

# Template Map for the Case Import File

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# **INSTRUCTIONS**

This template map provides variable names and acceptable values for the <u>case import file</u>. This import file is one of the two options for newborn screening programs to enter individual cases into the NewSTEPs Repository. The other option is to use the <u>online webform</u>.

The <u>case import file</u> contains the common demographic and screening variables that are asked for all conditions. It also contains a final diagnosis for certain conditions. General instructions to use the import file include:

- Required fields are indicated below; these variables must have an acceptable value entered in order for the import to work
- For fields that are not required, the variable or column is also not required
  - For non-required variables/columns included in the CSV file, enter an acceptable value or leave empty
- Variables/columns may be in any order
- Each row is unique to the case/baby; please be sure to select the correct condition, this includes secondary conditions

Download the <u>case import file</u>, enter the data that is being reported, and save the document as a CSV file to your desktop. To import the file into the repository, select **Choose File** on the righthand side of the screen. The File Explorer for your desktop will appear, and the desired file can be selected. Next, select **Submit CSV** to import the file. If the data isn't formatted correctly, the import will not be accepted.

Common errors in import files include:

- Abbreviation of the state or territory name; please spell out
- Conditions not spelled correctly or use the correct format; it is suggested that you copy and paste directly from this template map and only abbreviate conditions found on page 6
- NULL versus true zero: only enter zero when the value is a true zero, otherwise leave the cell empty



# INFANT DEMOGRAPHIC INFORMATION

state - name of the state/territorial newborn screening program, REQUIRED\* Acceptable values:

- Alabama •
- Alaska
- American Samoa
- Arizona •
- Arkansas •
- California •
- Colorado •
- Connecticut •
- Commonwealth of • the Northern Mariana Islands
- Delaware •
- **District of Columbia** •
- Florida •
- Georgia
- Guam •
- Hawaii •
- Idaho •
- Illinois •
- Indiana

- lowa •
- Kansas
- Kentucky •
- Louisiana •
- Maine •
- Maryland •
- Massachusetts •
- Michigan •
- Minnesota
- Mississippi •
- Missouri
- Montana
- Nebraska
- Nevada
- New Hampshire
- New Jersey
- New Mexico
- North Carolina
- North Dakota

- Ohio •
- Oklahoma •
- Oregon
- Pennsylvania
- Puerto Rico •
- Rhode Island
- South Carolina •
- South Dakota
- Tennessee
- Texas
- US Virgin Islands
- Utah
- Vermont
- Virginia
- Washington
- West Virginia
- Wisconsin
- Wyoming

birthYear - The year in which the birth occurred, REQUIRED\*

stateUniqueId - The unique identifier assigned to the case by the state, REQUIRED\*

#### condition - Name of condition, REQUIRED\* Acceptable values:

- 2,4 Dienoyl-CoA reductase deficiency DE RED
  - 2-Methyl-3-hydroxybutyric aciduria 2M3HBA
- 2-Methylbutyrylglycinuria 2MBG
- 3-Hydroxy-3-methyglutaric aciduria HMG
- 3-Methylcrotonyl-CoA carboxylase deficiency 3-MCC
- 3-Methylglutaconic aciduria 3MGA •
- Argininemia ARG •
- Argininosuccinic aciduria ASA •
- Beta-Ketothiolase deficiency BKT •
- Biopterin defect in cofactor biosynthesis BIOPT (BS) •
- Biopterin defect in cofactor regeneration BIOPT (RG) •
- **Biotinidase deficiency BIOT** •
- Carbamoyl phosphate synthetase I deficiency CPS
- Carnitine acylcarnitine translocase deficiency CACT
- Carnitine palmitoyltransferase type I deficiency CPT IA •
- Carnitine palmitoyltransferase type II deficiency CPT II



- New York

- Carnitine uptake defect/carnitine transport defect CUD
- Citrullinemia, type I CIT
- Citrullinemia, type II CIT II
- Classic galactosemia GALT
- Classic PKU & Hyperphe
- Congenital Toxoplasmosis TOXO
- Congenital adrenal hyperplasia CAH
- Congenital hypothyroidism CH
- Critical congenital heart disease CCHD
- Cystic fibrosis CF
- Cytomegalovirus CMV
- Duchenne Muscular Dystrophy DMD
- Ethylmalonic encephalopathy EME
- Fabry
- Formiminoglutamic acidemia FIGLU
- Galactoepimerase deficiency GALE
- Galactokinase deficiency GALK
- Gaucher
- Glucose-6-phosphate dehydrogenase deficiency G6PDD/G6PD
- Glutaric acidemia type I GA1
- Glutaric acidemia type II GA2
- Guanidinoacetate Methyltransferase GAMT
- Hb No structural variant
- Hearing loss HEAR
- Holocarboxylase synthetase deficiency MCD
- Homocystinuria HCY
- Human Immunodeficiency Virus HIV Exposure
- Hypermethioninemia MET
- Hyperornithinemia with Gyrate Deficiency Hyper ORN
- Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome HHH
- Isobutyrylglycinuria IBG
- Isovaleric acidemia IVA
- Krabbe Disease
- Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency LCHAD
- Malonic acidemia MAL
- Maple syrup urine disease MSUD
- Medium-chain acyl-CoA dehydrogenase deficiency MCAD
- Medium-chain ketoacyl-CoA thiolase deficiency MCKAT
- Medium/short-chain L-3-hydroxyacl-CoA dehydrogenase deficiency M/SCHAD
- Methylmalonic acidemia (cobalamin disorders) Cbl A,B
- Methylmalonic acidemia (methylmalonyl-CoA mutase) MUT
- Methylmalonic acidemia with homocystinuria Cbl C,D
- Mucopolysaccharidosis I MPS I
- Mucopolysaccharidosis II MPS II
- Niemann Pick



- Nonketotic Hyperglycinemia NKH
- Ornithine transcarbamylase deficiency OTC
- Pompe
- Presence of Hb S
- Presence of Other Hb Variant
- Prolinemia Type I/ Type II PRO
- Propionic acidemia PROP
- Pyroglutamic acidemia 5-OXO
- Severe Combined Immunodeficiencies SCID
- Short-chain acyl-CoA dehydrogenase deficiency SCAD
- Spinal Muscular Atrophy SMA
- T-cell related lymphocyte deficiencies
- Trifunctional protein deficiency TFP
- Tyrosinemia, type I TYR I
- Tyrosinemia, type II TYR II
- Tyrosinemia, type III TYR III
- Very long-chain acyl-CoA dehydrogenase deficiency VLCAD
- X-linked Adrenoleukodystrophy
- Zellweger Syndrome

*Note: The following condition abbreviations can be used instead of using the entire condition name:* 

- 3-MCC
- ASA
- BIOT
- BKT
- CAH
- Cbl A,B
- CCHD
- CF
- CH
- CIT
- CUD
- GA1
- GALT
- GAMT
- HCY

- HEAR
- HMG
- IVA
- LCHAD
- MCAD
- MCD
- MPS I
- MPS II
- MSUD
- MUT
- Pompe
- PROP
- TFP
- TYR I
- VLCAD

gestationalAge - the gestational age in weeks (please use whole numbers only)

birthWeight - the birth weight in grams

biologicalGender - the biological gender of the infant



Acceptable values: FEMALE, MALE, UNSPECIFIED, UNKNOWN

ethnicity - The ethnicity of the infant

Acceptable values:

- HISPANIC\_LATINO\_OR\_SPANISH
- NOT\_HISPANIC\_LATINO\_OR\_SPANISH
- NOT\_REPORTED
- UNKNOWN

Note: only one value should be specified

**race** - the race of the infant Acceptable race values:

- ISLANDER
- ASIAN
- NATIVE AMERICAN
- BLACK\_OR\_AFRICAN\_AMERICAN
- WHITE
- UNKNOWN
- NOT REPORTED

Note: If more than one value applies, separate each value with a colon (e.g., ISALNDER:WHITE) Note: ISLANDER = Native Hawaiian or other Pacific Islander

