



NewSTEPS

A Program of the Association of Public Health Laboratories™

Case Worksheets for Newborn Screening

Case Information Worksheet

Question:	Answer
INFANTDEMOGRAPHICS	
State Unique ID?(alphanumeric)	
Date of Birth?(mm/dd/yyyy)	
Gestational Age?(in weeks)	
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/Unknown
Ethnicity? (select one)	<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/Unknown
SCREENINGINFORMATION	
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"/> Don't Know
Which newborn screen result indicated this infant was at risk for the disorder?	<input type="checkbox"/> Initial Screen <input type="checkbox"/> 2nd Required Screen <input type="checkbox"/> Subsequent Screen
Was this individual diagnosed later in life (not identified by newborn screening)?	<input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
Date of <u>initial</u> specimen collection (mm/dd/yyyy)?	
Date of receipt by lab of <u>initial</u> specimen (mm/dd/yyyy)?	
Date of release of out of range results of <u>initial</u> specimen (mm/dd/yyyy)?	
Date of <u>subsequent</u> specimen collection (mm/dd/yyyy)?	
Date of receipt by lab of <u>subsequent</u> specimen (mm/dd/yyyy)?	
Date of release of out of range results of <u>subsequent</u> specimen (mm/dd/yyyy)?	
Date of intervention by appropriate medical provider (mm/dd/yyyy)?	
Date of confirmation of diagnosis (mm/dd/yyyy)?	

Newborn Screening Surveillance Case Definitions

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) or for β -Ketothiolase deficiency (BKT). These are forthcoming.

Metabolic Disorders

Organic Acid Disorders:

Glutaric Acidemia/ Aciduria Type I (GA1) Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were urine organic acids tested?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<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>Were serum organic acids tested?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<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>Were plasma acylcarnitines tested?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Was C5 -DC level</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p>Was a mutation analysis performed for Glutaric aciduria type I?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>What genes were included in the mutation analysis?</p> <input type="checkbox"/> GCDH gene <input type="checkbox"/> Other gene: _____

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<p>Were variants detected in the GCDH gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants detected in other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Was enzyme analysis for Glutaric Acidemia enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

**Isovaleric Acidemia/ Aciduria (IVA)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were urine organic acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>Were plasma acylcarnitines tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Was C5 level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p>Was a mutation analysis performed for Isovaleric aciduria?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>What genes were included in the mutation analysis?</p> <p><input type="checkbox"/> IVD gene</p> <p><input type="checkbox"/> Other gene: _____</p>

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<p>Were variants detected in the IVD gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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> ○ 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> ○ Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants detected in other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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> ○ 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> ○ Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Was enzyme analysis for isovaleric acidemia enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

**3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
Were urine organic acids tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was 3OH Isovaleric acid level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown Was 3-methylcrotonyl glycine level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Were plasma acylcarnitines tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was C5-OH level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was maternal 3-MCC level tested and ruled out? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	
Was a mutation analysis performed for 3-MCC? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	What genes were included in the mutation analysis? <input type="checkbox"/> MCCC1 gene <input type="checkbox"/> MCCC2 gene <input type="checkbox"/> Other gene: _____

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<p>Were variants detected in the MCCC1 gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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"/> 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"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants detected in the MCCC2 gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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"/> 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"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants detected in other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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"/> 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"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Was enzyme analysis for 3-MCC enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

MMA With Homocystinuria; (CbIC; CbID; CbIF; CbIDv1; CbIJ)
Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please Select One:

- A. Cobalamin C deficiency (CbIC)
- B. Cobalamin D deficiency (CbID)
- C. Cobalamin F deficiency (CbIF)
- D. Cobalamin Dv1 deficiency (CbIDv1)
- E. Cobalamin J deficiency (CbIJ)
- F. Other cobalamin deficiency not listed above: _____

Please answer the following as Yes/No/Don't Know	If Yes
Was serum MMA level tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was MMA level in serum: <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was urine MMA level tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was MMA level in urine: <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Were plasma acylcarnitines tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was C3: <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was maternal vitamin B12 levels tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was maternal vitamin B12 deficient? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown

<p>Were infant vitamin B12 levels tested?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Was infant vitamin B12 deficient?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
<p>Was total plasma homocysteine tested?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Was total plasma homocysteine:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p>Was mutation analysis done?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>What genes were included in the mutation analysis?</p> <input type="checkbox"/> C2ORF25 gene (cbID) <input type="checkbox"/> MMACHC gene <input type="checkbox"/> LMBRD1 gene (cbIF) <input type="checkbox"/> ABCD4 gene (cbIJ) <input type="checkbox"/> Other MMA associated gene: _____

MOLECULAR GENETICS REPORT

<p>Were variants found in C2ORF25 gene?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance ○ Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Were variants found in MMACHC gene?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance ○ Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
	<p><i>Allele 2:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance ○ Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown

<p>Were variants found in LMBRD1 gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants found in ABCD4 gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants found in other MMA related genes? _____</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were enzyme complementation studies completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

**MMA Without Homocystinuria; (CblA; CblB; mut-; mut0; CblDv2)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please Select One:

- G. Cobalamin A deficiency (CblA)
- H. Cobalamin B deficiency (CblB)
- I. Mutase (-) (mut-0)
- J. Mutase (0) (mut0)
- K. Cobalamin Dv2 (CblDv2)

Please answer the following as Yes/No/Don't Know	If Yes
Was serum MMA level tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was MMA level in serum: <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was urine MMA level tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was MMA level in urine: <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Were plasma acylcarnitines tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was C3: <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was maternal vitamin B12 levels tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was maternal vitamin B12 deficient? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown

