ERNDiM
Quality Assurance in Laboratory Testing for IEM

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Critical Errors in the 2024 Qualitative EQA schemes

All critical errors for the 2024 schemes were agreed at the SAB online meeting held on 28th and 29th November 2024.

EQA scheme							
Scheme Name[1]	**Sample Number**	**Scheme Year**	**Diagnosis**	**Critical Error**	**Number of Labs**	**No of participants[2]**	**% CE**
AAI	2024-2	2024	Lysinuric protein intolerance (LPI)	Overlooking the possibility of aminoacidopathy.	1	138	0.7%
ACDB Heidelberg	2024-A	2024	Carnitine palmitoyltransferase I deficiency	Reported either no abnormal results or elevated free carnitine in combination with other AC homologues not relevant to this sample.	1	40	2.5%
	2024-D	2024	Hydroxymethylglutaryl-CoA lyase deficiency	Reporting of isovaleric acidemia without any further recommendations.	1	40	2.5%
ACDB London	2024-C	2024	Glutaryl-CoA Dehydrogenase deficiency (Glutaric Aciduria type 1; GA1)	Failed to detect any abnormality.	2	43	4.7%
	2024-E	2024	Propionyl-CoA carboxylase deficiency (Propionic Acidaemia; PA)	Did not identify the correct abnormalities in this sample and provided misleading recommendations.	1	43	2.3%
	2024-F	2024	Medium Chain Acyl-CoA Dehydrogenase (MCAD) deficiency	Provided an incorrect diagnosis and misleading recommendations.	1	43	2.3%
ACDB Rome	2024-A	2024	CPT1	Diagnosis LCHAD.	1	42	2.4%
	2024-B	2024	GA1; GCDH	Reported as normal.	1	42	2.4%
CDG	2024-F	2024	Type 1 - PMM2-CDG	Failed to recognise an abnormal type I pattern and incorrectly reported the sample as normal.	2	53	3.8%
DPT CH	2024-C	2024	Mucopolysaccharidosis type IVA (OMIM #253000)	Normal GAG: proposed Mitochondrial respiratory chain defect.	1	21	4.8%
	2024-E	2024	GM1 gangliosidosis, beta-galactosidase-1 deficiency	Normal results, found elevation of succinylacetone and 4-OH-phenyllactic acid.	1	21	4.8%
DPT CZ	2024-B	2024	Alpha-mannosidosis due to alpha-mannosidase deficiency	The failure to recognize abnormal oligosaccharides profile, which would prevent establishing the correct diagnosis.	3	16	18.8%
DPT FR	-	2024	-	-	0	20	0.0%
DPT NL	2024-E	2024	LPI	Failure to report LPI as a possible diagnosis.	1	17	5.9%
DPT UK	2024-F	2024	Gyrate atrophy of retina and choroidea due to ornithine aminotransferase deficiency	Failure to identify the increased ornithine concentration.	2	20	10.0%
QLOU Barcelona	2024-B	2024	L-2-hydroxyglutaric aciduria	2 labs that reported as normal sample and one lab that gave the diagnosis of Maple syrup urine disease (MSUD).	3	70	4.3%
	2024-D	2024	Multiple acyl-CoA dehydrogenases deficiency	1 lab that reported as propionic academia, 1 lab that gave the diagnosis of hyperinsulinism, and 1 lab that reported as normal apparently due to a swap with sample E, and without additional specific recommendations.	3	70	4.3%
	2024-F	2024	Glutaric aciduria type I low excretor	1 lab that reported as 2-hydroxyglutaric aciduria, 1 lab that gave the diagnosis of lipoic acid defects and 1 lab that reported the diagnosis of 3-methylglutaconic aciduria.	3	70	4.3%
QLOU Heidelberg	2024-D	2024	Alkaptonuria (Homogentisate1,2-dioxygenase deficiency)	2 particip0ants reported a diagnosis of Malonic aciduria. 1 participant reported a diagnosis of dietary behaviour/paracetamol. 1 participant reported a normal result.	4	73	5.5%
	2024-F	2024	Methylmalonic aciduria due to deficient cobalamin adenosyltranferase	Diagnosed Alkaptonuria.	1	73	1.4%
QLOU Sheffield	2024-D	2024	Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)	Normal diagnosis given.	4	73	5.5%
UMPS	2024-A	2024	MPS-I	Reporting a normal profile.	1	83	1.2%
	2024-D	2024	MPS-III	Reporting a normal profile.	3	83	3.6%
	2024-E	2024	MPS-I	Reporting a normal profile.	1	83	1.2%
				Totals	**42**	**709**	**5.9%**

Notes

1. AAI = Amino Acids Interpretation; ACDB = Acylcarnitines in DBS; CDG = Congenital Disorders of Glycosylation; 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