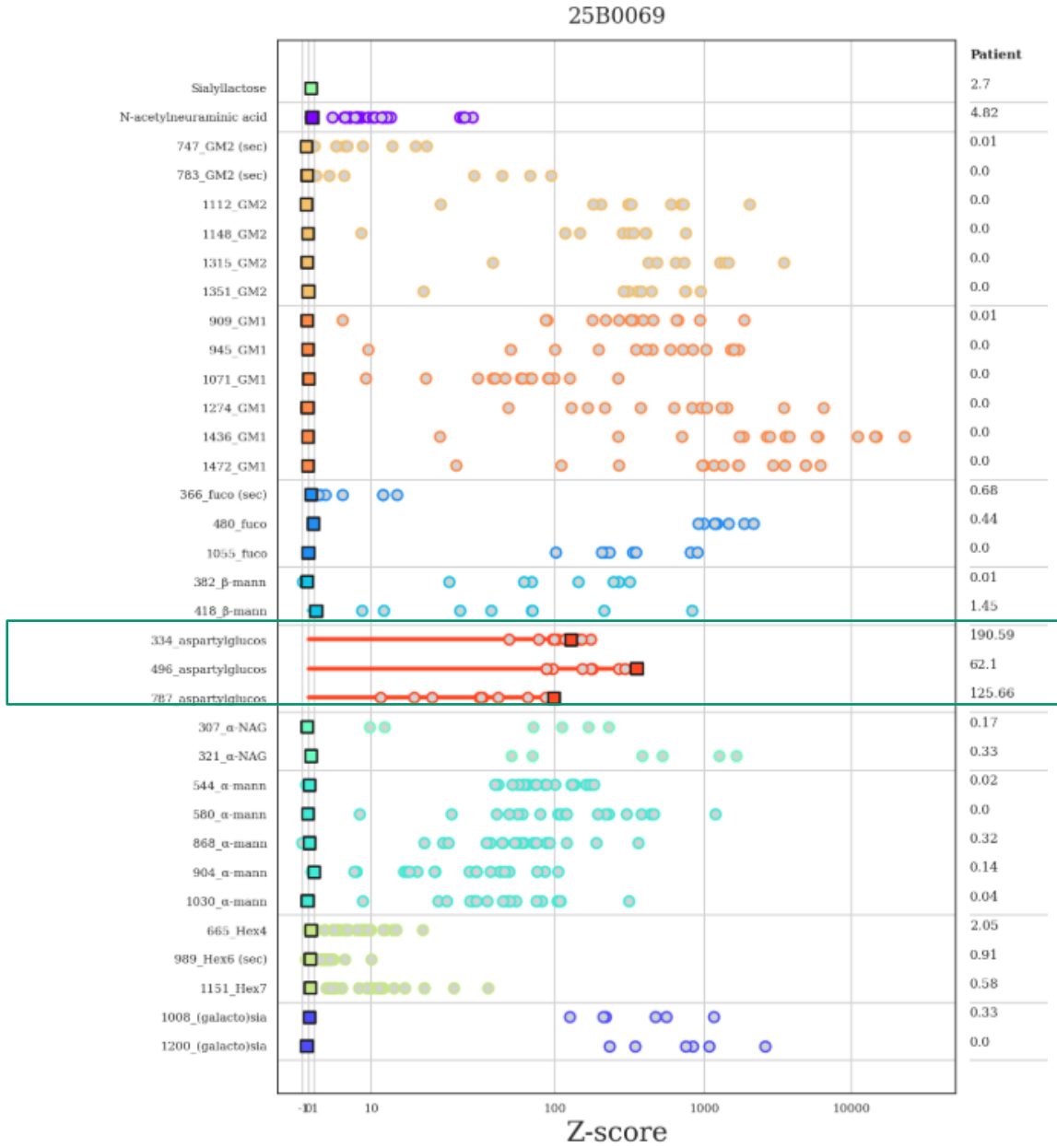
[2]Center for Lysosomal and Metabolic Diseases, Department of Pediatrics, Erasmus University Medical Center, Rotterdam, The Netherlands

Abstract
Oligosaccharidoses, sphingolipidoses and mucolipidoses are lysosomal storage disorders (LSDs) in which defective breakdown of glycan-side chains of glycosylated proteins and glycolipids leads to the accumulation of incompletely degraded oligosaccharides within lysosomes. In metabolic laboratories, these disorders are commonly diagnosed by thin-layer chromatography (TLC) but more recently also mass spectrometry based approaches have been published

