case_template.csv

- Required fields are indicated (and described first).
- For fields that are not required, the column is not required either.
- For non-required columns included in a csv file, enter an acceptable value or leave field empty.
- Columns may be in any order.

---

column – *description with acceptable values in* ***bold****.*

state - *One of 50 states, District of Columbia, Guam, or Puerto Rico, (****use common name*** *instead of abbreviation, e.g. "Virginia" instead of "VA" or "Commonweatlth of Virginia"), REQUIRED.*

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.* List of acceptable condition 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”**
- **"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 synthase 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”**

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

- **"3-MCC"**
- **"ASA"**

- **"BIOT"**
- **"BKT"**
- **"CAH"**
- **"CCHD"**
- **"CF"**
- **"CH"**
- **"CIT"**
- **"CUD"**
- **"Cbl A,B"**
- **"GA1"**
- **"GALT"**
- **"HCY"**
- **"HEAR"**
- **"HMG"**
- **"IVA"**
- **"LCHAD"**
- **"MCAD"**
- **"MCD"**
- **"MSUD"**
- **"MUT"**
- **"PROP"**
- **"TFP"**
- **"TYR I"**
- **"VLCAD"**

gestationalAge - *The gestational age in weeks.*

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. Only one value should be specified.* List of acceptable ethnicity values:

- **HISPANIC_LATINO_OR_SPANISH**
- **NOT_HISPANIC_LATINO_OR_SPANISH**
- **NOT_REPORTED**
- **UNKNOWN**

race - *The race of the infant. If more than one value applies, separate each value with a colon.* List of acceptable race values. (Note: ISLANDER = Native Hawaiian or other Pacific Islander):

- **UNKNOWN**
- **NOT_REPORTED**
- **ISLANDER**
- **ASIAN**
- **NATIVE_AMERICAN**
- **BLACK_OR_AFRICAN_AMERICAN**
- **WHITE**

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****.*

familyHistoryRisk - *Was there a family history that indicated that this infant was at risk for this disorder? Acceptable values:* ***true****,* ***false****.*

diagnosedAfterNewbornScreening - *Was this individual diagnosed later in life (not identified by newborn screening)? Acceptable values:* ***true****,* ***false.***

missedDiagnosisReason - *The reason this diagnosis was not identified by newborn screening (should only be answered if 'diagnosedAfterNewbornScreening' is true).* List of acceptable missed diagnosis reasons:

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

otherMissedDiagnosisReason - *Must (and should only) be specified if 'Other' given for 'missedDiagnosisReason'. Text description of the missed diagnosis reason up to 254 characters long.*

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****. When 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****. When 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****. When true signifies that the data available for the calculation of elapsed time included time as well as date.*

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****. When 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**. When 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**. When true signifies that the data available for the calculation of elapsed time included time as well asdate.*

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).*

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**. When true signifies that the data available for the calculation of elapsed time included time as well as date.*

Condition Specific columns

The condition specific templates include additional optional column to include information relevant to the diagnostic workup of the infant. Multiple conditions can be included in an import file. Leave columns that do not apply to a condition blank. It is OK to leave any of these columns blank or to not include the column in the import file. The columns may appear in any order.

Table of Contents:

endocrine_disorders_template.csv - Optional fields for Congenital Hypothyroidism and Congenital adrenal hyperplasia cases.

finalDiagnosis - *Final Diagnosis as determined by the endocrinologist or clinician performing the follow-up.*

- *CH Acceptable values:*
    - **Primary Congenital Hypothyroidism**
    - **Secondary Congenital Hypothyroidism**
    - **TBG Deficiency (Thyroxine Binding Globulin) or other protein binding defect**
- *CAH Acceptable values:*
    - **Classic 21-Hydroxylase Deficiency- Salt Wasting**
    - **Classic 21-Hydroxylase Deficiency- Simple Virilizing**
    - **Other Adrenal disorder**

otherFinalDiagnosisName - Specify the name for the “other” value when a value containing “other” is selected from in the finalDiagnosis column. (CAH only) Acceptable values: **ANY text**.

serumTshLevel - *What was the Serum TSH level if tested?* (CH only) *Acceptable values:*

ABOVE_UPPER_THRESHOLD	(description: TSH > 10 mU/L)
WITHIN_THRESHOLDS	(description: TSH 6-10 mU/L)
BELOW_LOWER_THRESHOLD	(description: TSH < 6 mU/L)
UNKNOWN	(description: TSH level unknown)

serumTshTestedBeforeTreatment - *Was Serum TSH tested before initiation of treatment? (CH only) Acceptable values:* ***TRUE, FALSE, UNKNOWN***

serumTotalTFourBelowReferenceRange - *Was Serum Total T4 below the age-established reference range? (CH only) Acceptable values:* ***TRUE, FALSE, UNKNOWN***

serumTotalTFourTestedBeforeTreatment - *Was Serum Total T4 tested before initiation of treatment? (CH only) Acceptable values:* ***TRUE, FALSE, UNKNOWN***

serumFreeTFourBelowReferenceRange - *Was Serum Free T4 below the age-established reference range? (CH only) Acceptable values:* ***TRUE, FALSE, UNKNOWN***

serumFreeTFourTestedBeforeTreatment - *Was Serum Free T4 tested before initiation of treatment? (CH only) Acceptable values:* ***TRUE, FALSE, UNKNOWN***

otherPituitaryHormoneDeficienciesPresent - *Does this infant have other pituitary hormone deficiencies? (CH only) Acceptable values:* ***TRUE, FALSE, UNKNOWN***

midlineDefectsPresent - *Does this infant have midline defects? (CH only) Acceptable values:* ***TRUE, FALSE, UNKNOWN***

tbgBelowReferenceRange - *Was TBG below the age established reference range? (CH only) Acceptable values:* ***TRUE, FALSE, UNKNOWN***

resinUptakeBelowReferenceRange - *Was T3 or T4 resin uptake above the age established reference range? (CH only) Acceptable values:* ***TRUE, FALSE, UNKNOWN***

societalGender - Societal Sex. (CAH only) Acceptable values: **FEMALE, MALE, UNSPECIFIED, UNKNOWN** confirmatorySerum17OhpLevelObtained - Was a confirmatory serum 17-OHP level obtained? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

serum17OhpLevel - Was there a value at baseline. (CAH only) Acceptable values:

- **GREATER_THAN_TEN_THOUSAND** (> 10,000 ng/dl)
- **BETWEEN_ONE_THOUSAND_AND_TEN_THOUSAND** (1000-10,000 ng/dl)
- **LESS_THAN_ONE_THOUSAND** (< 1000 ng/dl)
- **UNKNOWN**

serum17OhpLevelTested - Was it tested before initiation of treatment? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

serum17OhpAfterActh - Was there a result after ACTH stimulation. (CAH only) Acceptable values:

- **GREATER_THAN_TEN_THOUSAND** (> 10,000 ng/dl)
- **BETWEEN_ONE_THOUSAND_AND_TEN_THOUSAND** (1000-10,000 ng/dl)
- **LESS_THAN_ONE_THOUSAND** (< 1000 ng/dl)
- **UNKNOWN**

serum17OhpAfterActhTested - Was it tested before initiation of treatment? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

urineSteriodProfileObtained - Was tandem mass spectrometry urinary steroid profile obtained? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

urineSteroidProfile - Were the urinary spectrometry steroid profile results. (CAH only) Acceptable values:

