

INTAKE SURVEY: Homocystinuria (CBS deficiency, MTHFR, Cbl D variant 1, Cbl E, Cbl G)

Demographics

Unique registry ID (2digFIPS/2digBirthYr/1digCenter/4digAccession)	(2digFIPS/2digBirthYr/1digCenter/4digAccession)
Was Intake in IBEM-IS refused?	Yes, No
Permission to contact: I agree to be contacted with information on potential future research applicable to my/my child's inborn error of metabolism that becomes available.	Yes, No
Compensation: I agree that identifying information about me/my child may be used or disclosed as necessary to provide compensation if me/my child are eligible for compensation	Yes, No
Patient is enrolled in other research studies	Missing/unknown data, no, yes - other Region 4 emergency studies, yes - other Region 4 clinical/medication studies, yes - other Region 4 psychosocial studies, yes - studies related to this IBEM not conducted through Region 4, yes - other research
Is patient followed by >1 metabolic center?	Missing/unknown data, yes, no
If patient is followed by >1 metabolic center note which Metabolic Centers in which States (enter N/A if not applicable)	
If patient is followed by >1 metabolic center, did patient grant permission to share data via IBEM-IS between treating metabolic centers?	Missing/unknown data, N/A, Yes, No
If deceased, date of death (if N/A enter 01/01/1901)	
Biological mother's maiden name(enter N/A if unavailable)	
Specify ethnicity if ethnicity is listed as "other", enter N/A if not applicable	
Follow up status	Active, Inactive - Deceased, Inactive - Lost to follow up, Inactive - Moved to another State participating in IBEM-IS, Inactive - Moved to another State not participating in IBEM-IS, Inactive -Refused follow up, Inactive - Treatment deemed not necessary
Socioeconomic Status	
Maternal education: highest level of education	Missing/unknown data, 1-8 years, 9-12 years (no diploma), completed high school, training after high school, some college, college graduate, post-graduate
Paternal education: highest level of education	Missing/unknown data, 1-8 years, 9-12 years (no diploma), completed high school, training after high school, some college, college graduate, post-graduate
If patient >=18 years: highest level of education	Missing/unknown data, N/A-patient age < 18, 1-8 years, 9-12 years (no diploma), completed high school, training after high school, some college, college graduate, post-graduate
Parent/guardian considers patient Hispanic?	Missing/unknown data, yes, no
Is patient/primary caregiver proficient in written English?	Missing/unknown data, yes, no
Is patient/primary caregiver proficient in spoken English?	Missing/unknown data, yes, no

Socioeconomic Status

If >=18 years: was written/web-based information on this IBEM provided in patient's primary language? Missing/unknown data, N/A patient age <18 years, Yes, No

Was written/web-based information on this disorder provided in primary caregiver's primary language? Missing/Unknown data, yes, no

Family History

Consanguinity (defined as any common ancestor)? Missing/unknown data, known consanguinity, no known consanguinity

History of sibling death? Missing/unknown data, yes, no, N/A (only child)

If sibling death(s): enter sibling #, date (s) of death and cause(s) of death if known (enter 99999 if N/A)

Sibling #1 (oldest sibling) evaluated for this IBEM? Missing/unknown data, not tested, tested - affected (if affected and consented to IBEM-IS participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A

Sibling #2 (second oldest sibling) evaluated for this IBEM? Missing/unknown data, not tested, tested - affected (if affected and consented to IBEM-IS participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A

Sibling #3 (third oldest sibling) evaluated for this IBEM? Missing/unknown data, not tested, tested - affected (if affected and consented to IBEM-IS participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A

Sibling #4 (fourth oldest sibling) evaluated for this IBEM? Missing/unknown data, not tested, tested - affected (if affected and consented to IBEM-IS participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A

Sibling #5 (fifth oldest sibling) evaluated for this IBEM? Missing/unknown data, not tested, tested - affected (if affected and consented to IBEM-IS participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A

Sibling #6 (sixth oldest sibling) evaluated for this IBEM? Missing/unknown data, not tested, tested - affected (if affected and consented to IBEM-IS participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A

Sibling #7 (seventh oldest sibling) evaluated for this IBEM? Missing/unknown data, not tested, tested - affected (if affected and consented to IBEM-IS participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A

Sibling #8 (eighth oldest sibling) evaluated for this IBEM? Missing/unknown data, not tested, tested - affected (if affected and consented to IBEM-IS participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A

If sibling(s) affected with this IBEM enter sibling # and sibling unique registry ID number(s) here (enter 99999 if N/A)