Patient C; oligosaccharides - MS



Asn-GlcNAc-Gal-NANA
Asn-GlcNAc-Gal
Asn-GlcNAc

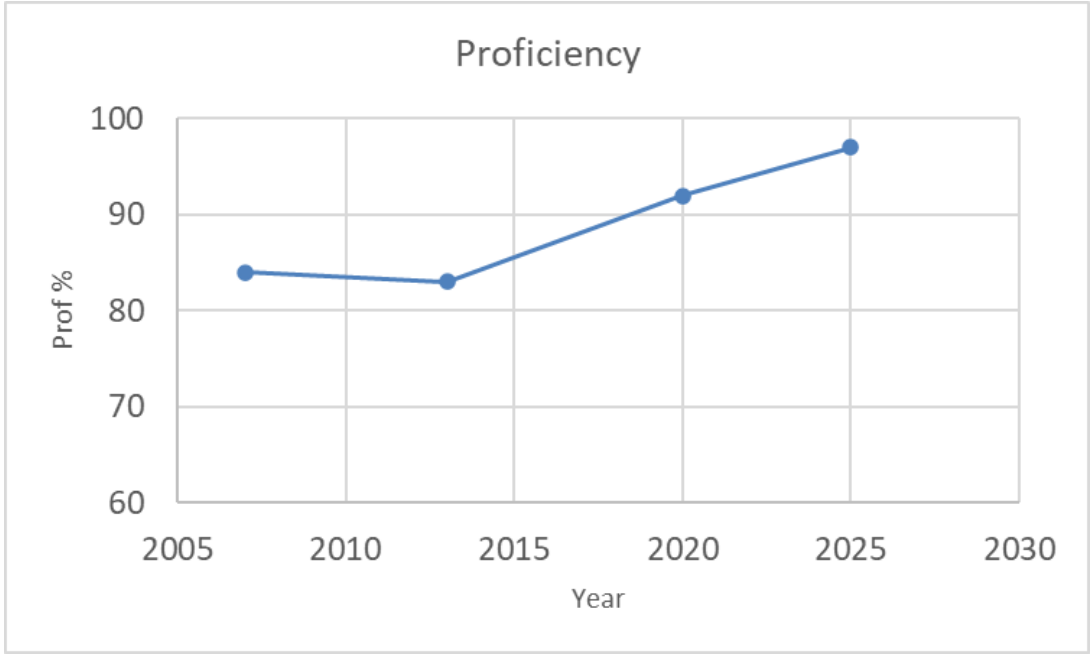
Patient C; recommendations

Further investigations reported:

Aspartylglucosaminidase activity in leu/fib
AGA mutations

Other AGU samples circulated:

	Proficiency
2007-G	84%
2013-E	83%
2020-B	92%
2025-C	97%



Patient C; Scoring

Performance item	Result	# points
Analytical	Elevated aspartylglucosamine and/or AGU oligosaccharide pattern Abnormal oligosaccharide pattern (incorrect/not specified)	2 1
Interpretation	Aspartylglucosaminuria Other oligosaccharidosis	2 1
Potential Critical error	Failure to detect oligosaccharidosis	

Conclusion:

- AGU deficiency detectable by amino acid- AND oligosaccharide analysis
- Overall proficiency 97%

Patient D

Clinical info: Boy aged 17 y. Slight anaemia and mild cognitive impairment.

Diagnosis: cblC deficiency (OMIM 277400)

Sample obtained from Erasmus MC

Diagnosed by WES (2 variants in MMACHC) confirmed by metabolite abnormalities

Participants:	18
Reports:	18
Correct diagnosis:	17
Proficiency:	94%

Patient D tests

Clinical info: Boy aged 17 y. Slight anaemia and mild cognitive impairment.