# SCREENING INFORMATION

**screeningIdentifyingRisk** - The screening result which indicated this infant was at risk for the disorder. Acceptable values:

- Initial Screen
- Second Required Screen
- Subsequent Screen

**prenatalTestForRisk** - Was prenatal testing done that indicated that this infant was at risk for this disorder? Acceptable values: TRUE, FALSE, UNKNOWN

**familyHistoryRisk** - Was there a family history that indicated that this infant was at risk for this disorder? Acceptable values: TRUE, FALSE, UNKNOWN

**diagnosedAfterNewbornScreening** - Was this individual identified outside of newborn screening? Acceptable values: TRUE, FALSE, UNKNOWN

**missedDiagnosisReason** - The reason this diagnosis was not identified by newborn screening. *Note: should only be answered if diagnosedAfterNewbornScreening is TRUE* Acceptable values:

- Parental Refusal
- Lost to follow-up after unsatisfactory specimen
- Biologic false negative / result within normal range
- Did not have a valid screen due to error



• Other

**otherMissedDiagnosisReason** - Text description of the missed diagnosis reason up to 254 characters long. *Note: should only be answered if missedDiagnosisReason is OTHER* 

# **INITIAL SPECIMEN COLLECTION INFORMATION**

**birthToInitialSpecimenCollection** - hours between birth and initial specimen collection. Integer value. *Not specified for conditions "Critical congenital heart disease - CCHD", "Hearing loss - HEAR"* 

**birthToInitialSpecimenCollectionIncludesTime** - Acceptable values: TRUE, FALSE *Note: TRUE signifies that the data available for the calculation of elapsed time included time as well as date* 

**birthToInitialReceiptByLab** - Time elapsed from birth until the initial NBS specimen was received by the lab, in days (as measured by 24-hour periods since the birth). Integer value. *Not specified for conditions "Critical congenital heart disease - CCHD", "Hearing loss - HEAR"* 

**birthToInitialReceiptByLabIncludesTime** - Acceptable value: TRUE, FALSE *Note: TRUE* signifies that the data available for the calculation of elapsed time included time as well as date

**birthToInitialResultRelease** - Time elapsed from birth until the release of out-of-range results as a result of the initial screen, in days (as measured by 24-hour periods since the birth). *Not specified for conditions "Critical congenital heart disease - CCHD", "Hearing loss - HEAR"* 

**birthToInitialResultReleaseIncludesTime** - Acceptable value: TRUE, FALSE. *Note: TRUE signifies that the data available for the calculation of elapsed time included time as well as date* 

# SUBSEQUENT SPECIMEN COLLECTION INFORMATION

**birthToSubsequentSpecimenCollection** - Time elapsed from birth until the subsequent NBS specimen was collected, in days (as measured by 24-hour periods since the birth). *Not specified for conditions "Critical congenital heart disease - CCHD", "Hearing loss - HEAR"* 

**birthToSubsequentSpecimenCollectionIncludesTime** - Acceptable value: TRUE, FALSE *Note: TRUE signifies that the data available for the calculation of elapsed time included time as well as date* 



**birthToSubsequentReceiptByLab** - Time elapsed from birth until the subsequent NBS specimen was received by the lab, in days (as measured by 24-hour periods since the birth). *Not specified for conditions "Critical congenital heart disease - CCHD", "Hearing loss - HEAR"* 

**birthToSubsequentReceiptByLabIncludesTime** - Acceptable value: TRUE, FALSE *Note: TRUE signifies that the data available for the calculation of elapsed time included time as well as date* 

**birthToSubsequentResultRelease** - Time elapsed from birth until the release of out-of-range results as a result of the subsequent screen, in days (as measured by 24-hour periods since the birth). *Not specified for conditions "Critical congenital heart disease - CCHD", "Hearing loss - HEAR"* 

**birthToSubsequentResultReleaseIncludesTime** - Acceptable value: TRUE, FALSE. *Note: TRUE signifies that the data available for the calculation of elapsed time included time as well as date* 

# **POINT-OF-CARE TEST INFORMATION**

**birthToPointOfCareTestInterval** - Time elapsed from birth in hours until the point of care screening test was performed. *Only specified for conditions "Critical congenital heart disease - CCHD", "Hearing loss - HEAR"* 

**birthToPointOfCareTestIntervalIncludesTime** - Acceptable value: TRUE, FALSE Note: true signifies that the data available for the calculation of elapsed time included time as well as date. Only specified for conditions "Critical congenital heart disease - CCHD", "Hearing loss - HEAR"

## **INTERVENTION, FOLLOW-UP, AND DIAGNOSIS**

**birthToIntervention** - Time elapsed from birth until intervention by an appropriate medical provider occurred, in days (as measured by 24-hour periods since the birth)

**birthToDiagnosisConfirmation** - Time elapsed from birth until confirmation of the diagnosis occurred, in days (as measured by 24-hour periods since the birth)

**treatmentInOtherState** - Is infant receiving treatment/care out-of-state? Acceptable values: TRUE, FALSE, UNKNOWN

**treatmentState** - state where infant receives treatment/care? *Note: should only be answered if treatmentInOtherState is TRUE* Acceptable values: see list provided for **state** 

**diagnosisReversed** - Is this diagnosis reversed? Note: this does not refer to the therapeutic interventions to address a condition (i.e., surgery, treatment, therapy, etc) Acceptable values: TRUE, FALSE, UNKNOWN



**diagnosisReversedYear** - year diagnosis reversed (*note: enter four-digit year*) Note: should only be answered if diagnosisReversed is TRUE

## **FINAL DIAGNOSIS**

**finalDiagnosis** - final diagnosis as determined by the medical provider performing the clinical diagnostic workup, REQUIRED\*

Note: not all conditions require a final diagnosis; please use the table to see what conditions need a final diagnosis and the associated acceptable values. The final diagnosis categories do NOT include any of the secondary or other conditions listed on the RUSP. These should be entered as a separate case (see **conditions**).

Condition	Acceptable Values
3-Methylcrotonyl-CoA carboxylase deficiency - 3-	3-Methylcrotonyl-CoA Carboxylase Deficiency
	- 3-MCC
MCC	Maternal MCC deficiency
	MT-ATP6 related mitochondrial disorders
	Unknown
	Argininosuccinic Acidemia/ Aciduria (ASA)
Argininosuccinic aciduria - ASA	Pyruvate carboxylase deficiency
	Unknown     Distinidase deficiency
Pietinidase deficiency PIOT	Protound Biotinidase deficiency
Biolinidase deliciency - BIOT	Partial Biotinidase deficiency
	Olikilowii     Oitrullinomia Tuna I
Citrullinomia type I CIT	Circuinternia, Type I     Dyruwete eerbewyleese deficiency/
Citulinemia, type i - Ch	
	Carniting Untake Deficiency (CUD)
Carnitine untake defect/carnitine transport defect	Maternal Carnitine Deficiency (COD)
	secondary)
	Unknown
	Classic phenylketonuria - PKU
	Benign hyperphenylalaninemia - H-PHE
	HyperPhe diet controlled
	Dihydropterine reductase deficiency (DHPR)
Classic PKU & Hyperphe	DNAJC12
	Parenteral nutrition
	Maternal PKU
	Unknown
	Classic Galactosemia
Classic galactosemia - GALT	Duarte variant galactosemia
	Unknown
Congenital hypothyroidism - CH	Primary Congenital Hypothyroidism
	Secondary Congenital Hypothyroidism
	TBG Deficiency (Thyroxine Binding Globulin)
	or other protein binding defect
	Transient Congenital Hypothyroidism
	• Unknown