Prenatal History

Was prenatal testing for this disorder done during this pregnancy? Missing/unknown data, yes, no

Method(s) if prenatal testing for this disorder done Missing/unknown data, N/A, amniocentesis (biochemical/enzyme), amniocentesis (DNA), chorionic villus (biochemical/enzyme), chorionic villus (DNA), fetal skin biopsy, periumbilical blood sampling (fetal blood), prenatal ultrasound (brain abnormality), prenatal ultrasound (renal abnormality), other

Neonatal History

Additional information about newborn period breastfed, distress, galactose containing formula, IV fluids, jaundiced, non-galactose containing formula, premature (<37 weeks gestation at birth), TPN, transfused

Measurements

Birth weight (enter 99999 if unknown) (kg)

Birth length(enter 99999 if unknown) (cm)

Birth head circumference (OFC), (enter 99999 if unknown) (cm)

Newborn Screening

Days of age at time primary or metabolic provider was notified of 1st abn newborn screen for this IBEM (365 x yrs or 30 x months or counted days)
 enter 99999 if N/A or unknown

State newborn screen serial number
 (enter 99999 if N/A or unknown)

Methionine level on FIRST newborn screen (enter 99999 if N/A) (umol/L)

Methionine/Phenylalanine ratio on FIRST newborn screen (enter 99999 if N/A)

2nd Tier Homocysteine level on FIRST newborn screen (enter 99999 if N/A) (umol/L)

Methionine level on SECOND newborn screen (enter 99999 if N/A) (umol/L)

Methionine/Phenylalanine ratio on SECOND newborn screen (enter 99999 if N/A)

2nd Tier Homocysteine level on SECOND newborn screen (enter 99999 if N/A) (umol/L)

Methionine level on THIRD newborn screen (enter 99999 if N/A) (umol/L)

Methionine/Phenylalanine ratio on THIRD newborn screen (enter 99999 if N/A)

2nd Tier Homocysteine level on THIRD newborn screen (enter 99999 if N/A) (umol/L)

Diagnostic Testing

Diagnosis (pick list: cystathionine beta-synthase (CBS) deficiency, methylene tetrahydrofolate reductase (MTHFR) deficiency, cobalamin D variant 1, cobalamin E, cobalamin G)

Molecular testing: Common or targeted mutation panel Missing/unknown data, not done, abnormal - compound heterozygote, abnormal - homozygote, abnormal - simple heterozygote, alteration(s) of unknown significance detected, no mutations detected

Molecular testing: Full sequencing Missing/unknown data, not done, abnormal - compound heterozygote, abnormal - homozygote, abnormal - simple heterozygote, alteration(s) of unknown significance detected, Presumed compound heterozygote-2nd mutation not identified, no mutations detected

Mutation description: Allele 1 (format example 985A>G)

Mutation description: Allele 2 (format example 985A>G)

Mother's mutation description: Allele 1 (format example 985A>G)

Mother's mutation description: Allele 2 (format example 985A>G)

Father's mutation description: Allele 1 (format example 985A>G)

Father's mutation description: Allele 2 (format example 985A>G)

Plasma amino acid profile Missing/unknown data, Not done, Abnormal, Normal, Non-diagnostic

Plasma total homocysteine level (enter 99999 if missing/unknown) (umol/L)

Plasma methionine level (umol/L)

Peripheral smear Missing/unknown data, not done, abnormal, normal, non-diagnostic

Platelet count (enter 99999 if missing/unknown) (10e9/L)

Serum methylmalonic acid level Missing/unknown data, not done, abnormally high, normal

Serum holo-transcobalamin II (holo-TC II) level Missing/unknown data, not done, abnormally low, normal

Serum vitamin B12 level Missing/unknown data, not done, abnormally low, normal

Urine homocysteine measurement	(ug/mg Cr)
Urine amino acid profile	Missing/unknown data, not done, abnormal, normal, non-diagnostic
Urine organic acids	Missing/unknown data, not done, abnormal, normal, non-diagnostic
Fibroblast C14-propionate tracer assay	Missing/unknown data, not done, Mut 0, Mut -, non-diagnostic
Complementation Analysis (fibroblasts)	Missing/unknown data, not done, normal, abnormal – Mut 0, abnormal - cblA, abnormal - cblB, abnormal-cblC, abnormal-cblD, abnormal- cblF, abnormal-cblH, non-diagnostic
Fibroblast Cobalamin Distribution	Missing/unknown data, not done, OH-cobalamin responsive, OH-cobalamin non-responsive
Maternal lab studies	Missing/unknown data, not done, serum vitamin B12 abnormally low, serum vitamin B12 normal, total plasma homocysteine abnormally high, total plasma homocysteine normal, urine MMA abnormally high, urine MMA normal

