NewSTEPs

A Program of the Association of Public Health Laboratories

Diagnostic Form Templates for Newborn Screening

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Case Information

State Unique ID	
Infant's Name	
Infant's Date of Birth (mm/dd/yyyy)	
Mom's Name	

Metabolic Disorders

Organic Acid Disorders

Elevated C5-DC Acylcarnitine (Glutaric Acidemia/ Aciduria Type I (GA1))

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ Glutaric Acidemia/ Aciduria Type I (GA1) ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:
Biochemical	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *3-OH Glutaric acid level:* ☐ Elevated ☐ Normal ☐ Unknown *Glutaric acid level:* ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ No ☐ Unknown	*C5-DC level:* ☐ Elevated ☐ Normal ☐ Unknown
Was urine glutarylcarnitine (C5-DC) tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was C5-DC level* ☐ Elevated ☐ Normal ☐ Unknown
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *GCDH* ☐ Other gene:

Metabolic Disorders

Organic Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metabolic Disorders

Organic Acid Disorders

Elevated C5-OH Acylcarnitine Result

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- ☐ Maternal 3-methylcrotonyl-CoA carboxylase deficiency
- ☐ 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)
- ☐ Holocarboxylase Synthetase Deficiency (MCD)
- ☐ 2-methyl-3-hydroxybutyric acidemia (MGBD)
- ☐ 3-methylglutaconic aciduria (MGA)
- ☐ MT-ATP6 related mitochondrial disorders
- ☐ Other Biotin Disorder (not biotinidase deficiency) ☐
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Other: ☐

Were urine organic acids tested on infant?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Were results indicative of a C5-OH related disease?

- ☐ Yes
- ☐ No
- ☐ Unknown

[IF YES above] Indicate elevated analytes

- ☐ 2-methyl-3-hydroxybutyric acid
- ☐ 2-methylacetoacetate
- ☐ 3-hydroxypropionic acid
- ☐ 3-hydroxyvaleric acid
- ☐ 3-hydroxy-3-methylglutaric acid
- ☐ 3-methylcrotonylglycine
- ☐ 3-methylglutaconic acid
- ☐ 3-methylglutaric acid
- ☐ Lactic acid
- ☐ Methylcitric acid
- ☐ Tiglylglycine
- ☐ Other ☐

Metabolic Disorders

Organic Acid Disorders

Biochemical — Urine Organic Acids (cont.)

Were urine organic acids tested on biological mother?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]
Were results indicative of a C5-OH related disease?

- ☐ Yes
- ☐ No
- ☐ Unknown

[IF YES above] Indicate elevated analytes

- ☐ 2-methyl-3-hydroxybutyric acid
- ☐ 2-methylacetoacetate
- ☐ 3-hydroxypropionic acid
- ☐ 3-hydroxyvaleric acid
- ☐ 3-hydroxy-3-methylglutaric acid
- ☐ 3-methylcrotonylglycine
- ☐ 3-methylglutaconic acid
- ☐ 3-methylglutaric acid
- ☐ Lactic acid
- ☐ Methylcitric acid
- ☐ Tiglylglycine
- ☐ Other _____

Biochemical — Plasma Acylcarnitines

Were plasma acylcarnitines tested on the infant?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]
Was C5-OH level

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Were plasma acylcarnitines tested on biological mother?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]
Was C5-OH level

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Biochemical — Enzyme Studies

Were biotinidase enzyme activity studies completed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]
Biotinidase enzyme activity was:

- ☐ Normal
- ☐ Abnormal
- ☐ Unknown

Metabolic Disorders

Organic Acid Disorders

Biochemical — Enzyme Studies (cont.)

Was holocarboxylase synthetase enzyme analysis completed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Enzyme activity was:

- ☐ Consistent with disease
- ☐ Normal activity (not consistent with disease)
- ☐ Unknown

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ *ACAT1*
- ☐ *AUH*
- ☐ *BTD*
- ☐ *HLCS*
- ☐ *HSD17B10*
- ☐ *HMGCL*
- ☐ *MCCC1*
- ☐ *MCCC2*
- ☐ Other gene: _____

[For each gene selected]

Check the types of variants found on:

Allele 1:

- ☐ Pathogenic variant
- ☐ Likely pathogenic variant
- ☐ Variant of uncertain significance
- ☐ Likely benign variant
- ☐ Benign variant
- ☐ No variant detected
- ☐ Unknown
- ☐ Other _____

Allele 2:

- ☐ Pathogenic variant
- ☐ Likely pathogenic variant
- ☐ Variant of uncertain significance
- ☐ Likely benign variant
- ☐ Benign variant
- ☐ No variant detected
- ☐ Unknown
- ☐ Other _____

Metabolic Disorders

Organic Acid Disorders

Molecular Genetics (cont.)	
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metabolic Disorders

Organic Acid Disorders

Elevated C5 Acylcarnitine (Isovaleric Acidemia/ Aciduria (IVA))

Final Outcome	
Final Outcome 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 ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:
Biochemical	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *3-OH Isovaleric acid level:* ☐ Elevated ☐ Normal ☐ Unknown *Isovalerylglycine level:* ☐ Elevated ☐ Normal ☐ Unknown *2-methylbutyrylglycine level:* ☐ Elevated ☐ Normal ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *C5 level:* ☐ Elevated ☐ Normal ☐ Unknown

Metabolic Disorders

Organic Acid Disorders

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ *ACADSB*
- ☐ *IVD*
- ☐ Other gene: _____

[For each gene selected]

Check the types of variants found on:

Allele 1:

- ☐ Pathogenic variant
- ☐ Likely pathogenic variant
- ☐ Variant of uncertain significance
- ☐ Likely benign variant
- ☐ Benign variant
- ☐ No variant detected
- ☐ Unknown
- ☐ Other _____

Allele 2:

- ☐ Pathogenic variant
- ☐ Likely pathogenic variant
- ☐ Variant of uncertain significance
- ☐ Likely benign variant
- ☐ Benign variant
- ☐ No variant detected
- ☐ Unknown
- ☐ Other _____

Was parental phasing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

The variants were found to be in:

- ☐ Cis
- ☐ Trans

Clinical Management

Was intervention initiated?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

(Check all that apply)

- ☐ Treatment already administered
- ☐ Monitoring protocols started
- ☐ Process for treatment administration started

Elevated C3 (Propionic Acidemia/ Aciduria (PROP) and Methylmalonic Acidemias (MUT, Cbls))

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ Propionic Acidemia (PROP) ☐ Maternal vitamin B12 deficiency ☐ Succinate-CoA ligase deficiency ☐ Mutase(-) (mut-) ☐ Mutase (0) (mut0) ☐ Cobalamin A deficiency (Cbl A) ☐ Cobalamin B deficiency (Cbl B) ☐ Cobalamin C deficiency (Cbl C) ☐ Cobalamin D deficiency (Cbl D) ☐ Cobalamin Dv1 deficiency (Cbl Dv1) ☐ Cobalamin Dv2 deficiency (Cbl Dv2) ☐ Cobalamin F deficiency (Cbl F) ☐ Cobalamin X deficiency (Cbl X) ☐ Other transcobalamin deficiency or defect ☐ Unclassified ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:

Metabolic Disorders

Organic Acid Disorders

Biochemical	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Please indicate which of the following metabolites were detected:* *Propionylglycine:* ☐ Yes ☐ No ☐ Unknown *Tiglyglycine:* ☐ Yes ☐ No ☐ Unknown *Methylcitrate:* ☐ Yes ☐ No ☐ Unknown *3-OH Propionic acid level:* ☐ Yes ☐ No ☐ Unknown *Methylmalonic acid (MMA):* ☐ Yes ☐ No ☐ Unknown *3-Methylcrotonylglycine:* ☐ Yes ☐ No ☐ Unknown
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was C3 level:* ☐ Elevated ☐ Normal ☐ Unknown
Were maternal vitamin B12 levels tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was biologic mom found to be vitamin B12 deficient?* ☐ Yes ☐ No ☐ Unknown

Metabolic Disorders

Organic Acid Disorders

Biochemical (cont.)	
Were infant vitamin B12 levels tested? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *Was infant found to be vitamin B12 deficient?* ☐ Yes ☐ No ☐ Unknown
Was total plasma homocysteine tested? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *Total plasma homocysteine:* ☐ Elevated ☐ Normal ☐ Unknown
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *What genes were included in the molecular analysis? (select all that apply)* ☐ *ABCD4* ☐ *C2ORF25* ☐ *HCFC1* ☐ *MMAA* ☐ *MMAB* ☐ *MMACHC* ☐ *MMADHC* ☐ *MMUT* ☐ *LMBRD1* ☐ *PCCA* ☐ *PCCB* ☐ Other gene: _____

