



# Case Worksheets for Newborn Screening

Last Updated: June 25, 2024

## Case Information Worksheet: Information Collected for ALL Cases

Infant Demographic Information	
State Unique ID? (alphanumeric)* <i>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.</i>	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) <i>If time of birth is not available, only enter the date</i>	
Year* <i>Year of birth is stored to calculate Quality Indicators</i>	<i>Automatically populated based on date of birth</i>
Birth Weight? (in grams)	
Biological Sex?	<input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Unspecified <input type="checkbox"/> Unknown
Race? (Select all that apply)	<input type="checkbox"/> White <input type="checkbox"/> Black or African American <input type="checkbox"/> American Indian or Alaskan Native <input type="checkbox"/> Asian <input type="checkbox"/> Native Hawaiian or other Pacific Islander <input type="checkbox"/> Not Reported <input type="checkbox"/> Unknown
Ethnicity?	<input type="checkbox"/> Hispanic, Latino(a) or Spanish origin <input type="checkbox"/> Not of Hispanic, Latino(a), or Spanish origin <input type="checkbox"/> Not Reported <input type="checkbox"/> Unknown
Screening Information	
Which newborn screen result indicated this infant was at risk for the disorder?	<input type="checkbox"/> Initial Screen <input type="checkbox"/> Subsequent Screen <input type="checkbox"/> Second Required Screen
Was prenatal testing done that indicated that this infant was at risk for this disorder?	<input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
Was there family history that indicated that this infant was at risk for this disorder?	<input type="checkbox"/> Yes <input type="checkbox"/> No

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

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

## Newborn Screening Surveillance Case Definitions:

### Case Confirmatory Diagnosis Follow-Up

Developed by the Health Resources and Services Administration (HRSA) and NewSTEPS in cooperation with the newborn screening medical sub-specialty community, standard surveillance case definitions for newborn screening conditions allow for determination of true prevalence and incidence of disorders, and for comparison of outcomes across states. The case definition forms can be found in the pages to follow, stratified by disorder type. Additionally, you can find case definition classification tables [linked here](#) that can be used as a reference resource.

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*Note: standard surveillance case definitions have not been developed for 3-Hydroxy-3-methylglutaric aciduria (HMG),  $\beta$ -Ketothiolase deficiency ( $\beta$ KT), Mucopolysaccharidosis Type II, and Guanidinoacetate methyltransferase deficiency (GAMT). These are forthcoming.*

# Metabolic Disorders

## Organic Acid Disorders

### Glutaric Acidemia/ Aciduria Type I (GA1)

Enzymatic	
<p><b>Were urine organic acids tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was 3-OH Glutaric acid level</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p>Was Glutaric acid level</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown	
<p><b>Were serum organic acids tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was 3-OH Glutaric acid level</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p>Was Glutaric acid level</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown	
<p><b>Were plasma acylcarnitines tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was C5 -DC level</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Was enzyme analysis for Glutaric Acidemia enzyme activity completed?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was enzyme activity:</p> <input type="checkbox"/> Consistent with disease <input type="checkbox"/> Normal activity (not consistent with disease) <input type="checkbox"/> Unknown
Molecular Genetics	

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



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

*\*Not Biotindase Deficiency*

### Final Diagnosis as determined by clinician performing follow-up:

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

Enzymatic	
<p><b>Were urine organic acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was 3OH Isovaleric acid level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was 3OH Propionic acid level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was 3-methylcrotonyl glycine level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were plasma acylcarnitines tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was C3 level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C5-OH level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>

<p><b>Were infant chemistries (biotinidase) studies completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Were infant chemistries (biotinidase) studies:</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Abnormal</p> <p><input type="checkbox"/> Unknown</p>
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<p><b>Was enzyme analysis for holocarboxylase synthetase deficiency enzyme activity completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme activity:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>
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**Molecular Genetics**

<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> <i>HLCS</i></p> <p><input type="checkbox"/> Other gene: _____</p>
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	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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## 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-methylbutyryl CoA dehydrogenase deficiency
- Unknown

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

	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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### 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	
<p><b>Were urine organic acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was 3OH Isovaleric acid level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was 3-methylcrotonyl glycine level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were plasma acylcarnitines tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was C5 -OH level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was maternal 3-MCC level tested and ruled out?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	
<p><b>Was enzyme analysis for 3-MCC enzyme activity completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme activity:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>
Molecular Genetics	
<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i> What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> <i>MCCC1</i></p> <p><input type="checkbox"/> <i>MCCC2</i></p> <p><input type="checkbox"/> Other gene: _____</p>

	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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## 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	
<b>Were urine organic acids tested?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<i>[IF YES]</i> Please indicate which of the following metabolites were detected:  Propionyl glycine? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown  Tiglylglycine? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown  Methylcitrate? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown  3OH Propionic acid level? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown  MMA? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown  Methylcrotonyl glycine? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown



<p><b>Were plasma acylcarnitines tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was C3 level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
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**Molecular Genetics**

<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> <i>PCCA</i></p> <p><input type="checkbox"/> <i>PCCB</i></p> <p><input type="checkbox"/> Other gene: _____</p>
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	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p> <p><i>Allele 2:</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
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## 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
- Does not differentiate Mutase(-) and Mutase(0)
- Unknown

Enzymatic	
<p><b>Was serum MMA level tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was MMA level in serum:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Was urine MMA level tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was MMA level in urine:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Were plasma acylcarnitines tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was C3 level</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Was maternal vitamin B12 levels tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was maternal vitamin B12 deficient?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
<p><b>Was infant vitamin B12 levels tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was infant vitamin B12 deficient?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
<p><b>Was total plasma homocysteine tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was total plasma homocysteine:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown

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

## 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
- Does not differentiate Cobalamin A deficiency and Cobalamin B deficiency
- Unknown

Enzymatic	
<p><b>Was serum MMA level tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was MMA level in serum:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Was urine MMA level tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was MMA level in urine:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Were plasma acylcarnitines tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was C3 level</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Was maternal vitamin B12 levels tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was maternal vitamin B12 deficient?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
<p><b>Was infant vitamin B12 levels tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was infant vitamin B12 deficient?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
<p><b>Was total plasma homocysteine tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was total plasma homocysteine:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown

