NewSTEPs
A Program of the Association of Public Health Laboratories™

Diagnostic Form Templates for Newborn Screening

Last Updated: November 10, 2025

Case Information Template:
Information Collected for ALL Cases

Infant Demographic Information	
State Unique ID? (alphanumeric)* *A state unique ID is a number and or letters that your program provides to tag or track each confirmed case and update information as needed.*	Unique IDs should only include numbers, letters, hyphens, and underscores
Gestational Age? (in weeks)	
Birth Information	
Date of Birth? (mm/dd/yyyy)*	
Time (hh:mm AM/PM) *If time of birth is not available, only enter the date*	
Year* *Year of birth is stored to calculate Quality Indicators*	*Automatically populated based on date of birth*
Birth Weight? (in grams)	
Biological Sex?	☐ Male ☐ Female ☐ Unspecified ☐ Unknown
Race? (Select all that apply)	☐ White ☐ Black or African American ☐ American Indian or Alaskan Native ☐ Asian ☐ Native Hawaiian or other Pacific Islander ☐ Not Reported ☐ Unknown
Ethnicity?	☐ Hispanic, Latino(a) or Spanish origin ☐ Not of Hispanic, Latino(a), or Spanish origin ☐ Not Reported ☐ Unknown
Screening Information	
Which newborn screen result indicated this infant was at risk for the disorder?	☐ First Screen ☐ Requested Subsequent Screen ☐ Routine Second Screen
Was prenatal testing done that indicated that this infant was at risk for this disorder?	☐ Yes ☐ No ☐ Unknown
Was there family history that indicated that this infant was at risk for this disorder?	☐ Yes ☐ No

	☐ Unknown
Was this individual identified outside of the newborn screening?	☐ Yes ☐ No ☐ Unknown
What was the reason the infant was missed? (IF individual was identified outside of newborn screening = Yes)	☐ Parental Refusal ☐ Lost to follow-up after unsatisfactory specimen ☐ Biologic false negative/result within normal range ☐ Did not have valid screen due to error ☐ Other (please describe below)
Initial & Subsequent Specimen Collection Information	
Specimen Collection	
Date of specimen collection (mm/dd/yyyy)?	
Time (hh:mm AM/PM)	
Time Elapsed Since Birth (in hours)	*Automatically calculated from birth and specimen collection dates; some states can enter directly*
Receipt by Lab	
Date of receipt by lab (mm/dd/yyyy)?	
Time (hh:mm AM/PM)	
Time Elapsed Since Birth (in days)	*Automatically calculated from birth and receipt date; some states can enter directly*
Release of Out-of-Range Results	
Date of release of out-of-range results (mm/dd/yyyy)?	
Time (hh:mm AM/PM)	
Time Elapsed Since Birth (in days)	*Automatically calculated from birth and report date; some states can enter directly*
Intervention, Follow-up, and Diagnosis	
Intervention by Appropriate Medical Provider	
Date of intervention by appropriate medical provider (mm/dd/yyyy)?	
Time (hh:mm AM/PM)	
Time Elapsed Since Birth (in days)	*Automatically calculated from birth and intervention date; some states can enter directly*
Confirmation of Diagnosis	
Date of confirmation of diagnosis (mm/dd/yyyy)?	

Time (hh:mm AM/PM)	
Time Elapsed Since Birth (in days)	*Automatically calculated from birth and diagnosis date; some states can enter directly*
Is infant receiving treatment/care out-of-state?	☐ Yes; enter where state receives care ☐ No ☐ Unknown
Is this diagnosis reversed (does not refer to the therapeutic interventions to address a condition (i.e., surgery, treatment, therapy, etc.)	☐ Yes; enter Year diagnosis reversed ☐ No ☐ Unknown

Table of Contents

Note: standard surveillance case definitions have not been developed for 3-Hydroxy-3-methyglutaric aciduria (HMG), ß-Ketothiolase deficiency (ßKT), Mucopolysaccharidosis Type II (MPS II), Guanidinoacetate methyltransferase deficiency (GAMT), and Infantile Krabbe Disease (Krabbe). These are forthcoming.

Metabolic Disorders

Organic Acid Disorders

Glutaric Acidemia/ Aciduria Type I (GA1)

Enzymatic	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was 3-OH Glutaric acid level ☐ Elevated ☐ Normal ☐ Unknown Was Glutaric acid level ☐ Elevated ☐ Normal ☐ Unknown
Were serum organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was 3-OH Glutaric acid level ☐ Elevated ☐ Normal ☐ Unknown Was Glutaric acid level ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C5 -DC level ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for Glutaric Acidemia enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	

Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *GCDH* ☐ Other gene:_____
	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Holocarboxylase Synthetase (Multiple Carboxylase) Deficiency (MCD) or Other Biotin Disorders

**Not Biotindase Deficiency*

Final Diagnosis as determined by clinician performing follow-up:

- ☐ Holocarboxyase Synthetase Deficiency (MCD)
- ☐ Maternal 3-methylcrotonyl-CoA carboxylase deficiency
- ☐ MT-ATP6 related mitochondrial disorders
- ☐ Other Biotin Disorder (not biotindase deficiency)
- ☐ Unknown

Enzymatic	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was 3OH Isovaleric acid level: ☐ Elevated ☐ Normal ☐ Unknown Was 3OH Propionic acid level: ☐ Elevated ☐ Normal ☐ Unknown Was 3-methylcrotonyl glycine level: ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C3 level ☐ Elevated ☐ Normal ☐ Unknown Was C5-OH level ☐ Elevated ☐ Normal ☐ Unknown

Were infant chemistries (biotinidase) studies completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Were infant chemistries (biotinidase) studies: ☐ Normal ☐ Abnormal ☐ Unknown

Was enzyme analysis for holocarboxylase synthetase deficiency enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *HLCS* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Isovaleric Acidemia/ Aciduria (IVA)

Final Diagnosis as determined by clinician performing follow-up:

- ☐ Isovaleric Acidemia/ Aciduria (IVA)
- ☐ Short/branched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2-methylbutyrl CoA dehydrogenase deficiency
- ☐ Unknown

Enzymatic	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was 3OH Isovaleric acid level ☐ Elevated ☐ Normal ☐ Unknown Was Isovaleryl glycine level ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C5 -DC level ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for Glutaric Acidemia enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *IVD* ☐ Other gene:_____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)

Final Diagnosis as determined by clinician performing follow-up:

- ☐ 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)
- ☐ Maternal MCC deficiency
- ☐ MT-ATP6 related mitochondrial disorders
- ☐ Unknown

Enzymatic	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was 3OH Isovaleric acid level ☐ Elevated ☐ Normal ☐ Unknown Was 3-methylcrotonyl glycine level ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C5 -OH level ☐ Elevated ☐ Normal ☐ Unknown
Was maternal 3-MCC level tested and ruled out? ☐ Yes ☐ No ☐ Unknown	
Was enzyme analysis for 3-MCC enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *MCCC1* ☐ *MCCC2* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Propionic Acidemia/ Aciduria (PROP)

Final Diagnosis as determined by clinician performing follow-up:

- ☐ Propionic Acidemia (PROP)
- ☐ Maternal vitamin B12 deficiency
- ☐ Succinate-CoA ligase deficiency
- ☐ Unknown