Metabolic Disorders

Organic Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metabolic Disorders

Fatty Acid Disorders

Primary Carnitine Deficiency/ Carnitine Uptake Deficiency (CUD)

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ Carnitine Uptake Deficiency (CUD) ☐ Maternal Carnitine Deficiency (primary and secondary) ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:
Biochemical	
Was urine carnitine tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was fractional excretion of free carnitine level:* ☐ Elevated ☐ Normal ☐ Unknown *Was 3-Methylcrotonylglycine 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	
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *SLC22A5* ☐ Other gene:

Metabolic Disorders

Fatty Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metabolic Disorders

Fatty Acid Disorders

Medium-chain acyl-CoA Dehydrogenase Deficiency (MCAD)

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ MCAD ☐ Medium-chain ketoacyl-CoA thiolase deficiency ☐ GAII ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:
Biochemical	
Were urine organic acids or acylglycines tested? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *Hexanoylglycine level:* ☐ Elevated ☐ Normal ☐ Unknown

Metabolic Disorders

Fatty Acid Disorders

Biochemical (cont.)

Were plasma acylcarnitines tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

C8 level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

C8/C10 level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

C8/C2 level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

C6 level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

C10 level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

C10:1 level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ *ACADM*
- ☐ Other gene: _____

Metabolic Disorders

Fatty Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Elevated C16-OH +/- and C18:1-OH (Trifunctional Protein Deficiency (TFP)/LCHAD)

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ Trifunctional Protein Deficiency (TFP) ☐ Long-chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:
Biochemical	
Were urine organic acids tested? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *C12-OH dicarboxylic acid level:* ☐ Elevated ☐ Normal ☐ Unknown *C10-OH dicarboxylic acid level:* ☐ Elevated ☐ Normal ☐ Unknown

Metabolic Disorders

Fatty Acid Disorders

Biochemical (cont.)	
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *C16-OH level:* ☐ Elevated ☐ Normal ☐ Unknown *C16:1-OH level:* ☐ Elevated ☐ Normal ☐ Unknown *C18-OH level:* ☐ Elevated ☐ Normal ☐ Unknown *C18:1-OH level:* ☐ Elevated ☐ Normal ☐ Unknown
Was TFP enzyme analysis completed? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Enzyme activity:* ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *HADHA* ☐ *HADHB* ☐ Other gene: _____

Metabolic Disorders

Fatty Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metabolic Disorders

Fatty Acid Disorders

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

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ VLCAD ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other: _____
Biochemical	
Were plasma acylcarnitines tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *C14:1 level:* ☐ Elevated (on more than one sample) ☐ Normal ☐ Unknown *C14:2-OH level:* ☐ Elevated ☐ Normal ☐ Unknown *C14 level:* ☐ Elevated ☐ Normal ☐ Unknown
Was VLCAD enzyme activity analysis completed? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Enzyme activity:* ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *ACADVL* ☐ Other gene: _____

Metabolic Disorders

Fatty Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metabolic Disorders

Amino Acid Disorders

Elevated Citrulline (Argininosuccinic Acidemia/ Aciduria (ASA), Citrullinemia Type I (CIT))

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ Argininosuccinic Acidemia/ Aciduria (ASA) ☐ Citrullinemia Type I ☐ Citrullinemia Type II ☐ Pyruvate carboxylase deficiency ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other: _____
Biochemical	
Were plasma amino acids tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Plasma ASA level:* ☐ Elevated ☐ Normal ☐ Unknown *Citrulline level:* ☐ Elevated ☐ Normal ☐ Unknown
Were urine amino acids tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Urine ASA level:* ☐ Elevated ☐ Normal ☐ Unknown *Urine Citrulline level:* ☐ Elevated ☐ Normal ☐ Unknown

Metabolic Disorders

Amino Acid Disorders

Biochemical (cont.)	
Was blood ammonia levels tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Blood ammonia levels:* ☐ Elevated ☐ Normal ☐ Unknown
Was ASA enzyme activity analysis completed? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Enzyme analysis results:* ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Was Citrullinemia Type I enzyme activity analysis completed? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Enzyme analysis results:* ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *ASL* ☐ *ASS1* ☐ *SLC25A13* ☐ Other gene: _____

Metabolic Disorders

Amino Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metabolic Disorders

Amino Acid Disorders

Classic Phenylketonuria (PKU) and Hyperphenylalaninemia (Hyperphe)