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

## 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
- Does not differentiate Cobalamin C deficiency and Cobalamin D deficiency
- Other cobalamin deficiency
- Unknown

Enzymatic	
<p><b>Was serum MMA level tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was MMA level in serum:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Was urine MMA level tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was MMA level in urine:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Were plasma acylcarnitines tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was C3 level</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p><b>Was maternal vitamin B12 levels tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was maternal vitamin B12 deficient?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
<p><b>Was infant vitamin B12 levels tested?</b></p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p><i>[IF YES]</i></p> <p>Was infant vitamin B12 deficient?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown

<p><b>Was total plasma homocysteine tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was total plasma homocysteine:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
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<p><b>Were enzyme complementation studies completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Were complementation studies:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>
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<b>Molecular Genetics</b>
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<p><b>Was mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> <i>MMACHC</i></p> <p><input type="checkbox"/> <i>MMADHC</i></p> <p><input type="checkbox"/> <i>LMBRD1</i></p> <p><input type="checkbox"/> <i>ABCD4</i></p> <p><input type="checkbox"/> <i>HCFC1</i></p> <p><input type="checkbox"/> <i>C2ORF25</i></p> <p><input type="checkbox"/> Other gene: _____</p>
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	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p> <p><i>Allele 2:</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
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## 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	
<p><b>Was urine carnitine tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was fractional excretion of free carnitine level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was 3-methylcrotonyl glycine level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were plasma carnitine levels tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was free carnitine (CO)</p> <p><input type="checkbox"/> Low</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were other causes for carnitine loss ruled out?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	
<p><b>Was enzyme analysis for carnitine deficiency enzyme activity completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme activity:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>
Molecular Genetics	
<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> <i>SCL22A5</i></p> <p><input type="checkbox"/> Other gene: _____</p>



	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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## Medium-chain acyl-CoA Dehydrogenase Deficiency (MCAD)

Enzymatic	
<p><b>Were urine organic acids or acylglycines tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was Hexanoylglycine level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were plasma acylcarnitines tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was C8 level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was repeat C8 level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C8&gt;C10 level:</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p>Was C8&gt;C6 level:</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p>Was C6 level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C10 level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>

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

## Trifunctional Protein Deficiency (TFP)

Enzymatic	
<p><b>Were urine organic acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was C12-OH dicarboxylic acid level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C10-OH dicarboxylic acid level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were plasma acylcarnitines tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was C16-OH level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C16:1-OH level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C18-OH level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C18:1-OH level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was enzyme analysis for TFP enzyme activity completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme activity:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>

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

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

Enzymatic	
<p><b>Were urine organic acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was C12-OH dicarboxylic acid level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C10-OH dicarboxylic acid level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were plasma acylcarnitines tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was C16-OH level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C16:1-OH level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C18-OH level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C18:1-OH level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was enzyme analysis for TFP enzyme activity completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme activity:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>

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

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

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



	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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## 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	
<p><b>Were plasma amino acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was plasma ASA level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was Citrulline level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were plasma urine acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was urine ASA level?</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was urine Citrulline level?</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was enzyme analysis for ASA enzyme activity completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme analysis:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>
Molecular Genetics	

<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> <i>ASL</i></p> <p><input type="checkbox"/> Other gene: _____</p>
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	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p> <p><i>Allele 2:</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
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## 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	
<p><b>Were plasma amino acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was plasma ASA level:</p> <p><input type="checkbox"/> Present</p> <p><input type="checkbox"/> Absent</p> <p><input type="checkbox"/> Unknown</p> <p>Was Citrulline level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was blood ammonia levels tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was blood ammonia level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was enzyme analysis for Citrullinemia type 1 enzyme activity completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme analysis:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>
Molecular Genetics	
<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> <i>ASS1</i></p> <p><input type="checkbox"/> Other gene: _____</p>

	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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## 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	
<p><b>Were plasma amino acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was Phe level:</p> <p><input type="checkbox"/> Elevated (&gt;120umol/L on unrestricted diet)</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was Phe/Tyr level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were biopterin studies done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Were biopterin studies:</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Abnormal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was enzyme analysis for Hyperphe (inclusive of classic PKU) enzyme activity completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme analysis:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>
Molecular Genetics	
<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> PAH</p> <p><input type="checkbox"/> Other gene: _____</p>

	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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## 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	
<p><b>Were plasma amino acids tested?</b></p> <p><input type="checkbox"/> Yes  <input type="checkbox"/> No  <input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was Methionine level:</p> <p><input type="checkbox"/> Elevated  <input type="checkbox"/> Normal  <input type="checkbox"/> Unknown</p>
<p><b>Was plasma Homocysteine tested?</b></p> <p><input type="checkbox"/> Yes  <input type="checkbox"/> No  <input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was plasma Homocysteine level:</p> <p><input type="checkbox"/> Elevated  <input type="checkbox"/> Normal  <input type="checkbox"/> Unknown</p>
<p><b>Was enzyme analysis for CBS enzyme activity completed?</b></p> <p><input type="checkbox"/> Yes  <input type="checkbox"/> No  <input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme analysis:</p> <p><input type="checkbox"/> Consistent with disease  <input type="checkbox"/> Normal activity (not consistent with disease)  <input type="checkbox"/> Unknown</p>
Molecular Genetics	
<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes  <input type="checkbox"/> No  <input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> CBS  <input type="checkbox"/> Other gene: _____</p>



	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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## Maple Syrup Urine Disease (MSUD)

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

- Maple Syrup Urine Disease, Type IA
- Maple Syrup Urine Disease, Type IB
- Maple Syrup Urine Disease, Type II
- Maple Syrup Urine Disease, Type III
- Hydroxyprolinemia
- Does not differentiate
- Unknown