<p>Were infant vitamin B12 levels tested?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Was infant vitamin B12 deficient?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
<p>Was total plasma homocysteine tested?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Was total plasma homocysteine:</p> <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
<p>Was mutation analysis done?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>What genes were included in the mutation analysis?</p> <input type="checkbox"/> METHYLMALONYL-CoA MUTASE <input type="checkbox"/> MMAA gene <input type="checkbox"/> MMAB gene <input type="checkbox"/> Other MMA associated gene:_____

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<p>Were variants found in the METHYLMALONYL-CoA MUTASE gene?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance ○ Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
	<p><i>Allele 2:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance ○ Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Were variants found in MMAA gene?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance ○ Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
	<p><i>Allele 2:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance ○ Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown

<p>Were variants found in MMAB gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Were variants found in other MMA related genes?_____</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Were enzyme complementation studies completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Were complementation studies:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Consistent with disease <input type="checkbox"/> Normal activity (not consistent with disease) <input type="checkbox"/> Unknown

**Propionic Acidemia/ Aciduria
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were urine organic acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Please indicate which of the following metabolites were detected:</p> <p>Propionyl glycine:</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p>Tiglylglycine:</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p>Methylcitrate:</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p>3OH propionic acid:</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p>MMA:</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p> <p>Methylcrotonyl glycine:</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>
<p>Were plasma acylcarnitines tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 a mutation analysis performed for Propionyl-CoA carboxylase (PCC)?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>What genes were included in the mutation analysis?</p> <p><input type="checkbox"/> PCCA</p> <p><input type="checkbox"/> PCCB</p> <p><input type="checkbox"/> Other gene: _____</p>
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MOLECULAR GENETICS REPORT

<p>Were variants detected in the PCCA gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
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<p>Were variants detected in the PCCB gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
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Were variants detected in other genes?

- Yes
- No
- Don't Know

Check the types of variants found on:

Allele 1:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

Allele 2:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

**Holocarboxylase Synthetase (Multiple Carboxylase) Deficiency or Other Biotin Disorders
(Not Biotinidase Deficiency) (MCD)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the question as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please choose one:

- A. Holocarboxylase deficiency
- B. Other biotin disorder (not biotinidase deficiency) _____

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were urine organic acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>Were plasma acylcarnitines tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>Were infant chemistries (biotinidase) studies completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>What were the Biotinadase results?</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Abnormal</p> <p><input type="checkbox"/> Untested/Unknown</p>

<p>Was a mutation analysis performed for Holocarboxylase Synthetase Deficiency?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>What genes were included in the mutation analysis?</p> <input type="checkbox"/> HLCS gene <input type="checkbox"/> Other gene: _____
MOLECULAR GENETICS REPORT	
<p>Were variants detected in the HLCS gene?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Were variants detected in other genes?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Was enzyme analysis for holocarboxylase synthetase deficiency enzyme activity completed?</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	<p>Was enzyme activity:</p> <input type="checkbox"/> Consistent with disease <input type="checkbox"/> Normal activity (not consistent with disease) <input type="checkbox"/> Unknown

Fatty Acid Disorders:

Primary Carnitine Deficiency/ Carnitine Uptake Deficiency (CUD)

Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
Was urine carnitine tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was fractional excretion of free carnitine level: <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Were plasma carnitine levels tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was free carnitine (C0) <input type="checkbox"/> Low <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Were other causes for carnitine loss ruled out? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	
Was a mutation analysis performed for carnitine transporter defects? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	What genes were included in the mutation analysis? <input type="checkbox"/> SLC22A5 gene <input type="checkbox"/> Other gene: _____
MOLECULAR GENETICS REPORT	
Were variants detected in the SLC22A5 gene? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Check the types of variants found on: <i>Allele 1:</i> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance ○ Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <i>Allele 2:</i> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance ○ Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown

<p>Were variants detected in other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Was enzyme analysis for carnitine deficiency enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

**Medium-chain acyl-CoA Dehydrogenase Deficiency (MCAD)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
Were urine organic acids or acylglycines tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was Hexanoylglycine level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Were plasma acylcarnitines tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was C8 level: <input type="checkbox"/> Elevated <input type="checkbox"/> Elevated on repeat testing <input type="checkbox"/> Normal <input type="checkbox"/> Unknown Was C8>C10 level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown Was C8>C6 level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown Was C6 level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown Was C10 level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was mutation analysis performed for MCAD? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	What genes were included in the mutation analysis? <input type="checkbox"/> ACADM <input type="checkbox"/> Other: _____

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<p>Were variants detected in the ACADM gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants detected in other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Was functional analysis of fatty acid oxidation in cultured fibroblasts performed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>
<p>Was enzyme analysis for MCAD enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

**Tri-Functional Protein Deficiency (TFP); Inclusive of LCHAD
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please select one:

- L. Trifunctional Protein Deficiency
- M. Long Chain Acyl CoA dehydrogenase deficiency (LCHAD)

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were urine organic acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p> <p>Were plasma acylcarnitines tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 level?</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was C16-OH 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 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>Was enzyme analysis for TFP enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>Was functional analysis of fatty acid oxidation in cultured fibroblasts performed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

Very Long-chain acyl-CoA Dehydrogenase Deficiency
Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were plasma acylcarnitines tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p> <p>Was mutation analysis performed for VLCAD?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 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>What genes were included in the mutation analysis?</p> <p><input type="checkbox"/> ACADVL</p> <p><input type="checkbox"/> Other: _____</p>

Molecular Genetics Report

Were variants detected in ACADVL gene?

- Yes
- No
- Don't Know

Were variants detected in Other genes?

- Yes
- No
- Don't Know

Check the types of variants found on:

Allele 1:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

Allele 2:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

Check the types of variants found on:

Allele 1:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

Allele 2:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

Was enzyme analysis for VLCAD enzyme activity completed?