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ Classic phenylketonuria (PKU)
- ☐ Benign hyperphenylalaninemia (H-PHE)
- ☐ HyperPhe diet controlled
- ☐ Dihydropterine reductase deficiency (DHPR)
- ☐ DNAJC12
- ☐ Maternal PKU
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Other:

Biochemical

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

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ *PAH*
- ☐ Other gene:

Metabolic Disorders

Amino Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metabolic Disorders

Amino Acid Disorders

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

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ Classic Homocystinuria ☐ Methionine Adenosyltransferase (MAT I/III Deficiency) ☐ Glycine n-methyltransferase (GNMT) ☐ Adenosylhomocysteine Hydrolase Deficiency ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other: _____
Biochemical	
Were plasma amino acids tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was Methionine level:* ☐ Elevated ☐ Normal ☐ Unknown *Was Homocysteine level:* ☐ Elevated ☐ Normal ☐ Unknown
Was plasma total Homocysteine tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was plasma total Homocysteine level:* ☐ Elevated ☐ Normal ☐ Unknown
Was CBS enzyme activity analysis completed? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was enzyme analysis:* ☐ Consistent with disease ☐ Normal activity (not consistent with disease) ☐ Unknown

Metabolic Disorders

Amino Acid Disorders

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ *CBS*
- ☐ *MTHFR*
- ☐ Other gene: _____

[For each gene selected]

Check the types of variants found on:

Allele 1:

- ☐ Pathogenic variant
- ☐ Likely pathogenic variant
- ☐ Variant of uncertain significance
- ☐ Likely benign variant
- ☐ Benign variant
- ☐ No variant detected
- ☐ Unknown
- ☐ Other _____

Allele 2:

- ☐ Pathogenic variant
- ☐ Likely pathogenic variant
- ☐ Variant of uncertain significance
- ☐ Likely benign variant
- ☐ Benign variant
- ☐ No variant detected
- ☐ Unknown
- ☐ Other _____

Was parental phasing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

The variants were found to be in:

- ☐ Cis
- ☐ Trans

Clinical Management

Was intervention initiated?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

(Check all that apply)

- ☐ Treatment already administered
- ☐ Monitoring protocols started
- ☐ Process for treatment administration started

Metabolic Disorders

Amino Acid Disorders

Maple Syrup Urine Disease (MSUD)

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ Classic ☐ Intermediate ☐ Thiamine-response ☐ Hydroxyprolinemia ☐ Unclassified ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:

Biochemical	
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

Metabolic Disorders

Amino Acid Disorders

Biochemical (cont.)	
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
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *DBT* ☐ *BCKDHB* ☐ *DLD* ☐ *BCKDHA* ☐ Other gene:

Metabolic Disorders

Amino Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metabolic Disorders

Amino Acid Disorders

Tyrosinemia Type I (TYR-1)

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ Tyrosinemia, Type I (hepatorenal) ☐ Transient Tyrosinemia of the neonate (TTN) ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:
Biochemical	
Were plasma amino acids tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *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 quantitative succinylacetone tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was the succinylacetone level:* ☐ Elevated ☐ Normal ☐ Unknown
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *FAH* ☐ Other gene:

Metabolic Disorders

Amino Acid Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Endocrine Disorders

Congenital Disorders

Congenital Hypothyroidism (CH)

Final Outcome	
Final Outcome 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 ☐ False Positive/Unaffected ☐ Other:
Biochemical	
Was Serum TSH tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What was the TSH 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 Serum Total T4 tested before initiation of treatment?* ☐ Yes ☐ No ☐ Unknown

Endocrine Disorders

Congenital Disorders

Biochemical (cont.)	
Was Serum Free T4 tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was Serum Free T4 below the age-established reference range?* ☐ Yes ☐ No ☐ Unknown *Was Serum Free T4 tested before initiation of treatment?* ☐ Yes ☐ No ☐ Unknown
Does this infant have any other pituitary hormone deficiencies? ☐ Yes ☐ No ☐ Unknown	
Was TBG tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Was TBG below the age established reference range?* ☐ Yes ☐ No ☐ Unknown
Was Imaging done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Were any of these found?* ☐ Thyroid agenesis ☐ Hypoplastic thyroid tissue ☐ Ectopic thyroid tissue
Were Anti-TSH receptor antibodies measured? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Were TRBAbs present?* ☐ Yes ☐ No ☐ Unknown
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Endocrine Disorders

Congenital Disorders

Congenital Adrenal Hyperplasia (CAH)