Enzymatic	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Please indicate which of the following metabolites were detected: Propionyl glycine? ☐ Yes ☐ No ☐ Unknown Tiglyglycine? ☐ Yes ☐ No ☐ Unknown Methylcitrate? ☐ Yes ☐ No ☐ Unknown 3OH Propionic acid level? ☐ Yes ☐ No ☐ Unknown MMA? ☐ Yes ☐ No ☐ Unknown Methylcrotonyl glycine? ☐ Yes ☐ No ☐ Unknown

Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C3 level: ☐ Elevated ☐ Normal ☐ Unknown

Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *PCCA* ☐ *PCCB* ☐ Other gene: _____
	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Methylmalonic Acidemia (methylmalonyl-CoA mutase; MUT)

Final Diagnosis as determined by metabolic geneticist or clinician performing follow-up:

- ☐ Mutase(-) (mut-)
- ☐ Mutase (0) (mut0)
- ☐ Maternal vitamin B12 deficiency
- ☐ Succinate-CoA ligase deficiency
- ☐ Unclassified
- ☐ Unknown

Enzymatic	
Was serum MMA level tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was MMA level in serum: ☐ Elevated ☐ Normal ☐ Unknown
Was urine MMA level tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was MMA level in urine: ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C3 level ☐ Elevated ☐ Normal ☐ Unknown
Was maternal vitamin B12 levels tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was maternal vitamin B12 deficient? ☐ Yes ☐ No ☐ Unknown
Was infant vitamin B12 levels tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was infant vitamin B12 deficient? ☐ Yes ☐ No ☐ Unknown
Was total plasma homocysteine tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was total plasma homocysteine: ☐ Elevated ☐ Normal ☐ Unknown

Were enzyme complementation studies completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Were complementation studies: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *METHYLMALONYL-CoA MUTASE* ☐ Other gene: _____
	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Methylmalonic Acidemia (cobalamin disorders; Cbl A, Cbl B, Cbl Dv2)

Final Diagnosis as determined by metabolic geneticist or clinician performing follow-up:

- ☐ Cobalamin A deficiency (Cbl A)
- ☐ Cobalamin B deficiency (Cbl B)
- ☐ Cobalamin Dv2 deficiency (Cbl Dv2)
- ☐ Maternal vitamin B12 deficiency
- ☐ Succinate-CoA ligase deficiency
- ☐ Unclassified
- ☐ Unknown

Enzymatic	
Was serum MMA level tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was MMA level in serum: ☐ Elevated ☐ Normal ☐ Unknown
Was urine MMA level tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was MMA level in urine: ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C3 level ☐ Elevated ☐ Normal ☐ Unknown
Was maternal vitamin B12 levels tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was maternal vitamin B12 deficient? ☐ Yes ☐ No ☐ Unknown
Was infant vitamin B12 levels tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was infant vitamin B12 deficient? ☐ Yes ☐ No ☐ Unknown
Was total plasma homocysteine tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was total plasma homocysteine: ☐ Elevated ☐ Normal ☐ Unknown

Were enzyme complementation studies completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Were complementation studies: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *MMAA gene* ☐ *MMAB gene* ☐ Other gene: _____
	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Methylmalonic Acidemia with Homocystinuria (Cbl C, Cbl D, Cbl F, Cbl Dv1, Cbl J)

**Secondary RUSP Condition*

Final Diagnosis as determined by metabolic geneticist or clinician performing follow-up:

- ☐ Cobalamin C deficiency (Cbl C)
- ☐ Cobalamin D deficiency (Cbl D)
- ☐ Cobalamin F deficiency (Cbl F)
- ☐ Cobalamin Dv1 deficiency (Cbl Dv1)
- ☐ Cobalamin J deficiency (Cbl J)
- ☐ Maternal vitamin B12 deficiency
- ☐ Succinate-CoA ligase deficiency
- ☐ Unclassified
- ☐ Other cobalamin deficiency
- ☐ Unknown

Enzymatic	
Was serum MMA level tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was MMA level in serum: ☐ Elevated ☐ Normal ☐ Unknown
Was urine MMA level tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was MMA level in urine: ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C3 level ☐ Elevated ☐ Normal ☐ Unknown
Was maternal vitamin B12 levels tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was maternal vitamin B12 deficient? ☐ Yes ☐ No ☐ Unknown
Was infant vitamin B12 levels tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was infant vitamin B12 deficient? ☐ Yes ☐ No ☐ Unknown

Was total plasma homocysteine tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was total plasma homocysteine: ☐ Elevated ☐ Normal ☐ Unknown

Were enzyme complementation studies completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Were complementation studies: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *MMACHC* ☐ *MMADHC* ☐ *LMBRD1* ☐ *ABCD4* ☐ *HCFC1* ☐ *C2ORF25* ☐ Other gene: _____
	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Fatty Acid Disorders

Primary Carnitine Deficiency/ Carnitine Uptake Deficiency (CUD)

Final Diagnosis as determined by metabolic geneticist or clinician performing follow-up:

- ☐ Carnitine Uptake Deficiency (CUD)
- ☐ Maternal Carnitine Deficiency (primary and secondary)
- ☐ Unknown

Enzymatic	
Was urine carnitine tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was fractional excretion of free carnitine level: ☐ Elevated ☐ Normal ☐ Unknown Was 3-methylcrotonyl glycine level ☐ Elevated ☐ Normal ☐ Unknown
Were plasma carnitine levels tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was free carnitine (C0) ☐ Low ☐ Normal ☐ Unknown
Were other causes for carnitine loss ruled out? ☐ Yes ☐ No ☐ Unknown	
Was enzyme analysis for carnitine deficiency enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *SCL22A5* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Medium-chain acyl-CoA Dehydrogenase Deficiency (MCAD)

Enzymatic	
Were urine organic acids or aclyglycines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was Hexanoylglycine level: ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C8 level: ☐ Elevated ☐ Normal ☐ Unknown Was repeat C8 level: ☐ Elevated ☐ Normal ☐ Unknown Was C8>C10 level: ☐ Yes ☐ No ☐ Unknown Was C8>C6 level: ☐ Yes ☐ No ☐ Unknown Was C6 level: ☐ Elevated ☐ Normal ☐ Unknown Was C10 level: ☐ Elevated ☐ Normal ☐ Unknown

Was functional analysis of fatty acid oxidation in cultured fibroblasts performed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was functional fibroblast analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Was enzyme analysis for MCAD enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *ACADM* ☐ Other gene: _____
	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Trifunctional Protein Deficiency (TFP)

Enzymatic	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C12-OH dicarboxylic acid level: ☐ Elevated ☐ Normal ☐ Unknown Was C10-OH dicarboxylic acid level: ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C16-OH level: ☐ Elevated ☐ Normal ☐ Unknown Was C16:1-OH level: ☐ Elevated ☐ Normal ☐ Unknown Was C18-OH level: ☐ Elevated ☐ Normal ☐ Unknown Was C18:1-OH level: ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for TFP enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown

Was functional analysis of fatty acid oxidation in cultured fibroblasts performed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was functional fibroblast analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *HADHA* ☐ *HADHB* ☐ Other gene: _____
	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)