- **HYDROXYLASE_DEFICIENCY** (Indicative of 21-Hydroxylase Deficiency CAH)
- **UNKNOWN**

serumSodiumLevelTested - Was serum sodium level measured before initiation of treatment? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

serumSodiumLevel - Was the sodium level. (CAH only) Acceptable values:

- **GREATER_THAN_THRESHOLD** (> 135 mEq/L)
- **LESS_THAN_THRESHOLD** (< 135 mEq/L)
- **UNKNOWN**

plasmaReninActivityMeasured - Was Plasma renin activity level measured at time of initiation of treatment? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

plasmaReninActivity - Was the Plasma renin activity normal for age? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

plasmaReninActivityTested - Was it tested before initiation of treatment? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

saltWastingEvidence - Is there evidence of salt wasting? (e.g. shock or severe failure to thrive) (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

supportiveEvidenceExists - Is there supportive clinical or laboratory evidence of CAH? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN** supportiveEvidence - Is the evidence: (check all that apply). (CAH only) Acceptable values (separated by ‘:’):

- **AMBIGUOUS_GENITALIA** (Ambiguous genitalia, with 46,XX karyotype)
- **NORMAL_GENITALIA** (Normal genitalia, with 46,XY karyotype)
- **OTHER_HORMONAL_EVIDENCE** (Other hormonal evidence of CAH)

mutationAnalysisDone - Was mutation analysis done? (CAH only) Acceptable values: **TRUE, FALSE, YES, NO, UNKNOWN**

cyp21a2Gene.alleleOne - CYP21A2 Check the types of variants found on: Allele 1. (CAH only) Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

cyp21a2Gene.alleleTwo - CYP21A2 Check the types of variants found on: Allele 2. (CAH only) Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

otherGeneName - Other gene name. (CAH only) Acceptable values: **ANY text**.

otherGene.alleleOne - Other Gene Check the types of variants found on: Allele 1. (CAH only) Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

otherGene.alleleTwo - Other Gene Check the types of variants found on: Allele 2. (CAH only) Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

hb_disorders_template.csv - Optional fields for each Hemoglobin disorder. Only the finalDiagnosis column is applicable for the three cases, the other columns are applicable for ‘Presence of Hb S’ and ’Presence of Other Hb Variant’ (when not specified).

finalDiagnosis - Final Diagnosis as determined by a clinician performing the follow-up.

- Hb - No structural variant, acceptable values:
  - **Alpha thalassemia major (Fetal Hydrops)**
  - **Hgb H disease**
  - **Beta thalassemia major (Cooley's anemia)**
- Presence of Hb S, acceptable values:
  - **S, Beta + thalassemia - Hb S/B+ Th**
  - **S,C disease - Hb S/C**
  - **S,S disease (Sickle cell anemia) - Hb SS**
  - **S, Beta 0-thalassemia - Hb S/B0Th**
  - **Not Known**
  - **S, other**
- Presence of Other Hb Variant, acceptable values:
  - **Hemoglobin C disease**
  - **Hemoglobin D disease**
  - **Hemoglobin E disease**
  - **Hemoglobin O-Arab disease**
  - **Other hemoglobin disease, please describe**

otherFinalDiagnosisName - Specify the name for the “other” value when a value containing “other” is selected from in the finalDiagnosis column. Acceptable values: **ANY text**

alphaThalassemiaPresent - Alpha thalassemia present? (Applicable ONLY to ‘Presence of Other Hb Variant’) Acceptable values: **TRUE, FALSE, UNKNOWN**

qualitativeTestResult - What were the qualitative (IEF or HPLC) test results? (You can use any text here to set other value)

- Presence of Hb S, acceptable values:
  - **FS**
  - **FSC**
  - **FSA**
  - **FSA2**
  - **FSAA2**
  - **OTHER**
  - **UNKNOWN**
- Presence of Other Hb Variant, acceptable values:
  - **FC**
  - **FD**
  - **FE**
  - **FO_ARAB**
  - **OTHER**
  - **UNKNOWN**

qualitativeTestResultRepeated - Test repeated? Acceptable values: **TRUE, FALSE** quantitativeTestResult - What were the quantitative (HPLC or electrophoresis) test results? (You can use any text here to set other value)

- Presence of Hb S, acceptable values:
  - **FS**
  - **FSC**
  - **FS_HIGH_A2**
  - **FSA_HIGH_A2**
  - **FSA**
  - **OTHER**
  - **UNKNOWN**
- Presence of Other Hb Variant, acceptable values:
  - **FC**
  - **FD**
  - **FE**
  - **FO_ARAB**
  - **OTHER**
  - **UNKNOWN**

alleleOneVariant - Type of variant found on allele 1 (You can use any text here to set other value)

- Presence of Hb S, acceptable values:
  - **S**
  - **C**
  - **BETA_PLUS_THAL**
  - **BETA_ZERO_THAL**
  - **OTHER**
  - **UNKNOWN**
- Presence of Other Hb Variant, acceptable values:
  - **C**
  - **D**
  - **E**
  - **O_ARAB**
  - **OTHER**
  - **UNKNOWN**

alleleTwoVariant - Type of variant found on allele 2 (You can use any text here to set other value)

- Presence of Hb S, acceptable values:
  - **S**
  - **C**
  - **BETA_PLUS_THAL**
  - **BETA_ZERO_THAL**
  - **OTHER**
  - **UNKNOWN**
- Presence of Other Hb Variant, acceptable values:
  - **C**
  - **D**
  - **E**
  - **O_ARAB**
  - **BETA_PLUS_THAL**
  - **BETA_ZERO_THAL**
  - **OTHER**
  - **UNKNOWN**

nbsResult - What was the NBS result? (You can use any text here to set other value)

- Presence of Hb S, acceptable values:
  - **FS**
  - **FSC**
  - **FSA**
  - **FSA2**
  - **OTHER**
  - **UNKNOWN**
- Presence of Other Hb Variant, acceptable values:
  - **FC**
  - **FD**
  - **FE**
  - **FO_ARAB**
  - **OTHER**
  - **UNKNOWN**

cbcResult- What were the CBS results? Acceptable values: **NORMAL, LOW, UNKNOWN** maternalStatus, paternalStatus - Were family studies (in parents) done? (You can use any text here to set other value)

- Presence of Hb S, acceptable values:
  - **CARRIER_S**
  - **CARRIER_C**
  - **CARRIER_BETA_PLUS_THAL**
  - **CARRIER_BETA_ZERO_THAL**
  - **OTHER**
  - **UNKNOWN**
- Presence of Other Hb Variant, acceptable values:
  - **CARRIER_C**
  - **CARRIER_D**
  - **CARRIER_E**
  - **CARRIER_O_ARAB**
  - **CARRIER_BETA_PLUS_THAL**
  - **CARRIER_BETA_ZERO_THAL**
  - **OTHER**
  - **UNKNOWN**

positiveFamilyHistory - Was there a positive family history? Acceptable values: **TRUE, FALSE, UNKNOWN**

hplcAndIefTestResult - Were HPLC & IEF tested on the same sample from the infant?

