

Interval Methylmalonic Acidemia (MMA)

Interval Propionic Acidemia (PA)

Demographics	Today's Action
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
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)	

Socioeconomic Status	Today's Action
Current insurance status	Missing/unknown data, commercial/private insurance, military insurance, none (self-pay), State program (newborn screening funds/special needs program/MCH block grant), State/Federal insurance (Medicaid/Medicare), Other

Measurements	Today's Action
Weight at this visit (enter 99999 if not measured)	(kg)
Height at this visit (enter 99999 if not measured)	(cm)
Head circumference (OFC) at this visit (enter 99999 if not measured)	(cm)

Past Health History	Today's Action
Date of last outpatient metabolic visit (if unknown enter 01/01/1901)	
Has patient had general anesthesia since the last outpatient metabolic visit?	Missing/unknown data, yes, no
Has patient had surgical procedure(s) since the last outpatient metabolic visit?	Missing/unknown data, yes, no
What type of surgical procedure(s) were done since the last outpatient metabolic visit? If not applicable enter N/A	
Complications associated with surgical procedure(s) done since the last outpatient metabolic visit? If not applicable enter N/A	

Emergency Management	Today's Action
Patient/primary caregiver knows how to reach the 24 hour on-call contact information for a metabolic provider	Missing/unknown data, yes, no
Patient/primary caregiver currently has a written emergency letter for this disorder?	Missing/unknown data, yes, no
Patient/primary caregiver currently has a sick day plan specific to this disorder?	Missing/unknown data, yes, no
Patient was enrolled in a web-based emergency alert program?	Missing/unknown data, yes - MEMSCIS, yes - other web-based program, no

Emergency Management	Today's Action
Has patient accessed their web-based emergency plan since the last outpatient metabolic visit?	Missing/unknown data, N/A - patient does not have a web-based emergency plan, yes - MEMSCIS, yes - other web-based emergency plan, no
Number of ER visits since last metabolic visit	Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10, >10
Number of ER visits METABOLIC RELATED since last outpatient metabolic visit	Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10, >10
Total number of ER visits with NO metabolic decompensation since last outpatient metabolic visit	Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10, >10
Number of hospital admissions (total) since last metabolic visit	Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10, >10
Total number of hospital admissions	Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10,

with NO metabolic decompensation since last outpatient metabolic visit	>10
Total number of hospital (inpatient) days METABOLIC RELATED since last outpatient metabolic visit	Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10,11-20,21-30,31-40, 41-50, 51-60,>60
Care Coordination	Today's Action
Other health services received currently	Missing/unknown data, None, Audiology, Behavioral/developmental Pediatrics, Cardiology, Dermatology, Endocrinology, Hematology/oncology, Home health care, Nephrology, Neurology, Neuropsychology, Occupational therapy, Ophthalmology, Orthopedics, Otolaryngology, Physical therapy, Psychiatry, Psychology, Public health nursing, Pulmonology, Respiratory therapy, Speech-language therapy, Transplant evaluation - kidney, Transplant evaluation-heart, Transplant evaluation - liver, Transplant received - kidney, Transplant received - heart, Transplant received-liver, Other
Community resources received currently	Missing/unknown data, none, daycare, family support group related to this IBEM, family support - other, Medical Home, nutritional services (WIC/MAC), personal care attendant (PCA), preschool, Head Start, respite care, social services - county, social services - medical, social services - developmental disability, waived services (CAC/CADI waiver /other waivers), other
Providers seen at this metabolic visit	Dietitian, genetic counselor, neuropsychologist, nurse, nurse practitioner, physician, physician assistant, psychologist, social worker, other (go to next question and explain)
Other providers seen at this metabolic visit(enter N/A if not applicable)	
Developmental Assessment	Today's Action
Developmental screening occurred at this visit?	Missing/unknown data, yes, no
Developmental screening tool used at this visit (if screening was done)	Missing/unknown data, N/A - no developmental screening occurred at this visit, Ages and Stages Questionnaire, Ages and Stages - Social/Emotional Questionnaire, Denver (DDST-II), PEDS Questionnaire, provider history, other standardized screening tool
Developmental milestones achieved at this time?	Missing/unknown data, N/A - no developmental screening occurred at this visit, yes, no
If developmental milestone(s) not achieved, which one(s) were not achieved?	Missing/unknown data, N/A - developmental screening occurred at this visit and all developmental milestones were achieved, N/A - no developmental screening occurred at this visit, Cognitive, Fine motor, Gross motor, Social-emotional, Speech-language
If developmental milestones were not achieved, was patient referred for further developmental evaluation?	Missing/unknown data, N/A - all milestones achieved, N/A - no developmental screening occurred at this visit, yes, no
Was neuropsychological evaluation done since last outpatient metabolic visit? (If yes, complete Neuropsych Survey)	Missing/unknown data, Yes, No
Overall neuropsychological testing impression (from most recent neuropsych evaluation)	Missing/unknown data, N/A - no neuropsych evaluation has been done on this patient, above average, average, below average
Are behavioral concerns suspected at this time?	Missing/unknown data, Yes (go to next question and explain), No
If behavioral concerns are suspected at this time, explain (enter N/A if no behavioral concerns suspected)	
If behavioral concerns are suspected at this time, was patient referred for further evaluation?	Missing/unknown data, N/A - no behavioral concerns suspected, Yes - to behavioral pediatrics, Yes - to neuropsychologist, Yes - to psychiatrist, Yes - to psychologist, Yes - to therapist/counselor, Yes - to other, No
Education	Today's Action
Was patient referred for Special Education evaluation at this time?	Missing/unknown data, yes, no
Are Special Educational services received by this patient currently?	Missing/unknown data, yes - IEP/IFSP, yes - 504 plan, yes - other, no
Special Educational services are received currently: age (in years) child	Missing/unknown data, N/A - no Special Ed services are currently received, N/A - patient is