Past Health History

Number of hospitalizations prior to Intake in IBEM-IS	0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10, Unknown
Initial diagnosis of this IBEM found by:	Missing/unknown data, abnormal newborn screen, clinical presentation, sibling of patient with IBEM, parent of patient with IBEM, affected mother of child who had abnormal newborn screen, prenatal testing, post-mortem testing
Days of age from birth to initiation of intervention for this IBEM (365 x yrs or 30 x months or counted days), enter 99999 if unknown	
Symptom(s) at time of initial metabolic contact	Missing/unknown data, none, acute liver failure, acute renal failure, alopecia, aneurysm , apnea, arrhythmia, ataxia, athetosis, autistic-like features, body odor, candidiasis, cardiomyopathy, cataract(s), cerebral edema, chorea, cirrhosis, coma, confusion, conjunctivitis, corneal erosion, dehydration, dermatitis, developmental delay(s), dolichostenomelia , dysarthria, dysmorphism, dysphagia, dystonia, ectopia lentis , eczema, edema, failure to thrive, fatigue, hearing loss, hemolytic uremic syndrome , hepatic encephalopathy, hepatomegaly, hypertension , hypertonia, hypotonia, hypothermia, infection/sepsis, irritability, jaundice, keratosis, lethargy, macrocephaly, malignant hyperthermia, microcephaly, multiorgan failure, myocardial infarction , myopathy, optic nerve atrophy, pancreatitis, peripheral neuropathy, photophobia, poor feeding, profuse sweating, pulmonary embolism , pulmonary hypertension , retinal hemorrhage, rickets, rigidity, seizure, splenomegaly, stomatitis, stridor, stroke, subdural hemorrhage, sudden death, syncope, tachycardia, tachypnea, thrombosis , tremors, vascular disease , vision loss, vomiting, other (go to next question to explain)
Other symptom(s) at time of initial metabolic contact (enter N/A if not applicable)	
Lab abnormalities at time patient or primary care provider (on behalf of patient) first contacts metabolic specialist.	Missing/unknown data, no abnormal labs, no labs done, yes-anemia, yes-bone marrow suppression, yes-elevated amylase, yes -elevated CK, yes-elevated lipase, yes -elevated liver function tests, yes-coagulopathy, yes-hematuria, yes -hyperammonemia, yes-hyperglycemia, yes-hyperglycinemia, yes-hypertriglyceridemia, yes - hyperuricemia, yes - hypoglycemia, yes-immunologic abnormalities, yes-ketonuria, yes – ketosis, yes-lactic acidosis, yes-low/absent ketones, yes - metabolic acidosis, yes -myoglobinuria, yes-plasma total carnitine elevation, yes-low plasma free carnitine, yes - low plasma total carnitine, yes-proteinuria, yes -renal tubular acidosis, yes - other (go to next question to explain)

History of transplant?

Missing/unknown data, yes (complete
Transplant survey now), no

Lab tests (other) at time of initial
metabolic contact (indicate type and if
WNL or Abn), enter N/A if not
applicable

Days of age at time of initial face to face
metabolic consultation (365 x yrs or 30
x months or counted days), enter 99999
if unknown

Was genetic counseling for this disorder
provided?

Missing/unknown data, yes, no

Date of last outpatient metabolic visit (if
unknown enter 01/01/1901)

Age at time of first abnormal dexa scan
(enter 99999 if N/A or unknown)

(years)

Echocardiogram results obtained prior
to Intake: enter date of echo and
explain results, enter N/A if not
applicable

Has patient received dialysis prior to
enrollment in the IBEM-IS?

Missing/unknown data, yes (complete Dialysis
survey now), no

Emergency Management

Patient was enrolled in a web-based
emergency alert program?

Missing/unknown data, yes - MEMSCIS, yes -
other web-based program, no

Patient/primary caregiver was given the
24 hour on-call contact information for a
metabolic provider

Missing/unknown data, yes, no

Patient/primary caregiver was given a
written emergency letter for this IBEM?

Missing/unknown data, yes, no

Patient/primary caregiver was given a
sick day plan specific to this IBEM?

Missing/unknown data, yes, no

Education

Are Special Educational services
received by this patient currently?

Missing/unknown data, yes - IEP/IFSP, yes -
504 plan, yes – other

Other Comments