ERNDiM
Quality Assurance in Laboratory Testing for IEM

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Critical Errors in the 2025 Qualitative EQA schemes

All critical errors for the 2025 schemes were agreed at the SAB online meeting held on 27th and 28th November 2025.

EQA scheme							
Scheme Name[1]	**Sample Number**	**Scheme Year**	**Diagnosis**	**Critical Error**	**Number of Labs**	**No of participants[2]**	**% CE**
AAI	2025-01	2025	OTC deficiency	Failure to recommend the determination of ammonia concentration and a diagnosis of a Urea Cycle Defect (UCD)	2	139	1.4%
ACDB-Heidelberg	2025-A	2025	VLCADD	1 lab did not detect elevated C14:1, and actively excluded VLCADD. Another lab failed to detect relevant metabolites and reported a normal profile.	2	40	5.0%
ACDB-London	2025-F	2025	Malonic aciduria	Did not identify increased C3DC (or C3DC+C4OH) and did not recommend further urine organic acid analysis	4	46	8.7%
CDG	2025-C	2025	RFT1-CDG	Failure to identify the abnormal sample and reporting it as normal	1	49	2.0%
	2025-D	2025	PMM2-CDG	Failure to identify the abnormal sample and reporting it as normal	1	49	2.0%
	2025-F	2025	PMM2-CDG	Failure to identify the abnormal sample and reporting it as normal	1	49	2.0%
DPT-CZ	2025-B	2025	3-methylglutaconic aciduria type IV	Failure to recognize abnormal excretion of 3-methylglutaconic acid	1	14	7.1%
	2025-C	2025	Cystinuria	Failure to recognize abnormal excretion of cystine	1	14	7.1%
DPT-FR	2025-C	2025	MSUD	Failure to diagnose MSUD	1	19	5.3%
	2025-F	2025	Glutaric aciduria type I low excretor	Failure to propose glutaric aciduria type I as first or alternative diagnosis	1	19	5.3%
DPT-NL	2025-B	2025	Beta-ketothiolase deficiency	Failure to report beta-ketothiolase as a diagnosis	1	18	5.6%
	2025-F	2025	Gyrate atrophy (OAT deficiency)	Failure to report ornithine aminotransferase deficiency as a diagnosis	1	18	5.6%
QLOU-Barcelona	2025-C	2025	Glutaric aciduria type I	Reported the case as glutaric aciduria type II but failed to detect the elevated levels of 3-hydroxyglutaric acid	1	71	1.4%
QLOU-Heidelberg	2025-C	2025	Mevalonic aciduria	One lab stated a normal organic acids profile	1	69	1.4%
	2025-D	2025	Methylmalonic aciduria mut0	One lab found elevated levels of 3-OH- and 4-OH-phenylacetic acids and 5-oxoproline and identified phenylketonuria as principal diagnosis	1	69	1.4%
	2025-E	2025	3-Hydroxy-3-methylglutaryl-CoA lyase deficie	One lab reported methylmalonic aciduria for this sample, and another lab stated 3-MGA1 without alternative or recommendations	2	69	2.9%
QLOU-Sheffield	2025-D	2025	Fumarate Hydratase Deficiency	No abnormality reported.	1	70	1.4%
	2025-E	2025	SSADH Deficiency	Failure to diagnose SSADH Deficiency	3	70	4.3%
UMPS	2025-A	2025	MPS-III	Reporting a normal profile.	3	83	3.6%
	2025-E	2025	MPS-IV	Reporting a normal profile.	2	83	2.4%
	2025-F	2025	MPS-II	Reporting a normal profile.	1	83	1.2%
				Total	32	618	5.2%

Notes

1. AAI = Amino Acids Interpretation; ACDB = Acylcarnitines in DBS; CDG = Congenital Disorders of Glycoslylation; DPT = Diagnostic Proficiency Testing; CH = Switzerland; CZ = Czech Republic; FR = France; NL = Netherlands; UK = United Kingdom; QLOU = Qualitative Organic Acid; UMPS = Urine Mucopolysaccharides

2. Number of participants = number of registered labs minus any Educational participants, non- or partial submitters and any labs that withdrew from the scheme