- Yes
- No
- Don't Know

Was enzyme analysis:

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

Was functional analysis of fatty acid oxidation in cultured fibroblasts performed?

- Yes
- No
- Don't Know

Was functional fibroblast analysis:

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

**Argininosuccinic Acidemia/ Aciduria (ASA)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were plasma amino acids tested?</p> <p><input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know</p>	<p>Was plasma ASA level</p> <p><input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown</p> <p>Was Citrulline level</p> <p><input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown</p>
<p>Were plasma urine acids tested?</p> <p><input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know</p>	<p>Was urine ASA level?</p> <p><input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown</p> <p>Was urine Citrulline level?</p> <p><input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown</p>
<p>Was a mutation analysis performed for ASA?</p> <p><input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know</p>	<p>What genes were included in the mutation analysis?</p> <p><input type="checkbox"/> ASL <input type="checkbox"/> Other gene: _____</p>

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<p>Were variants detected in the ASL gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants detected in other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Was enzyme analysis for ASA enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

**Citrullinemia Type I (CIT)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were plasma amino acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Was plasma ASA:</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>Was blood ammonia tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>Was a mutation analysis performed for Citrullinemia type I?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>What genes were included in the mutation analysis?</p> <p><input type="checkbox"/> ASS1 gene</p> <p><input type="checkbox"/> Other gene: _____</p>

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<p>Were variants detected in the ASS1 gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants detected in other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Was enzyme analysis for Cirtullinemia type-I enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

Cystathionine Beta-Synthase (CBS) Deficiency (Classic Homocystinuria)
Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
Were plasma amino acids tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was Methionine level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was plasma Homocysteine tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was plasma Homocysteine level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was a mutation analysis performed for CBS? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	What genes were included in the mutation analysis? <input type="checkbox"/> CBS gene <input type="checkbox"/> Other gene: _____

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<p>Were variants detected in the CBS gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Were variants detected in other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Was enzyme analysis for CBS enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Was enzyme activity:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Consistent with disease <input type="checkbox"/> Normal activity (not consistent with disease) <input type="checkbox"/> Unknown

**Maple Syrup Urine Disease (MSUD)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

- A. MAPLE SYRUP URINE DISEASE, TYPE IA
- B. MAPLE SYRUP URINE DISEASE, TYPE IB
- C. MAPLE SYRUP URINE DISEASE, TYPE II
- D. MAPLE SYRUP URINE DISEASE, TYPE III

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were plasma amino acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Was Alloisoleucine level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was Leucine level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was Isoeucine level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was Valine level</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was Leu>Val?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>

<p>Were Urine organic acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>Was maternal 3-MCC level tested and ruled out?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p style="background-color: black; color: black;">[REDACTED]</p>
<p>Was a mutation analysis performed for MSUD?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>What genes were included in the mutation analysis?</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>Other: _____</p>
<p>MOLECULAR GENETICS REPORT</p>	
<p>Were variants detected in <i>DBT</i>?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>

<p>Were variants detected in <i>BCKDHD</i>?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Were variants detected in <i>DLD</i>?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Were variants detected in <i>BCKDHA</i>?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown

<p>Were variants detected in Other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p><i>Allele 2:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
<p>Was enzyme analysis for MSUD enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Was enzyme activity:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Consistent with disease <input type="checkbox"/> Normal activity (not consistent with disease) <input type="checkbox"/> Unknown

**Hyperphenylalaninemia (HyperPHE) (Inclusive of Classic PKU)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please choose one:

- A. Classic PKU
- B. Benign HyperPhe
- C. HyperPhe diet controlled

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were plasma amino acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Was Phe level</p> <p><input type="checkbox"/> Elevated (>120umol/L on unrestricted diet)</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p> <p>Was Phe/Tyr ratio</p> <p><input type="checkbox"/> Elevated</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Unknown</p>
<p>Were bipterin studies done?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Were bipterin studies:</p> <p><input type="checkbox"/> Normal</p> <p><input type="checkbox"/> Abnormal</p> <p><input type="checkbox"/> Unknown</p>
<p>Was a mutation analysis performed for Hyperphenylalaninemia?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>What genes were included in the mutation analysis?</p> <p><input type="checkbox"/> PAH gene</p> <p><input type="checkbox"/> Other gene: _____</p>

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<p>Were variants detected in the PAH gene?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Were variants detected in other genes?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
<p>Was enzyme analysis for Hyperphe (inclusive of classic PKU) enzyme activity completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

Tyrosinemia Type I (TYR-1)
Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following as Yes/No/Don't Know	If Yes
<p>Were plasma organic acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p> <p>Were urine organic acids tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>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>Was mutation analysis performed for Tyrosinemia Type I?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>What genes were included in the mutation analysis?</p> <p><input type="checkbox"/> FAH</p> <p><input type="checkbox"/> Other: _____</p>

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Were variants detected in FAH?

- Yes
- No
- Don't Know

Check the types of variants found on:

Allele 1:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

Allele 2:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

Were variants detected in Other genes?

- Yes
- No
- Don't Know

Check the types of variants found on:

Allele 1:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

Allele 2:

- Variant known to be disease causing
- Variant of unknown significance
 - Predicted to be pathogenic
- Wild Type (Normal)
- Unknown

Was enzyme analysis for Tyrosinemia Type I enzyme activity completed?