Creatinine median value 5.1 mmol/L

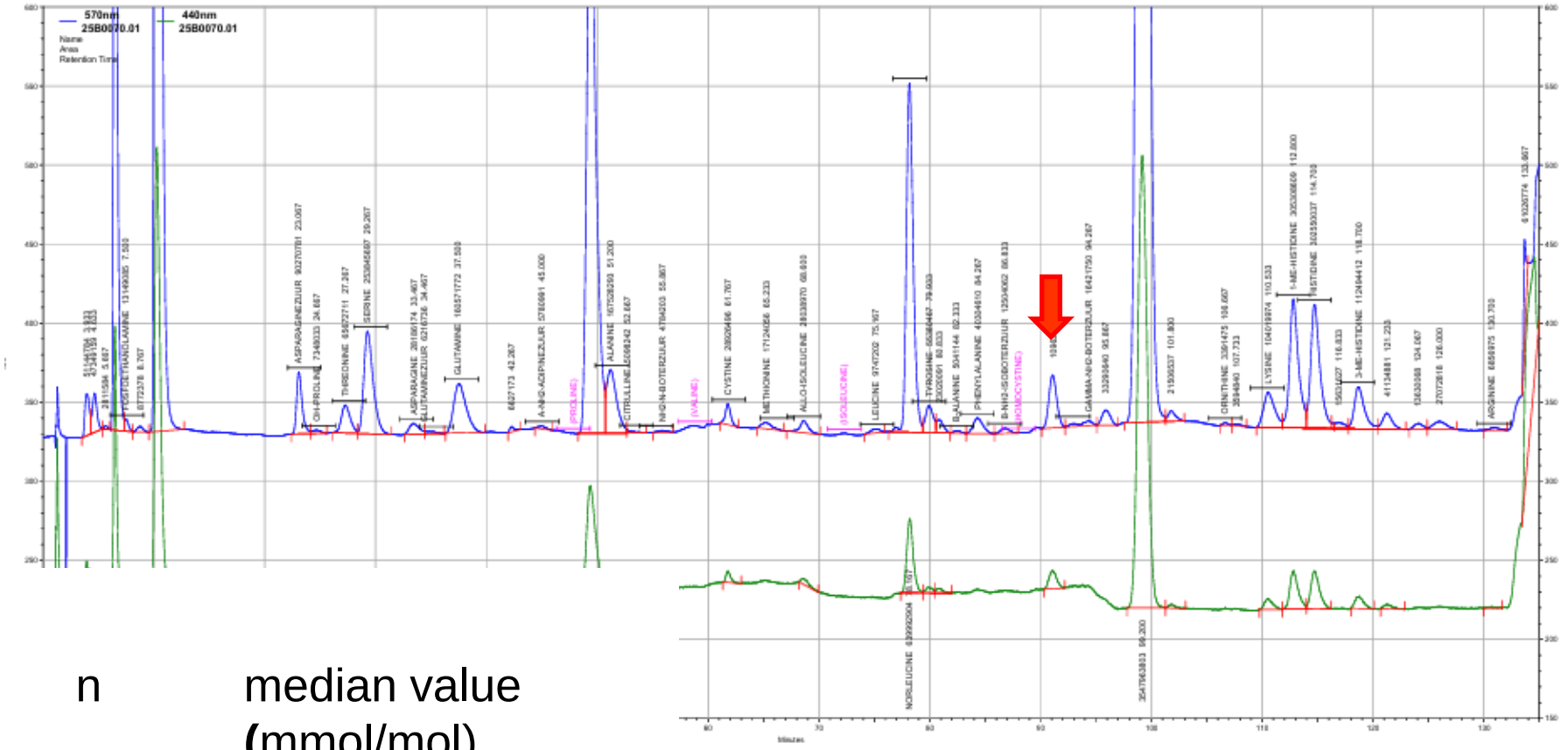
Clinical symptoms rather aspecific

→ Screening AA, OA GAG, oligosaccharides, PuPy

Patient D; results

MMA-uria

+

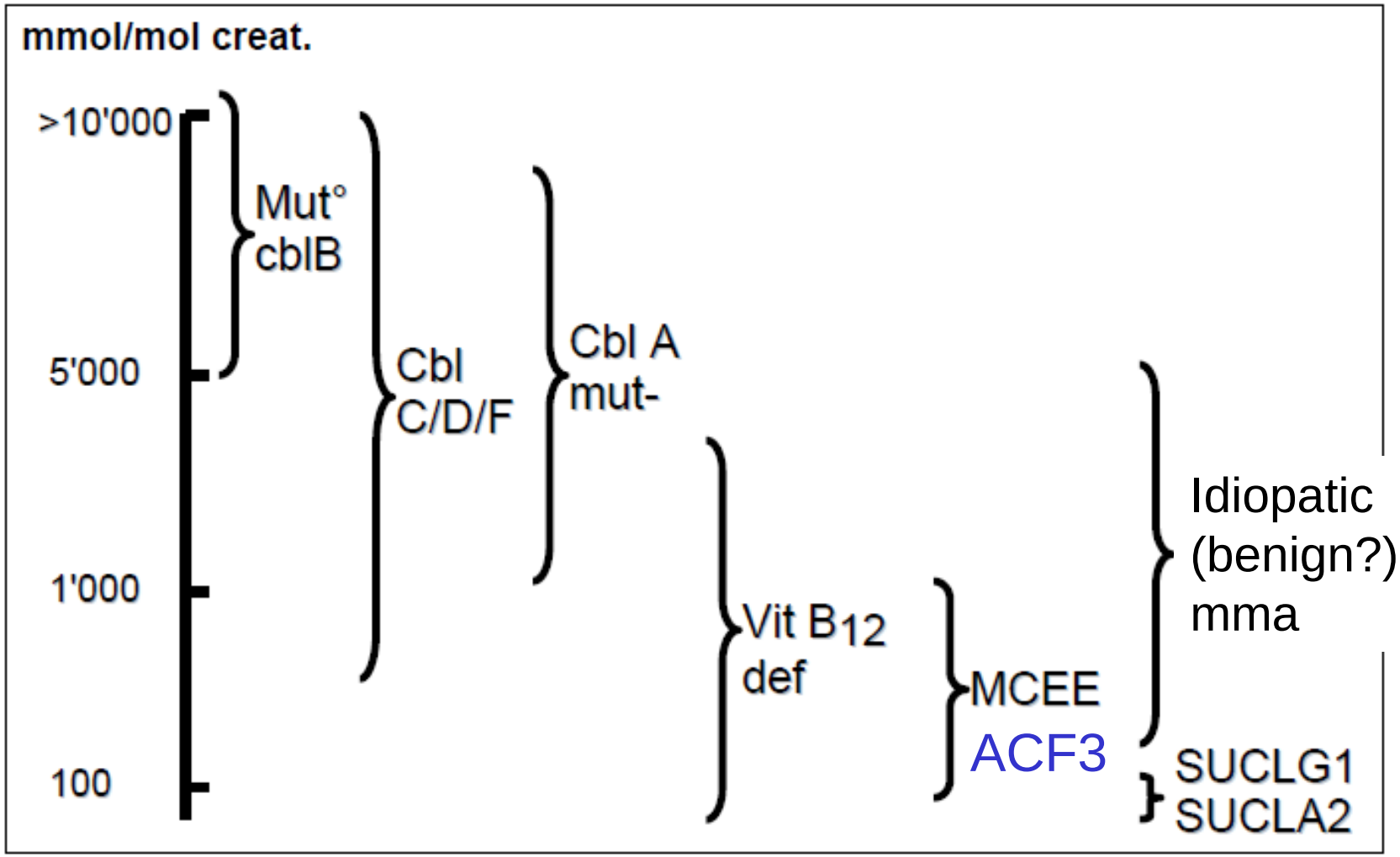


	n	median value (mmol/mol)
MMA elevated	18	321
Me-citrate elevated	8	6
Homocystine elevated	13	11
Hcys-cys elevated	3	9

Patient D; interpretation

Interpretation	Most likely	Alternative	Comment
B12 def or defect in B12 uptake/metab	4	11	
cblC	7	-	
CblC/D/F/J	3	1	
MMA	2	1	homocystine elevated
B12 def	2	3	
NO2 use	-	6	
SUCLA2	-	4	homocystine elevated
MUT	-	3	homocystine elevated

Urine MMA in various defects



ACF3: combined mma/malonic (ratio 2-10)

Adapted from: Fowler, Baumgartner, EMG Workshop 2007

Patient D; cblC adult phenotype

Kalantari *et al.*
Orphanet Journal of Rare Diseases (2022) 17:33
https://doi.org/10.1186/s13023-022-02179-y

Orphanet Journal of Rare Diseases

REVIEW Open Access

Adult-onset CblC deficiency: a challenging diagnosis involving different adult clinical specialists

Silvia Kalantari[1], Brigida Brezzi[2], Valeria Bracciamà[1], Antonella Barreca[3], Paolo Nozza[4], Tiziana Vaisitti[1], Antonio Amoroso[1,5], Silvia Deaglio[1,5], Marco Manganaro[2], Francesco Porta[6*] and Marco Spada[6]

Abstract

Background: Methylmalonic aciduria and homocystinuria, CblC type (OMIM #277400) is the most common disorder of cobalamin intracellular metabolism, an autosomal recessive disease, whose biochemical hallmarks are hyperhomocysteinemia, methylmalonic aciduria and low plasma methionine. Despite being a well-recognized disease for pediatricians, there is scarce awareness of its adult presentation. A thorough analysis and discussion of cobalamin C defect presentation in adult patients has never been extensively performed. This article reviews the published data and adds a new case of the latest onset of symptoms ever described for the disease.

Results: We present the emblematic case of a 45-year-old male, describing the diagnostic odyssey he ventured through to get to the appropriate treatment and molecular diagnosis. Furthermore, available clinical, biochemical and molecular data from 22 reports on cases and case series were collected, resulting in 45 adult-onset CblC cases, including our own. We describe the onset of the disease in adulthood, encompassing neurological, psychiatric, renal, ophthalmic and thromboembolic symptoms. In all cases treatment with intramuscular hydroxycobalamin was effective in reversing symptoms. From a molecular point of view adult patients are usually compound heterozygous carriers of a truncating and a non-truncating variant in the *MMACHC* gene.

