Newborn Screening Quality Assurance Program
Combined Disease-Specific Proficiency Testing Program
2026 Quarter 1 Report
Provided by the Newborn Screening and Molecular Biology Branch

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Issued: May 13, 2026 **Volume 1, No 1**

REPORT AUTHORIZATION
Dr. Stanimila Nikolova, Laboratory Chief of the Proficiency Testing and Reference Materials team, reviewed and authorized this report.

CONFIDENTIALITY STATEMENT
NSQAP participant information and evaluations are strictly confidential and only available by accessing the NSQAP Participant Portal.

Event Summary

This report summarizes ALD, CAH, LSD, TOXO, SMA and TREC proficiency testing (PT) data received during the 2026 Quarter 1 Newborn Screening Quality Assurance Program's (NSQAP) PT event. Data were collected in the NSQAP participant portal.
https://nbs.dynamics365portals.us/

On January 28, 2026, NSQAP distributed panels of five unknown dried blood spot (DBS) specimens to all active participants.

- The Adrenoleukodystrophy (ALDPT) panel contained predetermined concentrations of C24:0-lysophosphatidylcholine (24:0-LPC) and C26:0-lysophosphatidylcholine (26:0-LPC).
- The Second-tier Congenital Adrenal Hyperplasia (CAHPT) panel contained predetermined concentrations of 17 α-Hydroxyprogesterone (17OHP), 4-Androstenedione (4AD), Cortisol (Cort), 11-Deoxycortisol (11D), 21-Deoxycortisol (21D).
- The Lysosomal Storage Disorders (LSDPT) panel contained predetermined concentrations of Galactoceramidase (GALC), Acid α-Glucosidase (GAA), α-L-Iduronidase (IDUA), α-Galactosidase (GLA), β-Glucocerebrosidase (GBA), Acid Sphingomyelinase (ASM), Iduronate-2-Sulfatase (I2S).
- The anti-*Toxoplasma* Antibodies (TOXOPT) panel contained predetermined concentrations of *Toxoplasma gondii* IgM Antibodies (TOXO).
- The Spinal Muscular Atrophy (SMAPT) panel contained predetermined copies of Survival Motor Neuron 1 (*SMN1*) Exon 7.
- The T-cell Receptor Excision Circle (TRECPT) panel contained predetermined concentrations of T-cell Receptor Excision Circle (TREC).

We processed 7,541 results from 513 laboratories for this report.

Specimen Consensus

If more than 10 U.S. laboratories report results, we require at least 80% of those laboratory results to agree with the CDC result in order to evaluate a specimen. If fewer than 80% of the reported results agree with the CDC result, the specimen will be marked as “Not Evaluated”.

If fewer than 10 U.S. laboratories report results, we do not apply the 80% consensus threshold. In these cases, all submitted results are evaluated, and the specimen will be evaluated regardless of the level of agreement with the CDC result. NSQAP occasionally challenges cutoff levels by enriching samples near those levels. The NSQAP PT Committee closely reviews all specimen data. Specimens that are not evaluated are considered educational.

All specimens included in the 2026 Q1 Combined Disease-Specific PT event met the 80% criterion.

Evaluations

NSQAP evaluated each reported result as “Acceptable” or “Unacceptable.” For each analyte and specimen, the participating laboratory’s assessment must match the CDC certified assessment to achieve an “Acceptable” evaluation. When assessments differ, the evaluation will be “Unacceptable.” NSQAP does not identify “Unacceptable” results as “false negative” or “false positive.” It is the responsibility of the laboratory to categorize “Unacceptable” results according to their protocols and policies.

ALDPT panels were distributed to 99 laboratories; 89 laboratories submitted results. Of these, 40 were domestic and 48 were international. There were 15 unacceptable evaluations.

LSDPT panels were distributed to 88 laboratories; 79 laboratories submitted results. Of these, 36 were domestic and 43 were international. There were 12 unacceptable evaluations.

CAHPT panels were distributed to 57 laboratories; 46 laboratories submitted results. Of these, 8 were domestic and 38 were international. There were 11 unacceptable evaluations.

TOXOPT panels were distributed to 15 laboratories; 14 laboratories submitted results. Of these, 2 were domestic and 12 were international. There were 10 unacceptable evaluations.

SMAPT panels were distributed to 156 laboratories; 145 laboratories submitted results. Of these, 36 were domestic and 109 were international. There were 6 unacceptable evaluations.

TRECPT panels were distributed to 153 laboratories; 144 laboratories submitted results. Of these, 43 were domestic and 101 were international. There were 19 unacceptable evaluations.

Proficiency Testing Materials Preparation

NSQAP produces PT specimens from adult donor blood products and purchased umbilical cord blood, adjusted to 50% ± 1% hematocrit before applying to filter paper. NSQAP tests PT specimens for homogeneity, accuracy, stability, and suitability for newborn screening assays.

ALDPT specimens were prepared from human whole blood, which was adjusted to a hematocrit of 50 ± 1% and enriched with the biomarkers 24LPC and 26LPC. Expected values for each were determined by LC-MS/MS in units of µmol/L blood.
Clinical assessments were based on the NSQAP cut-off of 0.20 µmol/L blood for 24LPC and 0.15 µmol/L blood for 26LPC.

