Proposed Terms

NewSTEPs
A Program of the Association of Public Health Laboratories™

	CF	CAH	CH
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	Date of Presumptive Positive diagnosis: - Therapies may include Enzymes/ Salt - Documentation of phone conversation that changed the care of that infant - Clinic visit	Date of Clinic visit or hospital consultation to evaluate potential diagnosis of CAH: - Standard Confirmatory Testing: Clinician draws electrolytes and 17-OHP - Electrolytes may or may not indicate need for urgent intervention, however a decision is rendered based on laboratory results and clinical presentation of the infant - Advanced Confirmatory Testing: In cases in which exam or presentation strongly suggests diagnosis of CAH, additional adrenal testing may be warranted in consultation with endocrinologist	Date of intervention/ diagnosis: - Confirmatory serology demonstrates elevated TSH/Free T4 - Start of therapy - This typically occurs at the Clinic Visit/ Hospital Consultation
Date of Diagnosis	Date of diagnosis, with diagnosis confirmed upon: - Positive sweat chloride test (CI > 60 mmol/L) to confirm out-of-range screening result - Genotype/ sequencing to identify CFR mutations (on sample taken from the infant) - Result of Nasal Potential Difference Results	Date of diagnosis, with diagnosis confirmed upon: - Elevated 17-OHP (+/- other abnormal adrenal hormone abnormalities) and evaluation by endocrinologist	Date of diagnosis: - Confirmatory serology demonstrates elevated TSH/ Free T4 - Start of therapy - This typically occurs at Clinic Visit/ Hospital Consultation

Proposed Terms

NewSTEPs
A Program of the Association of Public Health Laboratories™

	Hemoglobinopathies		
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional		
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	Earliest time point: - Date that infant is clinically evaluated related to disease - Date that a decision was made to initiate penicillin (or to NOT initiate penicillin)		
Date of Diagnosis	Earliest time point: - Date result from confirmatory testing from a specimen drawn from the baby subsequent to the first newborn screening specimen was reported (biochemical or DNA) - Date results from family studies that confirm NBS result are reported		

Proposed Terms

NewSTEPs
A Program of the Association of Public Health Laboratories™

	PROP: Propionic acidemia	MUT: Methylmalonic acidemia (methylmalonyl-CoA mutase)	Cbl A,B: Methylmalonic acidemia (cobalamin disorders)
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	(this date may be before the NBS result is acted upon) Date diet change/drug and supplement therapy was initiated.	(this date may be before the NBS result is acted upon) Date diet change/drug and supplement therapy was initiated.	(this date may be before the NBS result is acted upon) Date diet change/drug and supplement therapy was initiated.
Date of Diagnosis	Date of the report reflecting that urine organic acids, plasma acylcarnitine profile results are consistent with diagnosis of Propionic Acidemia	Date of the report reflecting that urine organic acids, plasma acylcarnitine profile results, mutation analysis to know subtype are consistent with diagnosis of MMA	Date of the report reflecting that urine organic acids, plasma carnitine, acylcarnitine profile results, mutation analysis to know subtype, are consistent with diagnosis of CBL A,B

Proposed Terms

NewSTEPs
A Program of the Association of Public Health Laboratories™

	IVA: Isovaleric acidemia	3-MCC: 3-Methylcrotonyl-CoA carboxylase deficiency	HMG: 3-Hydroxy-3-methyglutaric aciduria
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	(this date may be before the NBS result is acted upon) Date diet change/drug and supplement therapy was initiated.	(this date may be before the NBS result is acted upon) Date diet change/drug and supplement therapy was initiated.	(this date may be before the NBS result is acted upon) Date diet change/drug and supplement therapy was initiated.
Date of Diagnosis	Date of the report reflecting that urine organic acids, plasma acylcarnitine, acylglycine profile results are consistent with diagnosis of Isovaleric acidemia	Date of the report reflecting that urine organic acids, plasma acylcarnitine profile results are consistent with diagnosis of 3-MCC	Date of the report reflecting that urine organic acids, plasma acylcarnitine profile results are consistent with diagnosis of HMG

