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Critical Errors in the 2023 Qualitative EQA schemes

All critical errors for the 2023 schemes were agreed at the SAB online meeting held on 30th November and 1st December 2023.

EQA scheme							
Scheme Name[1]	**Sample Number**	**Scheme Year**	**Diagnosis**	**Critical Error**	**Number of Labs**	**No of participants**[2]	**% CE**
AAI	2023-02	2023	Adenosine kinase deficiency	The participant interpreted the changes as a result of a liver dysfunction. The recommendations for further examinations did not take an inborn disorder of metabolism into consideration.	1	127	0.8%
	2023-05	2023	Remethylation defect (CblC disease)	Four critical errors were agreed for suggesting mild hyperhomocysteinaemia, a normal profile, or wrong diagnosis, without a further recommendation to clarify the findings	4	127	3.1%
ACDB Heidelberg	2023-A	2023	Glutaric acidemia type 1 (common sample)	Failure to report C5DC or a related ratio and did not report any diagnosis.	1	40	2.5%
	2023-B	2023	3-Methylglutaconic aciduria type I	Did not report elevated ACs also did not report a diagnosis.	2	40	5.0%
	2023-D	2023	Propionic acidemia, PA	Two participants stated the acylcarnitine profile not to be representative for an organoaciduria or fatty acid metabolism defect and provided alternative possible disorders. One failed to provide any diagnosis.	3	40	7.5%
	2023-E	2023	Very long-chain acyl CoA dehydrogenase (VLCAD) deficiency	One participant commented on slightly elevated C14 carnitine but categorized it as not relevant for a pathologic condition and opted for a normal acylcarnitine profile. Another participant reported several numeric concentrations for various acylcarnitines but did not categorize the findings or give a diagnosis.	2	40	5.0%
ACDB London	2023-A	2023	Glutaric Acidaemia Type 1 OMIM 231670	Did not detect glutaryl carnitine, did not suggest GA1 as a diagnosis.	1	42	2.4%
	2023-F	2023	Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000	Did not detect increased C3 carnitine.	1	42	2.4%
ACDB Rome	-	2023	-	-	0	43	0.0%
CDG	-	2023	-	-	0	52	0.0%
DPT CH	2023-C	2023	Alkaptonuria (OMIM #203500)	Incorrect diagnosis provided.	1	21	4.8%
DPT CZ	2023-E	2023	Phenylketonuria due to phenylalanine hydroxylase deficiency.	Failure to recognize abnormal excretion of phenylalanine and/or its metabolites, which prevented establishing the correct diagnosis.	1	19	5.3%
DPT FR	-	2023	-	-	0	19	0.0%
DPT NL	-	2023	-	-	0	18	0.0%
DPT UK	-	2023	-	-	0	19	0.0%
QLOU Barcelona	2023-A	2023	Medium chain acyl-CoA dehydrogenase (MCAD) deficiency	Reported a normal profile and made no recommendaitons for further testing.	4	74	5.4%
	2023-D	2023	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	Reported the diagnosis of 3-methylcrotonylglicinuria or 3-methylglutaconic aciduria type I and not identify the increase of 3-hydroxy-3-methylglutarate.	3	74	4.1%
	2023-E	2023	L-2-hydroxyglutaric aciduria	Reported the diagnosis of multyple acyl-CoA dehydrogenase deficiency without information regarding neither alternative diagnosis or recommendation.	1	74	1.4%
QLOU Heidelberg	2023-C	2023	MSUD	One participant suggested a ketogenesis defect as principal diagnosis, another lab reported lactic aciduria (although both labs managed to find two key metabolites each).	2	73	2.7%
	2023-E	2023	PKU	Reported only on elevated lactic acid, mentioned lactic acidemia with possible pyruvate dehydrogenase phosphatase deficiency	1	73	1.4%
QLOU Sheffield	2023-B	2023	Methylmalonic Aciduria	Incorrect analytical findings and diagnosis.	1	71	1.4%
	2023-F	2023	Alkaptonuria	Failed to detect the large peak of homogentisic acid and as a result did not make the correct diagnosis.	1	71	1.4%
UMPS	2023-C	2023	MPS-III	Reporting a normal profile.	1	81	1.2%
				Totals	**31**	**699**	**4.4%**

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