Enzymatic	
<p><b>Were plasma amino acids tested?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>Was Alloisoleucine level:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Elevated</li> <li><input type="checkbox"/> Normal</li> <li><input type="checkbox"/> Unknown</li> </ul> <p>Was Leucine level:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Elevated</li> <li><input type="checkbox"/> Normal</li> <li><input type="checkbox"/> Unknown</li> </ul> <p>Was Isoleucine level:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Elevated</li> <li><input type="checkbox"/> Normal</li> <li><input type="checkbox"/> Unknown</li> </ul> <p>Was Valine level:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Elevated</li> <li><input type="checkbox"/> Normal</li> <li><input type="checkbox"/> Unknown</li> </ul> <p>Was Leu&gt;Val level:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>

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

	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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## 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	
<p><b>Were plasma organic acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was plasma succinylacetone level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was plasma tyrosine level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were urine organic acids tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was urine succinylacetone level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was urine tyrosine level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was enzyme analysis for fumarylacetoacetate hydrolase completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was enzyme analysis:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>
Molecular Genetics	

<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> <i>FAH</i></p> <p><input type="checkbox"/> Other gene: _____</p>
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	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p> <p><i>Allele 2:</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
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# Endocrine Disorders

## Congenital Hypothyroidism (CH)

Final Diagnosis as determined by clinician performing follow-up:

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

Enzymatic	
<p><b>Was Serum TSH tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What was the level:</p> <p><input type="checkbox"/> TSH &gt; 10 mU/L</p> <p><input type="checkbox"/> TSH 6-10 mU/L</p> <p><input type="checkbox"/> TSH &lt;10 mU/L</p> <p><input type="checkbox"/> TSH &lt;6 mU/L</p> <p><input type="checkbox"/> Unknown</p> <p>Was it tested before initiation of treatment?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was Serum Total T4 tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was Serum Total T4 below the age-established reference range?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p>Was it tested before initiation of treatment?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>

<p><b>Was Serum Free T4 tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was Serum Free T4 below the age-established reference range?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p>Was it tested before initiation of treatment?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Does this baby have other pituitary hormone deficiencies?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	
<p><b>Does this baby have midline defects?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	
<p><b>Was TBG tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was TBG below the age established reference range?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was T3 or T4 resin uptake tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was T3 or T4 resin uptake above the age-established reference range?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>



## 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	
<p><b>Societal Sex</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Male</li> <li><input type="checkbox"/> Female</li> <li><input type="checkbox"/> Unknown</li> <li><input type="checkbox"/> Unspecified</li> </ul>	
<p><b>Was confirmatory serum 17-OHP level obtained?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>Was there a value at baseline:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> &gt;10,000 ng/dl</li> <li><input type="checkbox"/> 1000-10,000 ng/dl;</li> <li><input type="checkbox"/> &lt; 1000 ng/dl;</li> <li><input type="checkbox"/> Unknown</li> </ul> <p>Was it tested before initiation of treatment?</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> </ul> <p>Was there a result after ACTH stimulation:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> &gt;10,000 ng/dl</li> <li><input type="checkbox"/> 1000-10,000 ng/dl;</li> <li><input type="checkbox"/> &lt; 1000 ng/dl;</li> <li><input type="checkbox"/> Unknown</li> </ul> <p>Was it tested before initiation of treatment?</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> </ul>
<p><b>Was tandem mass spectrometry urinary steroid profile obtained?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>Were the urinary spectrometry steroid profile results:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Indicative of 21-Hydroxylase Deficiency CAH</li> <li><input type="checkbox"/> Unknown</li> </ul>

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

	<p><i>[For each gene selected]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><i>Allele 2</i></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (Normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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# 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	
<p><b>Was qualitative (IEF or HPLC) testing completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What were the results?</p> <p><input type="checkbox"/> FS</p> <p><input type="checkbox"/> FSC</p> <p><input type="checkbox"/> FSA</p> <p><input type="checkbox"/> FSA<sub>2</sub></p> <p><input type="checkbox"/> FSAA<sub>2</sub></p> <p><input type="checkbox"/> Other; other result name _____</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was quantitative (HPLC or electrophoresis) testing completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What were the results?</p> <p><input type="checkbox"/> FS</p> <p><input type="checkbox"/> FSC</p> <p><input type="checkbox"/> FS with high A<sub>2</sub></p> <p><input type="checkbox"/> FSA with high A<sub>2</sub></p> <p><input type="checkbox"/> FSA</p> <p><input type="checkbox"/> Other; other result name _____</p> <p><input type="checkbox"/> Unknown</p>

<p><b>Was mutation analysis performed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Check the type of variant found on:</p> <p><i>Allele 1</i></p> <p><input type="checkbox"/> S</p> <p><input type="checkbox"/> C</p> <p><input type="checkbox"/> <i>Beta + Thal</i></p> <p><input type="checkbox"/> <i>Beta<sup>0</sup> + Thal</i></p> <p><input type="checkbox"/> Other; _____</p> <p><input type="checkbox"/> Unknown</p> <p><i>Allele 2</i></p> <p><input type="checkbox"/> S</p> <p><input type="checkbox"/> C</p> <p><input type="checkbox"/> <i>Beta + Thal</i></p> <p><input type="checkbox"/> <i>Beta<sup>0</sup> + Thal</i></p> <p><input type="checkbox"/> Other; other name _____</p> <p><input type="checkbox"/> Unknown</p>
<p><b>NBS result</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What were the results?</p> <p><input type="checkbox"/> FS</p> <p><input type="checkbox"/> FSC</p> <p><input type="checkbox"/> FSA</p> <p><input type="checkbox"/> FSA<sub>2</sub></p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was a CBC performed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What were the results?</p> <p><input type="checkbox"/> Normal – high MCV</p> <p><input type="checkbox"/> Low MCV</p> <p><input type="checkbox"/> Unknown</p>