- Applicable ONLY to ‘Presence of Hb S’. Acceptable values:
  - **FS**
  - **FSC**
  - **FSA2**
  - **FSAA2**
  - **OTHER**
  - **UNKNOWN** (You can use any text here to set other value)

hbgTestResult - Were Hgb tests (electrophoresis or HPLC) performed on family members? Acceptable values: **ELEVATED, ABSENT, UNKNOWN**

other_disorders_template.csv - Optional fields for Cystic Fibrosis, Biotinidase deficiency - BIOT, Classic galactosemia - GALT, Critical congenital heart disease - CCHD, Severe Combined Immunodeficiencies - SCID cases.
finalDiagnosis – *Final Diagnosis as determined by a clinician performing the follow-up.* (CF, BIOT, SCID only)

- *CF Acceptable values:*
    - **Typical Cystic Fibrosis (CF)**
    - **CFTR-Related Metabolic Syndrome (CRMS)**
    - **CFTR Related Disease**
- *BIOT Acceptable values:*
    - **Profound Biotinidase deficiency**
    - **Partial Biotinidase deficiency**
- *SCID Acceptable values:*
    - **Classic SCID**
    - **Leaky SCID**
    - **Omenn syndrome**

nbsElevatedIrt - *Did the NBS result indicate an elevated IRT? (CF only)*
Acceptable values: **TRUE**, **FALSE, *YES, NO,* UNKNOWN**

cftrOnNewbornScreeningMutationPanel - *Were CFTR mutations detected on the newborn screening mutation panel? (CF only)*
Acceptable values: **TRUE**, **FALSE, *YES, NO,* UNKNOWN**

cftrDetected.alleleOne – Only answered when cftrOnNewbornScreeningMutationPanel is TRUE/YES. *What was the variant found on: Allele 1. (CF only)*
Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing in CFTR2)
- **LOWER_SWEAT_CHLORIDES** (Variant known to be disease causing in CFTR2. Shown to be associated with lower sweat chlorides)
- **UNCERTAIN_SIGNIFICANCE** (Neutral variant)
- **VARYING_SIGNIFICANCE** (Variant of varying clinical consequence in CFTR2)
- **NONE** (Wild Type (Normal))
- **UNKNOWN** (Unknown (not reported in CFTR2))

cftrDetected.alleleTwo - Only answered when cftrOnNewbornScreeningMutationPanel is TRUE/YES. *What was the variant found on: Allele 2. (CF only)*
Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing in CFTR2)
- **LOWER_SWEAT_CHLORIDES** (Variant known to be disease causing in CFTR2. Shown to be associated with lower sweat chlorides)
- **UNCERTAIN_SIGNIFICANCE** (Neutral variant)
- **VARYING_SIGNIFICANCE** (Variant of varying clinical consequence in CFTR2)
- **NONE** (Wild Type (Normal))
- **UNKNOWN** (Unknown (not reported in CFTR2))

meconiumIleusPresent - Did the child have meconium ileus? *(CF only)*
Acceptable values: **TRUE**, **FALSE, YES, NO, UNKNOWN**

sweatChlorideDone - Was a valid sweat chloride result available? *(CF only)*
Acceptable values: **TRUE**, **FALSE, YES, NO, UNKNOWN**

sweatChlorideResult – Only answered when sweatChlorideDone is TRUE/YES. What were the sweat test results (please report on the highest sweat chloride value from one sweat test)? *(CF only)*
Acceptable values:

- **GREATER_THAN_60** (>= 60 mmol/L (regardless of age))
- **LESS_THAN_30** (< 30 mmol/L (if age < 6 months))
- **BETWEEN_30_AND_59** (30-59 mmol/L (if age < 6 months))
- **LESS_THAN_40** (< 40 mmol/L (if age > 6 months))
- **BETWEEN_40_AND_59** (40 -59 mmol/L (if age > 6 months))
- **QUANTITY_NOT_SUFFICIENT** (Quantity Not Sufficient)

quantityNotSufficientSweatChloride - Only answered when sweatChlorideDone is TRUE/YES. If a valid sweat test was not available, were there attempts to obtain a sweat chloride that were quantity not sufficient (QNS)? *(CF only)*
Acceptable values: **TRUE**, **FALSE, YES, NO, UNKNOWN**

sweatChlorideRepeatDone - Was a sweat chloride repeated on a separate day (results from different arm on the same day should not be reported here)? *(CF only)*
Acceptable values: **TRUE**, **FALSE, YES, NO, UNKNOWN**

sweatChlorideRepeated - Only answered when sweatChlorideRepeatDone is TRUE/YES. What were the repeat sweat test results (please report on the highest sweat chloride value from one sweat test)? *(CF only)*
Acceptable values:

- **GREATER_THAN_60** (>= 60 mmol/L (regardless of age))
- **LESS_THAN_30** (< 30 mmol/L (if age < 6 months))
- **BETWEEN_30_AND_59** (30-59 mmol/L (if age < 6 months))
- **LESS_THAN_40** (< 40 mmol/L (if age > 6 months))
- **BETWEEN_40_AND_59** (40 -59 mmol/L (if age > 6 months))
- **QUANTITY_NOT_SUFFICIENT** (Quantity Not Sufficient)

cftrMutationPanelCompletedAfterNewbornScreening - Was a CFTR mutation panel completed after the newborn screening mutation panel? *(CF only)*
Acceptable values: **TRUE**, **FALSE**, **YES, NO, UNKNOWN**

cftrCompleted.alleleOne – Only answered when cftrMutationPanelCompletedAfterNewbornScreening is TRUE/YES. Type of variant found on: Allele 1. *(CF only)*
Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing in CFTR2)
- **LOWER_SWEAT_CHLORIDES** (Variant known to be disease causing in CFTR2. Shown to be associated with lower sweat chlorides)
- **UNCERTAIN_SIGNIFICANCE** (Neutral variant)
- **VARYING_SIGNIFICANCE** (Variant of varying clinical consequence in CFTR2)
- **NONE** (Wild Type (Normal))
- **UNKNOWN** (Unknown (not reported in CFTR2))

cftrCompleted.alleleTwo - Only answered when cftrMutationPanelCompletedAfterNewbornScreening is TRUE/YES. Type of variant found on: Allele 2. *(CF only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing in CFTR2)
- **LOWER_SWEAT_CHLORIDES** (Variant known to be disease causing in CFTR2. Shown to be associated with lower sweat chlorides)
- **UNCERTAIN_SIGNIFICANCE** (Neutral variant)
- **VARYING_SIGNIFICANCE** (Variant of varying clinical consequence in CFTR2)
- **NONE** (Wild Type (Normal))
- **UNKNOWN** (Unknown (not reported in CFTR2))

clinicalSymptomsPresent - If child was diagnosed after the newborn period, were clinical symptoms associated with CFTR Related Disease present? *(CF only)*

Acceptable values: **TRUE**, **FALSE**, **YES, NO, UNKNOWN**

cfClinicalSymptoms – Only answered when clinicalSymptomsPresent is TRUE/YES. List symptoms present. Specify multiply symptoms by separating values with colon (eg ,NASAL_POLYPOSIS:INFERTILITY,…). *(CF only)*

Acceptable values:

- **CBAVD**
- **RECURRENT_PANCREATITIS**
- **NASAL_POLYPOSIS**
- **INFERTILITY**
- **FOCAL_BILIARY_CIRRHOSIS**

biotinidaseActivityTested - Was enzyme analysis for biotinidase enzyme activity completed? *(BIOT only)*

Acceptable values: **TRUE**, **FALSE**, **YES, NO, UNKNOWN**

biotinidaseActivity – Only answered when biotinidaseActivityTested is TRUE/YES. What was the enzyme activity *(BIOT only)*

Acceptable values:

- **LESS_THAN_TEN_PERCENT_NORMAL** (<10%)
- **BETWEEN_TEN_AND_THIRTY_PERCENT_NORMAL** (10-30%)
- **NORMAL**
- **UNKNOWN**

galtLevelTested - Were GALT levels tested? *(GALT only)*

Acceptable values: **TRUE**, **FALSE**, **YES, NO, UNKNOWN**

galtLevel – Only answered when galtLevelTested is TRUE/YES. What was the GALT level? *(GALT only)*

Acceptable values:

- **LESS_THAN_TEN_PERCENT_NORMAL** (<10%)
- **BETWEEN_TEN_AND_THIRTY_PERCENT_NORMAL** (10-30%)
- **NORMAL**
- **UNKNOWN**

gal1PTested - Was Gal-1-P tested? *(GALT only)*

Acceptable values: **TRUE**, **FALSE**, **YES, NO, UNKNOWN**

gal1PLevel – Only answered when gal1PTested is TRUE/YES. What was the enzyme activity? *(GALT only)*

Acceptable values: **ELEVATED, NORMAL, UNKNOWN**

urineGalactitolTested - Was Urine Galactitol tested? *(GALT only)*
Acceptable values: **TRUE**, **FALSE**, **YES, NO, UNKNOWN**

urineGalactitolLevel – Only answered when urineGalactitolTested is TRUE/YES. What was Urine Galactitol level? *(GALT only)*
Acceptable values: **ELEVATED, NORMAL, UNKNOWN**

proteinPhenotypingCompleted - If Variant Galactosemia, was protein phenotyping completed? *(GALT only)*
Acceptable values: **TRUE**, **FALSE**, **YES, NO, UNKNOWN**

proteinPhenotypingResult – Only answered when proteinPhenotypingCompleted is TRUE/YES. What did protein phenotyping indicate?
Acceptable values:

- **CONSISTENT** (Phenotype consistent with variant)
- **INCONSISTENT** (Phenotype NOT consistent with variant)
- **UNKNOWN**

enzymeAnalysisCompleted - If Arginase Deficiency, were enzyme studies completed? *(GALT only)*
Acceptable values: **TRUE**, **FALSE**, **YES, NO, UNKNOWN**

enzymeAnalysisResult – Only answered when enzymeAnalysisCompleted is TRUE/YES. What did enzyme activity indicate? *(GALT only)*
Acceptable values:

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity - not consistent with disease)
- **UNKNOWN**

primaryScreeningTargets – Primary screening targets. Specify multiply symptoms by separating values with colon (eg ,TRICUSPID_ATRESIA:TRUNCUS_ARTERIOSUS,…). *(CCHD only)*
Acceptable values:

- **HYPOPLASTIC_LEFT_HEART_SYNDROME**
- **PULMONARY_ATRESIA_INTACT_WITH_SEPTUM**
- **TETRALOGY_OF_FALLOT**
- **TOTAL_ANOMALOUS_PULMONARY_VENOUS_RETURN**
- **TRANSPOSITION_OF_GREAT_ARTERIES**
- **TRICUSPID_ATRESIA**
- **TRUNCUS_ARTERIOSUS**

secondaryScreeningTargets – Secondary Screening Targets. Specify multiply symptoms by separating values with colon and no spaces, see example for primaryScreening targets above. *(CCHD only)*
Acceptable values:

- **COARCTATION_OF_AORTA**
- **DOUBLE_OUTLET_RIGHT_VENTRICLE**
- **EBSTEIN_ANOMALY**
- **INTERRUPTED_AORTIC_ARCH**
- **SINGLE_VENTRICLE**

mutationAnalysisDone - Was mutation analysis performed for the disorder? (*BIOT and GALT only)*
Acceptable values: **TRUE**, **FALSE**, **YES, NO, UNKNOWN**

btdGene.alleleOne – Only answered when mutationAnalysisDone is TRUE/YES. Type of variant found on BTD Gene, Allele 1. *(BIOT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **PROFOUND_ENZYME_DEFICIENCY** (Variant known to be associated with profound enzyme deficiency)
- **PARTIAL_ENZYME_DEFICIENCY** (Variant known to be associated with partial enzyme deficiency [‘mild’ mutation (D444H)]
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **NONE** (Wild type or normal)
- **UNKNOWN**

btdGene.alleleTwo – Only answered when mutationAnalysisDone is TRUE/YES. Type of variant found on BTD Gene, Allele 2. *(BIOT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **PROFOUND_ENZYME_DEFICIENCY** (Variant known to be associated with profound enzyme deficiency)
- **PARTIAL_ENZYME_DEFICIENCY** (Variant known to be associated with partial enzyme deficiency [‘mild’ mutation (D444H)]
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **NONE** (Wild type or normal)
- **UNKNOWN**

galactosemiaGene.alleleOne - Only answered when mutationAnalysisDone is TRUE/YES. Type of variant found on Galactosemia Gene, Allele 1. *(GALT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **PREDICTED_PATHOGENIC** (Variant predicted to be pathogenic)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **NONE** (Wild type or normal)
- **UNKNOWN**

galactosemiaGene.alleleTwo - Only answered when mutationAnalysisDone is TRUE/YES. Type of variant found on Galactosemia Gene, Allele 2. *(GALT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **PREDICTED_PATHOGENIC** (Variant predicted to be pathogenic)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **NONE** (Wild type or normal)
- **UNKNOWN**

otherGeneName - Only answered when mutationAnalysisDone is TRUE/YES. Name of other gene included in the mutation analysis. *(BIOT and GALT only)*

Acceptable values: ANY text.

otherGene.alleleOne - Only answered when mutationAnalysisDone is TRUE/YES. Type of variant found on Other Gene, Allele 1. *(BIOT and GALT only)*

Acceptable values for BIOT:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **PROFOUND_ENZYME_DEFICIENCY** (Variant known to be associated with profound enzyme deficiency)
- **PARTIAL_ENZYME_DEFICIENCY** (Variant known to be associated with partial enzyme deficiency [‘mild’ mutation (D444H)]
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **NONE** (Wild type or normal)
- **UNKNOWN**

Acceptable values for GALT:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **PREDICTED_PATHOGENIC** (Variant predicted to be pathogenic)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **NONE** (Wild type or normal)
- **UNKNOWN**

otherGene.alleleTwo - Only answered when mutationAnalysisDone is TRUE/YES. Type of variant found on Other Gene, Allele 2. *(BIOT and GALT only)*

Acceptable values for BIOT:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **PROFOUND_ENZYME_DEFICIENCY** (Variant known to be associated with profound enzyme deficiency)
- **PARTIAL_ENZYME_DEFICIENCY** (Variant known to be associated with partial enzyme deficiency [‘mild’ mutation (D444H)]
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **NONE** (Wild type or normal)
- **UNKNOWN**