Enzymatic	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C12-OH dicarboxylic acid level: ☐ Elevated ☐ Normal ☐ Unknown Was C10-OH dicarboxylic acid level: ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C16-OH level: ☐ Elevated ☐ Normal ☐ Unknown Was C16:1-OH level: ☐ Elevated ☐ Normal ☐ Unknown Was C18-OH level: ☐ Elevated ☐ Normal ☐ Unknown Was C18:1-OH level: ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for TFP enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown

Was functional analysis of fatty acid oxidation in cultured fibroblasts performed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was functional fibroblast analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *HADHA* ☐ *HADHB* ☐ Other gene: _____
	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Very Long-chain acyl-CoA Dehydrogenase Deficiency (VLCAD)

Enzymatic	
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was C14:1 level: ☐ Elevated (on more than one sample) ☐ Normal ☐ Unknown Was C14:2-OH level: ☐ Elevated ☐ Normal ☐ Unknown Was C14 level: ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for VLCAD enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Was functional analysis of fatty acid oxidation in cultured fibroblasts performed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was functional fibroblast analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *ACADVL* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Amino Acid Disorders

Argininosuccinic Acidemia/ Aciduria (ASA)

Final diagnosis as determined by a metabolic geneticist or clinician performing following-up:

- ☐ Argininosuccinic Acidemia/ Aciduria (ASA)
- ☐ Pyruvate carboxylase deficiency
- ☐ Unknown

Enzymatic	
Were plasma amino acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was plasma ASA level: ☐ Elevated ☐ Normal ☐ Unknown Was Citrulline level: ☐ Elevated ☐ Normal ☐ Unknown
Were plasma urine acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was urine ASA level? ☐ Elevated ☐ Normal ☐ Unknown Was urine Citrulline level? ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for ASA enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	

Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *ASL* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Citrullinemia, Type I (CIT)

Final diagnosis as determined by a metabolic geneticist or clinician performing following-up

- ☐ Citrullinemia, Type I
- ☐ Pyruvate Carboxylase Deficiency
- ☐ Unknown

Enzymatic	
Were plasma amino acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was plasma ASA level: ☐ Present ☐ Absent ☐ Unknown Was Citrulline level: ☐ Elevated ☐ Normal ☐ Unknown
Was blood ammonia levels tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was blood ammonia level: ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for Citrullinemia type 1 enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *ASS1* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Classic Phenylketonuria (PKU) and Hyperphenylalaninemia (Hyperphe)

Final diagnosis as determined by a metabolic geneticist or clinician performing following-up:

- ☐ Classic phenylketonuria (PKU)
- ☐ Benign hyperphenylalaninemia (H-PHE)
- ☐ HyperPhe diet controlled
- ☐ Dihydropterine reductase deficiency (DHPR)
- ☐ DNAJC12
- ☐ Parenteral nutrition
- ☐ Maternal PKU
- ☐ Unknown

Enzymatic	
Were plasma amino acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was Phe level: ☐ Elevated (>120umol/L on unrestricted diet) ☐ Normal ☐ Unknown Was Phe/Tyr level: ☐ Elevated ☐ Normal ☐ Unknown
Were biopterin studies done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Were biopterin studies: ☐ Normal ☐ Abnormal ☐ Unknown
Was enzyme analysis for Hyperphe (inclusive of classic PKU) enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *PAH* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Homocystinuria (Cystathionine Beta-Synthase (CBS) Deficiency; HCY)

Final diagnosis as determined by a metabolic geneticist or clinician performing following-up:

- ☐ Classic Homocystinuria
- ☐ Methionine Adenosyltransferase (MAT I/III Deficiency)
- ☐ Glycine n-methyltransferase (GNMT)
- ☐ Adenosylhomocysteine Hydrolase Deficiency
- ☐ Unknown

Enzymatic	
Were plasma amino acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was Methionine level: ☐ Elevated ☐ Normal ☐ Unknown
Was plasma Homocysteine tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was plasma Homocysteine level: ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for CBS enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *CBS* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Maple Syrup Urine Disease (MSUD)

Final diagnosis as determined by a metabolic geneticist or clinician performing following-up:

- ☐ Classic
- ☐ Intermediate
- ☐ Thiamine-response
- ☐ Hydroxyprolinemia
- ☐ Unclassified
- ☐ Unknown

Enzymatic	
Were plasma amino acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was Alloisoleucine level: ☐ Elevated ☐ Normal ☐ Unknown Was Leucine level: ☐ Elevated ☐ Normal ☐ Unknown Was Isoleucine level: ☐ Elevated ☐ Normal ☐ Unknown Was Valine level: ☐ Elevated ☐ Normal ☐ Unknown Was Leu>Val level: ☐ Yes ☐ No ☐ Unknown

Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was 2-ketoisocaproic acid level: ☐ Elevated ☐ Normal ☐ Unknown Was 2-OH Isovaleric acid level: ☐ Elevated ☐ Normal ☐ Unknown Was 2-ketomethyl valeric acid level ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for MSUD enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *DBT* ☐ *BCKDHB* ☐ *DLD* ☐ *BCKDHA* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Tyrosinemia Type I (TYR-1)

Final diagnosis as determined by a metabolic geneticist or clinician performing following-up:

- ☐ Tyrosinemia, Type I (hepatorenal)
- ☐ Transient Tyrosinemia of the neonate (TTN)
- ☐ Unknown

Enzymatic	
Were plasma organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was plasma succinylacetone level: ☐ Elevated ☐ Normal ☐ Unknown Was plasma tyrosine level: ☐ Elevated ☐ Normal ☐ Unknown
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was urine succinylacetone level: ☐ Elevated ☐ Normal ☐ Unknown Was urine tyrosine level: ☐ Elevated ☐ Normal ☐ Unknown
Was enzyme analysis for fumarylacetoacetate hydrolase completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme analysis: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	

Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *FAH* ☐ Other gene: _____

	[For each gene selected] Check the types of variants found on: *Allele 1:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Endocrine Disorders

Congenital Hypothyroidism (CH)

Final Diagnosis as determined by clinician performing follow-up:

- ☐ Primary Congenital Hypothyroidism
- ☐ Secondary Congenital Hypothyroidism
- ☐ Subclinical Congenital Hypothyroidism
- ☐ TBG Deficiency (Thyroxine Binding Globulin) or other protein binding defect
- ☐ Transient Congenital Hypothyroidism
- ☐ Unknown

Enzymatic	
Was Serum TSH tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the level: ☐ TSH > 10 mU/L ☐ TSH 6-10 mU/L ☐ TSH <10 mU/L ☐ TSH <6 mU/L ☐ Unknown Was it tested before initiation of treatment? ☐ Yes ☐ No ☐ Unknown
Was Serum Total T4 tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was Serum Total T4 below the age-established reference range? ☐ Yes ☐ No ☐ Unknown Was it tested before initiation of treatment? ☐ Yes ☐ No ☐ Unknown