Congenital adrenal hyperplasia - CAH       • Classic 21-Hydroxylase Deficiency- Salt Wasting         Congenital adrenal hyperplasia - CAH       • Classic 21-Hydroxylase Deficiency- Simple Virilizing         Critical congenital heart disease - CCHD       • Other Adrenal disorder         Critical congenital heart disease - CCHD       • CCHD         Cystic fibrosis - CF       • CFTR-Related Disease         Cystic fibrosis - CF       • Holocarboxylase synthetase deficiency (MCD)         Holocarboxylase synthetase deficiency - MCD       • Holocarboxylase synthetase deficiency (MCD)         Holocarboxylase synthetase deficiency - MCD       • Holocarboxylase synthetase deficiency (MCD)         Homocystinuria - HCY       • Classic Homocystinuria         Homocystinuria - HCY       • Classic Homocystinuria         Homocystinuria - HCY       • Istovaleric Acidemia/ Aciduria (IVA)         Isovaleric acidemia - IVA       • Istovaleric Acidemia/ Aciduria (IVA)         Krabbe Disease       • Unknown         Krabbe Disease       • Unknown         • Classic       • Infantile Onset Krabbe Disease         • Unknown       • Classic         Maple syrup urine disease - MSUD       • Infantile Onset Krabbe Disease         • Hydroxyprolinemia       • Unclassified         • Hydroxyprolinemia       • Unknown         Maple syrup urine disease - MSUD	Condition	Acceptable Values
Congenital adrenal hyperplasia - CAH         Wasting           Classic 21-Hydroxylase Deficiency- Simple Virilizing         Other Adrenal disorder           Unknown         Unknown           Critical congenital heart disease - CCHD         Non critical CCHD           Cystic fibrosis - CF         CFTR-Related Metabolic Syndrome (CRMS)           Cystic fibrosis - CF         CFTR-Related Metabolic Syndrome (CRMS)           Holocarboxylase synthetase deficiency - MCD         Maternal 3-methylcrotonyl-CoA carboxylase deficiency (MCD)           Holocarboxylase synthetase deficiency - MCD         Maternal 3-methylcrotonyl-CoA carboxylase deficiency (MCD)           Homocystinuria - HCY         Classic Homocystinuria           Homocystinuria - HCY         Classic Addemial Aciduria (IVA)           Isovaleric acidemia - IVA         Isovaleric Acidemia/ Aciduria (IVA)           Isovaleric acidemia - IVA         Isovaleric Acidemia/ Aciduria (IVA)           Maple syrup urine disease - MSUD         Infantile Onset Krabbe Disease           Maple syrup urine disease - MSUD         Classic           Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT         Mutase (-) (mut-)           Methylmalonic acidemia (cobalamin disorders) - Mutase) - MUT         Chalastified	Concepital adrenal hyperplasia - CAH	Classic 21-Hydroxylase Deficiency- Salt
Congenital adrenal hyperplasia - CAH       • Classic 21-Hydroxylase Deficiency-Simple Virilizing         Critical congenital heart disease - CCHD       • CCHD         Critical congenital heart disease - CCHD       • CCHD         Cystic fibrosis - CF       • CFTR-Related Metabolic Syndrome (CRMS)         Cystic fibrosis - CF       • CFTR-Related Disease         Holocarboxylase synthetase deficiency - MCD       • Holocarboxylase synthetase deficiency (MCD)         Holocarboxylase synthetase deficiency - MCD       • Holocarboxylase synthetase deficiency (MCD)         Homocystinuria - HCY       • Classic Homocystinuria         Homocystinuria - HCY       • Classic Homocystinuria         Isovaleric acidemia - IVA       • Storubransferase (GNMT)         Krabbe Disease       • MSUD         Maple syrup urine disease - MSUD       • Classic Homocystinuria         Maple syrup urine disease - MSUD       • Classic         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       • Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - Unknown       • Unknown         • Unknown       • Classic       • Intermediate         Maple syrup urine disease - MSUD       • Classic       • Intermediate         Methylmalonic acidemia (cobalamin disorders) - Unknown       • Unknown       • Unknown         • Chalamin A deficiency (Cbl A)		Wasting
Sengeninal database (percent)       Vinitizing         Vinitizing       Other Adrenal disorder         Unknown       CCHD         Critical congenital heart disease - CCHD       Non critical CCHD         Other       Unknown         Cystic fibrosis - CF       CFTR-Related Metabolic Syndrome (CRMS)         Cystic fibrosis - CF       CFTR-Related Metabolic Syndrome (CRMS)         Holocarboxylase synthetase deficiency - MCD       Maternal 3-methylcrotonyl-CoA carboxylase deficiency (MCD)         Holocarboxylase synthetase deficiency - MCD       Maternal 3-methylcrotonyl-CoA carboxylase deficiency (MCD)         Homocystinuria - HCY       Classic Homocystinuria         Homocystinuria - HCY       Classic Homocystinuria         Isovaleric acidemia - IVA       Short/branched chain acyl-CoA dehydrogenase deficiency (BCAD) or 2-methyltransferase (GNMT)         Krabbe Disease       Infantile Onset Krabbe Disease         Krabbe Disease       Uncertain Type/Onset         Maple syrup urine disease - MSUD       Classic         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       Mutase (n) (mut)         Methylmalonic acidemia (cobalamin disorders) - CoA dendricency (CbI A)       Mutase (D) (mut)         Methylmalonic acidemia (cobalamin disorders) - CoA dehydrogenase deficiency       Unknown         Krabbe Disease - MSUD       Classified       Intermedi		Classic 21-Hydroxylase Deficiency- Simple
• Other Adrenal disorder         Unknown         Critical congenital heart disease - CCHD         Non critical CCHD         Other         Unknown         Cystic fibrosis - CF         Holocarboxylase synthetase deficiency - MCD         Homocystinuria - HCY         Homocystinuria - HCY         Isovaleric acidemia - IVA         Krabbe Disease         Maple synup urine disease - MSUD         Maple synup urine disease - MSUD         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT         Methylmalonic acidemia (cobalamin disorders) - MUT         Methylmalonic acidemia (cobalamin disorders) - MUT         Methylmalonic acidemia (cobalamin disorders) - COA         Methylmalonic acidemia (cobalamin disorders) - Clabality         Methylmalonic acidemia (cobalamin disorders) - Clabality         Methylmalonic acidemia (cobalamin disorders) - Clabality         Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (CDI A)         Chabality         Maple synup urine disease - MSUD         Maternal (cobalamin disorders) - Cobalamin A deficiency (CDI A) <td></td> <td>Virilizing</td>		Virilizing
• Unknown         Critical congenital heart disease - CCHD         Critical congenital heart disease - CCHD         • Non critical CCHD         • Unknown         Cystic fibrosis - CF         • CFTR-Related Metabolic Syndrome (CRMS)         • CFTR-Related Disease         • CFTR-Related Disease         • Holocarboxylase synthetase deficiency - MCD         • Holocarboxylase synthetase deficiency - MCD         • Homocystinuria - HCY         • Classic Homocystinuria         • Homocystinuria - HCY         • Stovaleric acidemia - IVA         • Isovaleric acidemia - IVA         • Isovaleric acidemia - IVA         • Infantile Onset Krabbe Disease         • Later Onset Krabbe Disease         • Later Onset Krabbe Disease         • Unknown         • Classic         Maple syrup urine disease - MSUD         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT         • Mutase (-) (mut-)         • Maple syrup urine disease - MSUD         • Classified         • Unknown         • Classified		Other Adrenal disorder
Critical congenital heart disease - CCHD       Non critical CCHD         Other       Unknown         Cystic fibrosis - CF       CFTR-Related Metabolic Syndrome (CRMS)         Cystic fibrosis - CF       CFTR-Related Disease         Holocarboxylase synthetase deficiency - MCD       Holocarboxylase synthetase deficiency (MCD)         Holocarboxylase synthetase deficiency - MCD       Maternal 3-methylcrotonyl-CoA carboxylase deficiency         Homocystinuria - HCY       Classic Homocystinuria         Homocystinuria - HCY       Classic Homocystinuria         Isovaleric acidemia - IVA       Elsovaleric Acidemia / Aciduria (IVA)         Short/branched chain acyl-CoA dehydrogenase Deficiency       Unknown         Krabbe Disease       Isovaleric Acidemia / Aciduria (IVA)         Maple syrup urine disease - MSUD       Infantile Onset Krabbe Disease         Maple syrup urine disease - MSUD       Mataeral (Ometa)         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       Mataeral vitamin B12 deficiency         Methylmalonic acidemia (cobalamin disorders) - Cobalamin & deficiency (CDI P)       Mataeral vitamin B12 deficiency         Methylmalonic acidemia (cobalamin disorders) - Cobalamin & deficiency (CDI P)       Succinate-CoA ligase deficiency         Maple syrup urine disease - MSUD       Mataeral vitamin B12 deficiency         Methylmalonic acidemia (methylmalonyl-CoA       Mutase (O)		Unknown
Critical congenital heart disease - CCHD       • Non Chitca CCHD         Other       • Unknown         Cystic fibrosis - CF       • CFTR-Related Metabolic Syndrome (CRMS)         France       • CFTR-Related Disease         Typical Cystic Fibrosis (CF)       • Unknown         Holocarboxylase synthetase deficiency - MCD       • Holocarboxylase synthetase deficiency (MCD)         Homocystinuria - HCY       • Holocarboxylase synthetase deficiency         Homocystinuria - HCY       • Classic Homocystinuria         Homocystinuria - HCY       • Classic Homocystinuria         Homocystinuria - HCY       • Isovaleric Acidemia / Aciduria (IVA)         Isovaleric acidemia - IVA       • Isovaleric Acidemia/ Aciduria (IVA)         Krabbe Disease       • Infantile Onset Krabbe Disease         Krabbe Disease       • Unknown         Maple syrup urine disease - MSUD       • Classic         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       • Mutase (0) (mut0)         Methylmalonic acidemia (cobalamin disorders) - CoAlamin A deficiency (CbI A)       • Mutase (0) (Mut0)         Methylmalonic acidemia (cobalamin disorders) - CoAlamin A deficiency (CbI A)       • Cobalamin A deficiency (CbI A)		
Outer         Unknown         Unknown         Cystic fibrosis - CF         Holocarboxylase synthetase deficiency - MCD         Homocystinuria - HCY         Homocystinuria - HCY         Homocystinuria - HCY         Sovaleric acidemia - IVA         Homocystinuria - HCY         Isovaleric acidemia - IVA         Rabe Disease         Maternal S-methylicotonyl-CoA         Homocystinuria - HCY         Homocystinuria - HCY         Homocysteric acidemia - IVA         Sovaleric acidemia - IVA         Isovaleric acidemia - IVA         Homocysteric Acidemia / Aciduria (IVA)         Short/branched chain acyl-CoA         dehydrogenase Deficiency (SBCAD) or 2- methylbutyri CoA dehydrogenase deficiency         Unknown         Isovaleric acidemia - IVA         Maple syrup urine disease - MSUD         Maple syrup urine disease - MSUD         Maple syrup urine disease - MSUD         Methylinatonic acidemia (methylmatonyl-CoA         Methylimatonic acidemia (methylmatonyl-CoA	Critical congenital heart disease - CCHD	