LSDPT specimens were produced by combining heat-treated, doubly leuko-depleted human red blood cells with heat-treated charcoal stripped serum and adjusted to a 50% ± 1% hematocrit. The prepared blood was enriched with recombinant enzymes to achieve normal results for non-target analytes. Clinical assessments were based on the NSQAP cut-off of 1.50 (µmol/hr/L) for ABG, 1.35 (µmol/hr/L) for ASM, 1.80 (µmol/hr/L) for GAA, 0.60 (µmol/hr/L) for GALC, 4.90 (µmol/hr/L) for GLA, 1.35 (µmol/hr/L) for IDUA, and 5.30 (µmol/hr/L) for I2S.

2nd-tier CAHPT specimens were prepared at 50% ± 1% hematocrit, with different enrichments of five biomarkers for congenital adrenal hyperplasia (CAH): 17 α-hydroxyprogesterone (17OHP), 4-androstenedione (4AD), cortisol (Cort), 11-deoxycortisol (11D), and 21-deoxycortisol (21D). Expected values (sum of endogenous and enrichment values) were determined by LC-MS/MS. For determination of the Clinical Assessment (CA), NSQAP applied the formula: clinical ratio = (17OHP + 4AD)/CORT. A cutoff of 1.0 ng/mL was used to assess whether the specimen was classified as Within Normal Limits or Outside Normal Limits.

TOXOPT specimens were prepared from serum samples positive for Toxoplasma IgG and IgM purchased from SeraCare (Medford, Massachusetts) and from human serum positive for exposure to Toxoplasma gondii from a CDC specimen bank. All serum samples were mixed with washed red blood cells. A cutoff of 10 EIU/mL was used to assess specimens.

SMAPT specimens were prepared from human blood, including leukocyte-depleted blood, and leukocyte-depleted blood containing Epstein-Barr Virus (EBV) transduced lymphocytes from anonymous SMA patients, carriers, or unaffected individuals. Screen-positive and screen-negative status were determined based on *SMN1* and reference gene performance when screened using the LDT Real Time PCR - SMN1/TREC AND Reference Gene run in a single tube genotyping method.

TRECPT specimens were prepared from human blood, including umbilical cord blood from unaffected individuals, leukocyte-depleted blood, and leukocyte-depleted blood containing EBV-transduced lymphocytes. Screen-positive and screen-negative status were determined based on TREC and reference gene performance when screened using the “LDT Real Time PCR - SMN1/TREC AND Reference Gene run in a single tube” genotyping method.

Proficiency Testing Data Handling

Tables in this report include Specimen Certifications, Frequency Distribution of Participants’ Clinical Assessments, Overall Statistics, and Mean Reported Concentrations by Method. Confidential individual laboratory data certification and evaluations are in the Report Results section of the NSQAP Participant Portal. https://nbs.dynamics365portals.us/

Table of Contents

1. ALDPT (µmol/L)

WNL = Within Normal Limits; ONL = Outside Normal Limits; NE = Not Evaluated; EV = Expected Value

1.1. Specimen Certification

Specimen Number	24LPC EV	24LPC Assessment	26LPC EV	26LPC Assessment
20261002001	1.31	ONL	1.23	ONL
20261002002	0.10	WNL	0.02	WNL
20261002003	0.09	WNL	0.03	WNL
20261002004	1.59	ONL	1.52	ONL
20261002005	0.11	WNL	0.03	WNL

1.2. Frequency Distribution

Specimen Number	Analyte	ONL	WNL
20261002001	24LPC	40	3
20261002002	24LPC	0	43
20261002003	24LPC	0	43
20261002004	24LPC	39	4
20261002005	24LPC	1	42
20261002001	26LPC	89	0
20261002002	26LPC	1	88
20261002003	26LPC	3	86
20261002004	26LPC	89	0
20261002005	26LPC	3	86

1.3. Mean Reported Concentrations 24LPC - First Tier

ALDPT (µmol/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Specimen Number	Analyte	N	Mean	SD
20261002001	24LPC	42	1.77	0.62
20261002002	24LPC	42	0.37	0.20
20261002003	24LPC	42	0.47	0.26
20261002004	24LPC	42	1.93	0.62
20261002005	24LPC	42	0.52	0.26

1.4. Mean Reported Concentrations 24LPC - Second Tier

ALDPT (µmol/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Specimen Number	Analyte	N	Mean	SD
20261002001	24LPC	9	1.92	0.47
20261002002	24LPC	7	0.21	0.13
20261002003	24LPC	8	0.17	0.12
20261002004	24LPC	9	2.14	0.57
20261002005	24LPC	8	0.22	0.17

1.5. Mean Reported Concentrations 26LPC - First Tier

ALDPT (µmol/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Specimen Number	Analyte	N	Mean	SD
20261002001	26LPC	85	1.41	0.36
20261002002	26LPC	86	0.29	0.19
20261002003	26LPC	86	0.33	0.22
20261002004	26LPC	84	1.71	0.38
20261002005	26LPC	86	0.35	0.22