Was Serum Free T4 tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was Serum Free T4 below the age-established reference range? ☐ Yes ☐ No ☐ Unknown Was it tested before initiation of treatment? ☐ Yes ☐ No ☐ Unknown
Does this baby have other pituitary hormone deficiencies? ☐ Yes ☐ No ☐ Unknown	
Does this baby have midline defects? ☐ Yes ☐ No ☐ Unknown	
Was TBG tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was TBG below the age established reference range? ☐ Yes ☐ No ☐ Unknown
Was T3 or T4 resin uptake tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was T3 or T4 resin uptake above the age-established reference range? ☐ Yes ☐ No ☐ Unknown

Congenital Adrenal Hyperplasia (CAH)

Final Diagnosis as determined by clinician performing follow-up:

- ☐ Classic 21-Hydroxylase Deficiency-Salt Wasting
- ☐ Classic 21-Hydroxylase Deficiency-Simple Virilizing
- ☐ Other Adrenal disorder: other final diagnosis name _____
- ☐ Unknown

Enzymatic	
Societal Sex ☐ Male ☐ Female ☐ Unknown ☐ Unspecified	
Was confirmatory serum 17-OHP level obtained? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was there a value at baseline: ☐ >10,000 ng/dl ☐ 1000-10,000 ng/dl; ☐ < 1000 ng/dl; ☐ Unknown Was it tested before initiation of treatment? ☐ Yes ☐ No Was there a result after ACTH stimulation: ☐ >10,000 ng/dl ☐ 1000-10,000 ng/dl; ☐ < 1000 ng/dl; ☐ Unknown Was it tested before initiation of treatment? ☐ Yes ☐ No
Was tandem mass spectrometry urinary steroid profile obtained? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Were the urinary spectrometry steroid profile results: ☐ Indicative of 21-Hydroxylase Deficiency CAH ☐ Unknown

Was serum sodium level measured before initiation of treatment? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was the sodium level: ☐ < 135 mEq/L ☐ > 135 mEq/L ☐ Unknown
Was plasma renin activity level measured at time of initiation of treatment? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was plasma renin activity normal for age? ☐ Yes ☐ No ☐ Unknown Was it tested before initiation of treatment? ☐ Yes ☐ No
Clinical Results	
Is there evidence of salt wasting (e.g., shock or severe failure to thrive)? ☐ Yes ☐ No ☐ Unknown	
Is there supportive clinical or laboratory evidence of CAH? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Is the evidence (check all that apply): ☐ Ambiguous genitalia, with 46 XX karyotype ☐ Normal genitalia, with 46 XY karyotype ☐ Other hormonal evidence of CAH
Molecular Genetics	
Was mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *CYP21A2* ☐ Other gene:_____

	[For each gene selected] Check the types of variants found on: *Allele 1* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Hemoglobinopathies

Presence of Hb S

Final diagnosis as determined by a clinician performing the follow-up:

- ☐ S, Beta 0-thalassemia – HB S/B0Th
- ☐ S,S Disease (Sickle Cell Anemia) – HbSS
- ☐ S, Beta + Thalassemia – HbS/B + Th
- ☐ S,C Disease – Hb S/C
- ☐ S, Other; other result name _____
- ☐ Unknown

Diagnostic Workup	
Was qualitative (IEF or HPLC) testing completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ FS ☐ FSC ☐ FSA ☐ FSA_2 ☐ $FSAA_2$ ☐ Other; other result name_____ ☐ Unknown
Was quantitative (HPLC or electrophoresis) testing completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ FS ☐ FSC ☐ FS with high A_2 ☐ FSA with high A_2 ☐ FSA ☐ Other; other result name_____ ☐ Unknown

Was mutation analysis performed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Check the type of variant found on: *Allele 1* ☐ *S* ☐ *C* ☐ *Beta + Thal* ☐ *Beta^0 + Thal* ☐ Other; _____ ☐ Unknown *Allele 2* ☐ *S* ☐ *C* ☐ *Beta + Thal* ☐ *Beta^0 + Thal* ☐ Other; other name_____ ☐ Unknown
NBS result ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ FS ☐ FSC ☐ FSA ☐ FSA_2 ☐ Other ☐ Unknown
Was a CBC performed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ Normal – high MCV ☐ Low MCV ☐ Unknown

Were family studies (in parents) done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Maternal Status: what were the results? ☐ Carrier *S* ☐ Carrier *C* ☐ Carrier *Beta + Thal* ☐ Carrier *Beta^0 Thal* ☐ Other: _____ ☐ Unknown Paternal Status: what were the results? ☐ Carrier *S* ☐ Carrier *C* ☐ Carrier *Beta + Thal* ☐ Carrier *Beta^0 Thal* ☐ Other: _____ ☐ Unknown
Was there a positive family history? ☐ Yes ☐ No ☐ Unknown	
Were HPLC & IEF tested on the same sample from the infant? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ FS ☐ FSC ☐ FSA_2 ☐ $FSAA_2$ ☐ Other ☐ Unknown
Were Hgb tests (electrophoresis or HPLC) performed on family members? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ Positive ☐ Negative ☐ Unknown

Presence of Other Hb Variant

**This is a Secondary RUSP Condition*

Final diagnosis as determined by a clinician performing the follow-up:

- ☐ Hemoglobin C Disease
- ☐ Hemoglobin D Disease
- ☐ Hemoglobin E Disease
- ☐ Hemoglobin O-Arab Disease
- ☐ Other Hemoglobin Disease; please describe
- ☐ Unknown

Diagnostic Workup	
Alpha thalassemia present? ☐ Yes ☐ No ☐ Unknown	
Was qualitative (IEF or HPLC) testing completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ FC ☐ FD ☐ FE ☐ FO_{ARAB} ☐ Other; other result name_____ ☐ Unknown
Was quantitative (HPLC or electrophoresis) testing completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ FC ☐ FD ☐ FE ☐ FO_{ARAB} ☐ Other; other result name_____ ☐ Unknown

Was mutation analysis performed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Check the type of variant found on allele 1: ☐ *C* ☐ *D* ☐ *E* ☐ O_{ARAB} ☐ Other; other name_____ ☐ Unknown Check the type of variant found on allele 2: ☐ *C* ☐ *D* ☐ *E* ☐ O_{ARAB} ☐ *Beta + Thal* ☐ $Beta^{0}$ *+ Thal* ☐ Other; other name_____ ☐ Unknown
NBS result ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ FC ☐ FD ☐ FE ☐ FO_{ARAB} ☐ Other; other result name_____ ☐ Unknown
Was a CBC performed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ Normal – high MCV ☐ Low MCV

Were family studies (in parents) done? ☐ Yes ☐ No ☐ Unknown	Maternal Status: what were the results? ☐ Carrier *C* ☐ Carrier *D* ☐ Carrier *E* ☐ Carrier O_{Arab} ☐ Carrier *Beta + Thal* ☐ Carrier $Beta^0$ *Thal* ☐ Other: _____ ☐ Unknown Paternal Status: what were the results? ☐ Carrier *C* ☐ Carrier *D* ☐ Carrier *E* ☐ Carrier O_{Arab} ☐ Carrier *Beta + Thal* ☐ Carrier $Beta^0$ *Thal* ☐ Other: _____ ☐ Unknown
Was there a positive family history? ☐ Yes ☐ No ☐ Unknown	
Were Hgb tests (electrophoresis or HPLC) performed on family members? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the results? ☐ Positive ☐ Negative ☐ Unknown