Conclusion: Adult onset CblC disease is a rare disorder whose diagnosis can be delayed due to poor awareness regarding its presenting insidious symptoms and biochemical hallmarks. To avoid misdiagnosis, we suggest that

COLD SPRING HARBOR
Molecular Case Studies

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Neurodevelopmental and neuropsychiatric disorders in cobalamin C disease: a case report and review of the literature

Minh G. Nguyen[1,2], Lauren Tronick[3], Faraz Modirian[4], Rebecca Mardach[2,5] and Aaron D. Besterman[1,2,6]

Author Affiliations

Corresponding author: abesterman@health.ucsd.edu

Abstract

Cobalamin C disease is the most common complementation class of cobalamin disorders. Here, we present a case of a 14-yr-old male with early-onset cblC disease and autism spectrum disorder (ASD) admitted to our inpatient medical service for behavioral decompensation. We use this case to highlight key aspects of the neurodevelopmental and neuropsychiatric disorders associated with cblC disease. By incorporating a comprehensive review of existing literature, we highlight salient domains of psychological impairment in cblC disease, discuss the full range of neuropsychiatric presentations, and review clinical management implications unique to cblC disease.

Patient D; recommendations

Recommendations
Plasma B12
Plasma mma, hcys, amino acids
Plasma acylcarnitines
Sequence MMACHC
Sequence cbl genes etc (WES)

Supplementation B12, folate, betaine

Patient D; scoring

Performance item	Result	# points
Analytical	mma elevated homocystine and/or hcys-cys elevated	1 1
Interpretation	B12 deficiency, defect in B12 uptake or metabolism MMA/B12 deficiency with cbl not mentioned	2 1
Potential critical error	Failure to report increased mma	

Conclusion

- Clearly abnormal OA, AA
- Some participants did not detect/report homocystine
- Proficiency (94%)

Patient E

Clinical info: A 4 y old girl presenting with neuromotor regression, hypotonia and epilepsy.

Diagnosis: Adenylosuccinate lyase deficiency (OMIM 103050)

Sample provided by Prof Kayserili, Koc University Hospital, Istanbul

Diagnosed by 2 variants in ADSL (WES), confirmed by metabolite testing.

Participants:	18
Reports:	18
Correct diagnosis:	14
Proficiency:	78%

Previous survey (other samples):

2012-D; proficiency 62%

2023-B; proficiency 85%

Patient E; tests

Clinical info: A 4 y old girl presenting with neuromotor regression, hypotonia and epilepsy.

