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Critical Errors in the 2015 Qualitative EQA schemes

All critical errors for the 2015 schemes were agreed at the SAB meeting held on 17th March 2016 in Prague, Czech Republic.

EQA scheme			Diagnosis	Error	Number of CE	No of participants ²	% CE
Scheme Name ¹	Sample Number	Scheme Year					
AC DBS Heidelberg	2015.01	2015	Glutaric aciduria type I	missed detection of C5DC	1	50	2.0%
	2015.04	2015	3HMGCoA lyase def	missed detection of C5OH and C6DC	1	50	2.0%
	2015.06	2015	LCHAD def	missed detection of hydroxylated long chain acylcarnitines	1	50	2.0%
DPT CH	D [common sample]	2015	Homocystinuria due to cystathionine beta-synthase deficiency, OMIM #236200	to miss homocyst(e)ine without any other abnormality or appropriate recommendation for further test	1	21	4.8%
	F	2015	Glutaric acidaemia type I due to glutaryl-CoA dehydrogenase deficiency OMIM #231670	to miss critical metabolite on organic acid analysis	2	21	9.5%
DPT CZ	A	2015	Argininosuccinic aciduria	failure to recognize abnormal excretion of argininosuccinic acid and its anhydrides	1	20	5.0%
	D [common sample]	2015	Homocystinuria due to cystathionine beta-synthase deficiency, OMIM #236200	failure to recognize abnormal excretion of homocystine	1	20	5.0%
DPT FR	D	2015	Canavan disease	missed detection of N-acetylaspartic acid	1	23	4.3%
DPT NL	D [common sample]	2015	Homocystinuria due to cystathionine beta-synthase deficiency, OMIM #236200	missing any abnormal metabolite & recomm not specific	3	20	15.0%
	F	2015	GA I	3OHglutarate/C5DC not reported & no recomm	1	20	5.0%
DPT UK	A	2015	Homocystinuria	failed to detect increased homocysteine	1	23	4.3%
	B [common sample]	2015	Homocystinuria due to cystathionine beta-synthase deficiency, OMIM #236200	failed to detect increased homocysteine	1	23	4.3%
	E	2015	Argininosuccinic aciduria	failed to detect orotic acid and ASA	2	23	8.7%
Qual OA Heidelberg	223	2015	Canavan disease	missed detection of N-acetylaspartic acid	3	99	3.0%
	227	2015	MSUD	missed detection of branched chain oxo- and hydroxyacids	2	99	2.0%
Qual OA Sheffield	223	2015	MMA	missed the MMA peak	1	100	1.0%
UMPS	F	2015	MPS II	diagnosis 'normal'	1	94	1.1%
Totals					24	329	7.3%

Notes

1. AC DBS Heidelberg = Acylcarnitines in DBS Heidelberg; DPT CH = Diagnostic Proficiency Testing Switzerland; DPT CZ = DPT Czech Republic; DPT FR = DPT France; DPT NL = DPT Netherlands; DPT UK = DPT United Kingdom; Qual OA Heidelberg = Qualitative Organic Acids Heidelberg; Qual OA Sheffield = Qualitative Organic Acids Sheffield; 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