Acceptable values for GALT:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **PREDICTED_PATHOGENIC** (Variant predicted to be pathogenic)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **NONE** (Wild type or normal)
- **UNKNOWN**

fatty_acid_disorders_template.csv - Optional fields for Medium-chain acyl-CoA dehydrogenase deficiency, Carnitine uptake defect/carnitine transport defect, Very long-chain acyl-CoA dehydrogenase deficiency, Long-chain L- 3 hydroxyacyl-CoA dehydrogenase deficiency and Trifunctional protein deficiency cases.

urineOrganicAcidsOrAclyglycinesTested - *Were urine organic acids or aclyglycines tested? (MCAD only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

hexanoylglycineLevel - *Was Hexanoylglycine level. (MCAD only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

fibroblastsAnalysisPerformed - *Was functional analysis of fatty acid oxidation in cultured fibroblasts performed? (MCAD only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

fibroblastAnalysisResult - *Was functional fibroblast analysis. (MCAD only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

urineCarnitineTested - *Was urine carnitine tested? (CUD only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

urineCarnitineLevel - *Was fractional excretion of free carnitine level. (CUD only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

plasmaCarnitineLevelsTested - *Were plasma carnitine levels tested? (CUD only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

plasmaCarnitineLevel - *Was free carnitine (C0). (CUD only) Acceptable values:* **LOW, NORMAL, UNKNOWN**

secondaryLossRuledOut - *Were other causes for carnitine loss ruled out? (CUD only) Acceptable values:* **TRUE**, **FALSE, *YES, NO,* UNKNOWN**

functionalAnalysisPerformed - *Was functional analysis of fatty acid oxidation in cultured fibroblasts performed? (VLCAD, LCHAD, TFP only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

functionalFibroblastAnalysisResult - *Was functional fibroblast analysis. (VLCAD, LCHAD, TFP only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

urineOrganicAcids - *Were urine organic acids tested? (LCHAD, TFP only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

c12DicarboxylicAcidLevel - *Was C12-OH dicarboxylic acid level. (LCHAD, TFP only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

c10DicarboxylicAcidLevel - *Was C10-OH dicarboxylic level. (LCHAD, TFP only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

plasmaAcylcarnitines - *Were plasma acylcarnitines tested? Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

c8Level - *Was C8 level. (MCAD only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN*** c8LevelOnRepeatTesting - *Was repeat C8 level. (MCAD only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

c8GreaterThanC10 - *Was C8>C10 level. (MCAD only) Acceptable values:* **TRUE**, **FALSE**, ***YES, NO,*** **UNKNOWN**

c8GreaterThanC6 - *Was C8>C6 level. (MCAD only) Acceptable values:* **TRUE**, **FALSE**, ***YES, NO,*** **UNKNOWN**

c6Level - Was C6 level. *(MCAD only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

c10Level - Was C10 level. *(MCAD only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

c14Colon1LevelOnRepeatTesting - Was C14:1 level. *(VLCAD only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

c14Colon2Level - Was C14:2 level. *(VLCAD only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

c14Level - Was C14 level. *(VLCAD only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

c16LevelOnRepeatTesting - Was C16-OH level. *(LHCAD, TFP only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

c16Colon1Level - Was C16:1-OH level. *(LHCAD, TFP only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

c18Level - Was C18-OH level. *(LHCAD, TFP only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

c18Colon1Level - Was C18:1-OH level. *(LHCAD, TFP only) Acceptable values:* **ELEVATED**, **NORMAL**, ***UNKNOWN***

enzymeAnalysisCompleted - *Was enzyme analysis for MCAD enzyme activity completed? Acceptable values:* ***TRUE, FALSE, YES, NO, UNKNOWN***

mcadEnzymeAnalysisResult - *Was enzyme activity. (MCAD only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

cudEnzymeAnalysisResult - *Was enzyme activity. (CUD only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

vlcadEnzymeAnalysisResult - *Was enzyme activity. (VLCAD only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

tfpEnzymeAnalysisResult - *Was enzyme activity. (LHCAD, TFP only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

mutationAnalysisDone - *Was mutation analysis done? Acceptable values:* ***TRUE, FALSE, YES, NO, UNKNOWN*** acadmGene.alleleOne - ACADM *Check the types of variants found on: Allele 1. (MCAD only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

acadmGene.alleleTwo - ACADM *Check the types of variants found on: Allele 2. (MCAD only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

cudGene.alleleOne - SCL22A5 Gene *Check the types of variants found on: Allele 1. (CUD only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

cudGene.alleleTwo - SCL22A5 Gene *Check the types of variants found on: Allele 2. (CUD only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

acadvlGene.alleleOne - ACADVL Gene *Check the types of variants found on: Allele 1. (VLCAD only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

acadvlGene.alleleTwo - ACADVL Gene *Check the types of variants found on: Allele 2. (VLCAD only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

hadhAGene.alleleOne - HADHA Gene *Check the types of variants found on: Allele 1. (LHCAD, TFP only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

hadhAGene.alleleOne - HADHA Gene *Check the types of variants found on: Allele 2. (LHCAD, TFP only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

hadhBGene.alleleOne - HADHB Gene *Check the types of variants found on: Allele 1. (LHCAD, TFP only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

hadhBGene.alleleOne - HADHB Gene *Check the types of variants found on: Allele 2. (LHCAD, TFP only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

otherGeneName - Other gene name*. Acceptable values:* ***ANY text****.*

otherGene.alleleOne - Other Gene *Check the types of variants found on: Allele 1. Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

otherGene.alleleTwo - Other Gene *Check the types of variants found on: Allele 2. Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

amino_acid_disorders_template.csv - Optional fields for Argininosuccinic aciduria - ASA, Citrullinemia, type I - CIT, Classic PKU & Hyperphe, Homocystinuria - HCY, Maple syrup urine disease - MSUD, Tyrosinemia, type I - TYR I cases.

finalDiagnosis - *Final Diagnosis as determined by metabolic geneticist or clinician performing the follow-up. (Classic PKU & Hyperphe, only)*

- *Classic PKU & Hyperphe, acceptable values:*
  - **Classic phenylketonuria - PKU**
  - **Benign hyperphenylalaninemia - H-PHE**
  - **HyperPhe diet controlled**
- Maple syrup urine disease - MSUD, acceptable values:
  - **MAPLE SYRUP URINE DISEASE, TYPE IA**
  - **MAPLE SYRUP URINE DISEASE, TYPE IB**
  - **MAPLE SYRUP URINE DISEASE, TYPE II**
  - **MAPLE SYRUP URINE DISEASE, TYPE III**

plasmaAminoAcids - *Were plasma amino acids collected? (ASA, Classic PKU & Hyperphe, MSUD only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

plasmaOrganicAcidsTested - *Were plasma amino acids tested? (CIT only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

plasmaAsaLevel - *Was Plasma ASA level. (ASA, CIT only) Acceptable values:* **ELEVATED**, **NORMAL, UNKNOWN**

plasmaCitrullineLevel - *Was Citrulline level. (ASA only) Acceptable values:* **ELEVATED**, **NORMAL, UNKNOWN**