<p><b>Were family studies (in parents) done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Maternal Status: what were the results?</p> <p><input type="checkbox"/> Carrier S</p> <p><input type="checkbox"/> Carrier C</p> <p><input type="checkbox"/> Carrier <i>Beta + Thal</i></p> <p><input type="checkbox"/> Carrier <i>Beta<sup>0</sup> Thal</i></p> <p><input type="checkbox"/> Other: _____</p> <p><input type="checkbox"/> Unknown</p> <p>Paternal Status: what were the results?</p> <p><input type="checkbox"/> Carrier S</p> <p><input type="checkbox"/> Carrier C</p> <p><input type="checkbox"/> Carrier <i>Beta + Thal</i></p> <p><input type="checkbox"/> Carrier <i>Beta<sup>0</sup> Thal</i></p> <p><input type="checkbox"/> Other: _____</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was there a positive family history?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	
<p><b>Were HPLC &amp; IEF tested on the same sample from the infant?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What were the results?</p> <p><input type="checkbox"/> FS</p> <p><input type="checkbox"/> FSC</p> <p><input type="checkbox"/> FSA<sub>2</sub></p> <p><input type="checkbox"/> FSAA<sub>2</sub></p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Were Hgb tests (electrophoresis or HPLC) performed on family members?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What were the results?</p> <p><input type="checkbox"/> Positive</p> <p><input type="checkbox"/> Negative</p> <p><input type="checkbox"/> Unknown</p>

## Presence of Other Hb Variant

*\*This is a Secondary RUSP Condition*

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

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

Diagnostic Workup	
<b>Alpha thalassemia present?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	
<b>Was qualitative (IEF or HPLC) testing completed?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<i>[IF YES]</i> What were the results? <input type="checkbox"/> FC <input type="checkbox"/> FD <input type="checkbox"/> FE <input type="checkbox"/> FO <sub>ARAB</sub> <input type="checkbox"/> Other; other result name _____ <input type="checkbox"/> Unknown
<b>Was quantitative (HPLC or electrophoresis) testing completed?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<i>[IF YES]</i> What were the results? <input type="checkbox"/> FC <input type="checkbox"/> FD <input type="checkbox"/> FE <input type="checkbox"/> FO <sub>ARAB</sub> <input type="checkbox"/> Other; other result name _____ <input type="checkbox"/> Unknown

<p><b>Was mutation analysis performed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Check the type of variant found on allele 1:</p> <p><input type="checkbox"/> C</p> <p><input type="checkbox"/> D</p> <p><input type="checkbox"/> E</p> <p><input type="checkbox"/> O<sub>ARAB</sub></p> <p><input type="checkbox"/> Other; other name _____</p> <p><input type="checkbox"/> Unknown</p> <p>Check the type of variant found on allele 2:</p> <p><input type="checkbox"/> C</p> <p><input type="checkbox"/> D</p> <p><input type="checkbox"/> E</p> <p><input type="checkbox"/> O<sub>ARAB</sub></p> <p><input type="checkbox"/> Beta + Thal</p> <p><input type="checkbox"/> Beta<sup>0</sup> + Thal</p> <p><input type="checkbox"/> Other; other name _____</p> <p><input type="checkbox"/> Unknown</p>
<p><b>NBS result</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What were the results?</p> <p><input type="checkbox"/> FC</p> <p><input type="checkbox"/> FD</p> <p><input type="checkbox"/> FE</p> <p><input type="checkbox"/> FO<sub>ARAB</sub></p> <p><input type="checkbox"/> Other; other result name _____</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was a CBC performed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What were the results?</p> <p><input type="checkbox"/> Normal – high MCV</p> <p><input type="checkbox"/> Low MCV</p>



<p><b>Were family studies (in parents) done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><b>Maternal Status: what were the results?</b></p> <p><input type="checkbox"/> Carrier <i>C</i></p> <p><input type="checkbox"/> Carrier <i>D</i></p> <p><input type="checkbox"/> Carrier <i>E</i></p> <p><input type="checkbox"/> Carrier <i>O<sub>Arab</sub></i></p> <p><input type="checkbox"/> Carrier <i>Beta + Thal</i></p> <p><input type="checkbox"/> Carrier <i>Beta<sup>0</sup> Thal</i></p> <p><input type="checkbox"/> Other: _____</p> <p><input type="checkbox"/> Unknown</p> <p><b>Paternal Status: what were the results?</b></p> <p><input type="checkbox"/> Carrier <i>C</i></p> <p><input type="checkbox"/> Carrier <i>D</i></p> <p><input type="checkbox"/> Carrier <i>E</i></p> <p><input type="checkbox"/> Carrier <i>O<sub>Arab</sub></i></p> <p><input type="checkbox"/> Carrier <i>Beta + Thal</i></p> <p><input type="checkbox"/> Carrier <i>Beta<sup>0</sup> Thal</i></p> <p><input type="checkbox"/> Other: _____</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was there a positive family history?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	
<p><b>Were Hgb tests (electrophoresis or HPLC) performed on family members?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p><b>What were the results?</b></p> <p><input type="checkbox"/> Positive</p> <p><input type="checkbox"/> Negative</p> <p><input type="checkbox"/> Unknown</p>

# 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	
<p><b>Was enzyme activity tested?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>What was the enzyme level?</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Within lab known affected range</li> <li><input type="checkbox"/> Normal</li> <li><input type="checkbox"/> Unknown</li> </ul>
<p><b>Were urine GAGS tested?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>What was the urine GAG level?</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Elevated</li> <li><input type="checkbox"/> Normal</li> <li><input type="checkbox"/> Unknown</li> </ul>

<p><b>Clinical symptoms/lab findings?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Symptoms present and documented by specialists. Public health (PH) program continued to collect data through the development of symptoms</li> <li><input type="checkbox"/> 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)</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>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</i></p>
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Molecular Genetics	
<p><b>Were variants detected in genes known to be associated with MPS I?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Pathogenic variant and associated with SEVERE disease</li> <li><input type="checkbox"/> Pathogenic or likely pathogenic variant</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant known to be associated with ATTENUATED disease.</li> <li><input type="checkbox"/> Wild Type (Normal)</li> <li><input type="checkbox"/> Unknown</li> </ul> <p><i>Allele 2:</i></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Pathogenic variant and associated with SEVERE disease</li> <li><input type="checkbox"/> Pathogenic or likely pathogenic variant</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant known to be associated with ATTENUATED disease.</li> <li><input type="checkbox"/> Wild Type (Normal)</li> <li><input type="checkbox"/> Unknown</li> </ul>