- Yes
- No
- Don't Know

Was enzyme analysis:

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

Endocrine Disorders

Congenital Hypothyroidism (CH) Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please choose one:

- A. Primary Congenital Hypothyroidism
- B. Secondary Congenital Hypothyroidism
- C. TBG Deficiency (Thyroxine Binding Globulin) or other protein binding defect

Please answer the following as Yes/No/Don't Know	If Yes
Was Serum TSH tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	What was the level: <input type="checkbox"/> TSH > 10 mU/L <input type="checkbox"/> TSH 6-10 mU/L <input type="checkbox"/> TSH <6 mU/L <input type="checkbox"/> Unknown Was it tested before initiation of treatment? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
Was Serum Total T4 tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was Serum Total T4 below the age-established reference range? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown Was it tested before initiation of treatment? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
Was Serum <u>Free</u> T4 tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was Serum <u>Free</u> T4 below the age-established reference range? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown Was it tested before initiation of treatment? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown

<p>Does this baby have other pituitary hormone deficiencies?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	
<p>Does this baby have midline defects?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	
<p>Was TBG tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p> <p>Was T3 or T4 resin uptake tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>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)
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please choose one:

- A. Classic 21-Hydroxylase Deficiency – Salt Wasting
- B. Classic 21-Hydroxylase Deficiency – Simple Virilizing
- C. Other Adrenal disorder: (Please list) _____

Please answer the following as Yes/No/Don't Know	If Yes
<p>Was a confirmatory serum 17-OHP level obtained?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Was there a value at baseline:</p> <p><input type="checkbox"/> 10,000 ng/dl;</p> <p><input type="checkbox"/> 1000-10,000 ng/dl;</p> <p><input type="checkbox"/> < 1000 ng/dl;</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>Was there a result after ACTH stimulation:</p> <p><input type="checkbox"/> 10,000 ng/dl;</p> <p><input type="checkbox"/> 1000-10,000 ng/dl;</p> <p><input type="checkbox"/> < 1000 ng/dl;</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>Was tandem mass spectrometry urinary steroid profile obtained?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Were the urinary spectrometry steroid profile results:</p> <p><input type="checkbox"/> Indicative of 21-Hydroxylase Deficiency CAH</p> <p><input type="checkbox"/> Unknown</p>

<p>Was serum sodium level measured before initiation of treatment?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Was the sodium level:</p> <p><input type="checkbox"/> < 135 mEq/L</p> <p><input type="checkbox"/> > 135 mEq/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>Was Plasma renin activity level measured at time of initiation of treatment?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>Was the Plasma renin activity normal for age?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p>Was it tested before initiation of treatment?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p>

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<p>Was mutation analysis for 21-Hydroxylase deficiency (CYP21A2) performed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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 style="padding-left: 20px;"><input type="checkbox"/> 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 style="padding-left: 20px;"><input type="checkbox"/> Predicted to be pathogenic</p> <p><input type="checkbox"/> Wild Type (Normal)</p> <p><input type="checkbox"/> Unknown</p>
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CLINICAL RESULTS

<p>Is there evidence of salt wasting? (e.g. shock or severe failure to thrive)?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	
<p>Is there supportive clinical or laboratory evidence of CAH?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</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>

Hemoglobinopathies

Presence of Hb S Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information

Please answer the following:	If Yes
<p>Final Diagnosis as determined by a clinician performing the follow-up</p> <p><input type="checkbox"/> S, Beta + Thalassemia – Hb S/B + Th</p> <p><input type="checkbox"/> S, C disease – Hb S/C</p> <p><input type="checkbox"/> <i>Sickle Cell Disease, Hb S only</i></p> <p><input type="checkbox"/> S,S Disease (Sickle Cell Anemia) – Hb SS</p> <p><input type="checkbox"/> S, Beta 0-thalassemia – Hb S/B0Th</p> <p><input type="checkbox"/> Not Known</p> <p><input type="checkbox"/> S, other</p>	
<p>Was qualitative (IEF or HPLC) testing completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</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"/> FSA₂</p> <p><input type="checkbox"/> FSAA₂</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>
<p>Was quantitative (HPLC or electrophoresis) testing completed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</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 A₂</p> <p><input type="checkbox"/> FSA with high A₂</p> <p><input type="checkbox"/> FSA</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>

<p>Was mutation analysis performed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p>Check the type of variant found on allele 1:</p> <p><input type="checkbox"/> S</p> <p><input type="checkbox"/> C</p> <p><input type="checkbox"/> Beta + Thal</p> <p><input type="checkbox"/> Beta⁰ Thal</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p> <p>Check the type of variant found on allele 2:</p> <p><input type="checkbox"/> S</p> <p><input type="checkbox"/> C</p> <p><input type="checkbox"/> Beta + Thal</p> <p><input type="checkbox"/> Beta⁰ Thal</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>
<p>NBS result</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</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"/> FSA₂</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>
<p>Was a CBC performed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</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>Were family studies (in parents) done?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p>What were the results?</p> <p>Maternal Status:</p> <p><input type="checkbox"/> Carrier S</p> <p><input type="checkbox"/> Carrier C</p> <p><input type="checkbox"/> Carrier Beta + Thal</p> <p><input type="checkbox"/> Carrier Beta⁰ Thal</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p> <p>Paternal Status:</p> <p><input type="checkbox"/> Carrier S</p> <p><input type="checkbox"/> Carrier C</p> <p><input type="checkbox"/> Carrier Beta + Thal</p> <p><input type="checkbox"/> Carrier Beta⁰ Thal</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>

<p>Was there a positive family history?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	
<p>Were HPLC & IEF tested on the same sample from the infant?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p>What were the results?</p> <p><input type="checkbox"/> FS</p> <p><input type="checkbox"/> FSC</p> <p><input type="checkbox"/> FSA₂</p> <p><input type="checkbox"/> FSAA₂</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>
<p>Were Hgb tests (electrophoresis or HPLC) performed on family members?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</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
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

