NewSTEPs
A Program of the Association of Public Health Laboratories™

Template Map for the Case Import File

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INSTRUCTIONS

This template map provides variable names and acceptable values for the case import file. 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 online webform.

The case import file contains the common demographic and screening variables that are asked for all conditions. It also contains 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 case import file, 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 right-hand 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 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
- Iowa
- Kansas
- Kentucky
- Louisiana
- Maine
- Maryland
- Massachusetts
- Michigan
- Minnesota
- Mississippi
- Missouri
- Montana
- Nebraska
- Nevada
- New Hampshire
- New Jersey
- New Mexico
- New York
- 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

- 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-MCC	• 3-Methylcrotonyl-CoA Carboxylase Deficiency - 3-MCC • Maternal MCC deficiency • MT-ATP6 related mitochondrial disorders • Unknown
Argininosuccinic aciduria - ASA	• Argininosuccinic Acidemia/ Aciduria (ASA) • Pyruvate carboxylase deficiency • Unknown
Biotinidase deficiency - BIOT	• Profound Biotinidase deficiency • Partial Biotinidase deficiency • Unknown
Citrullinemia, type I - CIT	• Citrullinemia, Type I • Pyruvate carboxylase deficiency • Unknown
Carnitine uptake defect/carnitine transport defect - CUD	• Carnitine Uptake Deficiency (CUD) • Maternal Carnitine Deficiency (primary and secondary) • Unknown
Classic PKU & Hyperphe	• Classic phenylketonuria - PKU • Benign hyperphenylalaninemia - H-PHE • HyperPhe diet controlled • Dihydropterine reductase deficiency (DHPR) • DNAJC12 • Parenteral nutrition • Maternal PKU • Unknown
Classic galactosemia - GALT	• Classic Galactosemia • 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

Condition	Acceptable Values
Congenital adrenal hyperplasia - CAH	• Classic 21-Hydroxylase Deficiency- Salt Wasting • Classic 21-Hydroxylase Deficiency- Simple Virilizing • Other Adrenal disorder • Unknown
Critical congenital heart disease - CCHD	• CCHD • Non critical CCHD • Other • Unknown
Cystic fibrosis - CF	• CFTR-Related Metabolic Syndrome (CRMS) • CFTR-Related Disease • Typical Cystic Fibrosis (CF) • Unknown
Holocarboxylase synthetase deficiency - MCD	• Holocarboxylase synthetase deficiency (MCD) • Maternal 3-methylcrotonyl-CoA carboxylase deficiency • MT-ATP6 related mitochondrial disorders • Other biotin disorder • Unknown
Homocystinuria - HCY	• Classic Homocystinuria • Methionine Adenosyltransferase (MAT I/III Deficiency) • 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 • Later Onset Krabbe Disease • Uncertain Type/Onset • Unknown
Maple syrup urine disease - MSUD	• Classic • Intermediate • Thiamine-response • Hydroxyprolinemia • Unclassified • Unknown
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) • Cobalamin B deficiency (Cbl B)

Condition	Acceptable Values
	• Cobalamin Dv2 deficiency (Cbl Dv2) • Maternal vitamin B12 deficiency • Succinate-CoA ligase deficiency • Unclassified • Unknown
Methylmalonic acidemia with homocystinuria - Cbl C,D	• Cobalamin C deficiency (Cbl C) • Cobalamin D deficiency (Cbl D) • Cobalamin F deficiency (Cbl F) • Cobalamin Dv1 deficiency (Cbl Dv1) • Cobalamin J deficiency (Cbl J) • Maternal vitamin B12 deficiency • Succinate-CoA ligase deficiency • Other cobalamin deficiency • Unclassified • Unknown
Mucopolysaccharidosis I - MPS I	• MPS I - severe • MPS I - severity not determined • MPS I - attenuated • Uncertain Type/Onset • Unknown
Mucopolysaccharidosis II - MPS II	• Severe • Attenuated • Uncertain Type/Onset • Unknown
Pompe	• Infantile Onset (IO) Pompe Disease • Late Onset (LO) Pompe Disease • Uncertain Type/Onset • Unknown
Presence of Hb S	• S,S disease (Sickle cell anemia) - Hb SS • S, Beta 0-thalassemia - Hb S/B0Th • S, Beta + thalassemia - Hb S/B+ Th • S,C disease - Hb S/C • S, Other • Unknown
Presence of Other Hb Variant	• Hemoglobin C disease • 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 Syndrome

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

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 newbornSMN2MolecularTest 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 public health surveillance case definitions 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 (CblDv2) to Cobalamin Dv2 deficiency (CblDv2).
- Updated spacing in the following final diagnosis options
    - 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 Dv2 deficiency (Cbl Dv2)
    - Cobalamin D deficiency (Cbl D)
    - Cobalamin F deficiency (Cbl F)
    - Cobalamin Dv1 deficiency (Cbl Dv1)
    - Cobalamin J deficiency (Cbl J)
    - S,C disease- Hb S/C
    - S,S disease (Sickle cell anemia) - Hb SS
- Updated formatting
    - Argininosuccinic aciduria - ASA
    - Beta-Ketothiolase deficiency - BKT
    - Biotinidase deficiency - BIOT
    - Carbamoyl phosphate synthetase I deficiency - CPS
    - Carnitine acylcarnitine translocase deficiency - CACT
    - Carnitine uptake defect/carnitine transport defect - CUD
    - 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.