## 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	
<p><b>Was enzyme activity tested in blood (not DBS sample)?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>What was the enzyme level?</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Within lab known affected range for infantile onset (IO)</li> <li><input type="checkbox"/> Low (above affected range, for IO, may or may not be in late-onset (LO range), but should not be above LO range))</li> <li><input type="checkbox"/> Within lab known affected range for late onset (LO)</li> <li><input type="checkbox"/> Low (above affected range, for LO not normal)</li> <li><input type="checkbox"/> Unknown</li> </ul>
<p><b>Was enzyme activity tested in skin/muscle?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>What was the enzyme activity?</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Positive skin or muscle biopsy</li> <li><input type="checkbox"/> Unknown</li> </ul>
<p><b>Was there cardiac involvement consistent with Pompe?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>Findings:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Positive findings on chest X-ray/EKG/ECHO in newborn period</li> <li><input type="checkbox"/> Positive findings on chest X-ray/EKG/ECHO</li> </ul>
<p><b>Lab findings for CK/AST/ALT/LDH/Urine Hex4?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Elevated</li> <li><input type="checkbox"/> Not Present</li> <li><input type="checkbox"/> Unknown</li> <li><input type="checkbox"/> Untested</li> </ul>	

<p><b>Were there any clinical findings?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Symptoms present after one year of age and documented by specialists. PH program continue to collect data through the development of symptoms</li> <li><input type="checkbox"/> Symptoms present before one year of age, but no cardiac involvement</li> <li><input type="checkbox"/> Unknown or not reported to PH by the end of the follow-up period</li> </ul>	<p><i>Clinical symptoms consistent with Pompe Disease: progressive muscle weakness, need for respiratory assistance, swaying gait or waddle, Lordosis, kyphosis, or scoliosis</i></p>
<b>Molecular Genetics</b>	

<p><b>Were variants detected in genes known to be associated with Pompe Disease?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <p><input type="checkbox"/> Pathogenic</p> <p><input type="checkbox"/> Pathogenic variant and associated with infantile onset</p> <p><input type="checkbox"/> Novel variant that is likely pathogenic</p> <p><input type="checkbox"/> Pathogenic variant or likely pathogenic variant, with deletion or duplication consistent with infantile onset</p> <p><input type="checkbox"/> Pathogenic and associated with non-classical disease, or variant of uncertain significance</p> <p><input type="checkbox"/> Pathogenic or likely pathogenic variant, no other variants found; duplication/deletion testing not done or not known</p> <p><input type="checkbox"/> Pathogenic or likely pathogenic variant; no other variants found</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p> <p><i>Allele 2:</i></p> <p><input type="checkbox"/> Pathogenic</p> <p><input type="checkbox"/> Pathogenic variant and associated with infantile onset</p> <p><input type="checkbox"/> Novel variant that is likely pathogenic</p> <p><input type="checkbox"/> Pathogenic variant or likely pathogenic variant, with deletion or duplication consistent with infantile onset</p> <p><input type="checkbox"/> Pathogenic and associated with non-classical disease, or variant of uncertain significance</p> <p><input type="checkbox"/> Pathogenic or likely pathogenic variant, no other variants found; duplication/deletion testing not done or not known</p> <p><input type="checkbox"/> Pathogenic or likely pathogenic variant; no other variants found</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
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# 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	
<p><b>Was enzyme analysis for biotinidase enzyme activity completed?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>Was enzyme activity:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> &lt;10%</li> <li><input type="checkbox"/> 10-30%</li> <li><input type="checkbox"/> Normal</li> <li><input type="checkbox"/> Unknown</li> </ul>
Molecular Genetics	
<p><b>Was a mutation analysis performed for biotinidase deficiency?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i> What genes were included in the mutation analysis? (select all that apply)</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> BTD</li> <li><input type="checkbox"/> Other gene: _____</li> </ul>
	<p><i>[For all genes selected]</i></p> <p><b>Check the types of variants found on:</b></p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing (Unknown)</li> <li><input type="checkbox"/> Variant known to be disease causing (known to be associated with profound enzyme deficiency)</li> <li><input type="checkbox"/> Variant known to be disease causing (known to be associated with partial enzyme deficiency [“mild” mutation (D44H)])</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Wild Type (Normal)</li> <li><input type="checkbox"/> Unknown</li> </ul> <p><i>Allele 2</i></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing (Unknown)</li> <li><input type="checkbox"/> Variant known to be disease causing (known to be associated with profound enzyme deficiency)</li> <li><input type="checkbox"/> Variant known to be disease causing (known to be associated with partial enzyme deficiency [“mild” mutation (D44H)])</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Wild Type (Normal)</li> <li><input type="checkbox"/> Unknown</li> </ul>

## Galactosemia (GALT)

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

- Classic Galactosemia
- Duarte variant galactosemia
- Unknown

Enzymatic	
<p><b>Were GALT levels tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was GALT level:</p> <p><input type="checkbox"/> &lt;10%</p> <p><input type="checkbox"/> 10-30%</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was Gal-1-P tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was Gal-1-P level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>Was Urine Galactitol tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Was Urine Galactitol level:</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p><b>If Variant Galactosemia, was protein phenotyping completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p><input type="checkbox"/> Not applicable</p>	<p><i>[IF YES]</i></p> <p>Did result indicate:</p> <p><input type="checkbox"/> Phenotype consistent with variant</p> <p><input type="checkbox"/> Phenotype NOT consistent with variant</p> <p><input type="checkbox"/> Unknown</p>
<p><b>If Arginase Deficiency, were enzyme studies completed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p><input type="checkbox"/> Not applicable</p>	<p><i>[IF YES]</i></p> <p>Was enzyme activity:</p> <p><input type="checkbox"/> Consistent with disease</p> <p><input type="checkbox"/> Normal activity (not consistent with disease)</p> <p><input type="checkbox"/> Unknown</p>
Molecular Genetics	