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ Classic 21-Hydroxylase Deficiency-Salt Wasting ☐ Classic 21-Hydroxylase Deficiency-Simple Virilizing ☐ Non-Classic 21-Hydroxylase Deficiency ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:
Biochemical	
Sex assigned at birth ☐ Male ☐ Female ☐ Unknown ☐ Unspecified	
Was confirmatory serum 17-OHP level obtained? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *Baseline value:* ☐ >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

Endocrine Disorders

Congenital Disorders

Biochemical (cont.)	
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]* *(Check all that apply)* ☐ Ambiguous genitalia, with 46 XX karyotype ☐ Typical genitalia, with 46 XY karyotype ☐ Other hormonal evidence of CAH _____
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *CYP21A2* ☐ Other gene: _____

Endocrine Disorders

Congenital Disorders

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Presence of Hb S

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ S, Beta 0-thalassemia – HB S/B°Th
- ☐ S,S Disease (Sickle Cell Anemia) – HbSS
- ☐ S, Beta + Thalassemia – HbS/B+Th
- ☐ S,C Disease – Hb S/C
- ☐ Other: _____

Diagnostic Workup

Was qualitative (IEF or HPLC) testing completed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What were the results?

- ☐ FS
- ☐ FSC
- ☐ FSA
- ☐ FSA2
- ☐ FSAA2
- ☐ Other: _____
- ☐ Unknown

Was quantitative (HPLC or electrophoresis) testing completed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What were the results?

- ☐ FS
- ☐ FSC
- ☐ FS with high A2
- ☐ FSA with high A2
- ☐ FSA
- ☐ Other: _____
- ☐ Unknown

Hemoglobinopathies

Diagnostic Workup (cont.)

Was molecular genetic testing performed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Check the type of variant found on: Allele 1

- ☐ S
- ☐ C
- ☐ Beta + Thal
- ☐ Beta° + Thal
- ☐ Other _____
- ☐ Unknown

Allele 2

- ☐ S
- ☐ C
- ☐ Beta + Thal
- ☐ Beta° + Thal
- ☐ Other _____
- ☐ Unknown

Was a CBC performed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What were the results?

- ☐ Normal – high MCV
- ☐ Low MCV
- ☐ Unknown

Clinical Management

Was intervention initiated?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

(Check all that apply)

- ☐ Treatment already administered
- ☐ Monitoring protocols started
- ☐ Process for treatment administration started

Presence of Other Hb Variant

Final Outcome

Final Outcome as determined by clinician performing follow-up:

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

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
- ☐ FOARAB
- ☐ Other: _____
- ☐ Unknown

Was quantitative (HPLC or electrophoresis) testing completed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]
What were the results?

- ☐ FC
- ☐ FD
- ☐ FE
- ☐ FOARAB
- ☐ Other: _____
- ☐ Unknown

Hemoglobinopathies

Diagnostic Workup (cont.)

Was molecular genetic testing performed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Check the type of variant found on allele 1:

- ☐ C
- ☐ D
- ☐ E
- ☐ OARAB
- ☐ Other: _____
- ☐ Unknown

Check the type of variant found on allele 2:

- ☐ C
- ☐ D
- ☐ E
- ☐ OARAB
- ☐ Beta + Thal
- ☐ Beta° + Thal
- ☐ Other: _____
- ☐ Unknown

Was a CBC performed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What were the results?

- ☐ Normal – high MCV
- ☐ Low MCV

Clinical Management

Was intervention initiated?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

(Check all that apply)

- ☐ Treatment already administered
- ☐ Monitoring protocols started
- ☐ Process for treatment administration started

Mucopolysaccharidosis Type I (MPS I)

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ MPS I—Severe
- ☐ MPS I—Severity not determined
- ☐ MPS I—Attenuated
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Pseudodeficiency
- ☐ Other: _____

Biochemical

Was IDUA enzyme activity analysis completed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

IDUA enzyme level

- ☐ Within known affected range
- ☐ Normal
- ☐ Unknown

Were urine GAGs tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Urine GAG level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Were there clinical symptoms present at the time of initial evaluation?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

(Check all that apply)

- ☐ Hepatosplenomegaly
- ☐ Coarse facial features
- ☐ Hydrocephalus
- ☐ Skeletal abnormalities (dysostosis multiplex)
- ☐ Joint stiffness or limited mobility
- ☐ Macroglossia (large tongue)
- ☐ Prominent forehead (frontal bossing)
- ☐ Hearing loss
- ☐ Hernia
- ☐ No clinical symptoms observed
- ☐ Unknown