citrullineLevel - Was Citrulline level. *(CIT only) Acceptable values:* **ELEVATED**, **NORMAL, UNKNOWN**

pheLevel - *Was Phe level. (Classic PKU & Hyperphe only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

pheTyrRatio - Was Phe/Tyr ratio. *(Classic PKU & Hyperphe only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

alloisoleucineLevel - *Was Alloisoleucine level. (MSUD only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

leucineLevel - *Was Leucine level. (MSUD only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

isoleucineLevel - *Was Isoeucine level. (MSUD only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

valineLevel - *Was Valine level. (MSUD only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

leuGreaterThanVal - *Was Leu>Val? (MSUD only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

plasmaAminoAcidsTested - *Were plasma amino acids tested? (HCY only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN**

methionineLevel - *Was Methionine level. (HCY only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

plasmaHomocysteineTested - *Was plasma Homocysteine tested? (HCY only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,* UNKNOWN** homocysteineLevel - *Was plasma Homocysteine level. (HCY only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

serumOrganicAcids - *Were plasma amino acids tested? (TYR I only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,*** **UNKNOWN**

plasmaSuccinylacetoneLevel - *Was plasma succinylacetone level. (TYR I only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

plasmaTyrosineLevel - *Was plasma tyrosine level. (TYR I only) Acceptable values:* **ELEVATED, NORMAL, UNKNOWN**

urineAminoAcids - *Were plasma urine acids tested? (ASA only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,*** **UNKNOWN**

urineAsaLevel - *Was urine ASA level. (ASA only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

urineCitrullineLevel - *Was urine Citrulline level. (ASA only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

bloodAmmoniaLevelTested - *Was blood ammonia level tested? (CIT only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,*** **UNKNOWN**

bloodAmmoniaLevel - *Was blood ammonia level. (CIT only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

biopterinStudiesCompleted - *Were biopterin studies done? (Classic PKU & Hyperphe only) Acceptable values:* **TRUE**, **FALSE*, YES, NO,*** **UNKNOWN**

biopterinStudiesResult - Were biopterin studies*. (Classic PKU & Hyperphe only) Acceptable values:* **NORMAL*, ABNORMAL, UNKNOWN***

urineOrganicAcids - Were urine organic acids tested? *(MSUD, TYR I only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

twoKetoisocaproicAcidLevel - Was 2-ketoisocaproic acid level*. (MSUD only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

twoOhIsovalericAcidLevel - Was 2-OH Isovaleric acid level*. (MSUD only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

twoKetomethylValericAcidLevel - Was 2-ketomethyl valeric acid level*. (MSUD only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

urineSuccinylacetoneLevel - Was urine succinylacetone level*. (TYR I only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

urineTyrosineLevel - Was urine tyrosine level*. (TYR I only) Acceptable values:* **ELEVATED**, **NORMAL*, UNKNOWN***

aslEnzymeAnalysisTested - Was enzyme analysis for ASA enzyme activity completed? *(ASA only) Acceptable values:* ***TRUE**, **FALSE, YES, NO, UNKNOWN*** aslEnzymeAnalysisResult - *Was enzyme activity. (ASA only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

enzymeAnalysisCompleted - *Was enzyme analysis for activity completed? (all but ASA) Acceptable values:* ***TRUE, FALSE, YES, NO, UNKNOWN***

argininSynthaseEnzymeAnalysisResult - *Was enzyme activity. (CIT only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

pahEnzymeAnalysisResult - *Was enzyme activity. (Classic PKU & Hyperphe only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

cbsEnzymeAnalysisResult - *Was enzyme activity. (HCY only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

msudEnzymeAnalysisResult - *Was enzyme activity. (MSUD only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

fahEnzymeAnalysisResult - *Was enzyme activity. (MSUD only) Acceptable values:*

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity (not consistent with disease))
- **UNKNOWN**

mutationAnalysisDone - *Was mutation analysis done? Acceptable values:* ***TRUE, FALSE, YES, NO, UNKNOWN***

aslGene.alleleOne - ASL *Check the types of variants found on: Allele 1. (ASA only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

aslGene.alleleTwo - ASL *Check the types of variants found on: Allele 2. (ASA only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

ass1Gene.alleleOne - ASS1 Gene *Check the types of variants found on: Allele 1. (CIT only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

ass1Gene.alleleTwo - ASS1 Gene *Check the types of variants found on: Allele 2. (CIT only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

pahGene.alleleOne - PAH Gene *Check the types of variants found on: Allele 1. (Classic PKU & Hyperphe only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

pahGene.alleleTwo - PAH Gene *Check the types of variants found on: Allele 2. (Classic PKU & Hyperphe only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

cbsGene.alleleOne - CBS Gene *Check the types of variants found on: Allele 1. (HCY only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

cbsGene.alleleOne - CBS Gene *Check the types of variants found on: Allele 2. (HCY only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

dbtGene.alleleOne - DBT Gene *Check the types of variants found on: Allele 1. (MSUD only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

dbtGene.alleleOne - DBT Gene *Check the types of variants found on: Allele 2. (MSUD only) Acceptable values:*
- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

bckdhbGene.alleleOne - BCKDHB Gene *Check the types of variants found on: Allele 1. (MSUD only) Acceptable values:*
- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

bckdhbGene.alleleOne - BCKDHB Gene *Check the types of variants found on: Allele 2. (MSUD only) Acceptable values:*
- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

dldGene.alleleOne - DLD Gene *Check the types of variants found on: Allele 1. (MSUD only) Acceptable values:*
- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

dldGene.alleleOne - DLD Gene *Check the types of variants found on: Allele 2. (MSUD only) Acceptable values:*
- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

bckdhaGene.alleleOne - BCKDHA Gene *Check the types of variants found on: Allele 1. (MSUD only) Acceptable values:*
- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

bckdhaGene.alleleOne - BCKDHA Gene *Check the types of variants found on: Allele 2. (MSUD only) Acceptable values:*
- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

fahGene.alleleOne - FAH Gene *Check the types of variants found on: Allele 1. (TYR I only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

fahGene.alleleOne - FAH Gene *Check the types of variants found on: Allele 2. (TYR I only) Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

otherGeneName - Other gene name*. Acceptable values: **ANY text**.*

otherGene.alleleOne - Other Gene *Check the types of variants found on: Allele 1. Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

otherGene.alleleTwo - Other Gene *Check the types of variants found on: Allele 2. Acceptable values:*

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

Lysosomal_storage_disorders.csv – There are no diagnostic workup elements to be imported for lysosomal storage disorders at this time. Please use the case_template key for Infant Demographic and Screening Information import fields.

