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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 29<sup>th</sup> & 30<sup>th</sup> November 2019 in Leiden, Netherlands.

EQA scheme								
Scheme Name <sup>1</sup>	Sample Number	Scheme Year	Diagnosis	Critical Error	Number of Labs	No of participants <sup>2</sup>	% CE	
ACDB Heidelberg	2018-A	2018	Cobalamin A deficiency	Reporting a normal diagnosis without further recommendations	3	36	8.3%	
	2018-C	2018	Propionic aciduria	Reporting a normal diagnosis without further recommendations	3		8.3%	
ACDB London	2018-E	2018	Medium Chain acyl CoA dehydrogenase deficiency (MCADD)	Reporting a normal diagnosis without further recommendations	2	39	5.1%	
ACDB Rome	2018-D	2018	Short branched chain acyl-CoA dehydrogenase deficiency (SBCAD)	Failing to report an increase in C5 carnitine	1	37	2.7%	
	2018-E	2018	Methylmalonic aciduria, Vitamin B12-Responsive, due to defect in synthesis of Adenosylcobalamin, cblA	Failing to report an increase in C3 carnitine	1		2.7%	
CDG	-	2018	-	-	0	65	0.0%	
DPT CH	2018-A <i>[DPT Common Sample]</i>	2018	Dihydropyrimidine dehydrogenase (DPD) deficiency (thymine-uraciluria). OMIM # 274270	Failing to recognise abnormal excretion of uracil and thymine	1	22	4.5%	
	2018-F	2018	GM1-gangliosidosis # 230500	Failing to detect increased GAGs with no recommendation for oligosaccharide testing	1		4.5%	
DPT CZ	2018-D	2018	Glutaric aciduria type I	Failing to recognise abnormal excretion of 3-hydroxyglutarate and glutarate	2	20	10.0%	
DPT FR	2018-B	2018	Mucopolysaccharidosis type VII (Sly disease - GUSB gene)	Failing to diagnose a lysosomal storage disorder with no advice to perform mucopolysaccharides analysis	1	25	4.0%	
	2018-E	2018	Ornithine aminotransferase (OAT) deficiency ; gyrate atrophy of retina and choroidea	Failing to identify the increase in ornithine	1		4.0%	
DPT NL	2018-E	2018	Alfa-mannosidosis (OMIM 248500)	Failing to detect abnormal oligosaccharides or failing to perform oligosaccharide analysis with no recommendation to do so	1	21	4.8%	
DPT UK	2018-A <i>[DPT Common Sample]</i>	2018	Dihydropyrimidine dehydrogenase (DPD) deficiency (thymine-uraciluria). OMIM # 274270	Failing to recognise abnormal excretion of uracil and thymine	1	20	5.0%	
	2018-E	2018	Argininosuccinic aciduria (ASA).	Failing to detect both ASA and orotic acid	3		15.0%	
	2018-F	2018	Gyrate atrophy (ornithine aminotransferase deficiency)	Failing to detect increased ornithine	2		10.0%	
QLOU Barcelona	2018-B	2018	Normal	Reporting a diagnosis of PKU <i>(possible sample swap with sample 2018-C)</i>	1	68	1.5%	
	2018-C	2018	Phenylketonuria	Reporting a normal diagnosis <i>(possible sample swap with sample 2018-B)</i>	1		1.5%	
	2018-G	2018	Multiple acyl-CoA dehydrogenase deficiency	Reporting lactic acidosis due to biotinidase deficiency	1		1.5%	
	2018-I	2018	Isovaleric acidemia	Reporting Multiple Acyl Coa dehydrogenase deficiency (MADD)	1		1.5%	
QLOU Heidelberg	2018-B	2018	MCAD deficiency	Reporting a normal diagnosis with no further recommendations	7	71	9.9%	
QLOU Sheffield	2018-B	2018	Glutaric Aciduria Type 1	Reporting a normal diagnosis	3	72	4.2%	
	2018-D	2018	Methylmalonic Aciduria	Failing to identify highly increased MMA	2		2.8%	
	2018-E	2018	3-Methylglutaconic aciduria (NOS)	Failing to identify increased 3-MGC	2		2.8%	
	2018-F	2018	3-Methylglutaconic aciduria (NOS)	Failing to identify increased 3-MGC	1		1.4%	
	2018-G	2018	Propionic Acidaemia	Giving 2-Me3OHbutyric aciduria as primary diagnosis with no alternative diagnosis given	1		1.4%	
UMPS	2018.01	2018	MPS I Scheie	Reporting a normal diagnosis	2	92	2.2%	
					<b>Totals</b>	<b>45</b>	<b>588</b>	<b>7.7%</b>

#### Notes

1. 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