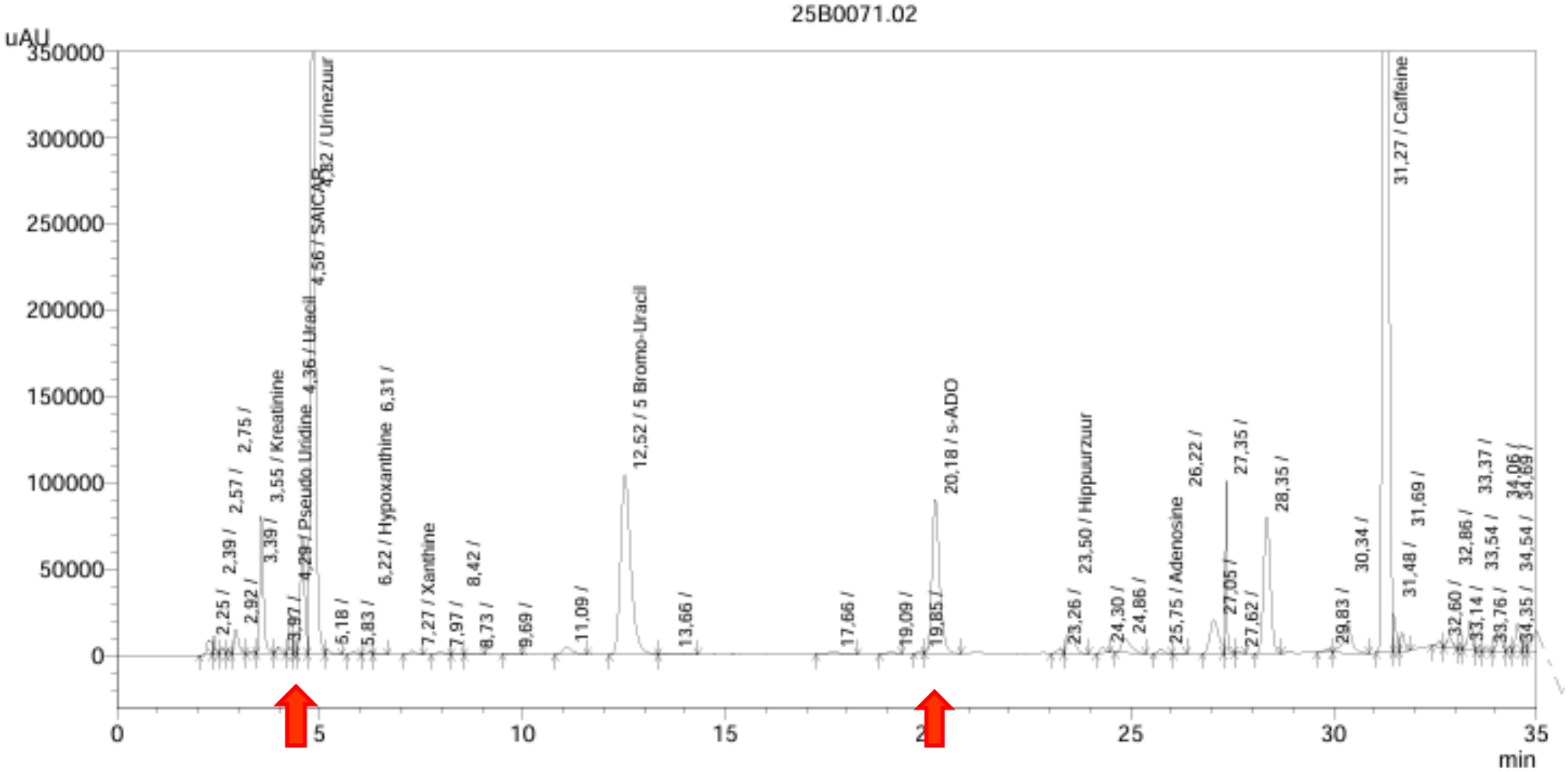
Creatinine median value 0.7 mmol/L

→ Retardation & convulsions tests

OA, AA, PuPy, GAG, Oligo

(+ Cre-Gua, alfa-AASA)

Patient E; purines-pyrimidines HPLC



Patient E; results purines, OA

	median	range (mmol/mol)	
SAICAr (n=7)	78	75-158	calibrator?
S-Ado (n=12)	144	38-173	

Other abnormalities

Low kreatinine	18		
3-OH-butyric	17		Ketosis due to fasting or treatment?
3-OH-isobutyric	3		approx 40 mmol/mol
3-OH-propionic	8		median 165 mmol/mol; source?
MeCitric	0	(1: normal)	trace (5 mmol/mol?)
Taurine	13		median 1638 mmol/mol; with ketogenic diet?
3-NH2-isobut	4		median 123 mmol/mol; =normal?
3-Alanine	2	(2: normal)	no Vigabatrin

Bratton Marshall (n=1): negative

SAICAr too low for positive Bratton-Marshall?

Test reported to have LOD of 1 umol/l ??

Patient E; Interpretation

	Diagnosis		
	Most likely	**Alternative**	**Comment**
ADSL def	14	-	
Propionic acidemia	1	1	Only 3-OH-propionic
SCOT def	1	-	
SCHAD def	1	-	
No diagnosis	1	-	
BKT or SCOT def	-	1	no BKT metabolites
creatine transporter def	-	1	Cre 1493 mmol/mol; elevated?
MCT1	-	1	
beta-alaninemia	-	2	beta-Ala borderline?

Patient E; recommendations

Further investigations:

ADSL enzyme analysis in Ery/Fib
ADSL mutations

ADSL: No effective causative treatment
Ketogenic diet?

Patient E; scoring

Performance item	Result	# points
Analytical	Elevated SAICAr and/or S-Ado	2
Interpretation	ADSL def Advice to analyse purines	2 1
Potential critical error	Sample not eligible ?	

Conclusions

- SAICAr and S-Ado concentrations not very high, but dilute urine sample
- PuPy analysis required to reach diagnosis
- Overall proficiency 78%

Patient F

Clinical info: A 10 year old male suffering from deteriorating visual acuity (on treatment).

Diagnosis: Ornithine aminotransferase deficiency (Gyrate atrophy; OMIM 258870)

