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 started 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 2025 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 2025)
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 2025 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 2025 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
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
Lathosterol	Lathosterolosis	Sterol biogenesis disorders	New
Sitosterol	Sitosterolemia	Sterol transport disorders	New
Ubiquinones			
Coenzyme Q10 (Ubiquinone)	Primary and secondary Coenzyme Q10 deficiencies	Mitochondrial disorders	SAS

END