Lysosomal Storage Disorders

Note: Case Confirmatory Diagnosis Follow-up for Mucopolysaccharidosis Type II (MPS II) is in development

Mucopolysaccharidosis Type I (MPS I)

Final Diagnosis as determined by metabolic geneticist or clinician performing follow-up:

- ☐ MPS I—Severe
- ☐ MPS I—Severity not determined
- ☐ MPS I—attenuated
- ☐ Uncertain Type/Onset
- ☐ Unknown

Enzymatic	
Was enzyme activity tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the enzyme level? ☐ Within lab known affected range ☐ Normal ☐ Unknown
Were urine GAGS tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the urine GAG level? ☐ Elevated ☐ Normal ☐ Unknown

Clinical symptoms/lab findings? ☐ Symptoms present and documented by specialists. Public health (PH) program continued to collect data through the development of symptoms ☐ No symptoms by the time the PH Program closes follow-up (either due to child being lost to follow-up OR program policy on follow-up time ☐ Unknown	*Clinical symptoms consistent with MPS-I include: Hepatosplenomegaly, Coarse facial features, Hydrocephalus, Skeletal deformities (dysostosis multiplex), Corneal clouding, Large tongue, Prominent forehead, Joint stiffness, Short stature, frequent ear infections and hearing loss, hernia*

Molecular Genetics	
Were variants detected in genes known to be associated with MPS I? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Check the types of variants found on: *Allele 1:* ☐ Pathogenic variant and associated with SEVERE disease ☐ Pathogenic or likely pathogenic variant ☐ Variant of unknown significance ☐ Variant known to be associated with ATTENUATED disease. ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Pathogenic variant and associated with SEVERE disease ☐ Pathogenic or likely pathogenic variant ☐ Variant of unknown significance ☐ Variant known to be associated with ATTENUATED disease. ☐ Wild Type (Normal) ☐ Unknown

Pompe Disease

Final Diagnosis as determined by metabolic geneticist or clinician performing follow-up:

- ☐ Infantile Onset (IO) Pompe Disease
- ☐ Late Onset (LO) Pompe Disease
- ☐ Uncertain Type/Onset
- ☐ Unknown

Enzymatic	
Was enzyme activity tested in blood (not DBS sample)? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the enzyme level? ☐ Within lab known affected range for infantile onset (IO) ☐ Low (above affected range, for IO, may or may not be in late-onset (LO range), but should not be above LO range)) ☐ Within lab known affected range for late onset (LO) ☐ Low (above affected range, for LO not normal) ☐ Unknown
Was enzyme activity tested in skin/muscle? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the enzyme activity? ☐ Positive skin or muscle biopsy ☐ Unknown
Was there cardiac involvement consistent with Pompe? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Findings: ☐ Positive findings on chest X-ray/EKG/ECHO in newborn period ☐ Positive findings on chest X-ray/EKG/ECHO
Lab findings for CK/AST/ALT/LDH/Urine Hex4? ☐ Elevated ☐ Not Present ☐ Unknown ☐ Untested	

Were there any clinical findings? ☐ Symptoms present after one year of age and documented by specialists. PH program continue to collect data through the development of symptoms ☐ Symptoms present before one year of age, but no cardiac involvement ☐ Unknown or not reported to PH by the end of the follow-up period	*Clinical symptoms consistent with Pompe Disease: progressive muscle weakness, need for respiratory assistance, swaying gait or waddle, Lordosis, kyphosis, or scoliosis*
Molecular Genetics	

Were variants detected in genes known to be associated with Pompe Disease? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Check the types of variants found on: *Allele 1:* ☐ Pathogenic ☐ Pathogenic variant and associated with infantile onset ☐ Novel variant that is likely pathogenic ☐ Pathogenic variant or likely pathogenic variant, with deletion or duplication consistent with infantile onset ☐ Pathogenic and associated with non-classical disease, or variant of uncertain significance ☐ Pathogenic or likely pathogenic variant, no other variants found; duplication/deletion testing not done or not known ☐ Pathogenic or likely pathogenic variant; no other variants found ☐ Wild Type (Normal) ☐ Unknown *Allele 2:* ☐ Pathogenic ☐ Pathogenic variant and associated with infantile onset ☐ Novel variant that is likely pathogenic ☐ Pathogenic variant or likely pathogenic variant, with deletion or duplication consistent with infantile onset ☐ Pathogenic and associated with non-classical disease, or variant of uncertain significance ☐ Pathogenic or likely pathogenic variant, no other variants found; duplication/deletion testing not done or not known ☐ Pathogenic or likely pathogenic variant; no other variants found ☐ Wild Type (Normal) ☐ Unknown

Other Disorders

Biotinidase Deficiency (BIOT)

Final Diagnosis as determined by metabolic geneticist or clinician performing follow-up:

- ☐ Profound Biotinidase deficiency
- ☐ Partial Biotinidase deficiency
- ☐ Unknown

Enzymatic	
Was enzyme analysis for biotinidase enzyme activity completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was enzyme activity: ☐ <10% ☐ 10-30% ☐ Normal ☐ Unknown
Molecular Genetics	
Was a mutation analysis performed for biotinidase deficiency? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ BTD ☐ Other gene:_____
	[For all genes selected] **Check the types of variants found on:** *Allele 1:* ☐ Variant known to be disease causing (Unknown) ☐ Variant known to be disease causing (known to be associated with profound enzyme deficiency) ☐ Variant known to be disease causing (known to be associated with partial enzyme deficiency [“mild” mutation (D44H)] ☐ Variant of unknown significance ☐ Wild Type (Normal) ☐ Unknown *Allele 2* ☐ Variant known to be disease causing (Unknown) ☐ Variant known to be disease causing (known to be associated with profound enzyme deficiency) ☐ Variant known to be disease causing (known to be associated with partial enzyme deficiency [“mild” mutation (D44H)] ☐ Variant of unknown significance ☐ Wild Type (Normal) ☐ Unknown

Galactosemia (GALT)

Final diagnosis as determined by a metabolic geneticist or clinician performing following-up:

- ☐ Classic Galactosemia
- ☐ Duarte variant galactosemia
- ☐ Unknown

Enzymatic	
Were GALT levels tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was GALT level: ☐ <10% ☐ 10-30% ☐ Normal ☐ Unknown
Was Gal-1-P tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was Gal-1-P level: ☐ Elevated ☐ Normal ☐ Unknown
Was Urine Galactitol tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Was Urine Galactitol level: ☐ Elevated ☐ Normal ☐ Unknown
If Variant Galactosemia, was protein phenotyping completed? ☐ Yes ☐ No ☐ Unknown ☐ Not applicable	*[IF YES]* Did result indicate: ☐ Phenotype consistent with variant ☐ Phenotype NOT consistent with variant ☐ Unknown
If Arginase Deficiency, were enzyme studies completed? ☐ Yes ☐ No ☐ Unknown ☐ Not applicable	*[IF YES]* Was enzyme activity: ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	

Was a mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ Galactosemia ☐ Other gene:_____

	[For each gene selected] **Check the types of variants found on:** *Allele 1* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown *Allele 2* ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (Normal) ☐ Unknown

Cystic Fibrosis

Final diagnosis as determined by a metabolic geneticist or clinician performing following-up:

- ☐ CFTR-Related Metabolic Syndrome (CRMS)
- ☐ CFTR-Related Disease
- ☐ Typical Cystic Fibrosis (CF)
- ☐ Unknown

Diagnostic Workup	
Did the NBS result indicate an elevated IRT? ☐ Yes ☐ No ☐ Unknown	
Were CFTR mutations detected on the <u>newborn screening</u> mutation panel? ☐ Yes ☐ No ☐ Unknown *Mutations seen in patients with CF have been classified as disease-causing, neutral, or varying clinical consequences through the CFTR2 project: http://cftr2.org/browse.php. Additional information about the mutation and the association with lower sweat chlorides can also be found at CFTR2.*	*[IF YES]* *Check the type of variant found on allele 1:* ☐ Variant known to be disease causing in CFTR2 ☐ Variant known to be disease causing in CFTR2 (shown to be associated with lower sweat chlorides) ☐ Neutral variant ☐ Variant of varying clinical consequence in CFTR2 ☐ Wild Type (Normal) ☐ Unknown (not reported in CFTR2) *Check the type of variant found on allele 1:* ☐ Varian known to be disease causing in CFTR2 ☐ Variant known to be disease causing in CFTR2 (shown to be associated with lower sweat chlorides) ☐ Neutral variant ☐ Variant of varying clinical consequence in CFTR2 ☐ Wild Type (Normal) ☐ Unknown (not reported in CFTR2)
Did the child have meconium ileus? ☐ Yes ☐ No ☐ Unknown	

Was a valid sweat chloride result available? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the sweat test results (please report on the highest sweat chloride value from one sweat test)? ☐ ≥60 mmol/L (regardless of age) ☐ <30 mmol/L (if age <6 months) ☐ 30-59 mmol/L (if age < 6 months) ☐ <40mmol/L (if age ≥6 months) ☐ 40-59 mmol/L (if age ≥6 months) ☐ Quantity not Sufficient *[IF NO]* If a valid sweat test was not available, were there attempts to obtain a sweat chloride that were quantity not sufficient (QNS)? ☐ Yes ☐ No ☐ Unknown
Was a sweat chloride repeated on a separate day? *(Results from different arm on the same day should NOT be reported here)* ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What were the repeat sweat test results (please report on the highest sweat chloride value from one sweat test)? ☐ ≥60 mmol/L (regardless of age) ☐ <30 mmol/L (if age <6 months) ☐ 30-59 mmol/L (if age < 6 months) ☐ <40mmol/L (if age ≥6 months) ☐ 40-59 mmol/L (if age ≥6 months) ☐ Quantity not sufficient (QNS)

Was a CFTR mutation panel completed <u>after</u> the newborn screening mutation panel? ☐ Yes ☐ No ☐ Unknown *Mutations seen in patients with CF have been classified as disease-causing, neutral, or varying clinical consequences through the CFTR2 project: http://cftr2.org/browse.php. Additional information about the mutation and the association with lower sweat chlorides can also be found at CFTR2.*	*[IF YES]* *Check the type of variant found on allele 1:* ☐ Variant known to be disease causing in CFTR2 ☐ Variant known to be disease causing in CFTR2 (shown to be associated with lower sweat chlorides) ☐ Neutral variant ☐ Variant of varying clinical consequence in CFTR2 ☐ Wild Type (Normal) ☐ Unknown (not reported in CFTR2) *Check the type of variant found on allele 2:* ☐ Variant known to be disease causing in CFTR2 ☐ Variant known to be disease causing in CFTR2 (shown to be associated with lower sweat chlorides) ☐ Neutral variant ☐ Variant of varying clinical consequence in CFTR2 ☐ Wild Type (Normal) ☐ Unknown (not reported in CFTR2)
If the child was diagnosed after the newborn period, were clinical symptoms associated with CFTR Related Disease present? *Select NA if the child was diagnosed during the newborn period.* ☐ Present ☐ Not Present ☐ Unknown ☐ Not applicable	*[IF PRESENT]* Select all symptoms included: ☐ CBAVD ☐ Recurrent pancreatitis ☐ Nasal polyposis ☐ Infertility ☐ Focal biliary cirrhosis with portal hypertension

Summary of common variants as reported on CFTR2 (this is not an exhaustive list; please visit www.CFTR2.org for the latest updated list).

Variant name - HGVS nomenclature	Protein name	Variant legacy name	On ACMG Screening Panel	CFTR2 final call	Associated with lower sweat chloride
c.3717+12191C>T	p.Phe316LeufsX12	**1078delT**	No	CF-causing	NO
c.579+3A>G	p.Phe342HisfsX28	**1154insTC**	No	CF-causing	NO
c.3454G>C	No protein name	**1717-1G->A**	Yes	CF-causing	NO
c.3208C>T	No protein name	**1811+1.6kbA->G**	No	CF-causing	NO
c.3154T>G	No protein name	**1898+1G->A**	Yes	CF-causing	NO
c.1585-1G>A	p.Leu671X	**2143delT**	No	CF-causing	NO
c.1680-1G>A	p.Lys684SerfsX38	**2183AA->G**	No	CF-causing	NO
c.1766+1G>A	p.Lys684AsnfsX38	**2184delA**	Yes	CF-causing	NO
c.2490+1G>A	p.Gln685ThrfsX4	**2184insA**	No	CF-causing	NO
c.2988+1G>A	p.Glu726ArgfsX4	**2307insA**	No	CF-causing	NO
c.1736A>G	No protein name	**2789+5G->A**	Yes	CF-causing	NO
c.1408A>G	No protein name	**3120+1G->A**	Yes	CF-causing	NO
c.1841A>G	No protein name	**3120G->A**	No	CF-causing	NO
c.2991G>C	No protein name	**3272-26A->G**	No	CF-causing	NO
c.489+1G>T	p.Lys1177SerfsX15	**3659delC**	Yes	CF-causing	NO
c.350G>A	No protein name	**3849+10kbC->T**	Yes	CF-causing	NO
c.4242+1G>T	p.Leu1258PhefsX7	**3905insT**	No	CF-causing	NO
c.3718-1G>A	p.Leu88IlefsX22	**394delTT**	No	CF-causing	NO
c.1240C>T	No protein name	**5T**	No	Indeterminate	YES
c.2260G>A	No protein name	**621+1G->T**	Yes	CF-causing	NO
c.1727G>C	No protein name	**711+1G->T**	Yes	CF-causing	NO
c.220C>T	No protein name	**711+5G->A**	No	CF-causing	NO
c.2834C>T	p.Ala455Glu	**A455E**	Yes	CF-causing	NO
c.1675G>A	p.Ala559Thr	**A559T**	No	CF-causing	NO
c.1127_1128insA	p.Ser18ArgfsX16	**CFTRdele2,3**	No	CF-causing	NO
c.1202G>A or c.1203G>A	p.Asp1152His	**D1152H**	No	Indeterminate	YES
c.1923_1931del9insA	p.Glu60X	**E60X**	No	CF-causing	NO
c.1679G>C	p.Phe508del	**F508del**	Yes	CF-causing	NO
c.3160C>G	p.Gly1244Glu	**G1244E**	No	CF-causing	NO
c.4046G>A	p.Gly178Glu	**G178R**	No	CF-causing	NO
c.4196_4197delTC	p.Gly542X	**G542X**	Yes	CF-causing	NO
c.3731G>A	p.Gly551Asp	**G551D**	Yes	CF-causing	NO
c.3197G>A	p.Gly85Glu	**G85E**	Yes	CF-causing	NO
c.2657+2_2657+3insA	p.Ile1027Thr	**I1027T**	No	Not CF-causing	NO
c.1673T>C	p.Ile148Thr	**I148T**	No	Not CF-causing	NO