Organic_acid_disorders.csv - Optional fields for:

- 3-Methylcrotonyl-CoA carboxylase deficiency - 3-MCC
- Glutaric acidemia type I - GA1
- Holocarboxylase synthase deficiency - MCD
- Isovaleric acidemia - IVA
- Methylmalonic acidemia (cobalamin disorders) - Cbl A,B
- Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT
- Methylmalonic acidemia with homocystinuria - Cbl C,D
- Propionic acidemia – PROP

finalDiagnosis – Final Diagnosis

- MCD *Acceptable Values*:
  - **Holocarboxylase Deficiency**
  - **Other biotin disorder (not biotinidase deficiency)**
- Methylmalonic acidemia (cobalamin disorders) - Cbl A,B *Acceptable Values*:
  - **Cobalamin A deficiency (CblA)**
  - **Cobalamin B deficiency (CblB)**
  - **Cobalamin Dv2 (CblDv2)**
- Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT *acceptable values*:
  - **Mutase (-) (mut-)**
  - **Mutase (0) (mut0)**
- Methylmalonic acidemia with homocystinuria - Cbl C,D *acceptable values*:
  - **Cobalamin C deficiency (CblC)**
  - **Cobalamin D deficiency (CblD)**
  - **Cobalamin F deficiency (CblF)**
  - **Cobalamin Dv1 deficiency (CblDv1)**
  - **Cobalamin J deficiency (CblJ)**
  - **Other cobalamin deficiency**

otherFinalDiagnosisName – Specify the name for the "other" value when a value containing "other" is selected from in the finalDiagnosis column *(MCD and Methylmalonic acidemia with homocystinuria - Cbl C,D only)*

maternalDeficiencyTested – Was maternal 3-MCC level tested and ruled out? *(3-MCC only)*
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

alphaMccEnzymeAnalysisTested - Was enzyme analysis for 3-MCC enzyme activity completed? *(3-MCC only)*
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

alphaMccEnzymeAnalysisResult **–** Only specified when alphaMccEnzymeAnalysisTested is true/yes. What was enzyme activity? *(3-MCC only)*
Acceptable Values:

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity, not consistent with disease)
- **UNKNOWN**

urineOrganicAcids – Were urine organic acids tested? *(3-MCC, GA1, MCD, PROP only)*
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

isovalericLevel - Only specified when urineOrganicAcids is true/yes. What was 3OH Isovaleric acid level? *(3-MCC only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

methylGlycineLevel - Only specified when urineOrganicAcids is true/yes. What was 3-methylcrotonyl glycine level? *(3-MCC only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

urineThreeOHGlutaricLevel - Only specified when urineOrganicAcids is true/yes. What Was 3-OH Glutaric acid level? *(GA1 only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

urineGlutaricLevel - Only specified when urineOrganicAcids is true/yes. What was the Glutaric acid level? *(GA1 only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

threeOhIsovalericAcidLevel – Only specified when urineOrganicAcids is true/yes. What was the 3OH Isovaleric acid level? *(MCD only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

threeOhPropionicAcidLevel – Only specified when urineOrganicAcids is true/yes. What was the 3OH Propionic acid level? *(MCD and IVAonly)*
MDC Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**
IVA Acceptable Values: **ABSENT, NORMAL, UNKNOWN**

threeMethylcrotonylGlycinthreeOHPropionicAcidLevel – Only specified when urineOrganicAcids is true/yes. What was 3-methylcrotonyl glycine acid level? *(MCD only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

isovalericAcidLevel – Only specified when urineOrganicAcids is true/yes. What was the 3OH Isovaleric acid level? *(IVA only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

isovalerylGlycineLevel – Only specified when urineOrganicAcids is true/yes. What was the Isovaleryl glycine level? *(IVA only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

propionylGlycineLevel – Only specified when urineOrganicAcids is true/yes. Were metabolites for Propionyl glycine detected? *(PROP only)*
Acceptable Values: **ABSENT, NORMAL, UNKNOWN**

tiglyglycineLevel – Only specified when urineOrganicAcids is true/yes. Were metabolites for Tiglyglycine detected? *(PROP only)*
Acceptable Values: **ABSENT, NORMAL, UNKNOWN**

methylcitrateLevel – Only specified when urineOrganicAcids is true/yes. Were metabolites for Methylcitrate detected? *(PROP only)*
Acceptable Values: **ABSENT, NORMAL, UNKNOWN**

mmaLevel – Only specified when urineOrganicAcids is true/yes. Were metabolites for MMA detected? *(PROP only)*
Acceptable Values: **ABSENT, NORMAL, UNKNOWN**

methylcrotonylGlycineLevel – Only specified when urineOrganicAcids is true/yes. Were metabolites for Methyl-crotonyl glycine detected? *(PROP only)*
Acceptable Values: **ABSENT, NORMAL, UNKNOWN**

plasmaOrganicAcidsTested - Were plasma amino acids tested? *(GA1 only)*
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

plasmaThreeOHGlutaricLevel - Only specified when plasmaOrganicAcidsTested is true/yes. What 3-OH Glutaric acid level? *(GA1 only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

plasmaGlutaricLevel - Only specified when plasmaOrganicAcidsTested is true/yes. What Glutaric acid level? *(GA1 only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

urineOrganicAcids - Were urine organic acids tested? *(GA1 only)*
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

urineThreeOHGlutaricLevel - Only specified when urineOrganicAcids is true/yes. What was 3-OH Glutaric acid level? *(GA1 only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

urineGlutaricLevel - Only specified when urineOrganicAcids is true/yes. What was Glutaric acid level? *(GA1 only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

plasmaAcylcarnitines - Were plasma acylcarnitines tested? *(GA1, MCD, and IVA only)*
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

c5dcLevel - Only specified when plasmaAcylcarnitines is true/yes. What was C5 -DC level? *(GA1 only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

c3Level - Only specified when plasmaAcylcarnitines is true/yes. What was C3 Level? *(MCD, "Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, "Methylmalonic acidemia with homocystinuria - Cbl C,D" and PROP only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

c5OhLevel - Only specified when plasmaAcylcarnitines is true/yes. What was C5-OH Level? *(MCD only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

c5Level - Only specified when plasmaAcylcarnitines is true/yes. What was C5 Level? *(IVA only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

plasmaAcylcarnitinesTested **-** Were plasma acylcarnitines tested? *(3-MCC, "Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, "Methylmalonic acidemia with homocystinuria - Cbl C,D", and PROP only)*
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

plasmaAcylcarnitinesLevel **-** Only specified when plasmaAcylcarnitinesTested is true/yes. What was C5-OH level? *(3-MCC only)*
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

enzymeAnalysisCompleted - Was enzyme analysis completed? *(GA1, MCD, IVA only)*

- For GA1 specifically Glutaric Acidemia enzyme activity
- For MCD specifically holocarboxylase synthetase deficiency enzyme activity
- For IVA specifically isovaleryl-CoA dehydrogenase enzyme activity

Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

glutarylCoAEnzymeAnalysisResult – Only specified when enzymeAnalysisCompleted is true/yes. What was enzyme activity *(GA1 only)*

Acceptable Values:

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity, not consistent with disease)
- **UNKNOWN**

pyruvateCarboxylaseEnzymeAnalysisResult – Only specified when enzymeAnalysisCompleted is true/yes. What was enzyme activity *(MCD only)*

Acceptable Values:

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity, not consistent with disease)
- **UNKNOWN**

isovalerylCoAEnzymeAnalysisResult – Only specified when enzymeAnalysisCompleted is true/yes. What was enzyme activity *(IVA only)*

Acceptable Values:

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity, not consistent with disease)
- **UNKNOWN**

biotinidaseStudyCompleted - Were infant chemistries (biotinidase) studies completed? *(MCD only)*

Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

biotinidaseStudyResult – Only specified when biotinidaseStudyCompleted is true/yes. What were infant chemistries (biotinidase) studies?