Please answer the following:	If Yes
Final Diagnosis as determined by a clinician performing the follow-up <input type="checkbox"/> Hemoglobin C disease <input type="checkbox"/> Hemoglobin D disease <input type="checkbox"/> Hemoglobin E disease <input type="checkbox"/> Hemoglobin O-Arab disease <input type="checkbox"/> Other hemoglobin disease, please describe	
Alpha thalassemia present? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	
Was qualitative (IEF or HPLC) testing completed? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	What were the results? <input type="checkbox"/> FC <input type="checkbox"/> FD <input type="checkbox"/> FE <input type="checkbox"/> FO _{ARAB} <input type="checkbox"/> Other <input type="checkbox"/> Unknown
Was quantitative (HPLC or electrophoresis) testing completed? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	What were the results? <input type="checkbox"/> FC <input type="checkbox"/> FD <input type="checkbox"/> FE <input type="checkbox"/> FO _{ARAB} <input type="checkbox"/> Other <input type="checkbox"/> Unknown

<p>Was mutation analysis performed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</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-Arab</p> <p><input type="checkbox"/> Other</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-Arab</p> <p><input type="checkbox"/> Beta + Thal</p> <p><input type="checkbox"/> Beta⁰ Thal</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>
<p>NBS result</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</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_{ARAB}</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>
<p>Was a CBC performed?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</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>Were family studies (in parents) done?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p>What were the results?</p> <p>Maternal Status:</p> <p><input type="checkbox"/> Carrier C</p> <p><input type="checkbox"/> Carrier D</p> <p><input type="checkbox"/> Carrier E</p> <p><input type="checkbox"/> Carrier O-Arab</p> <p><input type="checkbox"/> Carrier Beta + Thal</p> <p><input type="checkbox"/> Carrier Beta⁰ Thal</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p> <p>Paternal Status:</p> <p><input type="checkbox"/> Carrier C</p> <p><input type="checkbox"/> Carrier D</p> <p><input type="checkbox"/> Carrier E</p> <p><input type="checkbox"/> Carrier O-Arab</p> <p><input type="checkbox"/> Carrier Beta + Thal</p> <p><input type="checkbox"/> Carrier Beta⁰ Thal</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> Unknown</p>
<p>Was there a positive family history?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	
<p>Were Hgb tests (electrophoresis or HPLC) performed on family members?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</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>

Lysosomal Storage Disorders

Mucopolysaccharidosis Type I (MPS I) Case Confirmatory Diagnosis Follow-Up

Birth weight (grams):

Gestational age (weeks gestation at time of birth):

State of birth (state reporting the case):

Sex (male/female/unknown):

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please choose one:

1. Primary targets of NBS
 - a. MPS I –severe
 - b. MPS I – severity not determined
 - c. MPS I – attenuated

Please answer the following as Yes/No/Unknown	If Yes
Was enzyme activity tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	What was the enzyme level? <input type="checkbox"/> Within lab known affected range <input type="checkbox"/> Unknown
Were urine GAGS tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	What was the urine GAG level? <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown

<p>Clinical symptoms/ lab findings?</p> <p><input type="checkbox"/> Symptoms present and documented by specialists. Public health (PH) program continued to collect data through the development of symptoms</p> <p><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</p> <p><input type="checkbox"/> Unknown</p>	<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 REPORT

<p>Were variants detected in the genes known to be associated with MPS I?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown</p>	<p>Check the types of variant(s) found on:</p> <p>Allele 1:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Pathogenic variant and associated with SEVERE disease <input type="checkbox"/> Pathogenic or likely pathogenic variant <input type="checkbox"/> Variant known to be associated with ATTENUATED Disease <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Untested/Unknown <p>Allele 2:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Pathogenic variant and associated with SEVERE disease <input type="checkbox"/> Pathogenic or likely pathogenic variant <input type="checkbox"/> Variant of uncertain significance <input type="checkbox"/> Variant known to be associated with ATTENUATED Disease <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Untested/Unknown
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Pompe Disease Case Confirmatory Diagnosis Follow-Up

Birth weight (grams): _____

Gestational age (weeks gestation at time of birth): _____

State of birth (state reporting the case): _____

Sex (male/female/unknown): _____

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please choose one:

1. Primary targets of NBS
 - a. Infantile Onset (IO) Pompe Disease
 - b. Late Onset (LO) Pompe Disease

Please answer the following as Yes/No/Don't Know	If Yes
Was enzyme activity tested in blood (not DBS sample)? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	What was the enzyme level? <input type="checkbox"/> Within lab known affected range for infantile onset (IO) <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) <input type="checkbox"/> Within lab known affected range for late onset (LO) <input type="checkbox"/> Low (above affected range, for LO not normal) <input type="checkbox"/> Unknown
Was enzyme activity tested in skin/muscle? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<input type="checkbox"/> Positive skin or muscle biopsy <input type="checkbox"/> Unknown
Was there Cardiac involvement consistent with Pompe? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<input type="checkbox"/> Positive findings on Chest X-ray/EKG/ECHO in newborn period <input type="checkbox"/> Positive findings on Chest X-ray/EKG/ECHO

<p>Lab findings?</p> <ul style="list-style-type: none"> <input type="checkbox"/> Elevated CK/AST/ALT/LDH/Urine Hex4 <input type="checkbox"/> Not present <input type="checkbox"/> Unknown <input type="checkbox"/> Not done 	
<p>Clinical findings?</p> <ul style="list-style-type: none"> <input type="checkbox"/> Symptoms present after 1 year of age and documented by specialists. PH program continued to collect data through the development of symptoms <input type="checkbox"/> Symptoms present before 1 year of age but no cardiac involvement <input type="checkbox"/> Unknown or not reported to PH by the end of follow-up 	<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>