1.6. Mean Reported Concentrations 26LPC - Second Tier

ALDPT (µmol/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Specimen Number	Analyte	N	Mean	SD
20261002001	26LPC	29	1.28	0.37
20261002002	26LPC	19	0.06	0.08
20261002003	26LPC	21	0.07	0.09
20261002004	26LPC	29	1.67	0.51
20261002005	26LPC	21	0.07	0.11

1.7. Cutoff Statistics by Method - 24LPC First Tier

ALDPT (µmol/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Method	N	Mean	SD	Median	Min	Max
Non-derivatized - NeoBase 2 MS/MS	32	1.06	0.40	0.97	0.59	2.34
Non-derivatized - NeoMass AAAC 3.0	3	0.59	0.37	0.62	0.20	0.94
Other	3	0.91	0.68	1.10	0.16	1.47

1.8. Cutoff Statistics by Method 24LPC Second Tier

ALDPT (µmol/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Method	N	Mean	SD	Median	Min	Max
LC-MS/MS positive ion mode	4	0.31	0.17	0.26	0.18	0.55

1.9. Cutoff Statistics by Method - 26LPC First Tier

ALDPT (µmol/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Method	N	Mean	SD	Median	Min	Max
Flow Injection Analysis (FIA) - MS/MS non-derivatized non-kit	4	0.60	0.36	0.45	0.37	1.13
LC-MS/MS negative ion mode	7	0.16	0.02	0.16	0.13	0.18
Non-derivatized - NeoBase 2 MS/MS	59	0.58	0.23	0.52	0.33	1.19
Non-derivatized - NeoMass AAAC 3.0	3	1.38	0.79	1.80	0.47	1.87
Other	6	0.43	0.38	0.31	0.11	1.15

1.10. Cutoff Statistics by Method - 26LPC Second Tier

ALDPT (µmol/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Method	N	Mean	SD	Median	Min	Max
LC-MS/MS negative ion mode	12	0.17	0.04	0.16	0.12	0.26
LC-MS/MS non-kit	3	0.28	0.12	0.30	0.15	0.38
LC-MS/MS positive ion mode	8	0.22	0.09	0.20	0.10	0.40

1.11. Method Algorithms Reported by ≥3 Laboratories

First-Tier Method	Second-Tier Method	Number of Labs
Non-derivatized - NeoBase 2 MS/MS	N/A	41
Non-derivatized - NeoBase 2 MS/MS	LC-MS/MS negative ion mode	9
LC-MS/MS negative ion mode	N/A	8
Non-derivatized - NeoBase 2 MS/MS	LC-MS/MS positive ion mode	6
Other	N/A	5
Non-derivatized - NeoBase 2 MS/MS	Non-derivatized - NeoBase 2 MS/MS	3
Non-derivatized - NeoMass AAAC 3.0	N/A	3

2. CAHPT (ng/mL serum)

WNL = Within Normal Limits; ONL = Outside Normal Limits; NE = Not Evaluated; EV = Expected Value

2.1. Specimen Certification

Specimen Number	11D EV	17OHP EV	21D EV	4AD EV	Clinical Ratio EV	CORT EV	Clinical Assessment
20261010001	11.0	91.6	41.6	41.4	6.4	20.8	ONL
20261010002	21.0	66.6	1.6	26.4	0.8	120.8	WNL
20261010003	11.0	51.6	1.6	26.4	0.8	100.8	WNL
20261010004	51.0	11.6	1.6	21.4	0.8	40.8	WNL
20261010005	51.0	11.6	11.6	21.4	0.8	40.8	WNL

2.2. Frequency Distribution

Specimen Number	ONL	WNL	Not Reported
20261010001	44	2	0
20261010002	3	43	0
20261010003	3	43	0
20261010004	1	43	2
20261010005	2	42	2

2.3. Clinical Ratio Cutoff Values

CAHPT ((17OHP+4AD)/CORT)

Lab Type	Mean	Mode	Min	Max
All Laboratories	1.70	1	0.4	8.5
Domestic	1.06	1	0.9	1.4
International	1.83	1	0.4	8.5

2.4. Overall Statistics

CAHPT (ng/mL serum), SD = Standard Deviation, N < 3 participating laboratories are not shown

Specimen Number	Analyte	N	Mean	SD
20261010001	11D	38	10.33	2.70
20261010002	11D	38	19.20	4.46
20261010003	11D	38	9.32	2.75
20261010004	11D	37	51.25	12.49
20261010005	11D	37	45.10	12.15

Specimen Number	Analyte	N	Mean	SD
20261010001	17OHP	46	83.52	26.38
20261010002	17OHP	46	59.75	18.34
20261010003	17OHP	46	59.83	20.32
20261010004	17OHP	43	8.11	2.85
20261010005	17OHP	43	8.05	6.17
20261010001	21D	39	30.61	8.56
20261010002	21D	33	0.35	0.60
20261010003	21D	34	0.41	0.55
20261010004	21D	34	0.32	0.53
20261010005	21D	37	6.71	2.63
20261010001	4AD	45	36.12	10.84
20261010002	4AD	45	22.37	6.61
20261010003	4AD	45	24.01	7.22
20261010004	4AD	43	19.13	5.73
20261010005	4AD	43	17.90	5.15
20261010001	CORT	45	19.20	6.12
20261010002	CORT	45	114.59	30.97
20261010003	CORT	45	100.07	23.21
20261010004	CORT	41	37.64	9.55
20261010005	CORT	42	41.12	10.96