<p><b>Was a mutation analysis done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What genes were included in the mutation analysis? (select all that apply)</p> <p><input type="checkbox"/> Galactosemia</p> <p><input type="checkbox"/> Other gene: _____</p>
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	<p><i>[For each gene selected]</i></p> <p><b>Check the types of variants found on:</b></p> <p><i>Allele 1</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p> <p><i>Allele 2</i></p> <p><input type="checkbox"/> Variant known to be disease causing</p> <p><input type="checkbox"/> Variant of unknown significance</p> <p><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
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## 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	
<p><b>Did the NBS result indicate an elevated IRT?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	
<p><b>Were CFTR mutations detected on the <u>newborn screening</u> mutation panel?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p><i>Mutations seen in patients with CF have been classified as disease-causing, neutral, or varying clinical consequences through the CFTR2 project: <a href="http://cftr2.org/browse.php">http://cftr2.org/browse.php</a>. Additional information about the mutation and the association with lower sweat chlorides can also be found at CFTR2.</i></p>	<p><i>[IF YES]</i></p> <p><i>Check the type of variant found on allele 1:</i></p> <p><input type="checkbox"/> Variant known to be disease causing in CFTR2</p> <p><input type="checkbox"/> Variant known to be disease causing in CFTR2 (shown to be associated with lower sweat chlorides)</p> <p><input type="checkbox"/> Neutral variant</p> <p><input type="checkbox"/> Variant of varying clinical consequence in CFTR2</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown (not reported in CFTR2)</p> <p><i>Check the type of variant found on allele 1:</i></p> <p><input type="checkbox"/> Variant known to be disease causing in CFTR2</p> <p><input type="checkbox"/> Variant known to be disease causing in CFTR2 (shown to be associated with lower sweat chlorides)</p> <p><input type="checkbox"/> Neutral variant</p> <p><input type="checkbox"/> Variant of varying clinical consequence in CFTR2</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown (not reported in CFTR2)</p>
<p><b>Did the child have meconium ileus?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	

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

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

Summary of common variants as reported on CFTR2 (this is not an exhaustive list; please visit [www.CFTR2.org](http://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	<b>1078delT</b>	No	CF-causing	NO
c.579+3A>G	p.Phe342HisfsX28	<b>1154insTC</b>	No	CF-causing	NO
c.3454G>C	No protein name	<b>1717-1G-&gt;A</b>	Yes	CF-causing	NO
c.3208C>T	No protein name	<b>1811+1.6kbA-&gt;G</b>	No	CF-causing	NO
c.3154T>G	No protein name	<b>1898+1G-&gt;A</b>	Yes	CF-causing	NO
c.1585-1G>A	p.Leu671X	<b>2143delT</b>	No	CF-causing	NO
c.1680-1G>A	p.Lys684SerfsX38	<b>2183AA-&gt;G</b>	No	CF-causing	NO
c.1766+1G>A	p.Lys684AsnfsX38	<b>2184delA</b>	Yes	CF-causing	NO
c.2490+1G>A	p.Gln685ThrfsX4	<b>2184insA</b>	No	CF-causing	NO
c.2988+1G>A	p.Glu726ArgfsX4	<b>2307insA</b>	No	CF-causing	NO
c.1736A>G	No protein name	<b>2789+5G-&gt;A</b>	Yes	CF-causing	NO
c.1408A>G	No protein name	<b>3120+1G-&gt;A</b>	Yes	CF-causing	NO
c.1841A>G	No protein name	<b>3120G-&gt;A</b>	No	CF-causing	NO
c.2991G>C	No protein name	<b>3272-26A-&gt;G</b>	No	CF-causing	NO
c.489+1G>T	p.Lys1177SerfsX15	<b>3659delC</b>	Yes	CF-causing	NO
c.350G>A	No protein name	<b>3849+10kbC-&gt;T</b>	Yes	CF-causing	NO
c.4242+1G>T	p.Leu1258PhefsX7	<b>3905insT</b>	No	CF-causing	NO
c.3718-1G>A	p.Leu881lefsX22	<b>394delTT</b>	No	CF-causing	NO
c.1240C>T	No protein name	<b>5T</b>	No	Indeterminate	YES
c.2260G>A	No protein name	<b>621+1G-&gt;T</b>	Yes	CF-causing	NO
c.1727G>C	No protein name	<b>711+1G-&gt;T</b>	Yes	CF-causing	NO
c.220C>T	No protein name	<b>711+5G-&gt;A</b>	No	CF-causing	NO
c.2834C>T	p.Ala455Glu	<b>A455E</b>	Yes	CF-causing	NO
c.1675G>A	p.Ala559Thr	<b>A559T</b>	No	CF-causing	NO
c.1127_1128insA	p.Ser18ArgfsX16	<b>CFTRdele2,3</b>	No	CF-causing	NO
c.1202G>A or c.1203G>A	p.Asp1152His	<b>D1152H</b>	No	Indeterminate	YES
c.1923_1931del9insA	p.Glu60X	<b>E60X</b>	No	CF-causing	NO
c.1679G>C	p.Phe508del	<b>F508del</b>	Yes	CF-causing	NO
c.3160C>G	p.Gly1244Glu	<b>G1244E</b>	No	CF-causing	NO
c.4046G>A	p.Gly178Glu	<b>G178R</b>	No	CF-causing	NO
c.4196_4197delTC	p.Gly542X	<b>G542X</b>	Yes	CF-causing	NO
c.3731G>A	p.Gly551Asp	<b>G551D</b>	Yes	CF-causing	NO
c.3197G>A	p.Gly85Glu	<b>G85E</b>	Yes	CF-causing	NO
c.2657+2_2657+3insA	p.Ile1027Thr	<b>I1027T</b>	No	Not CF-causing	NO
c.1673T>C	p.Ile148Thr	<b>I148T</b>	No	Not CF-causing	NO