Cystic fibrosis - CF       CFTR-Related Metabolic Syndrome (CRMS)         Cystic fibrosis - CF       CFTR-Related Disease         Holocarboxylase synthetase deficiency - MCD       Unknown         Holocarboxylase synthetase deficiency - MCD       Maternal 3-methylcrotonyl-CoA carboxylase deficiency         Holocarboxylase synthetase deficiency - MCD       Maternal 3-methylcrotonyl-CoA carboxylase deficiency         Homocystinuria - HCY       Classic Homocystinuria         Homocystinuria - HCY       Classic Homocystinuria         Isovaleric acidemia - IVA       Glycine n-methyltransferase (GNMT)         Adenosylhomocysteine Hydrolase Deficiency       Unknown         Isovaleric acidemia - IVA       Isovaleric Acidemia/ Aciduria (IVA)         Short/branched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2-methylbutyl CoA dehydrogenase Deficiency (SBCAD) or 2-methylbutyl CoA dehydrogenase deficiency         Maple syrup urine disease - MSUD       Infartile Onset Krabbe Disease         Maple syrup urine disease - MSUD       Intermediate         Maternal (methylmalonyl-CoA mutase) - MUT       Mutase (0) (mut0)         Methylmalonic acidemia (cobalamin disorders) - CoA lagase deficiency       Unknown         Classified       Unknown         Maple syrup urine disease - MSUD       Mutase (0) (mut0)         Methylmalonic acidemia (methylmalonyl-CoA       Mutase (0) (mut0) <td< td=""><td>, , , , , , , , , , , , , , , , , , ,</td><td></td></td<>	, , , , , , , , , , , , , , , , , , ,	
Cystic fibrosis - CF       CFTR-Related Disease         Ypical Cystic Fibrosis (CF)       Unknown         Holocarboxylase synthetase deficiency - MCD       Holocarboxylase synthetase deficiency (MCD)         Maternal 3-methylorotonyl-CoA carboxylase deficiency       MT-ATP6 related mitochondrial disorders         Other biotin disorder       Unknown         Homocystinuria - HCY       Classic Homocystinuria         Homocystinuria - HCY       Classic Homocystinuria         Isovaleric acidemia - IVA       Classic Homocystinuria         Krabbe Disease       Isovaleric Acidemia / Aciduria (IVA)         Krabbe Disease       Isovaleric Acidemia / Aciduria (IVA)         Maple syrup urine disease - MSUD       Classic         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       Maternal vitamin B12 deficiency         Methylmalonic acidemia (cobalamin disorders) - Unknown       Mutase (0) (mut)         Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)       Classified         Unclassified       Unknown       Mutase (0) (mut0)         Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)       Cobalamin A deficiency (Cbl A)		CETR Related Metabolic Syndrome (CRMS)
Cystic fibrosis - CF       • Of Tirkindo Disease         • Typical Cystic Fibrosis (CF)         • Holocarboxylase synthetase deficiency - MCD         • Holocarboxylase synthetase deficiency - MCD         • Holocarboxylase synthetase deficiency - MCD         • Maternal 3-methylcrotonyl-CoA carboxylase deficiency         • MT-ATP6 related mitochondrial disorders         • Other biotin disorder         • Unknown         • Classic Homocystinuria         Homocystinuria - HCY         • Isovaleric acidemia - IVA         • Isovaleric acidemia - IVA         • Isovaleric acidemia - IVA         • Infantile Onset Krabbe Disease         • Unknown         • Classic         • Intermediate         • Typical cystic (mut-)         • Watese (-) (mut-)         • Unknown         • Intermediate         • Intermediate         • Hydroxprolinemia         • Unclassified         • Unknown         • M		CFTR-Related Disease
Holocarboxylase synthetase deficiency - MCD       • Holocarboxylase synthetase deficiency (MCD)         Holocarboxylase synthetase deficiency - MCD       • Holocarboxylase synthetase deficiency (MCD)         Maternal 3-methylcrotonyl-CoA carboxylase deficiency       • MT-ATP6 related mitochondrial disorders         • Unknown       • Unknown         Classic Homocystinuria       • Methionine Adenosyltransferase (MAT I/III Deficiency)         Homocystinuria - HCY       • Classic Homocystinuria         Isovaleric acidemia - IVA       • Isovaleric Acidemia/ Aciduria (IVA)         Short/branched chain acyl-CoA dehydrogenase Deficiency       • Unknown         Isovaleric acidemia - IVA       • Isovaleric Acidemia/ Aciduria (IVA)         Short/branched chain acyl-CoA dehydrogenase deficiency       • Unknown         Krabbe Disease       • Infantile Onset Krabbe Disease         Krabbe Disease       • Unknown         Maple syrup urine disease - MSUD       • Classic         Maple syrup urine disease - MSUD       • Classic         Maple syrup urine disease - MSUD       • Mutase (-) (mut-)         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       • Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - CoA ilgase deficiency       • Unknown         • Unknown       • Classified       • Unknown         • Classified       • Unknown <td>Cystic fibrosis - CF</td> <td>Typical Cystic Eibrosis (CE)</td>	Cystic fibrosis - CF	Typical Cystic Eibrosis (CE)
Holocarboxylase synthetase deficiency (MCD)         Holocarboxylase synthetase deficiency (MCD)         Maternal 3-methylcrotonyl-CoA carboxylase deficiency         MT-ATP6 related mitochondrial disorders         Other biotin disorder         Unknown         Classic Homocystinuria         Homocystinuria - HCY         Glycine n-methyltransferase (MAT I/III Deficiency)         Glycine n-methyltransferase (GNMT)         Adenosylhomocysteine Hydrolase Deficiency         Unknown         Isovaleric acidemia - IVA         Krabbe Disease         Maple syrup urine disease - MSUD         Mathylmalonic acidemia (methylmalonyl-CoA mutase) - MUT         Methylmalonic acidemia (cobalamin disorders) - Chl A B		
Holocarboxylase synthetase deficiency - MCD       • Mdternal 3-methylcrotonyl-CoA carboxylase         Holocarboxylase synthetase deficiency - MCD       • Mdternal 3-methylcrotonyl-CoA carboxylase         deficiency       • MT-ATP6 related mitochondrial disorders         • Other biotin disorder       • Unknown         Classic Homocystinuria       • Classic Homocystinuria         Homocystinuria - HCY       • Classic Homocystinuria         Isovaleric acidemia - IVA       • Short/branched chain acyl-CoA         Isovaleric acidemia - IVA       • Isovaleric Acidemia/ Aciduria (IVA)         Krabbe Disease       • Infantile Onset Krabbe Disease         Krabbe Disease       • Infantile Onset Krabbe Disease         Maple syrup urine disease - MSUD       • Classic         Methylmalonic acidemia (methylmalonyl-CoA       • Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - (CDI A)       • Cobalamin A deficiency (CDI A)         Methylmalonic acidemia (cobalamin disorders) - COA       • Unknown		<ul> <li>Unknown</li> <li>Heleserbevylese synthetese deficiency (MCD)</li> </ul>
Holocarboxylase synthetase deficiency - MCD       Materinal S-Interlifetolohyl-COA Carboxylase         Holocarboxylase synthetase deficiency - MCD       MT-ATP6 related mitochondrial disorders         Homocystinuria - HCY       Classic Homocystinuria         Homocystinuria - HCY       Classic Homocystinuria         Isovaleric acidemia - IVA       Short/branched chain acyl-CoA         Isovaleric acidemia - IVA       Isovaleric Acidemia/Aciduria (IVA)         Krabbe Disease       Infantile Onset Krabbe Disease         Unknown       Infantile Onset Krabbe Disease         Krabbe Disease       Later Onset Krabbe Disease         Maple syrup urine disease - MSUD       Classic         Methylmalonic acidemia (methylmalonyl-CoA       Unknown         Methylmalonic acidemia (cobalamin disorders) - MUT       Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)       Cobalamin A deficiency (Cbl A)		<ul> <li>Holocal boxylase synthetase denciency (MCD)</li> <li>Meternal 2 methylaratanyl CoA parhayllase</li> </ul>
Honocarboxylase synthetase deliciency - MCD <ul> <li>Mathematical deliciency - MCD</li> <li>Mathematical deliciency</li> <li>MT-ATP6 related mitochondrial disorders</li> <li>Other biotin disorder</li> <li>Unknown</li> </ul> Homocystinuria - HCY <ul> <li>Classic Homocystinuria</li> <li>Methionine Adenosyltransferase (MAT I/III)</li> <li>Deficiency)</li> <li>Glycine n-methyltransferase (GNMT)</li> <li>Adenosylhomocysteine Hydrolase Deficiency</li> <li>Unknown</li> </ul> Isovaleric acidemia - IVA <ul> <li>Isovaleric Acidemia/Aciduria (IVA)</li> <li>Short/branched chain acyl-CoA</li> <li>dehydrogenase Deficiency (SBCAD) or 2-methylbutyrl CoA dehydrogenase deficiency</li> <li>Unknown</li> </ul> Krabbe Disease <ul> <li>Infantile Onset Krabbe Disease</li> <li>Later Onset Krabbe Disease</li> <li>Uncertain Type/Onset</li> <li>Unclassified</li> <li>Unknown</li> </ul> Maple syrup urine disease - MSUD <ul> <li>Mutase (-) (mut.)</li> <li>Mutase (-) (mut.)</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Unclassified</li> <li>Unknown</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Unclassified</li> <li>Unknown</li> </ul>	Listeenthewylese synthetees deficiency MCD	<ul> <li>Maternal 5-methylcrotonyi-CoA carboxylase</li> <li>deficiency</li> </ul>
• MI - A IPb related mitochondrial disorders         • Other biotin disorder         • Uhknown         • Classic Homocystinuria         • Methionine Adenosyltransferase (MAT I/III Deficiency)         • Glycine n-methyltransferase (GNMT)         • Adenosylhomocysteine Hydrolase Deficiency         • Uhknown         Isovaleric acidemia - IVA         * Isovaleric Acidemia / Aciduria (IVA)         • Short/branched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2-methylbutyrl CoA dehydrogenase deficiency         • Unknown         Krabbe Disease         Maple syrup urine disease - MSUD         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT         Methylmalonic acidemia (cobalamin disorders) - MUT         Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (CbI A)         • Other solution of the other is a cidemia (cobalamin disorders) - Cobalamin A deficiency (CbI A)	Holocarboxylase synthetase deliciency - MCD	denciency
<ul> <li>Other bloth disorder</li> <li>Unknown</li> <li>Classic Homocystinuria</li> <li>Methionine Adenosyltransferase (MAT I/III Deficiency)</li> <li>Glycine n-methyltransferase (GNMT)</li> <li>Adenosylhomocysteine Hydrolase Deficiency</li> <li>Unknown</li> <li>Isovaleric acidemia - IVA</li> <li>Isovaleric Acidemia/ Aciduria (IVA)</li> <li>Short/branched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2- methylbutyrl CoA dehydrogenase deficiency</li> <li>Unknown</li> <li>Infantile Onset Krabbe Disease</li> <li>Later Onset Krabbe Disease</li> <li>Uncertain Type/Onset</li> <li>Unknown</li> <li>Classic</li> <li>Intermediate</li> <li>Thiamine-response</li> <li>Hydroxyprolinemia</li> <li>Unclassified</li> <li>Unknown</li> <li>Mutase (-) (mut-)</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Unclassified</li> <li>Unknown</li> </ul>		MI-ATP6 related millochondrial disorders
<ul> <li>Unknown</li> <li>Classic Homocystinuria</li> <li>Classic Homocystinuria</li> <li>Methionine Adenosyltransferase (MAT I/III Deficiency)</li> <li>Glycine n-methyltransferase (GNMT)</li> <li>Adenosylhomocysteine Hydrolase Deficiency</li> <li>Unknown</li> <li>Isovaleric Acidemia / Aciduria (IVA)</li> <li>Short/branched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2- methylbutyrl CoA dehydrogenase deficiency</li> <li>Unknown</li> <li>Infantile Onset Krabbe Disease</li> <li>Later Onset Krabbe Disease</li> <li>Uncertain Type/Onset</li> <li>Unknown</li> <li>Classic</li> <li>Intermediate</li> <li>Thiamine-response</li> <li>Hydroxyprolinemia</li> <li>Unclassified</li> <li>Unknown</li> <li>Mutase (-) (mut-)</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Unclassified</li> <li>Unknown</li> </ul>		Other blotin disorder
<ul> <li>Classic Homocystinuria</li> <li>Methionine Adenosyltransferase (MAT I/III Deficiency)</li> <li>Glycine n-methyltransferase (GNMT)</li> <li>Adenosylhomocysteine Hydrolase Deficiency</li> <li>Unknown</li> <li>Isovaleric acidemia - IVA</li> <li>Short/Vranched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2- methylbutyrl CoA dehydrogenase deficiency</li> <li>Unknown</li> <li>Infantile Onset Krabbe Disease</li> <li>Uncertain Type/Onset</li> <li>Unknown</li> <li>Classic</li> <li>Intermediate</li> <li>Thiamine-response</li> <li>Hydroxyprolinemia</li> <li>Unclassified</li> <li>Unknown</li> <li>Mutase (-) (mut.)</li> <li>Mut</li></ul>		