c.3763T>C	p.Ile336Lys	**I336K**	No	CF-causing	NO
c.1558G>T	p.Ile507del	**I507del**	Yes	CF-causing	NO
c.3230T>C	p.Leu1077Pro	**L1077P**	No	CF-causing	NO
c.1040G>A	p.Leu206Trp	**L206W**	No	CF-causing	NO
c.3302T>A	p.Met1101Lys	**M1101K**	No	CF-causing	NO
c.274G>A	p.Asn1303Lys	**N1303K**	Yes	CF-causing	NO
c.617T>G	p.Pro67Leu	**P67L**	No	CF-causing	NO
c.2764_2765insAG	p.Gln220X	**Q220X**	No	CF-causing	NO
c.1973_1985del13insAGAAA	p.Gln493X	**Q493X**	No	CF-causing	NO
c.3196C>T	p.Arg1066Cys	**R1066C**	No	CF-causing	NO
c.4296_4297insGA	p.Arg1158X	**R1158X**	No	CF-causing	NO
c.1692delA	p.Arg1162X	**R1162X**	Yes	CF-causing	NO
c.1055G>A	p.Arg117Cys	**R117C**	No	CF-causing	NO
c.1466C>A	p.Arg117His	**R117H**	Yes	Indeterminate	YES
c.1013C>T	p.Arg334Trp	**R334W**	Yes	CF-causing	NO
c.532G>A	p.Arg347His	**R347H**	Yes	CF-causing	NO
c.1040G>C	p.Arg347Pro	**R347P**	No	CF-causing	NO
c.2908G>C	p.Arg352Gln	**R352Q**	No	CF-causing	NO
c.2424_2425insAT	p.Arg553X	**R553X**	Yes	CF-causing	NO
c.2780T>C	p.Arg560Thr	**R560T**	Yes	CF-causing	NO
c.349C>T	p.Ser1251Asn	**S1251N**	No	CF-causing	NO
c.1000C>T	p.Ser549Asn	**S549N**	No	CF-causing	NO
c.3752G>A	p.Ser945Leu	**S945L**	No	CF-causing	NO
c.1645A>C or c.1647T>G	p.Val520Phe	**V520F**	No	CF-causing	NO
c.274G>T	p.Trp1282X	**W1282X**	Yes	CF-causing	NO
c.2128A>T	p.Tyr1092X	**Y1092X**	No	CF-causing	NO
c.2195T>G	p.Tyr122X	**Y122X**	No	CF-causing	NO

Severe Combined Immunodeficiencies (SCID)

Final diagnosis as determined by a metabolic geneticist or clinician performing follow-up:

- ☐ Classic SCID
- ☐ Leaky SCID
- ☐ Omenn Syndrome
- ☐ Unknown

Diagnostic Workup	
Was the CD3 T cell level tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the CD3 T cell level? ☐ <300 autologous T cells, undetectable or very few naïve T cells ☐ 300-1500, few naïve T cells, oligoclonal T cells, or poor T cell diversity ☐ >80% CD45RO+ ☐ Any number (not zero) ☐ Untested/Unknown
Was proliferation to PHA test done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Proliferation to PHA: ☐ <10% of normal ☐ 10-50% of normal PHA ☐ 10-30% normal PHA or Absent to Candida/TT ☐ <30% of normal ☐ Any/Unknown
Was maternal engraftment documented? ☐ Yes ☐ No ☐ Unknown	
Molecular Genetics	

Was mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* **Were variants detected in the genes known to be associated with SCID?** ☐ Yes ☐ No ☐ Unknown *[IF YES]* *Check the type of variant found on allele 1:* ☐ Pathogenic variant in a known SCID gene ☐ Pathogenic variant in a known SCID gene on X chromosome in a male ☐ Pathogenic variant in a known SCID gene known to be associated with leaky SCID (previously reported or in a gene previously associated with combined immunodeficiency) ☐ Wild Type (Normal) ☐ Untested/Unknown *Check the type of variant found on allele 2:* ☐ Pathogenic variant in a known SCID gene ☐ Pathogenic variant in a known SCID gene known to be associated with leaky SCID (previously reported or in a gene previously associated with immunodeficiency) ☐ Wild Type (Normal) ☐ Untested/Unknown
	[IF variants detected=YES] **Was *22q1* deletion assessed?** ☐ Yes ☐ No ☐ Unknown
	[IF variants detected=YES] **Were homozygous or compound heterozygous *FOXN1* mutations assessed?** ☐ Yes ☐ No ☐ Unknown

	[IF variants detected=YES] **Were heterozygous *TBX1* variants assessed?** ☐ Yes ☐ No ☐ Unknown

Critical Congenital Heart Disease (CCHD)

What was the final diagnosis?

- ☐ CCHD
- ☐ Non-critical CCHD
- ☐ Other
- ☐ Unknown

Diagnostic Workup	
	[IF CCHD SELECTED] ☐ Truncus Arteriosus ☐ Total Anomalous Pulmonary Venous Connection ☐ Tetralogy of fallot ☐ Pulmonary Atresia ☐ Ebstein's Anomaly ☐ Hypoplastic Left Heart Syndrome ☐ Single ventricle ☐ Tricuspid atresia ☐ Transposition of the great arteries ☐ Double outlet right ventricle ☐ Coarctation of aorta ☐ Interrupted arch ☐ Aortic valve disease If Other selected; please specify_____

Please answer the following:	If Yes, what were the results of the postnatal echocardiogram? *(select all that apply)*
Was a Postnatal Echocardiogram Completed? ☐ Yes ☐ No ☐ Unknown	**Truncus Arteriosus** ☐ Truncus arteriosus ☐ Truncus arteriosus + Interrupted aortic arch **Total Anomalous Pulmonary Venous Connection (TAPVC)** ☐ Type1 (supracardiac) ☐ Type 2 (cardiac) ☐ Type 3 (infracardiac) ☐ Type 4 (mixed)

	Tetralogy of Fallot (TOF) ☐ TOF ☐ TOF, Pulmonary stenosis ☐ TOF, AVCanal (AVSD) ☐ TOF, Absent pulmonary valve