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ *IDUA*
- ☐ Other gene: _____

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ Pseudodeficiency variant ☐ No variant detected ☐ Unknown ☐ Other **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ Pseudodeficiency variant ☐ No variant detected ☐ Unknown ☐ Other
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Mucopolysaccharidosis Type II (MPS II)

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ MPS II—Severe (neuropathic)
- ☐ MPS II—Attenuated (non-neuropathic)
- ☐ Multiple sulfatase deficiency (MSD)
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Pseudodeficiency
- ☐ Other: _____

Biochemical

Was I2S enzyme activity tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

I2S enzyme level

- ☐ Within known affected range
- ☐ Normal
- ☐ Unknown

Were urine GAGs tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Urine GAG level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Were urine sulfatides tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Urine sulfatide level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Were there clinical symptoms present at the time of initial evaluation?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

(Check all that apply)

- ☐ Hepatosplenomegaly
- ☐ Coarse facial features
- ☐ Large head (macrocephaly)
- ☐ Joint stiffness or limited mobility
- ☐ Macroglossia (large tongue)
- ☐ Hearing loss
- ☐ Hernia
- ☐ No clinical symptoms observed
- ☐ Unknown

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ I2S
- ☐ SUMF1
- ☐ Other gene: _____

[For each gene selected]

Check the

Allele 1:

- ☐ Pathogenic variant
- ☐ Likely pathogenic variant
- ☐ Variant of uncertain significance
- ☐ Likely benign variant
- ☐ Benign variant
- ☐ Pseudodeficiency variant
- ☐ No variant detected
- ☐ Unknown
- ☐ Other

Allele 2:

- ☐ Pathogenic variant
- ☐ Likely pathogenic variant
- ☐ Variant of uncertain significance
- ☐ Likely benign variant
- ☐ Benign variant
- ☐ Pseudodeficiency variant
- ☐ No variant detected
- ☐ Unknown
- ☐ Other

Was parental phasing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

The variants were found to be in:

- ☐ Cis
- ☐ Trans

Clinical Management

Was intervention initiated?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

(Check all that apply)

- ☐ Treatment already administered
- ☐ Monitoring protocols started
- ☐ Process for treatment administration started

Krabbe Disease

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ Infantile Krabbe Disease
- ☐ Late-Onset Krabbe Disease
- ☐ Uncertain Type/Onset
- ☐ Saposin A deficiency
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Pseudodeficiency
- ☐ Other: _____

Biochemical

Was GALC enzyme activity tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

GALC enzyme level

- ☐ Within known affected range
- ☐ Normal
- ☐ Unknown

Was psychosine tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Psychosine level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ GALC
- ☐ PSAP
- ☐ Other gene: _____

Molecular Genetics (cont.)	
	[For each gene selected] Check the **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ Pseudodeficiency variant ☐ No variant detected ☐ Unknown ☐ Other **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ Pseudodeficiency variant ☐ No variant detected ☐ Unknown ☐ Other
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Metachromatic Leukodystrophy (MLD)

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ Late-Infantile MLD
- ☐ Early-Juvenile MLD
- ☐ Late-Juvenile MLD
- ☐ Adult MLD
- ☐ Multiple sulfatase deficiency (MSD)
- ☐ Saposin B deficiency
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Pseudodeficiency
- ☐ Other: _____

Biochemical

Was ARSA enzyme activity tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

ARSA enzyme level

- ☐ Within known affected range
- ☐ Normal
- ☐ Unknown

Were urine sulfatides tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Urine sulfatide level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Were urine GAGs tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Urine GAG level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ ARSA
- ☐ PSAP
- ☐ SUMF1
- ☐ Other gene: _____

Molecular Genetics (cont.)	
	[For each gene selected] Check the **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ Pseudodeficiency variant ☐ No variant detected ☐ Unknown ☐ Other **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ Pseudodeficiency variant ☐ No variant detected ☐ Unknown ☐ Other
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Pompe Disease

Final Outcome

Final Outcome as determined by clinician performing follow-up:	☐ Infantile Onset (IO) Pompe Disease ☐ Late Onset (LO) Pompe Disease ☐ Uncertain Type/Onset ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Pseudodeficiency ☐ Other:

Biochemical

Was GAA enzyme activity in leukocytes tested? ☐ Yes ☐ No ☐ Unknown	**[If YES]** *Leukocyte GAA enzyme level:* ☐ Within affected range for infantile onset (IO) ☐ Within lab known affected range for late onset (LO) ☐ Low (generally) ☐ Unknown
Was enzyme activity tested in skin/muscle? ☐ Yes ☐ No ☐ Unknown	**[If YES]** *Results:* ☐ 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
Was Urine Hex4 level tested? ☐ Yes ☐ No ☐ Unknown	**[If YES]** *Urine Hex4 Level:* ☐ Elevated ☐ Normal ☐ Unknown

Biochemical (cont.)	
Were CK levels tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *CK levels:* ☐ Elevated ☐ Normal ☐ Unknown
Were LDH/ALT/AST tested? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *LDH/ALT/AST Enzyme levels:* ☐ Elevated ☐ Normal ☐ Unknown
Were there clinical symptoms present at the time of initial evaluation? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Muscle weakness (hypotonia) ☐ Respiratory insufficiency ☐ Feeding difficulties
Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ GAA ☐ Other gene:

Molecular Genetics (cont.)	
	[For each gene selected] Check the **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ Pseudodeficiency variant ☐ No variant detected ☐ Unknown ☐ Other **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ Pseudodeficiency variant ☐ No variant detected ☐ Unknown ☐ Other
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Biotinidase Deficiency (BIOT)

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ Profound Biotinidase deficiency
- ☐ Partial Biotinidase deficiency
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Other: _____

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 molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ *BTD*
- ☐ Other gene: _____

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Galactosemia (GALT)

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ Classic Galactosemia
- ☐ Duarte variant galactosemia
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Other: _____

Biochemical

Were GALT enzyme levels tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]
Was GALT level:

- ☐ <10%
- ☐ 10-30%
- ☐ Normal
- ☐ Unknown

Was Galactose-1-phosphate (Gal-1-P) tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]
Was Gal-1-P level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]
What genes were included in the molecular analysis? (select all that apply)

- ☐ *GALT*
- ☐ Other gene: _____

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Cystic Fibrosis

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ CFTR-Related Metabolic Syndrome/CF Screen Positive, Inconclusive Diagnosis (CRMS/CFSPID)
- ☐ Cystic Fibrosis (CF)
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Other: _____

Diagnostic Workup

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?

- ☐ ≥60 mmol/L
- ☐ 30-59 mmol/L
- ☐ ≤29mmol/L

[If NO]

Were there attempts to obtain a sweat chloride that were quantity not sufficient (QNS)?

- ☐ Yes
- ☐ No
- ☐ Unknown

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ *CFTR*
- ☐ Other gene: _____

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *The variants were found to be in:* ☐ Cis ☐ Trans
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Severe Combined Immunodeficiencies (SCID)

Final Outcome

Final Outcome as determined by clinician performing follow-up:	☐ Classic SCID ☐ Leaky SCID ☐ Omenn Syndrome ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:

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	

Other Disorders

Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *ADA* ☐ *DCLRE1C* ☐ *IL2RG* ☐ *IL7R* ☐ *JAK3* ☐ *RAG1* ☐ *RAG2* ☐ *RMRP* ☐ Other gene: _____
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans

Other Disorders

Molecular Genetics (cont.)	
Was 22q1 deletion assessed? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *22q deletion results:* ☐ Deletion found ☐ No deletion found ☐ Unknown
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Critical Congenital Heart Disease (CCHD)

Final Outcome	
Final Outcome as determined by clinician performing follow-up:	☐ CCHD ☐ Non-critical CCHD ☐ Carrier/Heterozygote ☐ False Positive/Unaffected ☐ Other:
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 ☐ Other; please specify
Was a Postnatal Echocardiogram Completed? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *What were the results? (select all that apply)*
	Truncus Arteriosus ☐ Truncus arteriosus ☐ Truncus arteriosus + Interrupted aortic arch
	Total Anomalous Pulmonary Venous Connection (TAPVC) ☐ Type 1 (supracardiac) ☐ Type 2 (cardiac) ☐ Type 3 (infracardiac) ☐ Type 4 (mixed)

Other Disorders

Diagnostic Workup (cont.)	
	Tetralogy of Fallot (TOF) ☐ TOF ☐ TOF, Pulmonary stenosis ☐ TOF, AVCanal (AVSD) ☐ TOF, Absent pulmonary valve
	Pulmonary Atresia ☐ 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 + TAPVC
	Tricuspid Atresia ☐ Single ventricle, Tricuspid atresia
	Transposition of the Great Arteries (TGA) ☐ d-TGA, IVS ☐ d-TGA, IVS-LVOTO ☐ d-TGA, VSD ☐ d-TGA, VSD-LVOTO

Other Disorders

Diagnostic Workup (cont.)	
	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
	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
	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
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

X-Linked Adrenoleukodystrophy (X-ALD)

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ X-Linked Adrenoleukodystrophy (in males)
- ☐ X-Linked Adrenoleukodystrophy (in females)
- ☐ Peroxisomal Spectrum Disorder
- ☐ Aicardi-Goutières syndrome (AGS)
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Other: _____

Diagnostic Workup

Was plasma VLCFA tested?