Homocystinuria - HCY       Methonine Adenosyltransferase (MA1 1/III Deficiency)         Homocystinuria - HCY       Glycine n-methyltransferase (GNMT) Adenosylhomocysteine Hydrolase Deficiency Unknown         Isovaleric acidemia - IVA       Isovaleric Acidemia/ Aciduria (IVA) Short/branched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2- methylbutyrl CoA dehydrogenase deficiency Unknown         Krabbe Disease       Infantile Onset Krabbe Disease Uncertain Type/Onset         Maple syrup urine disease - MSUD       Infantile Onset Krabbe Disease Uncertain Type/Onset         Maple syrup urine disease - MSUD       Infantile Onset Krabbe Disease Uncertain Type/Onset         Maple syrup urine disease - MSUD       Maternal vitamine-response Hydroxyprolinemia Unclassified         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       Mutase (-) (mut-) Mutase (0) (mut0)         Methylmalonic acidemia (cobalamin disorders) - Chl A B       Cobalamin A deficiency (Cbl A) Cobalamin A deficiency (Cbl A)		Classic Homocystinuria
Homocystinuria - HCY <ul> <li>Glycine n-methyltransferase (GNMT)</li> <li>Adenosylhomocysteine Hydrolase Deficiency</li> <li>Unknown</li> </ul> Isovaleric acidemia - IVA <ul> <li>Isovaleric Acidemia/ Aciduria (IVA)</li> <li>Short/branched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2- methylbutyrl CoA dehydrogenase deficiency</li> <li>Unknown</li> </ul> Krabbe Disease <ul> <li>Infantile Onset Krabbe Disease</li> <li>Later Onset Krabbe Disease</li> <li>Uncertain Type/Onset</li> <li>Unknown</li> </ul> Maple syrup urine disease - MSUD <ul> <li>Classic</li> <li>Intermediate</li> <li>Thiamine-response</li> <li>Hydroxyprolinemia</li> <li>Unclassified</li> <li>Unknown</li> </ul> Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT <ul> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Unclassified</li> <li>Unknown</li> </ul> Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)		Methionine Adenosyltransferase (MAT I/III     Definition on )
<ul> <li>Glyche n-methyltransterase (GNMT)</li> <li>Adenosylhomocysteine Hydrolase Deficiency</li> <li>Unknown</li> <li>Isovaleric Acidemia / Aciduria (IVA)</li> <li>Short/branched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2- methylbutyrl CoA dehydrogenase deficiency</li> <li>Unknown</li> <li>Infantile Onset Krabbe Disease</li> <li>Later Onset Krabbe Disease</li> <li>Uncertain Type/Onset</li> <li>Unknown</li> <li>Classic</li> <li>Intermediate</li> <li>Thiamine-response</li> <li>Hydroxyprolinemia</li> <li>Unclassified</li> <li>Unknown</li> <li>Mutase (-) (mut-)</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Unclassified</li> <li>Unknown</li> </ul>	Homocystinuria - HCY	Deficiency)
Adenosylhomocysteine Hydrolase Dericlency         Unknown         Isovaleric acidemia - IVA         Isovaleric Acidemia/Aciduria (IVA)         Isovaleric Acidemia/Aciduria (IVA)         Isovaleric Acidemia/Aciduria (IVA)         Isovaleric Acidemia - IVA         Isovaleric Acidemia/Aciduria (IVA)         Isovaleric Acidemia - IVA         I		Glycine n-methyltransferase (GNMT)
Isovaleric acidemia - IVA       Isovaleric Acidemia/ Aciduria (IVA)         Isovaleric acidemia - IVA       Isovaleric Acidemia/ Aciduria (IVA)         Short/branched chain acyl-CoA       dehydrogenase Deficiency (SBCAD) or 2-methylbutyrl CoA dehydrogenase deficiency         Krabbe Disease       Unknown         Krabbe Disease       Infantile Onset Krabbe Disease         Uncertain Type/Onset       Uncertain Type/Onset         Unknown       Classic         Maple syrup urine disease - MSUD       Classic         Maple syrup urine disease - MSUD       Classic         Maple syrup urine disease - MSUD       Mutase         Methylmalonic acidemia (methylmalonyl-CoA       Mutase		Adenosylnomocysteine Hydrolase Deliciency
Isovaleric Acidemia/ Aciduma (IVA)         Isovaleric Acidemia/ Aciduma (IVA)         Isovaleric Acidemia/ Aciduma (IVA)         Short/branched chain acyl-CoA dehydrogenase Deficiency (SBCAD) or 2- methylbutyrl CoA dehydrogenase deficiency         Krabbe Disease         Krabbe Disease         Maple syrup urine disease - MSUD         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT         Methylmalonic acidemia (cobalamin disorders) - ChL A B         Methylmalonic acidemia (cobalamin disorders) - ChL A B		Unknown     Lassalaria Asialaraia ( Asialuria (I) (A)
<ul> <li>Snort/branched chain ac/J-COA dehydrogenase Deficiency (SBCAD) or 2- methylbutyrl CoA dehydrogenase deficiency Unknown</li> <li>Infantile Onset Krabbe Disease</li> <li>Later Onset Krabbe Disease</li> <li>Uncertain Type/Onset</li> <li>Unknown</li> <li>Classic</li> <li>Intermediate</li> <li>Thiamine-response</li> <li>Hydroxyprolinemia</li> <li>Unclassified</li> <li>Unknown</li> <li>Mutase (-) (mut-)</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Unclassified</li> <li>Unknown</li> </ul>		<ul> <li>Isovaleric Acidemia/ Aciduria (IVA)</li> <li>Chart/knowshad a bain and Cat</li> </ul>
Maple syrup urine disease - MSUD <ul> <li>Maple syrup urine disease - MSUD</li> <li>Maple syrup urine disease - MSUD</li> <li>Maple syrup urine disease - MSUD</li> </ul> <ul> <li>Infantile Onset Krabbe Disease</li> <li>Later Onset Krabbe Disease</li> <li>Uncertain Type/Onset</li> <li>Unknown</li> </ul> Maple syrup urine disease - MSUD <ul> <li>Classic</li> <li>Intermediate</li> <li>Thiamine-response</li> <li>Hydroxyprolinemia</li> <li>Unclassified</li> <li>Unknown</li> </ul> Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT <ul> <li>Mutase (-) (mut-)</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Succinate-CoA ligase deficiency</li> <li>Unclassified</li> <li>Unknown</li> </ul>	Isovaleric acidemia - IVA	<ul> <li>Snort/branched chain acyl-CoA debydrogonoso Deficioney (SBCAD) or 2</li> </ul>
Krabbe Disease       • Unknown         Krabbe Disease       • Later Onset Krabbe Disease         • Unknown       • Uncertain Type/Onset         • Unknown       • Unknown         Maple syrup urine disease - MSUD       • Classic         • Intermediate       • Thiamine-response         • Hydroxyprolinemia       • Unclassified         • Unknown       • Unknown         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       • Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)       • Cobalamin A deficiency (Cbl A)		methylbutyrl CoA dehydrogenase deficiency
Krabbe Disease       Infantile Onset Krabbe Disease         Later Onset Krabbe Disease       Uncertain Type/Onset         Uncertain Type/Onset       Unknown         Classic       Intermediate         Thiamine-response       Hydroxyprolinemia         Unclassified       Unknown         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - Chl A B       Succinate-CoA ligase deficiency (Cbl A)		Unknown
Krabbe Disease       Later Onset Krabbe Disease         Maple syrup urine disease - MSUD       Classic         Intermediate       Thiamine-response         Hydroxyprolinemia       Unclassified         Unknown       Unclassified         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - Chl A B       Succinate-CoA ligase deficiency (Cbl A)		Infantile Onset Krabbe Disease
Krabbe Disease       Uncertain Type/Onset         Uncertain Type/Onset       Uncertain Type/Onset         Unknown       Classic         Intermediate       Thiamine-response         Hydroxyprolinemia       Unclassified         Unknown       Mutase (-) (mut-)         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       Mutase (0) (mut0)         Methylmalonic acidemia (cobalamin disorders) - Chl A B       Cobalamin A deficiency (Cbl A)		Later Onset Krabbe Disease
<ul> <li>Maple syrup urine disease - MSUD</li> <li>Classic</li> <li>Intermediate</li> <li>Thiamine-response</li> <li>Hydroxyprolinemia</li> <li>Unclassified</li> <li>Unknown</li> <li>Mutase (-) (mut-)</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Succinate-CoA ligase deficiency</li> <li>Unclassified</li> <li>Unclassified</li> <li>Unknown</li> </ul>	Krabbe Disease	Uncertain Type/Onset
<ul> <li>Maple syrup urine disease - MSUD</li> <li>Classic</li> <li>Intermediate</li> <li>Thiamine-response</li> <li>Hydroxyprolinemia</li> <li>Unclassified</li> <li>Unknown</li> <li>Mutase (-) (mut-)</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Succinate-CoA ligase deficiency</li> <li>Unclassified</li> <li>Unclassified</li> <li>Unclassified</li> <li>Unknown</li> <li>Methylmalonic acidemia (cobalamin disorders) -</li> <li>Cobalamin A deficiency (Cbl A)</li> <li>Cobalamin A deficiency (Cbl A)</li> </ul>		
Maple syrup urine disease - MSUD       Intermediate         Maple syrup urine disease - MSUD       Thiamine-response         Hydroxyprolinemia       Unclassified         Unclassified       Unknown         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - Chl A B       Succinate-CoA ligase deficiency (Cbl A)		
Maple syrup urine disease - MSUD       • Thiamine-response         • Hydroxyprolinemia       • Unclassified         • Unclassified       • Unknown         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       • Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)       • Cobalamin A deficiency (Cbl A)		Intermediate
Maple syrup urine disease - MSUD       Hindmine roopende         Hydroxyprolinemia       Unclassified         Unclassified       Unknown         Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       Mutase (-) (mut-)         Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)       Unclassified		Thiamine-response
<ul> <li>Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT</li> <li>Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)</li> <li>Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)</li> </ul>	Maple syrup urine disease - MSUD	Hydroxyprolinemia
<ul> <li>Onecosined</li> <li>Unknown</li> <li>Mutase (-) (mut-)</li> <li>Mutase (0) (mut0)</li> <li>Maternal vitamin B12 deficiency</li> <li>Succinate-CoA ligase deficiency</li> <li>Unclassified</li> <li>Unknown</li> <li>Methylmalonic acidemia (cobalamin disorders) -</li> <li>Cobalamin A deficiency (Cbl A)</li> <li>Cobalamin B deficiency (Cbl A)</li> </ul>		Inclassified
Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT Mutase (-) (mut-) Mutase (0) (mut0) Maternal vitamin B12 deficiency Succinate-CoA ligase deficiency Unclassified Unknown Methylmalonic acidemia (cobalamin disorders) - Cbl A B Cobalamin A deficiency (Cbl A)		Unknown
Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT Methylmalonic acidemia (cobalamin disorders) - Chl A B Mutase (0) (mut0) Mutase (0	Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT	Mutase (-) (mut-)
Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT       • Maternal vitamin B12 deficiency         • Mutase (o) (mido)       • Maternal vitamin B12 deficiency         • Mutase (o) (mido)       • Maternal vitamin B12 deficiency         • Mutase (o) (mido)       • Maternal vitamin B12 deficiency         • Mutase (o) (mido)       • Maternal vitamin B12 deficiency         • Mutase (o) (mido)       • Succinate-CoA ligase deficiency         • Unclassified       • Unknown         Methylmalonic acidemia (cobalamin disorders) - Cobalamin A deficiency (Cbl A)       • Cobalamin B deficiency (Cbl A)		<ul> <li>Mutase (0) (mut0)</li> </ul>
mutase) - MUT     • Succinate-CoA ligase deficiency       • Unclassified     • Unknown       Methylmalonic acidemia (cobalamin disorders) - Cbl A B     • Cobalamin A deficiency (Cbl A)		Maternal vitamin B12 deficiency
<ul> <li>Methylmalonic acidemia (cobalamin disorders) -</li> <li>Cobalamin A deficiency (Cbl A)</li> <li>Cobalamin B deficiency (Cbl B)</li> </ul>		Succinate-CoA linase deficiency
Methylmalonic acidemia (cobalamin disorders) - Chl A B Cobalamin R deficiency (Chl R) Cobalamin R deficiency (Chl R)		
Methylmalonic acidemia (cobalamin disorders) - Chl A B Cobalamin R deficiency (Chl R) Cobalamin R deficiency (Chl R)		
Chi A B	Methylmalonic acidemia (cobalamin disorders)	Cobalamin A deficiency (Cbl A)
	Cbl A.B	Cobalamin R deficiency (Cbl R)