c.3763T>C	p.Ile336Lys	<b>I336K</b>	No	CF-causing	NO
c.1558G>T	p.Ile507del	<b>I507del</b>	Yes	CF-causing	NO
c.3230T>C	p.Leu1077Pro	<b>L1077P</b>	No	CF-causing	NO
c.1040G>A	p.Leu206Trp	<b>L206W</b>	No	CF-causing	NO
c.3302T>A	p.Met1101Lys	<b>M1101K</b>	No	CF-causing	NO
c.274G>A	p.Asn1303Lys	<b>N1303K</b>	Yes	CF-causing	NO
c.617T>G	p.Pro67Leu	<b>P67L</b>	No	CF-causing	NO
c.2764_2765insAG	p.Gln220X	<b>Q220X</b>	No	CF-causing	NO
c.1973_1985del13insAGAA A	p.Gln493X	<b>Q493X</b>	No	CF-causing	NO
c.3196C>T	p.Arg1066Cys	<b>R1066C</b>	No	CF-causing	NO
c.4296_4297insGA	p.Arg1158X	<b>R1158X</b>	No	CF-causing	NO
c.1692delA	p.Arg1162X	<b>R1162X</b>	Yes	CF-causing	NO
c.1055G>A	p.Arg117Cys	<b>R117C</b>	No	CF-causing	NO
c.1466C>A	p.Arg117His	<b>R117H</b>	Yes	Indeterminate	YES
c.1013C>T	p.Arg334Trp	<b>R334W</b>	Yes	CF-causing	NO
c.532G>A	p.Arg347His	<b>R347H</b>	Yes	CF-causing	NO
c.1040G>C	p.Arg347Pro	<b>R347P</b>	No	CF-causing	NO
c.2908G>C	p.Arg352Gln	<b>R352Q</b>	No	CF-causing	NO
c.2424_2425insAT	p.Arg553X	<b>R553X</b>	Yes	CF-causing	NO
c.2780T>C	p.Arg560Thr	<b>R560T</b>	Yes	CF-causing	NO
c.349C>T	p.Ser1251Asn	<b>S1251N</b>	No	CF-causing	NO
c.1000C>T	p.Ser549Asn	<b>S549N</b>	No	CF-causing	NO
c.3752G>A	p.Ser945Leu	<b>S945L</b>	No	CF-causing	NO
c.1645A>C or c.1647T>G	p.Val520Phe	<b>V520F</b>	No	CF-causing	NO
c.274G>T	p.Trp1282X	<b>W1282X</b>	Yes	CF-causing	NO
c.2128A>T	p.Tyr1092X	<b>Y1092X</b>	No	CF-causing	NO
c.2195T>G	p.Tyr122X	<b>Y122X</b>	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	
<p><b>Was the CD3 T cell level tested?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i> What was the CD3 T cell level?</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> &lt;300 autologous T cells, undetectable or very few naïve T cells</li> <li><input type="checkbox"/> 300-1500, few naïve T cells, oligoclonal T cells, or poor T cell diversity</li> <li><input type="checkbox"/> &gt;80% CD45RO+</li> <li><input type="checkbox"/> Any number (not zero)</li> <li><input type="checkbox"/> Untested/Unknown</li> </ul>
<p><b>Was proliferation to PHA test done?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i> Proliferation to PHA:</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> &lt;10% of normal</li> <li><input type="checkbox"/> 10-50% of normal PHA</li> <li><input type="checkbox"/> 10-30% normal PHA or Absent to Candida/TT</li> <li><input type="checkbox"/> &lt;30% of normal</li> <li><input type="checkbox"/> Any/Unknown</li> </ul>
<p><b>Was maternal engraftment documented?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	
Molecular Genetics	

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



	<p><i>[IF variants detected=YES]</i></p> <p><b>Were heterozygous <i>TBX1</i> variants assessed?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>
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## Critical Congenital Heart Disease (CCHD)

What was the final diagnosis?

- CCHD
- Non-critical CCHD
- Other
- Unknown

Diagnostic Workup	
	<p><i>[IF CCHD SELECTED]</i></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Truncus Arteriosus</li> <li><input type="checkbox"/> Total Anomalous Pulmonary Venous Connection</li> <li><input type="checkbox"/> Tetralogy of fallot</li> <li><input type="checkbox"/> Pulmonary Atresia</li> <li><input type="checkbox"/> Ebstein’s Anomaly</li> <li><input type="checkbox"/> Hypoplastic Left Heart Syndrome</li> <li><input type="checkbox"/> Single ventricle</li> <li><input type="checkbox"/> Tricuspid atresia</li> <li><input type="checkbox"/> Transposition of the great arteries</li> <li><input type="checkbox"/> Double outlet right ventricle</li> <li><input type="checkbox"/> Coarctation of aorta</li> <li><input type="checkbox"/> Interrupted arch</li> <li><input type="checkbox"/> Aortic valve disease</li> </ul> <p>If Other selected; please specify _____</p>

Please answer the following:	If Yes, what were the results of the postnatal echocardiogram? <i>(select all that apply)</i>
<p><b>Was a Postnatal Echocardiogram Completed?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><b>Truncus Arteriosus</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Truncus arteriosus</li> <li><input type="checkbox"/> Truncus arteriosus + Interrupted aortic arch</li> </ul> <hr/> <p><b>Total Anomalous Pulmonary Venous Connection (TAPVC)</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Type1 (supracardiac)</li> <li><input type="checkbox"/> Type 2 (cardiac)</li> <li><input type="checkbox"/> Type 3 (infracardiac)</li> <li><input type="checkbox"/> Type 4 (mixed)</li> </ul>