	Pulmonary Artesia ☐ Pulmonary atresia ☐ Pulmonary atresia, IVS ☐ Pulmonary atresia, VSD (Including TOF, PA) ☐ Pulmonary atresia, VSD-MAPCA
	Ebstein's Anomaly ☐ Ebstein's anomaly
	Hypoplastic Left Heart Syndrome (HLHS) ☐ Hypoplastic left heart syndrome
	Single Ventricle ☐ Single ventricle, DILV ☐ Single ventricle, DIRV ☐ Single ventricle, Mitral atresia ☐ Single ventricle, Unbalanced AV canal ☐ Single ventricle, Heterotaxia syndrome ☐ Single ventricle, Other ☐ Single ventricle + Total anomalous pulmonary venous connection (TAPVC)
	Tricuspid Artesia ☐ Single ventricle, Tricuspid atresia
	Transposition of the Great Arteries (TGA) ☐ d-TGA, IVS ☐ d-TGA, IVS-LVOTO ☐ d-TGA, VSD ☐ d-TGA, VSD-LVOTO
	Double Outlet Right Ventricle (DORV) ☐ DORV, VSD type ☐ DORV, TOF type ☐ DORV, TGA type ☐ DORV, Remote VSD (uncommitted VSD) ☐ DORV + AVSD (AV Canal) ☐ DORV, IVS ☐ DORV, Remote VSD (uncommitted VSD)
	Coarctation of Aorta ☐ Coarctation of aorta ☐ Aortic arch hypoplasia ☐ VSD + Aortic arch hypoplasia ☐ VSD + Coarctation of aorta

	Interrupted Arch ☐ Interrupted aortic arch ☐ Interrupted aortic arch + VSD ☐ Interrupted aortic arch + AP window (aortopulmonary window)

	Aortic Valve Disease ☐ Aortic Stenosis receiving intervention in first 30 days of life ☐ Pulmonary Stenosis receiving intervention in the first 30 days of life
Was a Prenatal Echocardiogram Completed? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Did the Prenatal Echo findings suggest CCHD? ☐ Yes ☐ No ☐ Unknown

X-Linked Adrenoleukodystrophy (X-ALD)

Final diagnosis as determined by a metabolic geneticist or clinician performing following-up:

- ☐ X-Linked Adrenoleukodystrophy (in males)
- ☐ Contiguous ABCD1 DXS1357E deletion syndrome (CADDS)
- ☐ X-Linked Adrenoleukodystrophy (in females)
- ☐ Peroxisomal Disorder
- ☐ Acyl-CoA Oxidase Deficiency
- ☐ D-Bifunctional Protein Deficiency
- ☐ Dyamin-like protein 1 (DLP1)
- ☐ ABDC5
- ☐ Non-peroxisomal Disorder
- ☐ Uncertain Type/Onset
- ☐ Unknown

Diagnostic Workup	
Was plasma VLCFA tested? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the VLCFA level? ☐ Elevated ☐ Slightly elevated ☐ Normal ☐ Low ☐ Unknown *"Elevated" signifies in pathogenic range, while "slightly elevated" signifies above normal, but not in the pathogenic range*
Clinical symptoms? ☐ Present ☐ Not present ☐ Not present at birth ☐ Unknown	*Symptoms may include: neonatal hypotonia, neonatal seizures, liver disease, neonatal cholestasis, sensorineural deafness, failure to thrive, craniofacial abnormalities*
Was plasmalogen testing done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Plasmalogen level? ☐ Normal ☐ Low ☐ Unknown

Family History done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Family history results: ☐ Family history present ☐ Family VLCFA studies suggestive of X-linked ALD ☐ Family history not present ☐ Unknown
Were fibroblast studies done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Fibroblast study results: ☐ Consistent with Zellweger Spectrum Disorder ☐ Consistent with Acyl-CoA Oxidase Deficiency ☐ Consistent with D-Bifunctional Protein ☐ Consistent with DLP1 ☐ Consistent with ABCD5 ☐ Unknown
Molecular Genetics	
Was mutation analysis done? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What genes were included in the mutation analysis? (select all that apply) ☐ *ABCD1* ☐ *PEX1* ☐ *ACOX1* ☐ *HSD17B4* ☐ 1 of the 7 known genes for Aicardi-Goutières Syndrome ☐ Other gene
	[IF ABCD1] **Check the type of variations found:** ☐ Pathogenic variant ☐ Deletion/duplication identified ☐ No mutation on sequencing, deletion/duplication not done ☐ No mutation on sequencing, deletion/duplication not toned; rule out other disorders of peroxisomal beta oxidation ☐ Variant of unknown significance ☐ Deletion identified in ABCD1 and DXS1357 ☐ Unknown

	[IF PEX1] **Check the type of variations found on:** **Allele 1** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown **Allele 2** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown
	[IF ACOX1] **Check the type of variations found on:** **Allele 1** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown **Allele 2** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown

	*[IF HSD17b4]***Check the type of variations found on:** **Allele 1** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown **Allele 2** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown
	[IF 1 of the 7 known genes for Aicardi-Goutières Syndrome] **Check the type of variations found on:** **Allele 1** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown **Allele 2** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown

	[IF Other Gene Selected] **Other Gene Name**;_____ **Check the type of variations found on:** **Allele 1** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown **Allele 2** ☐ Variant known to be disease causing ☐ Variant of unknown significance ☐ Variant of unknown significance (predicted to be pathogenic) ☐ Wild Type (normal) ☐ Unknown

Spinal Muscular Atrophy (SMA)

Diagnostic Workup	
Newborn Screen Molecular Test for SMN1? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the result? ☐ Zero copies of *SMN1* (presumed homozygous deletion/conversion)* ☐ Zero copies of *SMN1* (presumed homozygous deletion/conversion)* - observed on two independently collected NBS specimens ☐ 2 pathogenic variants ☐ 2 pathogenic variants observed on two independently collected NBS specimens ☐ 1 pathogenic variant and 1 variant of unknown significance ☐ 2 variants of unknown significance ☐ Unknown/ Not Done/Screen Negative ** true deletion of exon 7 (or larger) or for which there has been a gene conversion of exon 7 (or*
Newborn Screen Molecular Test for SMN2? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* SMN2 Copy Number? ☐ One ☐ Two ☐ Two or more ☐ Unknown/Not Done
Post-Newborn Screen Molecular Test for SMN1? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the result? ☐ Zero copies of *SMN1* (presumed homozygous deletion/conversion)* ☐ Zero copies of *SMN1* (presumed homozygous deletion/conversion)* - observed on two independently collected specimens ☐ 2 pathogenic variants ☐ 2 pathogenic variants observed on two independently collected specimens ☐ 1 pathogenic variant and 1 variant of unknown significance ☐ 2 variants of unknown significance ☐ Unknown/ Not Done/Screen Negative ** true deletion of exon 7 (or larger) or for which there has been a gene conversion of exon 7 (or more)*

Post-Newborn Screen Molecular Test for SMN2? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* *SMN2* Copy Number? ☐ One ☐ Two ☐ Two or more ☐ Unknown/Not Done
Parental Molecular Testing Family History/Parental Genetic Testing? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* What was the result? ☐ Phasing is complete and confirms that variants are in trans or both parents are known to be carriers of the pathogenic variants identified ☐ Both parents are known carriers of *SMN1* deletion ☐ Unknown/Not Done
Clinical symptoms? ☐ Present ☐ Not present ☐ Unknown	*Symptoms may include: Electromyography evidence of motor neuron disease, Absent reflexes, Fasciculations, Feeding difficulty, Hypotonia, Respiratory Difficulty, Weakness*
Was treatment started? ☐ Yes ☐ No ☐ Unknown	*[IF YES]* Type of treatment? (Check all that apply) ☐ Gene Therapy ☐ Nusinersin ☐ Other: please describe_____ ☐ Unknown