2.4.1. Clinical Ratio LC-MS/MS

CAHPT ((17OHP+4AD)/CORT), SD = Standard Deviation, N < 3 participating laboratories are not shown

Specimen Number	Clinical Ratio N	Clinical Ratio Mean	Clinical Ratio SD	Clinical Ratio Min	Clinical Ratio Max
20261010001	45	6.35	1.76	0.13	9.17
20261010002	44	0.70	0.18	0.02	1.00
20261010003	44	0.83	0.11	0.66	1.10
20261010004	41	0.77	0.13	0.29	1.11
20261010005	41	0.69	0.12	0.43	0.97

3. LSDPT (µmol/hr/L)

WNL = Within Normal Limits; ONL = Outside Normal Limits; NE = Not Evaluated; EV = Expected Value

3.1. Expected Values

	I2S EV	ASM EV	IDUA EV	ABG EV	GAA EV	GALC EV	GLA EV
Specimen Number	LC-MS/MS	FIA-MS/MS	FIA-MS/MS	FIA-MS/MS	FIA-MS/MS	FIA-MS/MS	FIA-MS/MS
20261013001	11.04	2.39	5.03	5.88	7.40	6.53	17.44
20261013002	21.66	7.35	5.12	6.21	0.26	5.68	13.71
20261013003	22.06	7.10	5.04	5.73	10.94	0.08	12.64
20261013004	20.79	7.82	0.05	5.82	10.54	5.96	12.43
20261013005	0.07	6.96	4.77	5.91	10.95	5.80	10.87

3.2. Expected Clinical Assessments

Specimen Number	I2S Assessment	ASM Assessment	IDUA Assessment	ABG Assessment	GAA Assessment	GALC Assessment	GLA Assessment
20261013001	WNL	WNL	WNL	WNL	WNL	WNL	WNL
20261013002	WNL	WNL	WNL	WNL	ONL	WNL	WNL
20261013003	WNL	WNL	WNL	WNL	WNL	ONL	WNL
20261013004	WNL	WNL	ONL	WNL	WNL	WNL	WNL
20261013005	ONL	WNL	WNL	WNL	WNL	WNL	WNL

3.3. Domestic Laboratories - Frequency Distribution of Reported Clinical Assessments

WNL = Within Normal Limits; ONL = Outside Normal Limits

Analyte	Assessment	Specimen 20261013001	Specimen 20261013002	Specimen 20261013003	Specimen 20261013004	Specimen 20261013005
ABG	ONL	0	0	0	0	0
ABG	WNL	8	8	8	8	8
ASM	ONL	0	0	0	0	0
ASM	WNL	5	5	5	5	5
GAA	ONL	0	36	0	0	0
GAA	WNL	36	0	36	36	36
GALC	ONL	0	0	15	0	0
GALC	WNL	17	17	2	17	17
GLA	ONL	0	0	0	0	0

Analyte	Assessment	Specimen 20261013001	Specimen 20261013002	Specimen 20261013003	Specimen 20261013004	Specimen 20261013005
GLA	WNL	11	11	11	11	11
I2S	ONL	0	0	0	0	18
I2S	WNL	18	18	18	18	0
IDUA	ONL	0	0	0	35	0
IDUA	WNL	35	35	35	0	35

3.4. International Laboratories - Frequency Distribution of Reported Clinical Assessments

WNL = Within Normal Limits; ONL = Outside Normal Limits

Analyte	Assessment	Specimen 20261013001	Specimen 20261013002	Specimen 20261013003	Specimen 20261013004	Specimen 20261013005
ABG	ONL	0	0	0	0	0
ABG	WNL	32	32	32	32	32
ASM	ONL	5	0	0	0	0
ASM	WNL	23	28	28	28	28
GAA	ONL	0	35	0	0	0
GAA	WNL	36	1	36	36	36
GALC	ONL	0	0	25	0	0
GALC	WNL	26	26	1	26	26
GLA	ONL	0	0	0	0	0
GLA	WNL	33	33	33	33	33
I2S	ONL	1	0	0	0	14
I2S	WNL	13	14	14	14	0
IDUA	ONL	1	0	0	38	0
IDUA	WNL	38	39	39	1	39

3.5. Overall Statistics

LSDPT (µmol/hr/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Specimen Number	Analyte	EV	N	Mean	SD
20261013001	ABG	5.88	41	19.18	80.03
20261013002	ABG	6.21	41	7.30	2.58
20261013003	ABG	5.73	41	8.38	8.59