MOLECULAR GENETICS REPORT

<p>Were variants detected in the genes known to be associated with Pompe Disease?</p> <ul style="list-style-type: none"> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown 	<p>Check the types of variant(s) found on:</p> <p>Allele 1:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Pathogenic <input type="checkbox"/> Pathogenic variant and associated with infantile onset <input type="checkbox"/> Novel variant that is likely pathogenic <input type="checkbox"/> Pathogenic variant or likely pathogenic variant, with deletion or duplication consistent with infantile onset <input type="checkbox"/> Pathogenic and associated with non-classical disease, or variant of uncertain significance <input type="checkbox"/> Pathogenic or likely pathogenic variant, no other variants found, dup/del testing not done or not known <input type="checkbox"/> Pathogenic or likely pathogenic variant, no other variants found <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <p>Allele 2:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Pathogenic <input type="checkbox"/> Pathogenic variant and associated with infantile onset <input type="checkbox"/> Novel variant that is likely pathogenic <input type="checkbox"/> Pathogenic variant or likely pathogenic variant, with deletion or duplication consistent with infantile onset <input type="checkbox"/> Pathogenic and associated with non-classical disease, or variant of uncertain significance <input type="checkbox"/> Pathogenic or likely pathogenic variant, no other variants found, dup/del testing not done or not known <input type="checkbox"/> Pathogenic or likely pathogenic variant, no other variants found <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
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Other Disorders

Biotinidase Deficiency Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. Attach a copy of the laboratory reports supporting these findings. The reports will be stored within the newborn screening program for future reference.

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

- D. Profound Biotinidase deficiency
- E. Partial Biotinidase deficiency

Was enzyme analysis for biotinidase enzyme activity completed? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	What as the enzyme activity? <input type="checkbox"/> <10% normal activity <input type="checkbox"/> 10-30% normal activity <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was mutation analysis performed for Biotinidase deficiency? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Check the types of variants found on: <i>Allele 1:</i> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Known to be associated with profound enzyme deficiency <input type="checkbox"/> Known to be associated with partial enzyme deficiency ['mild' mutation (D444H)] <input type="checkbox"/> Variant of unknown significance <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <i>Allele 2:</i> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Known to be associated with profound enzyme deficiency <input type="checkbox"/> Known to be associated with partial enzyme deficiency ['mild' mutation (D444H)] <input type="checkbox"/> Variant of unknown significance <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) Unknown

**Galactosemia
Case Confirmatory Diagnosis Follow-Up**

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing after newborn screening. Attach a copy of the laboratory reports supporting these findings. The reports will be stored within the newborn screening program for future reference.

Please answer the following as Yes/No/Don't Know	If Yes
Were GALT levels tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was GALT level <input type="checkbox"/> <10% <input type="checkbox"/> 10-30% <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Were Gal-1-P tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was Gal-1-P level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was Urine Galactitol tested? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Was Gal-1-P level <input type="checkbox"/> Elevated <input type="checkbox"/> Normal <input type="checkbox"/> Unknown
Was a mutation analysis performed for Galactosemia? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Check the types of variants found on: <i>Allele 1:</i> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown <i>Allele 2:</i> <input type="checkbox"/> Variant known to be disease causing <input type="checkbox"/> Variant of unknown significance <ul style="list-style-type: none"> <input type="checkbox"/> Predicted to be pathogenic <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Unknown
If Variant Galactosemia, was protein phenotyping completed? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know <input type="checkbox"/> N/A	Did result indicate <input type="checkbox"/> phenotype consistent with variant <input type="checkbox"/> phenotype NOT consistent with variant <input type="checkbox"/> Unknown

<p>If Arginase Deficiency, were enzyme studies completed?</p> <ul style="list-style-type: none"><input type="checkbox"/> Yes<input type="checkbox"/> No<input type="checkbox"/> Don't Know<input type="checkbox"/> N/A	<p>Was enzyme activity:</p> <ul style="list-style-type: none"><input type="checkbox"/> Consistent with disease<input type="checkbox"/> Normal activity (not consistent with disease)<input type="checkbox"/> Unknown
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Cystic Fibrosis Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. Attach a copy of the laboratory reports supporting these findings. The reports will be stored within the newborn screening program for future reference.

Please choose one:

- D. Typical Cystic Fibrosis (CF)
- E. CFTR-Related Metabolic Syndrome (CRMS)
- F. CFTR-Related Disease

Please answer the following as Yes/No/Don't Know	If Yes
<p>Did the NBS result indicate an elevated IRT?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	
<p>Were CFTR mutations detected on the newborn screening mutation panel?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p> <p>(* Mutations seen in patients with CF have been classified as disease-causing, neutral, or varying clinical consequences through the CFTR2 project: http://cftr2.org/browse.php. Additional information about the mutation and the association with lower sweat chlorides can also be found at CFTR2.)</p>	<p>Check the types of variants found on:</p> <p><i>Allele 1:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing in CFTR2 <ul style="list-style-type: none"> <input type="checkbox"/> 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><i>Allele 2:</i></p> <ul style="list-style-type: none"> <input type="checkbox"/> Variant known to be disease causing in CFTR2 <ul style="list-style-type: none"> <input type="checkbox"/> 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>Did the child have meconium ileus?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	

<p>Was a valid sweat chloride result available?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>What were the sweat test results (please report the highest sweat chloride value from one sweat test)?</p> <p><input type="checkbox"/> ≥ 60 mmol/L (regardless of age)</p> <p>If < 60 mmol/L</p> <p> If age < 6 months</p> <p> <input type="checkbox"/> < 30 mmol/L</p> <p> <input type="checkbox"/> 30-59 mmol/L</p> <p> If age ≥ 6 months</p> <p> <input type="checkbox"/> < 40 mmol/L</p> <p> <input type="checkbox"/> 40 -59 mmol/L</p> <p><input type="checkbox"/> Quantity Not Sufficient</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"/> Don't Know</p>	
<p>Was a sweat chloride repeated on a separate day (results from different arm on the same day should not be reported here)?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>	<p>What were the repeat sweat test results (please report the highest sweat chloride value from one sweat test)?</p> <p><input type="checkbox"/> ≥ 60 mmol/L (regardless of age)</p> <p>If < 60 mmol/L</p> <p> If age < 6 months</p> <p> <input type="checkbox"/> < 30 mmol/L</p> <p> <input type="checkbox"/> 30-59 mmol/L</p> <p> If age ≥ 6 months</p> <p> <input type="checkbox"/> < 40 mmol/L</p> <p> <input type="checkbox"/> 40 -59 mmol/L</p>

Was a CFTR mutation panel completed after the newborn screening mutation panel?

