

INTAKE SURVEY: MALONIC ACIDEMIA.DRAFT3

Demographics	
Unique registry ID (2digFIPS/2digBirthYr/1digCenter/4digAssession)	(2digFIPS/2digBirthYr/1digCenter/4digAssession)
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

History of Cardiomyopathy Yes, No, Unknown – no imaging done
Brain malformations (heterotopias/pachygyria) Yes, No, Unknown – no imaging done

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)
C3DC on FIRST newborn screen (enter 99999 if N/A) (umol/L)
C3DC on SECOND newborn screen (enter 99999 if N/A)
C3DC on THIRD newborn screen (enter 99999 if N/A)

Diagnostic Testing

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)

Diagnostic Testing

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 acylcarnitine profile Missing/unknown data, not done, abnormal, normal, non-diagnostic
Urine acylcarnitines Missing/unknown data, not done, abnormal, normal, non-diagnostic
Urine organic acids Missing/unknown data, not done, abnormal, normal, non-diagnostic

MMA elevation Missing/unknown data, not done, abnormal, normal, non-diagnostic
Plasma carnitine levels Missing/unknown data, not done, abnormally low, normal

Comment [RO1]: Common malformations

Comment [RO2]: Sometimes there is a combined phenotype with MA and MMA that seems to be more benign

<p>Malonyl-CoA decarboxylase (MCD) enzyme assay from fibroblasts</p>	<p>Missing/unknown data, not done, abnormally low as compared with control, normal, non-diagnostic</p>
<p>Past Health History</p>	
<p>Number of hospitalizations prior to Intake in IBEM-IS</p>	<p>0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10, Unknown</p>
<p>Initial diagnosis of this IBEM found by:</p>	<p>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</p>
<p>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</p>	
<p>Symptom(s) at time of initial metabolic contact</p>	<p>Missing/unknown data, none, acute liver contact failure, acute renal failure, renal dysplasia, alopecia, 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), dysarthria, dysmorphism, dysphagia, dystonia, eczema, edema, failure to thrive, fatigue, hearing loss, hepatic encephalopathy, hepatomegaly, hypertonia, hypotonia, hypothermia, infection/sepsis, irritability, jaundice, keratosis, lethargy, macrocephaly, malignant hyperthermia, microcephaly, multiorgan failure, myopathy, optic nerve atrophy, pancreatitis, peripheral neuropathy, photophobia, poor feeding, profuse sweating, retinal hemorrhage, rickets, rigidity, seizure, splenomegaly, stomatitis, stridor, stroke, brain abnormalities, subdural hemorrhage, sudden death, syncope, tachycardia, tachypnea, tremors, vision loss, vomiting, other (go to next question to explain)</p>
<p>Other symptom(s) at time of initial metabolic contact (enter N/A if not applicable)</p>	
<p>Lab abnormalities at time patient or primary care provider (on behalf of patient) first contacts metabolic specialist.</p>	<p>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-ketoneuria, 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)</p>
<p>Lab tests (other) at time of initial metabolic contact (indicate type and if WNL or Abn), enter N/A if not applicable</p>	
<p>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</p>	

Was genetic counseling for this disorder provided?	Missing/unknown data, yes, no
Date of last outpatient metabolic visit (if unknown enter 01/01/1901)	
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, no
Imaging Studies	
If abnormal dexa scan since last outpatient metabolic visit (z-score = - 4 to - 3), specify site	Missing/unknown data, N/A, hip, heel, pelvis, spine, total body, wrist, other
Other	
Other Comments	

