ERNDIM
QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

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Critical Errors in the 2019 Qualitative EQA schemes

All critical errors for the 2019 schemes were agreed at the SAB meeting held on 21st and 22nd November 2019 in Manchester, UK.

EQA scheme							
Scheme Name[1]	**Sample Number**	**Scheme Year**	**Diagnosis**	**Critical Error**	**Number of Labs**	**No of participants[2]**	**% CE**
ACDB Heidelberg	2019-F	2019	Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	Failing to detect elevated concentrations of hydroxylated long-chain acylcarnitines	3	40	7.5%
ACDB London	2019-D	2019	Propionic acidaemia (OMIM 6060540)	Failing to recognise the abnormality	1	39	2.6%
ACDB Rome	-	2019	-	-	0	41	0.0%
CDG	-	2019	-	-	0	64	0.0%
DPT CH	-	2019	-	-	0	20	0.0%
DPT CZ	2019-F	2019	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	Failing to recognize abnormal excretion of orotate and homocitrulline	1	20	5.0%
DPT FR	2019-C	2019	Hyperprolinaemia type II due to delta 1-pyrroline-5-carboxylate (P5C) dehydrogenase deficiency (ALDH4A1 gene).	Failing to perform amino acid analysis and failing to identify N-(pyrrole-2-carboxyl) glycine in the organic acid profile	2	24	8.3%
	2019-F	2019	Argininemia	non-identification of an increase of orotic acid without recommendation to perform plasma amino acid analysis	1		4.2%
DPT NL	2019-C	2019	MSUD (OMIM 248600)	Failing to conclude MSUD/DLD	1	21	4.8%
DPT UK	2019-C	2019	Lysinuric protein intolerance	Failing to detect the increased orotic acid	1	22	4.5%
QLOU Barcelona	2019-E	2019	Maple Syrup Urine Disease (MSUD).	Failing to identify MSUD and report the alterations secondary to hepatic failure or Tyrosinemia	3	69	4.3%
QLOU Heidelberg	-	2019	-	-	0	70	0.0%
QLOU Sheffield	2019-A	2019	Citrullinaemia	Failing to identify increased orotic acid excretion	10	71	14.1%
	2019-F	2019	Isovaleric Acidaemia	Failing to identify IVA	2		2.8%
UMPS	2019.02	2019	MPS II	Giving a diagnosis of normal	1	88	1.1%
	2019.06	2019	Normal	Diagnosing an MPS disorder	1		1.1%
				Totals	**27**	**589**	**4.6%**

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