Specimen Number	Analyte	EV	N	Mean	SD
20261013004	ABG	5.82	41	6.81	2.55
20261013005	ABG	5.91	41	7.15	2.78
20261013001	ASM	2.39	34	2.66	0.65
20261013002	ASM	7.35	34	8.58	2.23
20261013003	ASM	7.10	34	8.58	2.36
20261013004	ASM	7.82	34	8.68	2.42
20261013005	ASM	6.96	34	7.91	2.07
20261013001	GAA	7.40	73	10.21	8.79
20261013002	GAA	0.26	71	0.30	0.59
20261013003	GAA	10.94	73	17.09	11.57
20261013004	GAA	10.54	73	16.14	10.88
20261013005	GAA	10.95	73	16.65	11.21
20261013001	GALC	6.53	44	7.29	1.70
20261013002	GALC	5.68	44	6.29	1.52
20261013003	GALC	0.08	44	0.82	3.85
20261013004	GALC	5.96	44	6.62	1.72
20261013005	GALC	5.80	44	6.41	1.57
20261013001	GLA	17.44	45	21.85	13.84
20261013002	GLA	13.71	45	18.00	15.52
20261013003	GLA	12.64	45	17.69	14.92
20261013004	GLA	12.43	45	16.81	15.41
20261013005	GLA	10.87	45	15.13	14.49
20261013001	I2S	11.04	32	17.68	11.81
20261013002	I2S	21.66	32	30.72	20.40
20261013003	I2S	22.06	32	31.60	22.60
20261013004	I2S	20.79	32	30.70	22.66
20261013005	I2S	0.07	32	0.63	1.29
20261013001	IDUA	5.03	75	7.68	6.18
20261013002	IDUA	5.12	75	9.28	8.11
20261013003	IDUA	5.04	75	9.47	8.25

Specimen Number	Analyte	EV	N	Mean	SD
20261013004	IDUA	0.05	73	0.35	0.95
20261013005	IDUA	4.77	75	9.24	8.96

3.6. Summary of Reported Results by Method

LSDPT (µmol/hr/L), SD = Standard Deviation, Only N ≥3 participating laboratories are shown

Analyte: ABG

Analyte	Specimen Number	Method	N	Mean	SD
ABG	20261013001	LC-MS/MS non-kit	5	6.99	1.74
ABG	20261013001	NeoLSD MSMS	27	6.29	1.61
ABG	20261013002	LC-MS/MS non-kit	5	6.96	1.92
ABG	20261013002	NeoLSD MSMS	28	6.45	1.66
ABG	20261013003	LC-MS/MS non-kit	5	6.79	1.57
ABG	20261013003	NeoLSD MSMS	27	6.34	1.62
ABG	20261013004	LC-MS/MS non-kit	5	6.76	1.68
ABG	20261013004	NeoLSD MSMS	28	5.98	1.49
ABG	20261013005	LC-MS/MS non-kit	5	7.25	1.88
ABG	20261013005	NeoLSD MSMS	28	6.15	1.46

Analyte: ASM

Analyte	Specimen Number	Method	N	Mean	SD
ASM	20261013001	LC-MS/MS non-kit	5	3.03	0.66
ASM	20261013001	NeoLSD MSMS	25	2.68	0.53
ASM	20261013002	LC-MS/MS non-kit	5	9.64	3.26
ASM	20261013002	NeoLSD MSMS	25	8.48	1.48
ASM	20261013003	LC-MS/MS non-kit	5	9.77	3.05
ASM	20261013003	NeoLSD MSMS	25	8.56	1.77
ASM	20261013004	LC-MS/MS non-kit	5	10.13	3.49
ASM	20261013004	NeoLSD MSMS	25	8.54	1.65
ASM	20261013005	LC-MS/MS non-kit	5	9.41	2.75
ASM	20261013005	NeoLSD MSMS	25	7.80	1.46

Analyte: GAA

Analyte	Specimen Number	Method	N	Mean	SD
GAA	20261013001	Digital Microfluidic Fluorescence	9	24.20	3.08
GAA	20261013001	Fluorometric manual LSD - non-kit	5	2.48	2.69
GAA	20261013001	LC-MS/MS non-kit	12	6.67	1.61
GAA	20261013001	NeoLSD MSMS	40	7.61	1.01
GAA	20261013002	Digital Microfluidic Fluorescence	7	1.53	0.38
GAA	20261013002	Fluorometric manual LSD - non-kit	5	0.09	0.12
GAA	20261013002	LC-MS/MS non-kit	13	0.09	0.08
GAA	20261013002	NeoLSD MSMS	39	0.14	0.08
GAA	20261013003	Digital Microfluidic Fluorescence	9	39.46	4.13
GAA	20261013003	Fluorometric manual LSD - non-kit	5	4.55	4.81
GAA	20261013003	LC-MS/MS non-kit	12	11.74	2.97
GAA	20261013003	NeoLSD MSMS	39	13.02	2.16
GAA	20261013004	Digital Microfluidic Fluorescence	9	38.77	5.34
GAA	20261013004	Fluorometric manual LSD - non-kit	5	4.47	4.87
GAA	20261013004	LC-MS/MS non-kit	12	11.16	2.72
GAA	20261013004	NeoLSD MSMS	40	12.63	1.53
GAA	20261013005	Digital Microfluidic Fluorescence	9	39.95	6.26
GAA	20261013005	Fluorometric manual LSD - non-kit	5	4.13	4.08
GAA	20261013005	LC-MS/MS non-kit	12	11.77	3.16
GAA	20261013005	NeoLSD MSMS	40	13.02	1.60