Proposed Terms

NewSTEPs
A Program of the Association of Public Health Laboratories™

	MCD: Holocarboxylase synthase deficiency	ßKT: ß-Ketothiolase deficiency	GA1: Glutaric acidemia type I
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	(this date may be before the NBS result is acted upon) Date biotin therapy was initiated.	(this date may be before the NBS result is acted upon) Date diet change/drug and supplement therapy was initiated.	(this date may be before the NBS result is acted upon) Date diet change/drug/supplement therapy was initiated.
Date of Diagnosis	Date of the report reflecting that urine organic acids, plasma acylcarnitine, biotinidase profile results are consistent with diagnosis of MCD. Additional molecular testing might be required to determine the defect.	Date of the report reflecting that urine organic acids, plasma acylcarnitine profile results are consistent with diagnosis of BKT	Date of the report reflecting that urine/plamsa organic acids, urine/plasma acylcarnitine profile results are consistent with diagnosis of GA1

Proposed Terms

NewSTEPs
A Program of the Association of Public Health Laboratories™

	CUD: Carnitine uptake defect/carnitine transport defect	MCAD: Medium-chain acyl-CoA dehydrogenase deficiency	VLCAD: Very long-chain acyl-CoA dehydrogenase deficiency
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	(this date may be before the NBS result is acted upon) Date carnitine therapy was initiated.	(this date may be before the NBS result is acted upon) Date family is informed to prevent fasting	(this date may be before the NBS result is acted upon) Date family is informed to prevent fasting.
Date of Diagnosis	*Date of the report reflecting that urine and plasma carnitine profile results and mutation analysis are consistent with diagnosis of CUD*	Date of the report reflecting that urine organic acids/ acylglycine, plasma acylcarnitine/ profile results are consistent with diagnosis of MCAD	Date of the report reflecting that plasma acylcarnitine profile results and mutation analysis are consistent with diagnosis of VLCAD

Proposed Terms

NewSTEPs
A Program of the Association of Public Health Laboratories™

	LCHAD: Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency	TFP: Trifunctional protein deficiency	ASA: Argininosuccinic aciduria
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	(this date may be before the NBS result is acted upon) Date family was informed to prevent fasting	(this date may be before the NBS result is acted upon) Date family was informed to prevent fasting	(this date may be before the NBS result is acted upon) Date diet change/drug/supplement therapy was initiated.
Date of Diagnosis	Date of the report reflecting that urine organic acids, plasma acylcarnitine profile results and mutation analysis are consistent with diagnosis of LCHAD	Date of the report reflecting that urine organic acids, plasma acylcarnitine profile results and mutation analysis are consistent with diagnosis of TFP	Date of the report reflecting that urine / plasma amino acid profile results are consistent with diagnosis of ASA

Proposed Terms

NewSTEPs
A Program of the Association of Public Health Laboratories™

	CIT: Citrullinemia, type I	MSUD: Maple syrup urine disease	HCY: Homocystinuria
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	(this date may be before the NBS result is acted upon) Date diet change/drug/supplement therapy was initiated.	(this date may be before the NBS result is acted upon) Date diet change/drug/supplement therapy was initiated.	Date diet change/drug/supplement therapy was initiated.
Date of Diagnosis	Date of the report reflecting that plasma amino acid profile results (and mutation analysis for milder cases) are consistent with diagnosis of CIT	Date of the report reflecting that plasma amino acid and urine organic acid profile results are consistent with diagnosis of MSUD	Date of the report reflecting that plasma amino acid and total homocysteine profile results are consistent with diagnosis of HCY

Proposed Terms

NewSTEPs
A Program of the Association of Public Health Laboratories™

	PKU: Classic phenylketonuria	TYR I: Tyrosinemia, type I	GALT: Classic galactosemia
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	Date diet change was initiated.	Date diet change/drug therapy was initiated.	Date diet change was initiated.
Date of Diagnosis	Date of the report reflecting that plasma amino acid profile results are consistent with diagnosis of PKU	Date of the report reflecting that urine / plasma amino acid profile results are consistent with diagnosis of TYR I	Date of the report reflecting that enzyme levels are consistent with diagnosis of GALT

Draft Terms- still in progress

NewSTEPs
A Program of the Association of Public Health Laboratories™

	MPS I: Mucopolysaccharidosis Type 1	X-ALD: X-linked Adrenoleukodystrophy	GSD II: Glycogen Storage /Pompe: Type II Disease
Notification from PH Lab of abnormal NBS Result	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional	Release of out-of-range results to notification of appropriate medical professional
Date that care of the infant changed (earliest point at which a clinical action was rendered based on follow-up on the newborn screening results; This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation)	(this date may be before the NBS result is acted upon) Date infant is evaluated by specialist	(this date may be before the NBS result is acted upon) Date infant is evaluated by specialist	• Date an evaluation for cardiac status is initiated • ** Double check this one
Date of Diagnosis	*Enzyme + molecular*	• *VLCFA*	Enzyme + molecular