Condition	Acceptable Values
	Cobalamin Dv2 deficiency (Cbl Dv2)
	Maternal vitamin B12 deficiency
	Succinate-CoA ligase deficiency
	Cobalamin C deficiency (Cbl C)
	<ul> <li>Cobalamin D deficiency (Cbl D)</li> </ul>
	<ul> <li>Cobalamin E deficiency (Cbl E)</li> </ul>
	<ul> <li>Cobalamin T deficiency (Cbl Dv1)</li> <li>Cobalamin Dv1 deficiency (Cbl Dv1)</li> </ul>
Methylmalonic acidemia with homocystinuria - Chl	Cobalamin L deficiency (Cbl L)
	Maternal vitamin B12 deficiency
0,2	<ul> <li>Succinate-CoA ligase deficiency</li> </ul>
	Other cobalamin deficiency
	Unclassified
	Unknown
	MPS I - severe
	MPS I - severity not determined
Mucopolysaccharidosis I - MPS I	<ul> <li>MPS I - attenuated</li> </ul>
	Uncertain Type/Onset
	Unknown
	Severe
Mucopolysaccharidosis II - MPS II	Attenuated
	Uncertain Type/Onset
	Unknown
	Infantile Onset (IO) Pompe Disease
	Late Onset (LO) Pompe Disease
Pompe	Uncertain Type/Onset
	• Unknown
	S.S disease (Sickle cell anemia) - Hb SS
	<ul> <li>S. Beta 0-thalassemia - Hb S/B0Th</li> </ul>
	<ul> <li>S, Beta + thalassemia - Hb S/B+ Th</li> </ul>
Presence of HD S	• S,C disease - Hb S/C
	• S, Other
	Unknown
	Hemoglobin C disease
Presence of Other Hb Variant	Hemoglobin D disease
	Hemoglobin E disease
	Hemoglobin O-Arab disease
	Other hemoglobin disorder
	Unknown
Hb - No structural variant	Alpha thalassemia major (Fetal Hydrops)
	Beta thalassemia major (Cooley's anemia)
	Hgb H disease
	Unknown
Propionic acidemia - PROP	Propionic Acidemia (PROP)
	Maternal vitamin B12 deficiency
	Succinate-CoA ligase deficiency
	Unknown
Severe Combined Immunodeficiencies - SCID	Classic SCID
	Leaky SCID
	Omenn Svndrome