- Yes
- No
- Don't Know

(* Mutations seen in patients with CF have been classified as disease-causing, neutral, or varying clinical consequences through the CFTR2 project: <http://cftr2.org/browse.php>. Additional information about the mutation and the association with lower sweat chlorides can also be found at CFTR2.)

Check the types of variants found on:

Allele 1:

- Variant known to be disease causing in CFTR2
 - Shown to be associated with lower sweat chlorides
- Neutral variant
- Variant of varying clinical consequence in CFTR2
- Wild Type (Normal)
- Unknown (not reported in CFTR2)

Allele 2:

- Variant known to be disease causing in CFTR2
 - Shown to be associated with lower sweat chlorides
- Neutral variant
- Variant of varying clinical consequence in CFTR2
- Wild Type (Normal)
- Unknown (not reported in CFTR2)

Question Below to be answered only if child was diagnosed after the newborn period

If child was diagnosed after the newborn period, were clinical symptoms associated with CFTR Related Disease present?

- Yes
- No
- Don't Know

Symptoms include:

- CBAVD
- Recurrent pancreatitis
- Nasal polyposis
- Infertility
- Focal biliary cirrhosis with portal hypertension

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

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

c.3763T>C	p.Ile336Lys	I336K	No	CF-causing	NO
c.1558G>T	p.Ile507del	I507del	Yes	CF-causing	NO
c.3230T>C	p.Leu1077Pro	L1077P	No	CF-causing	NO
c.1040G>A	p.Leu206Trp	L206W	No	CF-causing	NO
c.3302T>A	p.Met1101Lys	M1101K	No	CF-causing	NO
c.274G>A	p.Asn1303Lys	N1303K	Yes	CF-causing	NO
c.617T>G	p.Pro67Leu	P67L	No	CF-causing	NO
c.2764_2765insAG	p.Gln220X	Q220X	No	CF-causing	NO
c.1973_1985del13insAGAA A	p.Gln493X	Q493X	No	CF-causing	NO
c.3196C>T	p.Arg1066Cys	R1066C	No	CF-causing	NO
c.4296_4297insGA	p.Arg1158X	R1158X	No	CF-causing	NO
c.1692delA	p.Arg1162X	R1162X	Yes	CF-causing	NO
c.1055G>A	p.Arg117Cys	R117C	No	CF-causing	NO
c.1466C>A	p.Arg117His	R117H	Yes	Indeterminate	YES
c.1013C>T	p.Arg334Trp	R334W	Yes	CF-causing	NO
c.532G>A	p.Arg347His	R347H	Yes	CF-causing	NO
c.1040G>C	p.Arg347Pro	R347P	No	CF-causing	NO
c.2908G>C	p.Arg352Gln	R352Q	No	CF-causing	NO
c.2424_2425insAT	p.Arg553X	R553X	Yes	CF-causing	NO
c.2780T>C	p.Arg560Thr	R560T	Yes	CF-causing	NO
c.349C>T	p.Ser1251Asn	S1251N	No	CF-causing	NO
c.1000C>T	p.Ser549Asn	S549N	No	CF-causing	NO
c.3752G>A	p.Ser945Leu	S945L	No	CF-causing	NO
c.1645A>C or c.1647T>G	p.Val520Phe	V520F	No	CF-causing	NO
c.274G>T	p.Trp1282X	W1282X	Yes	CF-causing	NO
c.2128A>T	p.Tyr1092X	Y1092X	No	CF-causing	NO
c.2195T>G	p.Tyr122X	Y122X	No	CF-causing	NO

Severe Combined Immunodeficiencies Case Confirmatory Diagnosis Follow-Up

Birth weight (grams): _____

Gestational age (weeks gestation at time of birth): _____

State of birth (state reporting the case): _____

Sex (male/female/unknown): _____

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. You may need to consult with relevant clinician(s) and/or medical staff to obtain this information.

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

Please choose one:

1. Primary targets of NBS
 - a. Classic SCID
 - b. Leaky SCID
 - c. Omenn Syndrome

Please answer the following as Yes/No/Don't Know	If Yes
<p>Was the CD3 T cell level tested?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown/untested</p>	<p>What was the CD3 T cell level?</p> <p><input type="checkbox"/> <300 autologous T cells, undetectable or very few naïve T cells</p> <p><input type="checkbox"/> 300-1500, few naïve T cells, oligoclonal T cells or poor T cell diversity</p> <p><input type="checkbox"/> >80% CD45RO+</p> <p><input type="checkbox"/> Any number (not zero)</p> <p><input type="checkbox"/> Untested/Unknown</p>
<p>Was proliferation to PHA test done?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown/untested</p>	<p>Proliferation to PHA:</p> <p><input type="checkbox"/> <10% of normal</p> <p><input type="checkbox"/> 10-50% of normal PHA</p> <p><input type="checkbox"/> 10-30% normal PHA or Absent to Candida/TT</p> <p><input type="checkbox"/> <30% of normal</p> <p><input type="checkbox"/> Any/Unknown</p>