<p><b>Tetralogy of Fallot (TOF)</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> TOF</li> <li><input type="checkbox"/> TOF, Pulmonary stenosis</li> <li><input type="checkbox"/> TOF, AVCanal (AVSD)</li> <li><input type="checkbox"/> TOF, Absent pulmonary valve</li> </ul>
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<p><b>Pulmonary Artesia</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Pulmonary atresia</li> <li><input type="checkbox"/> Pulmonary atresia, IVS</li> <li><input type="checkbox"/> Pulmonary atresia, VSD (Including TOF, PA)</li> <li><input type="checkbox"/> Pulmonary atresia, VSD-MAPCA</li> </ul>
<p><b>Ebstein’s Anomaly</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Ebstein's anomaly</li> </ul>
<p>Hypoplastic Left Heart Syndrome (HLHS)</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Hypoplastic left heart syndrome</li> </ul>
<p><b>Single Ventricle</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Single ventricle, DILV</li> <li><input type="checkbox"/> Single ventricle, DIRV</li> <li><input type="checkbox"/> Single ventricle, Mitral atresia</li> <li><input type="checkbox"/> Single ventricle, Unbalanced AV canal</li> <li><input type="checkbox"/> Single ventricle, Heterotaxia syndrome</li> <li><input type="checkbox"/> Single ventricle, Other</li> <li><input type="checkbox"/> Single ventricle + Total anomalous pulmonary venous connection (TAPVC)</li> </ul>
<p><b>Tricuspid Artesia</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Single ventricle, Tricuspid atresia</li> </ul>
<p><b>Transposition of the Great Arteries (TGA)</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> d-TGA, IVS</li> <li><input type="checkbox"/> d-TGA, IVS-LVOTO</li> <li><input type="checkbox"/> d-TGA, VSD</li> <li><input type="checkbox"/> d-TGA, VSD-LVOTO</li> </ul>
<p><b>Double Outlet Right Ventricle (DORV)</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> DORV, VSD type</li> <li><input type="checkbox"/> DORV, TOF type</li> <li><input type="checkbox"/> DORV, TGA type</li> <li><input type="checkbox"/> DORV, Remote VSD (uncommitted VSD)</li> <li><input type="checkbox"/> DORV + AVSD (AV Canal)</li> <li><input type="checkbox"/> DORV, IVS</li> <li><input type="checkbox"/> DORV, Remote VSD (uncommitted VSD)</li> </ul>
<p><b>Coarctation of Aorta</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Coarctation of aorta</li> <li><input type="checkbox"/> Aortic arch hypoplasia</li> <li><input type="checkbox"/> VSD + Aortic arch hypoplasia</li> <li><input type="checkbox"/> VSD + Coarctation of aorta</li> </ul>

	<p><b>Interrupted Arch</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Interrupted aortic arch</li> <li><input type="checkbox"/> Interrupted aortic arch + VSD</li> <li><input type="checkbox"/> Interrupted aortic arch + AP window (aortopulmonary window)</li> </ul>
	<p><b>Aortic Valve Disease</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Aortic Stenosis receiving intervention in first 30 days of life</li> <li><input type="checkbox"/> Pulmonary Stenosis receiving intervention in the first 30 days of life</li> </ul>
<p><b>Was a Prenatal Echocardiogram Completed?</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>	<p><i>[IF YES]</i></p> <p>Did the Prenatal Echo findings suggest CCHD?</p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> Unknown</li> </ul>

## 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 (CADD5)
- 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	
<p><b>Was plasma VLCFA tested?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>What was the VLCFA level?</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Slightly elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Low</p> <p><input type="checkbox"/> Unknown</p> <p><i>“Elevated” signifies in pathogenic range, while “slightly elevated” signifies above normal, but not in the pathogenic range</i></p>
<p><b>Clinical symptoms?</b></p> <p><input type="checkbox"/> Present</p> <p><input type="checkbox"/> Not present</p> <p><input type="checkbox"/> Not present at birth</p> <p><input type="checkbox"/> Unknown</p>	<p><i>Symptoms may include: neonatal hypotonia, neonatal seizures, liver disease, neonatal cholestasis, sensorineural deafness, failure to thrive, craniofacial abnormalities</i></p>
<p><b>Was plasmalogen testing done?</b></p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p><i>[IF YES]</i></p> <p>Plasmalogen level?</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Low</p> <p><input type="checkbox"/> Unknown</p>

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

	<p><b>[IF PEX1] Check the type of variations found on:</b></p> <p><b>Allele 1</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li> <li><input type="checkbox"/> Wild Type (normal)</li> <li><input type="checkbox"/> Unknown</li> </ul> <p><b>Allele 2</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li> <li><input type="checkbox"/> Wild Type (normal)</li> <li><input type="checkbox"/> Unknown</li> </ul>
	<p><b>[IF ACOX1] Check the type of variations found on:</b></p> <p><b>Allele 1</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li> <li><input type="checkbox"/> Wild Type (normal)</li> <li><input type="checkbox"/> Unknown</li> </ul> <p><b>Allele 2</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li> <li><input type="checkbox"/> Wild Type (normal)</li> <li><input type="checkbox"/> Unknown</li> </ul>

	<p><i>[IF HSD17b4]</i><b>Check the type of variations found on:</b></p> <p><b>Allele 1</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li> <li><input type="checkbox"/> Wild Type (normal)</li> <li><input type="checkbox"/> Unknown</li> </ul> <p><b>Allele 2</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li> <li><input type="checkbox"/> Wild Type (normal)</li> <li><input type="checkbox"/> Unknown</li> </ul>
	<p><i>[IF 1 of the 7 known genes for Aicardi-Goutières Syndrome]</i> <b>Check the type of variations found on:</b></p> <p><b>Allele 1</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li> <li><input type="checkbox"/> Wild Type (normal)</li> <li><input type="checkbox"/> Unknown</li> </ul> <p><b>Allele 2</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Variant known to be disease causing</li> <li><input type="checkbox"/> Variant of unknown significance</li> <li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li> <li><input type="checkbox"/> Wild Type (normal)</li> <li><input type="checkbox"/> Unknown</li> </ul>



	<p><i>[IF Other Gene Selected]</i></p> <p><b>Other Gene Name;</b> _____</p> <p><b>Check the type of variations found on:</b></p> <p><b>Allele 1</b></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (normal)</li><li><input type="checkbox"/> Unknown</li></ul> <p><b>Allele 2</b></p> <ul style="list-style-type: none"><li><input type="checkbox"/> Variant known to be disease causing</li><li><input type="checkbox"/> Variant of unknown significance</li><li><input type="checkbox"/> Variant of unknown significance (predicted to be pathogenic)</li><li><input type="checkbox"/> Wild Type (normal)</li><li><input type="checkbox"/> Unknown</li></ul>
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## Spinal Muscular Atrophy (SMA)

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

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