Sample provided by Sheffield Childrens Hospital

Participants:	18
Reports:	18
Correct diagnosis:	17
Proficiency:	94%

Patient F; tests

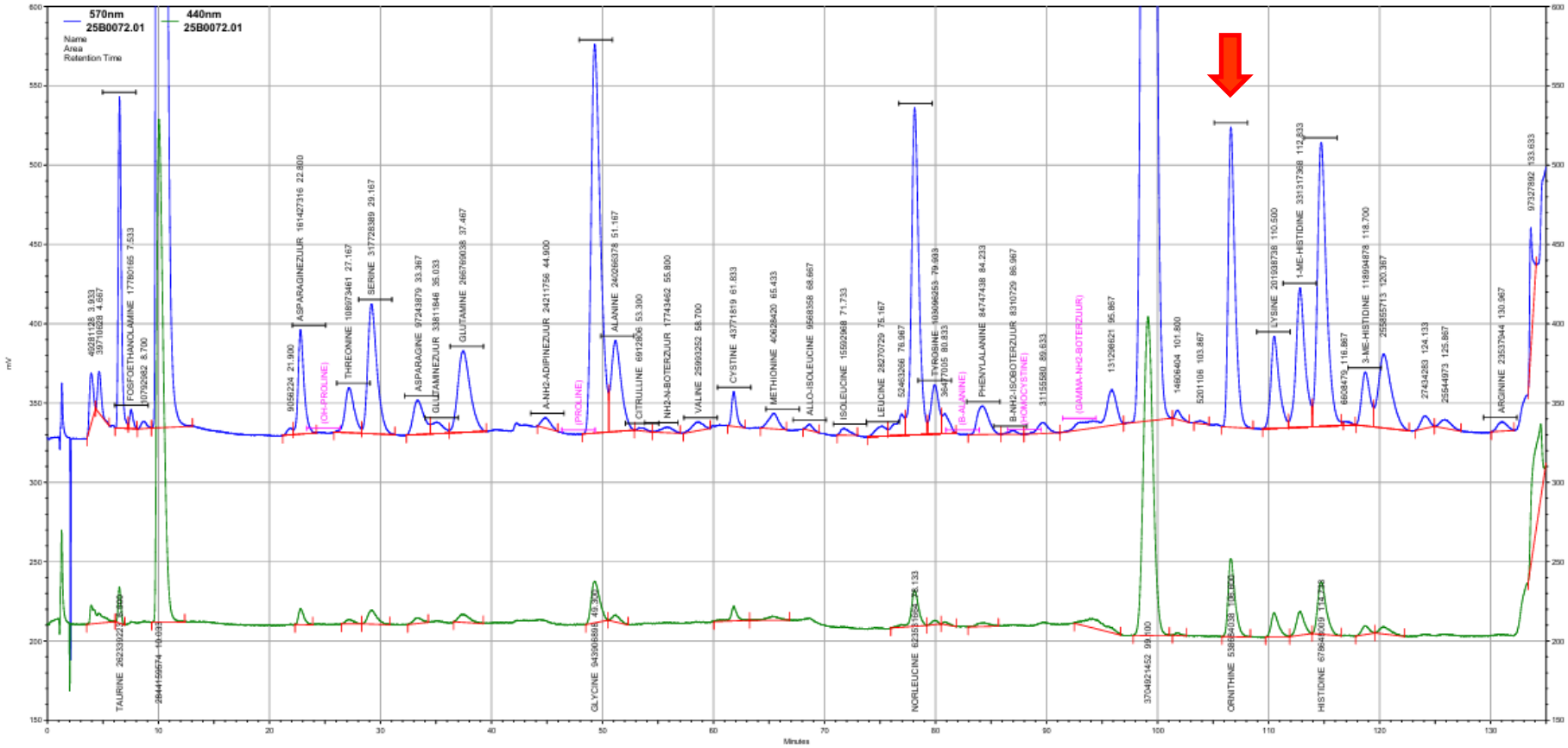
Clinical info: A 10 year old male suffering from deteriorating visual acuity (on treatment).

Creatinine median value 3.5 mmol/L

?? Problems with cornea, lens, retina? But no other symptoms…

→ Broad screening

Patient F; amino acids

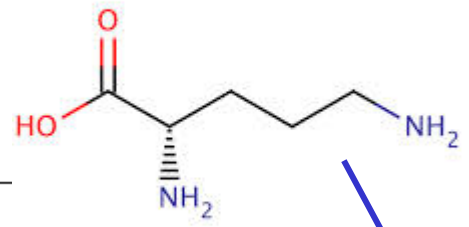


High ornithine in urine:

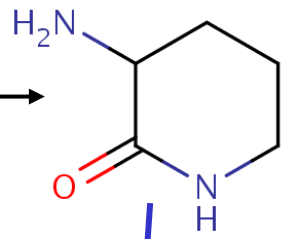
OAT	all other normal (+aminopiperidon)
LPI	+ lys, arg (cystine)
Cystinuria	+ cystine, lys, arg
HHH	+ homocitrulline
Hyperlysinemia	+ lys (saccharopine)