<p>Was Maternal engraftment documented?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown/Untested</p>	
<p>Was a mutation analysis performed in the genes known to be associated with SCID?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown/untested</p>	
<p>MOLECULAR GENETICS REPORT</p>	
<p>Were variants detected in the genes known to be associated with SCID?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Unknown/untested</p>	<p>Check the types of variant(s) found on:</p> <p>Allele 1:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Pathogenic variant in a known SCID gene <input type="checkbox"/> Pathogenic variant in a known SCID gene on X chromosome in a male <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) <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Untested/Unknown <p>Allele 2:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Pathogenic variant in a known SCID gene <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) <input type="checkbox"/> Wild Type (Normal) <input type="checkbox"/> Untested/Unknown
	<p>Was 22q11 deletion ruled out?</p> <p><input type="checkbox"/> Yes</p> <p><input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't Know</p>

	<p>Were homozygous or compound heterozygous FOXN1 mutations ruled out?</p> <p><input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know</p>
	<p>Were heterozygous TBX1 variants ruled out?</p> <p><input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know</p>

Critical Congenital Heart Disease Case Confirmatory Diagnosis Follow-Up

Instructions: Please answer the questions as fully as possible. All testing refers to confirmatory testing following newborn screening. Attach a copy of the laboratory reports supporting these findings. The reports will be stored within the newborn screening program for future reference.

What was the Final Diagnosis? (Check all that apply)

- CCHD (Specify)
 - Truncus Arteriosus
 - Total Anomalous Pulmonary Venous Connection
 - Tetralogy of Fallot
 - Pulmonary Atresia
 - Ebstein's Anomaly
 - Hypoplastic Left Heart Syndrome
 - Single Ventricle
 - Tricuspid Atresia
 - Transposition of the Great Arteries
 - Double Outlet Right Ventricle
 - Coarctation of Aorta
 - Interrupted Arch
 - Aortic Valve Disease
- Non critical CHD (Please specify)
- Other (Please specify)

Please answer the following:	If Yes, what were the results of the echocardiogram?	
Was a Postnatal Echocardiogram Completed? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown/untested	Truncus Arteriosus <input type="checkbox"/> Truncus arteriosus <input type="checkbox"/> Truncus arteriosus + Interrupted aortic arch	
	Total Anomalous Pulmonary Venous Connection (TAPVC) <input type="checkbox"/> Type1 (supracardiac) <input type="checkbox"/> Type 2 (cardiac) <input type="checkbox"/> Type 3 (infracardiac) <input type="checkbox"/> Type 4 (mixed)	
	Tetralogy of Fallot (TOF) <input type="checkbox"/> TOF <input type="checkbox"/> TOF, Pulmonary stenosis <input type="checkbox"/> TOF, AVCanal (AVSD) <input type="checkbox"/> TOF, Absent pulmonary valve <input type="checkbox"/> Interrupted aortic arch + AP window (aortopulmonary window)	
	Pulmonary Artesia <input type="checkbox"/> Pulmonary atresia <input type="checkbox"/> Pulmonary atresia, IVS <input type="checkbox"/> Pulmonary atresia, VSD (Including TOF, PA) <input type="checkbox"/> Pulmonary atresia, VSD-MAPCA	
	Ebstein's Anomaly <input type="checkbox"/> Ebstein's anomaly	
	Hypoplastic Left Heart Syndrome (HLHS) <input type="checkbox"/> Hypoplastic left heart syndrome	
	Single Ventricle <input type="checkbox"/> Single ventricle, DILV <input type="checkbox"/> Single ventricle, DIRV <input type="checkbox"/> Single ventricle, Mitral atresia <input type="checkbox"/> Single ventricle, Unbalanced AV canal	<input type="checkbox"/> Single ventricle, Heterotaxia syndrome <input type="checkbox"/> Single ventricle, Other <input type="checkbox"/> Single ventricle + Total anomalous pulmonary venous connection (TAPVC)
	Tricuspid Artesia <input type="checkbox"/> Single ventricle, Tricuspid atresia	
	Transposition of the Great Arteries (TGA) <input type="checkbox"/> d-TGA, IVS <input type="checkbox"/> d-TGA, IVS-LVOTO <input type="checkbox"/> d-TGA, VSD <input type="checkbox"/> d-TGA, VSD-LVOTO	

	Double Outlet Right Ventricle (DORV) <input type="checkbox"/> DORV, VSD type <input type="checkbox"/> DORV, TOF type <input type="checkbox"/> DORV, TGA type	<input type="checkbox"/> DORV, Remote VSD (uncommitted VSD) <input type="checkbox"/> DORV + AVSD (AV Canal) <input type="checkbox"/> DORV, IVS
	Coarctation of Aorta <input type="checkbox"/> Coarctation of aorta <input type="checkbox"/> Aortic arch hypoplasia <input type="checkbox"/> VSD + Aortic arch hypoplasia <input type="checkbox"/> VSD + Coarctation of aorta	
	Interrupted Arch <input type="checkbox"/> Interrupted aortic arch <input type="checkbox"/> Interrupted aortic arch + VSD <input type="checkbox"/> Interrupted aortic arch + AP window (aortopulmonary window)	
	Aortic Valve Disease <input type="checkbox"/> Aortic Stenosis receiving intervention in first 30 days of life <input type="checkbox"/> Pulmonary Stenosis receiving intervention in the first 30 days of life	
Please answer the following:	If Yes	
Was a Prenatal Echocardiogram Completed? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Don't Know	Did the Prenatal Echo findings suggest CCHD? <input type="checkbox"/> Yes <input type="checkbox"/> No	