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Critical Errors in the 2020 Qualitative EQA schemes

All critical errors for the 2020 schemes were agreed at the SAB online meeting held on 19th and 20th November 2020.

EQA scheme							
Scheme Name[1]	**Sample Number**	**Scheme Year**	**Diagnosis**	**Critical Error**	**Number of Labs**	**No of participants[2]**	**% CE**
ACDB Heidelberg	2020-A	2020	long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	Failure to detect elevated concentrations of hydroxylated long-chain acylcarnitines	2	40	5.0%
	2020-C	2020	Common sample: Long chain hydroxyacyl CoA dehydrogenase deficiency (LCHAI	Failure to detect elevated concentrations of hydroxylated long-chain acylcarnitines	2	40	5.0%
ACDB London	2020-C	2020	Common sample: Long chain hydroxyacyl CoA dehydrogenase deficiency (LCHAI	Failure to detect elevated concentrations of hydroxylated long-chain acylcarnitines	2	41	4.9%
ACDB Rome	2020-B	2020	medium chain acyl-CoA dehydrogenase deficiency (OMIM 201450)	Diagnosis of CPT1 deficiency	1	38	2.6%
	2020-C	2020	Common sample: Long chain hydroxyacyl CoA dehydrogenase deficiency (LCHAI	Diagnosis of Isovaleryl-CoA dehydrogenase deficiency (IVA) or 2-Methybutyryl-CoA dehydrogenase deficiency (2MBD/SBCAD) a	4	38	10.5%
CDG	2020-A	2020	PMM2-CDG	1) sample swap with sample B (normal) 2) misdiagnosis	2	65	3.1%
	2020-C	2020	Normal	Diagnosed as abnormal	1	65	1.5%
	2020-E	2020	Alcohol abuse	Abnormal pattern not detected and a diagnosis of normal given	1	65	1.5%
DPT CH	-	2020	-	-	0	22	0.0%
DPT CZ	-	2020	-	-	0	19	0.0%
DPT FR	2020-B	2020	Alkaptonuria (homogentisate 1,2-dioxygenase deficiency)	Not performing organic acids and not advising this should be done	1	23	4.3%
	2020-C	2020	Morquio disease A (mucopolysaccharidosis type IVA, galactosamine-6-sulfate su	Not performing quantification or fractionation of glycosaminoglycans and not advising these should be done	2	23	8.7%
DPT NL	2020-A	2020	Common sample: Phenylketonuria due to PAH mutations.	Failure to report elevated phenylalanine, abnormal organic acids and PKU	1	19	5.3%
DPT UK	-	2020	-	-	0	20	0.0%
QLOU Barcelona	2020-B	2020	L-2-hydroxyglutaric aciduria	Failing to identify 2-hydroxyglutaric aciduria	3	68	4.4%
	2020-D	2020	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	Failing to identify 3-hydroxy-3-methylglutaryl-CoA lyase deficiency	4	68	5.9%
	2020-G	2020	Multiple acyl-CoA dehydrogenase deficiency (MADD).	Diagnosing Methylmalonic aciduria	1	68	1.5%
QLOU Heidelberg	2020-A	2020	Canavan disease	1) Misdiagnosis with no further recommendations for testing (n = 3); or 2) Recommendations not specific enough to ensure a diagnosis will be made later (n = 3)	6	71	8.5%
	2020-C	2020	D-2-hydroxyglutaric aciduria	Failure to identify abnormal amounts of 2-hydroxyglutaric acid	6	71	8.5%
	2020-D	2020	ornithine transcarbamylase (OTC) deficiency	Diagnosing PKU or hyperphenylalaninaemia	2	71	2.8%
	2020-G	2020	glutaric aciduria type I (low excretor)	Reporting a normal diagnosis without suggesting adequate further examination	5	71	7.0%
	2020-I	2020	succinic semialdehyde dehydrogenase (SSADH) deficiency	1) Giving a normal diagnosis 2) Giving a diagnosis of UCD	2	71	2.8%
QLOU Sheffield	2020-G	2020	MCADD	Missing MCADD	1	73	1.4%
	2020-I	2020	Fumarate Hydratase	Failing to identify fumarate as an abnormal metabolite	1	73	1.4%
UMPS	2020-C	2020	MPS II	Diagnosed as 'normal'	1	89	1.1%
	2020-E	2020	MPS IV A	Reporting 'normal' as the most likely diagnosis	4	89	4.5%
				Totals	**55**	**588**	**9.4%**

Notes

1. 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