Condition	Acceptable Values
	Unknown
Tyrosinemia, type I - TYR I	<ul> <li>Tyrosinemia, Type I (hepatorenal)</li> <li>Transient Tyrosinemia of the neonate (TTN)</li> <li>Unknown</li> </ul>
X-linked Adrenoleukodystrophy	<ul> <li>X-Linked Adrenoleukodystrophy (in Males)</li> <li>X-Linked Adrenoleukodystrophy (in Females)</li> <li>Contiguous ABCD1 DXS1357E deletion syndrome (CADDS)</li> <li>Peroxisomal Disorder</li> <li>Acyl-CoA Oxidase Deficiency</li> <li>D-Bifunctional Protein Deficiency</li> <li>Dyamin-like protein 1 (DLP1)</li> <li>ABDC5</li> <li>Non-peroxisomal Disorder</li> <li>Uncertain Type/Onset</li> <li>Unknown</li> </ul>

**otherFinalDiagnosisName -** Specify the name for the other final diagnosis when the value "OTHER" is entered for *finalDiagnosis* 

## If condition is Presence of Other Hb Variant

**alphaThalassemiaPresent**- Alpha thalassemia present? Acceptable values: TRUE, FALSE, UNKNOWN *Note: must only be entered when condition is "Presence of Other Hb Variant"* 