- ○ Yes
- ○ No
- ○ Unknown

[If YES]

What was the VLCFA level?

- ○ Elevated
- ○ Slightly elevated
- ○ Normal
- ○ Low
- ○ Unknown

Were there clinical symptoms present at the time of initial evaluation?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

(Check all that apply)

- ☐ Hypotonia
- ☐ Seizures
- ☐ Liver disease
- ☐ Cholestasis
- ☐ Craniofacial abnormalities

Was plasmalogen testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Plasmalogen level?

- ☐ Normal
- ☐ Low
- ☐ Unknown

Family History assessed?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Family history results:

- ☐ Family history of X-ALD present
- ☐ Family VLCFA studies suggestive of X-linked ALD
- ☐ Family history not present
- ☐ Unknown

Other Disorders

Molecular Genetics	
Was molecular genetic testing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *What genes were included in the molecular analysis? (select all that apply)* ☐ *ABCD1* ☐ *PEX1* ☐ *ACOX1* ☐ *HSD17B4* ☐ Aicardi-Goutières Syndrome genes ☐ Other gene:
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other
Was parental phasing done? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *The variants were found to be in:* ☐ Cis ☐ Trans

Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	*[If YES]* *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started

Spinal Muscular Atrophy (SMA)

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ SMA
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Other: _____

Diagnostic Workup

Was SMN1 analysis done after newborn screening?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What was the result?

- ☐ Zero copies of SMN1
- ☐ 2 pathogenic variants
- ☐ 1 pathogenic variant and 1 variant of unknown significance
- ☐ 2 variants of unknown significance
- ☐ Unknown/Not Done/Screen Negative

Was SMN2 analysis done after newborn screening?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

SMN2 Copy Number?

- ☐ One
- ☐ Two
- ☐ Three
- ☐ Four
- ☐ Five or More
- ☐ Unknown/Not Done

Were there clinical symptoms present at the time of initial evaluation?

- ○ Yes
- ○ No
- ○ Unknown

[If YES]

(Check all that apply)

- ☐ Electromyography evidence of motor neuron disease
- ☐ Absent reflexes
- ☐ Fasciculations
- ☐ Feeding difficulty
- ☐ Hypotonia
- ☐ Respiratory difficulties
- ☐

Clinical Management

Was intervention started?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

Type of treatment? (Check all that apply)

- ☐ Gene Therapy
- ☐ Nusinersen
- ☐ Other: please describe _____
- ☐ Unknown

Duchenne Muscular Dystrophy (DMD)

Final Outcome

Final Outcome as determined by clinician performing follow-up:

- ☐ Duchenne Muscular Dystrophy
- ☐ Becker Muscular Dystrophy
- ☐ DMD-associated dilated cardiomyopathy
- ☐ Carrier/Heterozygote
- ☐ False Positive/Unaffected
- ☐ Other: _____

Biochemical

Was serum CK levels tested?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

CK level:

- ☐ Elevated
- ☐ Normal
- ☐ Unknown

Molecular Genetics

Was molecular genetic testing done?

- ☐ Yes
- ☐ No
- ☐ Unknown

[If YES]

What genes were included in the molecular analysis? (select all that apply)

- ☐ *DMD*
- ☐ Other gene: _____

Molecular Genetics (cont.)	
	[For each gene selected] Check the types of variants found on: **Allele 1:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____ **Allele 2:** ☐ Pathogenic variant ☐ Likely pathogenic variant ☐ Variant of uncertain significance ☐ Likely benign variant ☐ Benign variant ☐ No variant detected ☐ Unknown ☐ Other _____
Clinical Management	
Was intervention initiated? ☐ Yes ☐ No ☐ Unknown	***[If YES]*** *(Check all that apply)* ☐ Treatment already administered ☐ Monitoring protocols started ☐ Process for treatment administration started