Acceptable Values: **ABNORMAL, NORMAL, UNKNOWN**

serumMmaLevelTested - Was serum MMA level tested? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D" only)*

Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

serumMmaLevel - Only specified when serumMmaLevelTested is true/yes. What was MMA level in serum? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D" only)*

Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

urineMmaLevelTested - Was urine MMA level tested? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D" only)*

Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

urineMmaLevel - Only specified when urineMmaLevelTested is true/yes. What was MMA level in urine? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D"* only)
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

maternalB12LevelTested – Were maternal vitamin B12 levels tested? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D"* only)
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

maternalB12Level - Only specified when maternalB12LevelTested is true/yes. What was maternal vitamin B12 level? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D"* only)
Acceptable Values: **LOW, NORMAL, UNKNOWN**

infantB12LevelTested – Were infant vitamin B12 levels tested? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D"* only)
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

infantB12Level - Only specified when infantB12LevelTested is true/yes. What was infant vitamin B12 level? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D"* only)
Acceptable Values: **LOW, NORMAL, UNKNOWN**

plasmaHomocysteineLevelTested - Was total plasma homocysteine tested? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D"* only)
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

plasmaHomocysteineLevel - Only specified when plasmaHomocysteineLevelTested is true/yes. What was total plasma homocysteine? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D"* only)
Acceptable Values: **ELEVATED, NORMAL, UNKNOWN**

enzymeComplementationStudyCompleted - Were enzyme complementation studies completed? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D"* only)
Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

enzymeComplementationStudyResult - Only specified when enzymeComplementationStudyCompleted is true/yes. What were the results of the complementation studies? *("Methylmalonic acidemia (cobalamin disorders) - Cbl A,B", Methylmalonic acidemia (methylmalonyl-CoA mutase) – MUT, and "Methylmalonic acidemia with homocystinuria - Cbl C,D"* only)
Acceptable Values:

- **CONSISTENT** (Consistent with disease)
- **INCONSISTENT** (Normal activity, not consistent with disease)
- **UNKNOWN**

mutationAnalysisDone - Was mutation analysis done?

Acceptable Values: **TRUE, FALSE, YES, NO, UNKNOWN**

mccc1Gene.alleleOne – Only specified when mutationAnalysisDone is true/yes. Variant found on MCCC1 Gene, allele *1. (3-MCC only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

mccc1Gene.alleleTwo – Only specified when mutationAnalysisDone is true/yes. Variant found on MCCC1 Gene, allele *2. (3-MCC only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

mccc2Gene.alleleOne – Only specified when mutationAnalysisDone is true/yes. Variant found on MCCC2 Gene, allele *1. (3-MCC only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

mccc2Gene.alleleTwo – Only specified when mutationAnalysisDone is true/yes. Variant found on MCCC2 Gene, allele *2. (3-MCC only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

gcdhGene.alleleOne - Only specified when mutationAnalysisDone is true/yes. Variant found on GCDH Gene, allele 1 *(GA1 only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

gcdhGene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on GCDH Gene, allele 2 *(GA1 only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

hlcsGene.alleleOne - Only specified when mutationAnalysisDone is true/yes. Variant found on HLCS Gene, allele 1 *(MCD only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

hlcsGene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on hlcs Gene, allele 2 *(MCD only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

ivdGene.alleleOne - Only specified when mutationAnalysisDone is true/yes. Variant found on IVD Gene, allele 1 *(IVA only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

ivdGene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on IVD Gene, allele 2 *(IVA only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

methylmalonylCoaMutaseGene.alleleOne - Only specified when mutationAnalysisDone is true/yes. Variant found on METHYLMALONYL-CoA MUTASE Gene, allele 1 *(Methylmalonic acidemia (cobalamin disorders) - Cbl A,B and Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

methylmalonylCoaMutaseGene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on METHYLMALONYL-CoA MUTASE Gene, allele 2 *(Methylmalonic acidemia (cobalamin disorders) - Cbl A,B and Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

mmaaGene.alleleOne - Only specified when mutationAnalysisDone is true/yes. Variant found on MMAA Gene, allele 1 *(Methylmalonic acidemia (cobalamin disorders) - Cbl A,B and Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

mmaaGene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on MMAA Gene, allele 2 *(Methylmalonic acidemia (cobalamin disorders) - Cbl A,B and Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

mmabGene.alleleOne - Only specified when mutationAnalysisDone is true/yes. Variant found on MMAB Gene, allele 1 *(Methylmalonic acidemia (cobalamin disorders) - Cbl A,B and Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

mmabGene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on MMAB Gene, allele 2 *(Methylmalonic acidemia (cobalamin disorders) - Cbl A,B and Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

pccaGene.alleleOne - Only specified when mutationAnalysisDone is true/yes. Variant found on PCCA Gene, allele 1 *(PROP only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

pccaGene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on PCCA Gene, allele 2 *(PROP only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

pccbGene.alleleOne - Only specified when mutationAnalysisDone is true/yes. Variant found on PCCB Gene, allele 1 *(PROP only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

pccbGene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on PCCB Gene, allele 2 *(PROP only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

c3orf25Gene.alleleOne- Only specified when mutationAnalysisDone is true/yes. Variant found on C2ORF25 Gene, allele 1 *(Methylmalonic acidemia with homocystinuria - Cbl C,D only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

c3orf25Gene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on C2ORF25 Gene, allele 2 *(Methylmalonic acidemia with homocystinuria - Cbl C,D only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

mmachcGene.alleleOne- Only specified when mutationAnalysisDone is true/yes. Variant found on MMACHC Gene, allele 1 *(Methylmalonic acidemia with homocystinuria - Cbl C,D only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

mmachcGene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on MMACHC Gene, allele 2 *(Methylmalonic acidemia with homocystinuria - Cbl C,D only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

lmbrd1Gene.alleleOne- Only specified when mutationAnalysisDone is true/yes. Variant found on LMBRD1 Gene, allele 1 *(Methylmalonic acidemia with homocystinuria - Cbl C,D only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

lmbrd1Gene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on LMBRD1 Gene, allele 2 *(Methylmalonic acidemia with homocystinuria - Cbl C,D only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

abcd4Gene.alleleOne- Only specified when mutationAnalysisDone is true/yes. Variant found on ABCD4 Gene, allele 1 *Methylmalonic acidemia with homocystinuria - Cbl C,D only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

abcd4Gene.alleleTwo - Only specified when mutationAnalysisDone is true/yes. Variant found on ABCD4 Gene, allele 2 *(Methylmalonic acidemia with homocystinuria - Cbl C,D only)*

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

otherGeneName - Only specified when mutationAnalysisDone is true/yes. Name of other gene.
Acceptable values: any text.

otherGene.alleleOne – Only specified when mutationAnalysisDone is true/yes. Variant found on Other Gene, allele 1.

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**

otherGene.alleleTwo – Only specified when mutationAnalysisDone is true/yes. Variant found on Other Gene, allele 2.

Acceptable values:

- **DISEASE_CAUSING** (Variant known to be disease causing)
- **UNCERTAIN_SIGNIFICANCE** (Variant of unknown significance)
- **PREDICTED_PATHOGENIC** (Variant of unknown significance predicted to be pathogenic)
- **NONE** (Wild Type (Normal))
- **UNKNOWN**