Analyte: GALC

Analyte	Specimen Number	Method	N	Mean	SD
GALC	20261013001	LC-MS/MS non-kit	7	7.98	1.99
GALC	20261013001	NeoLSD MSMS	32	7.47	1.01
GALC	20261013002	LC-MS/MS non-kit	7	6.74	1.94
GALC	20261013002	NeoLSD MSMS	32	6.44	0.68
GALC	20261013003	LC-MS/MS non-kit	7	0.13	0.06
GALC	20261013003	NeoLSD MSMS	29	0.14	0.08
GALC	20261013004	LC-MS/MS non-kit	7	7.03	2.31

Analyte	Specimen Number	Method	N	Mean	SD
GALC	20261013004	NeoLSD MSMS	32	6.80	0.87
GALC	20261013005	LC-MS/MS non-kit	7	6.76	2.01
GALC	20261013005	NeoLSD MSMS	32	6.58	0.76

Analyte: GLA

Analyte	Specimen Number	Method	N	Mean	SD
GLA	20261013001	Fluorometric manual LSD - non-kit	3	16.86	6.18
GLA	20261013001	LC-MS/MS non-kit	6	19.27	4.95
GLA	20261013001	NeoLSD MSMS	30	18.00	2.42
GLA	20261013002	Fluorometric manual LSD - non-kit	3	13.80	10.22
GLA	20261013002	LC-MS/MS non-kit	6	13.11	3.69
GLA	20261013002	NeoLSD MSMS	30	13.73	2.03
GLA	20261013003	Fluorometric manual LSD - non-kit	3	11.98	7.62
GLA	20261013003	LC-MS/MS non-kit	6	12.70	2.84
GLA	20261013003	NeoLSD MSMS	30	14.08	3.84
GLA	20261013004	Fluorometric manual LSD - non-kit	3	11.69	7.75
GLA	20261013004	LC-MS/MS non-kit	6	12.33	3.23
GLA	20261013004	NeoLSD MSMS	30	12.83	1.68
GLA	20261013005	Fluorometric manual LSD - non-kit	3	9.70	5.61
GLA	20261013005	LC-MS/MS non-kit	6	10.75	2.64
GLA	20261013005	NeoLSD MSMS	30	11.35	1.53

Analyte: I2S

Analyte	Specimen Number	Method	N	Mean	SD
I2S	20261013001	Fluorometric manual LSD - non-kit	5	30.38	22.01
I2S	20261013001	LC-MS/MS non-kit	19	16.76	7.65
I2S	20261013002	Fluorometric manual LSD - non-kit	5	55.31	36.71
I2S	20261013002	LC-MS/MS non-kit	19	28.85	12.86
I2S	20261013003	Fluorometric manual LSD - non-kit	5	57.31	35.62
I2S	20261013003	LC-MS/MS non-kit	19	29.50	18.05
I2S	20261013004	Fluorometric manual LSD - non-kit	5	57.96	38.61

Analyte	Specimen Number	Method	N	Mean	SD
I2S	20261013004	LC-MS/MS non-kit	19	28.49	15.93
I2S	20261013005	Fluorometric manual LSD - non-kit	5	3.09	1.90
I2S	20261013005	LC-MS/MS non-kit	19	0.21	0.19

Analyte: IDUA

Analyte	Specimen Number	Method	N	Mean	SD
IDUA	20261013001	Digital Microfluidic Fluorescence	9	19.22	3.54
IDUA	20261013001	Fluorometric manual LSD - non-kit	3	4.14	1.61
IDUA	20261013001	LC-MS/MS non-kit	12	5.75	1.78
IDUA	20261013001	NeoLSD MSMS	45	5.32	0.86
IDUA	20261013002	Digital Microfluidic Fluorescence	9	22.36	1.68
IDUA	20261013002	Fluorometric manual LSD - non-kit	3	6.97	2.05
IDUA	20261013002	LC-MS/MS non-kit	11	6.04	1.55
IDUA	20261013002	NeoLSD MSMS	45	6.08	1.04
IDUA	20261013003	Digital Microfluidic Fluorescence	9	21.97	2.86
IDUA	20261013003	Fluorometric manual LSD - non-kit	3	6.64	2.09
IDUA	20261013003	LC-MS/MS non-kit	11	6.10	1.10
IDUA	20261013003	NeoLSD MSMS	44	6.28	1.13
IDUA	20261013004	Digital Microfluidic Fluorescence	7	1.81	0.28
IDUA	20261013004	Fluorometric manual LSD - non-kit	3	0.03	0.14
IDUA	20261013004	LC-MS/MS non-kit	12	0.10	0.13
IDUA	20261013004	NeoLSD MSMS	44	0.09	0.08
IDUA	20261013005	Digital Microfluidic Fluorescence	9	24.51	2.72
IDUA	20261013005	Fluorometric manual LSD - non-kit	3	6.29	1.83
IDUA	20261013005	LC-MS/MS non-kit	11	5.79	1.38
IDUA	20261013005	NeoLSD MSMS	45	5.74	1.06

