ERNDiM
Quality Assurance in Laboratory Testing for IEM

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Critical Errors in the 2021 Qualitative EQA schemes

All critical errors for the 2021 schemes were agreed at the SAB online meeting held on 25th and 26th November 2021.

EQA scheme							
Scheme Name[1]	**Sample Number**	**Scheme Year**	**Diagnosis**	**Critical Error**	**Number of Labs**	**No of participants[2]**	**% CE**
ACDB Heidelberg	-	2021	-	-	0	40	0.0%
ACDB London	2021-C	2021	LCHADD, OMIM 609016	Failure to detect the elevated long chain hydroxyacyl carnitines	1	40	2.5%
	2021-E	2021	Methylmalonyl CoA mutase deficiency, OMIM 251000	Failure to recognise the grossly elevated C3 carnitine	1	40	2.5%
	2021-F	2021	Biotin responsive multiple carboxylase deficiency, holocarboxylase synthetase deficiency, OMIM 253270	Failure to report the significant elevation of hydroxyacylcarnitine species of chain length C14-C18	1	40	2.5%
ACDB Rome	2021-F	2021	Biotin responsive multiple carboxylase deficiency, holocarboxylase synthetase deficiency, OMIM 253270	Failure to report the significant elevation of hydroxyacylcarnitine species of chain length C14-C18	1	40	2.5%
CDG	2021-A	2021	Alcohol Abuse	A normal profile interpretation without additional Diagnostic Suggestions	1	62	1.6%
	2021-B	2021	MAN1B1-CDG	A normal profile interpretation without additional Diagnostic Suggestions	1	62	1.6%
	2021-E	2021	SLC35A2-CDG	A normal profile interpretation without additional Diagnostic Suggestions	2	62	3.2%
DPT CH	2021-A	2021	Alfa-mannosidosis (OMIM 248500)	Missed the correct diagnosis and did not recommend any suitable further testing	2	21	9.5%
DPT CZ	2021-A	2021	Alfa-mannosidosis (OMIM 248500)	Failure to carry out oligosaccharide analysis and to recommend this mandatory test	1	19	5.3%
	2021-E	2021	Hyper IgD syndrome	Failure to recognize abnormal excretion of mevalonolactone and to recommend mevalonolactone analysis based on clinical information	1	19	5.3%
DPT FR	2021-A	2021	Alfa-mannosidosis (OMIM 248500)	Failure to carry out oligosaccharide analysis and to recommend this mandatory test	1	20	5.0%
	2021-F	2021	Hyperprolinaemia type II due to delta 1-pyrroline-5-carboxylate (P5C) dehydrogenase deficiency (ALDH4A1 gene)	Failure to detect an increase of proline	1	20	5.0%
DPT NL	2021-A	2021	Alfa-mannosidosis (OMIM 248500)	Failure to report abnormal oligosaccharides or failure to perform and recommend oligosaccharide analysis	1	18	5.6%
DPT UK	2021-A	2021	Alfa-mannosidosis (OMIM 248500)	Failure to recommend any suitable further testing	1	21	4.8%
	2021-D	2021	HMG CoA lyase deficiency	Failure to detect the increased 3 hydroxy 3 methylglutarate	1	21	4.8%
	2021-F	2021	Citrullinaemia Type 1	Failure to detect the increased concentration of citrulline	1	21	4.8%
QLOU Barcelona	2021-E	2021	Glutaric aciduria type I low excretor	Failure to identify this condition	2	63	3.2%
QLOU Heidelberg	2021-A	2021	Short-chain enoyl-CoA hydratase deficiency	Reporting "normal" without adequate further recommendations	2	71	2.8%
	2021-C	2021	Alkaptonuria	Failure to identify Alkaptonuria	2	71	2.8%
	2021-D	2021	3-methylcrotonyl-CoA carboxylase deficiency	incorrect abnormal diagnosis	2	71	2.8%
	2021-F	2021	Medium-chain acyl-CoA dehydrogenase deficiency	Failure to identify the relevant glycine conjugates	5	71	7.0%
QLOU Sheffield	2021-D	2021	3-Hydroxyisobutyric aciduria due to Methylmalonate semialdehyde dehydrogenase deficiency	Failure to identify this condition	1	71	1.4%
	2021-E	2021	Malonic aciduria due to Malonyl-CoA decarboxylase deficiency	Failure to identify the increased Malonic acid	1	71	1.4%
UMPS	2021-B	2021	MPS VI	Reporting as normal/no diagnosis	2	83	2.4%
	2021-D	2021	MPS II	Reporting a normal DS excretion	1	83	1.2%
	2021-F	2021	MPS-IIIA	Reporting a normal profile	2	83	2.4%
				Totals	**38**	**570**	**6.7%**

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