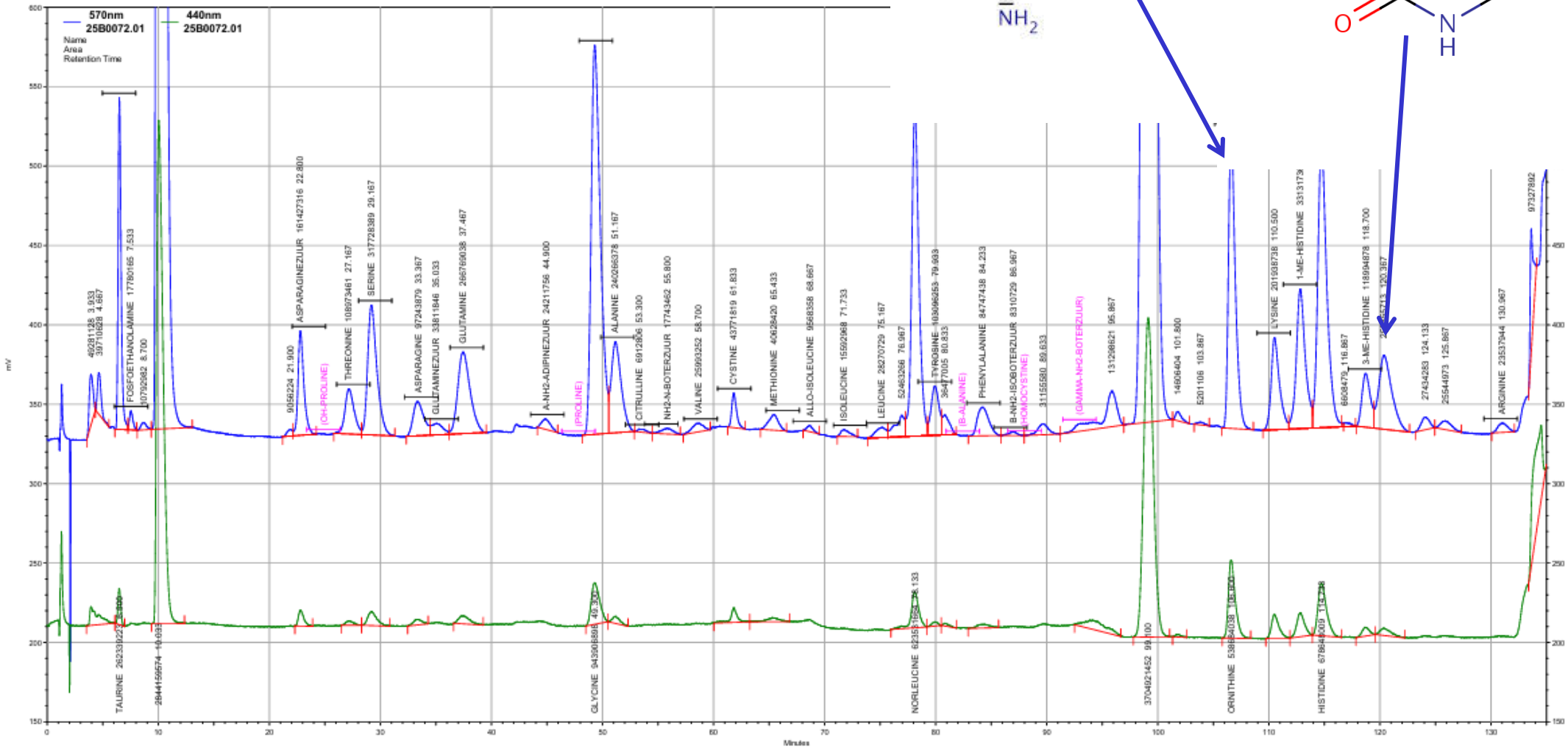
Patient F; aminopiperidone

ornithine



aminopiperidone





Patient F; interpretation & recommendations

	Most likely	Other possible	Comment
OAT	17	-	
Peroxisomal disorder?	1	-	
HHH	-	5 (2 ‘less likely’)	homocitrulline normal
UCD (NAGS, CPS1, OTC)	-	3 (1 ‘less likely’)	glutamine, orotic acid normal
LPI	-	3	Lys, Arg normal
cystinuria	-	1	Cystine, Lys, Arg normal

Further investigations:

Plasma amino acids
OAT activity/mutations
Plasma creatine (useful?)

Patient F; scoring

Performance item	Result	# points
Analytical	Elevated ornithine	2
Interpretation	OAT deficiency	2
Potential critical error	Failure to report OAT deficiency	

ERNDIM
QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

2026

Planning of 2026 scheme

- Sample dispatch: February 2026
- Survey 2026-1: March 2026
- Interim report 1: May 2026
- Survey 2026-2: June 2026
- Interim report 2: August 2026
- Annual report: Q1 2027
-

Next meeting: ERNDIM workshop 2026, Helsinki? date & location will follow in due time