3.7. Cutoff Statistics

LSDPT (µmol/hr/L), SD = Standard Deviation, N < 3 participating laboratories are not shown

Analyte	Method	N	Mean	SD	Median	Min	Max
ABG	LC-MS/MS non-kit	5	2.07	0.67	2.04	1.30	3.10

Analyte	Method	N	Mean	SD	Median	Min	Max
ABG	NeoLSD MSMS	28	2.09	0.90	2.15	0.00	4.04
ASM	LC-MS/MS non-kit	5	1.22	0.39	1.38	0.56	1.52
ASM	NeoLSD MSMS	25	1.63	0.68	1.46	0.21	3.20
GAA	Digital Microfluidic Fluorescence	9	8.48	1.45	9.00	6.00	10.00
GAA	Fluorometric manual LSD - non-kit	5	0.64	0.62	0.63	0.03	1.61
GAA	LC-MS/MS non-kit	13	1.74	0.46	1.70	1.20	2.73
GAA	NeoLSD MSMS	39	1.88	0.95	1.75	0.00	5.44
GAA	Other	5	2.27	1.22	2.00	1.17	4.00
GALC	LC-MS/MS non-kit	7	0.75	0.57	0.45	0.40	2.00
GALC	NeoLSD MSMS	31	0.81	0.38	0.74	0.00	1.90
GLA	Fluorometric manual LSD - non-kit	3	1.57	0.69	1.81	0.79	2.10
GLA	LC-MS/MS non-kit	6	1.35	0.39	1.26	0.89	2.00
GLA	NeoLSD MSMS	30	2.75	1.13	2.96	0.00	5.36
GLA	Other	3	4.68	3.75	2.75	2.30	9.00
I2S	Fluorometric manual LSD - non-kit	5	14.57	10.53	20.00	1.84	26.00
I2S	LC-MS/MS non-kit	19	5.57	4.19	4.11	0.80	15.73
I2S	Other	7	3.07	1.68	2.40	1.30	5.32
IDUA	Digital Microfluidic Fluorescence	9	4.91	1.17	5.00	3.50	7.00
IDUA	Fluorometric manual LSD - non-kit	3	1.57	0.84	2.00	0.60	2.10
IDUA	LC-MS/MS non-kit	12	1.56	0.76	1.62	0.62	2.73
IDUA	NeoLSD MSMS	44	1.67	0.89	1.74	0.00	4.40
IDUA	Other	5	3.17	2.84	2.67	0.50	8.00

4. TOXOPT

TOXO = anti Toxoplasma Antibodies; EIU = Enzyme International Units; R = Reactive; NR = Not Reactive; US = Unsatisfactory Sample; NE = Not Evaluated
SD = Standard Deviation

4.1. Specimen Certification (EIU/mL serum)

Specimen Number	Expected Value	SD	Clinical Assessment
20261014001	0.3	6.7	Toxoplasmosis antibody non-reactive
20261014002	201.2	22.8	Toxoplasmosis antibody reactive
20261014003	0.0	6.9	Toxoplasmosis antibody non-reactive
20261014004	1,363.6	135.2	Toxoplasmosis antibody reactive
20261014005	4.2	6.9	Toxoplasmosis antibody non-reactive

4.2. Frequency Distribution

Specimen Number	NR	R
20261014001	10	4
20261014002	0	14
20261014003	12	2
20261014004	0	14
20261014005	10	4

4.3. Mean Reported Concentrations

SD = Standard Deviation, N < 3 participating laboratories are not shown
Method/Antibody and units: TOXO Enzyme Immunoassay (EIA) IgM Absorbance (OD)
Mean Reported Cutoff: 0.166
Cutoff Range: 0.100–0.350

Method	Specimen Number	N	Mean	SD
TOXO Enzyme Immunoassay (EIA)	20261014001	8	0.025	0.036
TOXO Enzyme Immunoassay (EIA)	20261014002	8	0.635	0.512
TOXO Enzyme Immunoassay (EIA)	20261014003	8	0.038	0.060
TOXO Enzyme Immunoassay (EIA)	20261014004	8	2.261	0.951
TOXO Enzyme Immunoassay (EIA)	20261014005	8	0.035	0.047

5. SMAPT

5.1. Specimen Certification

Specimen Number	Specimen Description: Copies of SMN1 (exon 7 present)	Specimen Description: Copies of SMN2	Specimen Description: Reference Gene	Clinical Assessment
20261017001	1 (Carrier)	2	Within Normal Limits	Screen Negative (no follow up required)
20261017002	2	2	Within Normal Limits	Screen Negative (no follow up required)
20261017003	Outside Normal Limits or Undetermined	Outside Normal Limits or Undetermined	Outside Normal Limits or Undetermined	Unsatisfactory sample (SMN1 and reference gene out of range)
20261017004	0	4	Within Normal Limits	Screen Positive (SMN1 out of range, reference gene in range)
20261017005	0	2	Within Normal Limits	Screen Positive (SMN1 out of range, reference gene in range)