#### If condition is Critical congenital heart disease - CCHD and finalDiagnosis is CCHD

cchdFinalDiagnosesDetails- Specify type of CCHD diagnosed.

Acceptable values:

- TRUNCUS\_ARTERIOSUS
- TOTAL\_ANOMALOUS\_PULMONARY\_VENOUS\_CONNECTION
- TETRALOGY\_OF\_FALLOT
- PULMONARY\_ATRESIA
- EBSTEIN\_ANOMALY
- HYPOPLASTIC\_LEFT\_HEART\_SYNDROME
- SINGLE\_VENTRICLE
- TRICUSPID\_ATRESIA
- TRANSPOSITION\_OF\_GREAT\_ARTERIES
- DOUBLE\_OUTLET\_RIGHT\_VENTRICLE
- COARCTATION\_OF\_AORTA
- INTERRUPTED\_AORTIC\_ARCH
- AORTIC\_VALVE\_DISEASE



Note: must only be entered when CCHD FinalDiagnosis is CCHD; can add multiple selections by using a colon to separate each acceptable value (e.g., TRUNCUS\_ARTERIOSUS:PULMONARY ATRESIA:SINGLE\_VENTRICLE)

If condition is Spinal Muscular Atrophy - SMA

**newbornSMN2MolecularTest** - newborn screen molecular test for SMN2? Acceptable values: TRUE, FALSE, UNKNOWN *Note: only enter if condition is "Spinal Muscular Atrophy - SMA"* 

**newbornSMN2MolecularTestValue** - SMN2 copy number? Acceptable values: ONE, TWO, TWO\_OR\_MORE, UNKNOWN Note: only enter if condition is Spinal Muscular Atrophy - SMA" and newbornSMN2MolecularTest is TRUE

**postNewbornSMN2MolecularTest** - post-newborn screen molecular test for SMN2? Acceptable values: TRUE, FALSE, UNKNOWN *Note: only enter if condition is "Spinal Muscular Atrophy - SMA"* 

**postNewbornSMN2MolecularTestValue** - SMN2 copy number? Acceptable values: ONE, TWO, TWO\_OR\_MORE, UNKNOWN *Note: only enter if condition is Spinal Muscular Atrophy - SMA" and postNewbornSMN2MolecularTest is TRUE* 

# **CHANGE LOG**

#### Modifications made from August 2023 version

- Removed all diagnostic variables
  - Note: Collecting diagnostic information was well intended to standardize the identification and classification of disorders, aligning with the <u>public health</u> <u>surveillance case definitions</u> for newborns in the United States. However, it was decided that the cons (i.e., staff time, poor feedback loop with clinicians, inability to obtain all diagnostic information to feed into the classification system) outweigh the benefits of collecting this information. NewSTEPs will continue to collect demographic and newborn screening information for individual cases, which will continue to help inform our birth prevalence, health equity, and quality improvement practices.

#### Modifications made from January 2024 version

• Updated spelling from Holocarboxylase synthase deficiency - MCD to Holocarboxylase synthetase deficiency - MCD

#### Modifications made from March 2024 version



• Updated language for diagnosedAfterNewbornScreening to clarify that the diagnosis was made outside of newborn screening. *Note: this was just a change to the variable label and does not impact queries.* 

#### Modifications from May 2024 version

- Final diagnosis category is now a required field
- Unknown was added to the final diagnosis for all conditions that have a final diagnosis
  option

#### Modifications from June 2024 version

- In the case template map, separated NOT\_REPORTED and UNKNOWN for ethnicity. This was a typo only in the case template map as these two categories were accidentally on the same line.
- Updated spelling from Cobalamin Dv2 (CbIDv2) to Cobalamin Dv2 deficiency (CbIDv2).
- Updated spacing for the following final diagnosis options to accept the following values:
  - Classic 21-Hydroxylase Deficiency- Salt Wasting
  - Classic 21-Hydroxylase Deficiency- Simple Virilizing
  - Cobalamin A deficiency (Cbl A)
  - Cobalamin B deficiency (Cbl B)
  - Cobalamin C deficiency (Cbl C)
  - Cobalamin D deficiency (Cbl D)
  - Cobalamin Dv1 deficiency (Cbl Dv1)
  - Cobalamin Dv2 deficiency (Cbl Dv2)
  - Cobalamin F deficiency (Cbl F)
  - Cobalamin J deficiency (Cbl J)
  - S,C disease Hb S/C
  - S,S disease (Sickle cell anemia) Hb SS
- Updated formatting for the following condition options to accept the following values:
  - Argininosuccinic aciduria ASA
  - o Beta-Ketothiolase deficiency BKT
  - Biotinidase deficiency BIOT
  - Carbamoyl phosphate synthetase I deficiency CPS
  - o Carnitine acylcarnitine translocase deficiency CACT
  - o Carnitine uptake defect/carnitine transport defect CUD
  - o Citrullinemia, type I CIT
  - Citrullinemia, type II CITII
  - Classic galactosemia GALT
  - Congenital Toxoplasmosis TOXO
  - Congenital adrenal hyperplasia CAH
  - Congenital hypothyroidism CH
  - Critical congenital heart disease CCHD
  - Cystic fibrosis CF
  - Cytomegalovirus CMV
  - Ethylmalonic encephalopathy EME
  - Formiminoglutamic acidemia FIGLU
  - Galactoepimerase deficiency GALE
  - Galactokinase deficiency GALK
  - Guanidinoacetate Methyltransferase GAMT
  - Hb No structural variant
  - Hearing loss HEAR



- Holocarboxylase synthetase deficiency MCD
- Homocystinuria HCY
- Hypermethioninemia MET
- Spinal Muscular Atrophy SMA

#### Modifications from September 2024 version

- Added clarification that gestational age should be in whole numbers.
- Updated spelling for SMA molecular variables from UKNOWN to UNKNOWN.

#### Modification from December 2024

- Updated language from Was this individual **not** identified by newborn screening to Was this individual identified **outside** of newborn screen?
- Add Duchenne Muscular Dystrophy DMD as an acceptable condition.
- Updated final diagnosis for Maple syrup urine disease by removing Type IA III, and adding classic, intermediate, thiamine-response.
- Added final diagnosis of unclassified to Maple syrup urine disease, Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT, Methylmalonic acidemia (cobalamin disorders) - Cbl A, B, and Methylmalonic acidemia with homocystinuria – Cbl C,D.
- Added final diagnosis of uncertain type/onset to Mucopolysaccharidosis I MPS I, Pompe, and X-linked Adrenoleukodystrophy.
- Added final diagnoses to Krabbe Disease of infantile onset Krabbe disease, later onset Krabbe disease, uncertain type/onset, and unknown.
- Added final diagnoses to Mucopolysaccharidosis II MPS II of severe, attenuated, uncertain type/onset, and unknown.

