

QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

Critical Errors in the 2017 Qualitative EQA schemes

All critical errors for the 2017 schemes were agreed at the SAB meetings held on 23rd November 2017, Leiden and 22nd & 23rd March 2018 in Barcelona, Spain.

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EQA scheme			1				
Scheme Name ¹	Sample Number	Scheme Year	Diagnosis	Error	Number of CE	No of participants ²	% CE
ACDB Heidelberg	2017.04	2017	cobalamin A deficiency	failing to identify elevated C3-carnitine or any noticeable ratios and giving a normal diagnosis without recommending organic acid analysis in urine	1	37	2.7%
ACDB London	-	2017	-	-	0	39	0.0%
ACDB Zurich	-	2017	-	-	0	36	0.0%
CDG	2017.01	2017	PGM1-CDG OMIM: 171900	failure to make the correct diagnosis	1	62	1.6%
DPT CH	-	2017	-	-	0	21	0.0%
DPT CZ	С	2017	aspartylglucosaminuria	failure to recognize excretion of aspartylglucosamine in OLS and/or am	1	20	5.0%
	D		MCAD deficiency	failure to recognize abnormal excretion of glycine conjugates without i	1		5.0%
DPT FR	D	2017	GM1 gangliosidosis - beta-galactosidase deficiency (GLB1 gene - OMIM *611458)	diagnosing HHH syndrome without mentioning the possibility of a lysosomal storage disorder on the clinical presentation	1	24	4.2%
DPT NL	2017.05	2017	GM1	failure to detect abnormal oligo or to recommend oligo analysis	1	20	5.0%
DPT UK	F	2017	Tyrosinaemia Type 1	failing to detect succinylacetone	2	22	9.1%
QLOU Heidelberg	-	2017	-	-	0	109	0.0%
QLOU Sheffield	248	2017	MCADD	failed to identify the MCADD profile	4	104	3.8%
UMPS	-	2017	-	-	0	92	0.0%
				Totals	12	586	2.0%

Notes

 ACDB = Acylcarnitines in DBS Heidelberg; ACDB London = Acylcarnitines in DBS London; ACDB Zurich = Acylcarnitines in DBS Zurich DPT CH = Diagnostic Proficiency Testing Switzerland; DPT CZ = DPT Czech Republic; DPT FR = DPT France; DPT NL = DPT Netherlands; DPT UK = DPT United Kingdom; QLOU Heidelberg = Qualitative Organic Acids Heidelberg; QLOU Sheffield = Qualitative Organic Acids Sheffield; UMPS = Urine Mucopolysaccharides
Number of participants = number of registered labs minus any Educational participants, non- or partial submitters and any labs that withdrew from the scheme