5.2. Reported Laboratory Genotyping Methods for SMA

N < 3 participating laboratories are not shown

Primary Method	Number of Laboratories
Revvity Eonis™ SCID-SMA kit	47
LDT Real Time PCR - SMN1/TREC AND Reference Gene run in a single tube	27
Revvity NeoMDx RUO	14
ImmunoIVD SPOT-it™ TREC & SMN1 Screening Kit	13
LDT Real Time PCR - SMN1 AND Reference Gene run in a single tube	7
ZenTech Targeted qPCR SMA	7
Trimaris SMA Real-Time PCR Assay	4
ImmunoIVD SPOT-it™ TREC, KREC & SMN1 Screening Kit	3
Other	23

5.3. Reported DNA Extraction Methods

N < 3 participating laboratories are not shown

DNA Extraction Method	Number of Laboratories
Revvity DNA Extraction Solution	60
ImmunoIVD SPOT-it™ TREC & SMN1 Screening Kit reagents	13
Extracta™ DBS	12
ZenTech Targeted qPCR SMA kit reagents	7

DNA Extraction Method	Number of Laboratories
Generation™ DNA Purification and Elution Solutions (S1/S2)	6
In situ/on card (DNA is NOT extracted)	5
Generation™ DNA Elution Solution (S2 only)	4
Trimaris DBS DNA Extraction Kit	4
Other	34

5.4. Reported Reference Genes

N < 3 participating laboratories are not shown

Reference Gene	Number of Laboratories
RNaseP subunit (RPP30)	94
Beta-actin (ACTB)	22
TaqMan™ RNase P Control Reagents Kit	8
Other	21

5.5. Reported Clinical Assessments and Misclassifications

Specimen Number	Screen Negative (no follow up required)	Screen Positive (SMN1 out of range, reference gene in range)	Unsatisfactory sample (SMN1 and reference gene out of range)	Incorrect Clinical Assessments
20261017001	145	0	0	0
20261017002	145	0	0	0
20261017003	4	1	140	5
20261017004	1	144	0	1
20261017005	0	145	0	0

6. TRECPT

6.1. Specimen Certification

Specimen Number	Specimen Description: TREC	Specimen Description: Reference Gene	Clinical Assessment
20261015001	Within Normal Limits	Within Normal Limits	Screen Negative (no follow up required)
20261015002	Outside Normal Limits or Undetermined	Outside Normal Limits or Undetermined	Unsatisfactory sample (TREC and reference gene out of range)
20261015003	Within Normal Limits	Within Normal Limits	Screen Negative (no follow up required)
20261015004	Absent or Below Normal Limits	Within Normal Limits	Screen Positive (TREC out of range, reference gene in range)
20261015005	Absent or Below Normal Limits	Within Normal Limits	Screen Positive (TREC out of range, reference gene in range)

6.2. Reported Laboratory Genotyping Methods for TREC

N < 3 participating laboratories are not shown

Primary Method	Number of Laboratories
Revvity Eonis™ SCID-SMA kit	45
LDT Real Time PCR - TREC/SMN1 AND Reference Gene run in a single tube	23
ImmunoIVD SPOT-it™ TREC & SMN1 Screening Kit	14
Revvity NeoMDx RUO TREC/SMN1 kit	14
EnLite™ Neonatal TREC kit	12
LDT Real Time PCR - TREC AND Reference Gene run in a single tube	9
ImmunoIVD SPOT-it™ TREC, KREC & SMN1 Screening Kit	6
LDT Real Time PCR - TREC only (reference gene run separately)	3
Other	18

6.3. Reported DNA Extraction Methods

N < 3 participating laboratories are not shown

DNA Extraction Method	Number of Laboratories
Revvity DNA Extraction Solutions	56
ImmunoIVD SPOT-it™ TREC & SMN1 Screening Kit reagents	15
EnLite™ (DNA is NOT extracted)	12
Extracta™ DBS with one wash	9
Generation™ DNA Purification and Elution Solutions (S1/S2)	6
In situ/on card (DNA is NOT extracted)	6

DNA Extraction Method	Number of Laboratories
Generation™ DNA Elution Solution (S2 only)	5
ImmunoIVD SPOT-it™ TREC, KREC & SMN1 Screening Kit	5
Other	30

6.4. Reported Reference Genes

N < 3 participating laboratories are not shown

Reference Gene	Number of Laboratories
RNaseP subunit (RPP30)	83
Beta-actin (ACTB)	44
TaqMan™ RNase P Control Reagents Kit	8
Other	9

6.5. Reported Clinical Assessments and Misclassifications

Specimen Number	Screen Negative (no follow up required)	Screen Positive (TREC out of range, reference gene in range)	Unsatisfactory sample (TREC and reference gene out of range)	Incorrect Clinical Assessments
20261015001	144	0	0	0
20261015002	1	4	139	5
20261015003	144	0	0	0
20261015004	1	141	2	3
20261015005	1	133	10	11

This *NEWBORN SCREENING QUALITY ASSURANCE PROGRAM* report is an internal publication distributed to program participants and selected program colleagues. The laboratory quality assurance program is a project cosponsored by the Centers for Disease Control and Prevention (CDC) and the Association of Public Health Laboratories.

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