Lipids in Serum Pilot

Scheme Code: LIS

Aim:	Comparison of assays outcomes for various lipids in serum relevant to the diagnosis of inborn errors of metabolism, in respect to median and target values.
Status:	New pilot scheme starting in 2024

General

Eligibility Requirements:	Participants must produce their own results and cannot send samples to a sub-contracted (or cluster) laboratory **The number of participants for 2024 is limited**
Use of cluster labs allowed?	No
No of organising centres:	One
Geographic area:	Worldwide
Price:	0 Euro / 0 GBP / 0 USD.
Scientific Advisor:	Dr Susanna Goorden, admin@erndim.org
Scheme Organiser:	MCA Laboratory, Netherlands

EQA Samples

Sample volume:	1ml
Sample type:	Matrix of human serum spiked with commercially available analytes
Analytes:	See Appendix 2 (page 3) for details

Scheme Design

Sample design/selection:	Scientific Advisor and deputy Scientific Advisor
Sample manufacture subcontracted to:	MCA Laboratory
Sample aliquoting subcontracted to:	MCA Laboratory
Sample Dispatch subcontracted to:	MCA Laboratory, one dispatch per year (Feb 2024)
Country samples will be dispatched from:	Netherlands
No of samples/year:	8

Results Submission

No of submission deadlines/year:	8
Submission of results:	Online (ERNDIM-MCA website)
Results to be submitted:	Quantitative (all components) & qualitative (only N-Palmitoyl-O-Phosphocholineserine [PPCS]) - participants will be asked to report if PPCS values are increased according to local reference values.

Scoring of results:

Analysis:	Yes
Interpretation:	Yes

Reports:

Interim Reports	Published 14 days after each submission deadline
Individual Lab Annual Reports	Published 14 days after the last submission deadline
Scheme Annual Report (AR)	Published in Jan-Feb of the following year

Appendix 1: Provisional 2024 Calendar

Year	Month	LIS Pilot
-1	Sep	R
	Oct	R
	Nov	R
	Dec	
Scheme Year	Jan	
	Feb	D
	Mar	S
	Apr	S
	May	S
	Jun	S
	Jul	S
	Aug	S
	Sep	S
	Oct	S
	Nov	ILAR
	Dec	
+1	Jan	AR

R = Registration open
D = Sample Dispatch
S = Submission Deadline
ILAR = Individual Laboratory Annual Report
AR = Annual Report published

Appendix 2: Analytes included in the 2024 pilot scheme

Metabolite	Disorder	Group of disorders	SAS/new
Lysosphingolipids			
Lyso-sphingomyelin (Lyso-SM)	Niemann Pick Disease type A/B (NPA/B)	Sphingolipidoses	SAS
Lyso-globotriaosylceramide (Lyso-Gb3)	Fabry Disease	Sphingolipidoses	SAS
Glucosylsphingosine	Gaucher Disease	Sphingolipidoses	SAS
N-palmitoyl-O-phosphocholineserine (PPCS); (previously known as Lysosphingomyelin-509)	Niemann Pick Disease type C (NPC) and Niemann Pick Disease type A/B (NPA/B)	Sphingolipidoses	New
Lyso-monosialoganglioside 1 (Lyso-GM1)	GM1 gangliosidosis	Sphingolipidoses	New
Lyso-monosialoganglioside 2 (Lyso-GM2)	GM2 gangliosidoses (Tay Sachs and Sandhoff disease)	Sphingolipidoses	New
Oxysterols			
Cholestane-3β,5α,6β-triol	Niemann Pick Disease type C (NPC)	Sphingolipidoses	SAS
7-ketocholesterol (7-KC)	Niemann Pick Disease type C (NPC)	Sphingolipidoses	SAS
Lysophospatidylcholines			
C26:0-lysophospatidylcholine (C26:0-lysoPC)	X-linked adrenoleukodystrophy (X-ALD), D-bifunctional protein (DBP) deficiency, peroxisomal acyl-CoA type 1 (ACOX1) deficiency and Zellweger Spectrum Disorders (ZSD)	Peroxisomal disorders	SAS (pilot in 2023)
Sterols			
Cholestanol	Cerebrotendinous Xanthomatotic (CTX)	Sterol biogenesis disorders	SAS
7-dehydrocholesterol (7-DHC)	Smith Lemli Opitz Syndrome (SLO)	Sterol biogenesis disorders	SAS
Desmosterol	Desmosterolosis	Sterol biogenesis disorders	New
Sitosterol	Sitosterolemia	Sterol transport disorders	New

END