qualified for services?	>21 years old, <1, >1-2, >2-3, >3-4, >4-5, 6-10, 11-17, 18+
Reason Special Educational services are received currently?	Missing/unknown data, N/A (no Special Ed services are currently received), cognitive disability, fine motor disability, gross motor disability, learning disability, social-emotional disability, speech-language disability, other health impaired (OHI), other
Home Monitoring	
Today's Action	
Patient does home monitoring?	Missing/unknown data, no, yes-urine ketones
Frequency of home monitoring.	Missing/unknown data, N/A, only when symptomatic, weekly, every 2 weeks, monthly, other frequency
Frequency of home monitoring	Missing/unknown data, N/A, only when symptomatic, routinely
Number of home monitoring samples reported since last outpatient metabolic visit (enter 99999 if N/A)	
Laboratory Studies	
Today's Action	
Molecular testing: Common or targeted mutation panel done at this visit(enter specific mutation(s) on Enrollment Survey)?	Missing/unknown data, no, yes - abnormal: compound heterozygote, yes - abnormal: homozygote, yes - abnormal: simple heterozygote, yes - alteration(s) of unknown significance detected, yes but no mutations were detected
Molecular testing: Full sequencing done at this visit(enter specific mutation (s) on Enrollment Survey)?	Missing/unknown data, no, yes - abnormal: compound heterozygote, yes - Presumed compound heterozygote – 2nd mutation not identified, yes - abnormal: homozygote, yes - abnormal: simple heterozygote, yes - alteration (s) of unknown significance detected, yes but no mutations were detected
Lab tests chemistry collected at this visit	Missing/unknown data, none, ABG-WNL, visit ABG-Abn, Anion Gap-WNL, Ammonia – WNL, Ammonia – Abn, Anion Gap-Abn, beta-hydroxybutyrate – WNL, beta-hydroxybutyrate-Abn, Ca++-WNL, Ca++-Abn, Cl-WNL, Cl-Abn, CO2-WNL, CO2-Abn, cortisol-WNL, cortisol-Abn, Gluc-WNL, Gluc-Abn, insulin-WNL, insulin-Abn, K+-WNL, K+-Abn, LDH - WNL, LDH - Abn, Magnesium - WNL, Magnesium - Abn, Na+-WNL, Na+-Abn, Phosphorus - WNL, Phosphorus - Abn, Total Cholesterol(fasting) - WNL, Total Cholesterol (fasting) - Abn, Total Cholesterol(random) - WNL, Total Cholesterol(random) - Abn, Uric Acid - WNL, Uric Acid - Abn, Other
Lab tests hematology collected at this visit	Missing/unknown data, none, Fibrinogen - WNL, Fibrinogen - Abn, Hct-WNL, Hct-Abn, Hgb-WNL, Hgb-Abn, INR-PTT - WNL, INR-PTT - Abn, Peripheral blood smear-WNL, Peripheral blood smear-Abn, Plat-WNL, Plt-Abn, RBC-WNL, RBC-Abn, WBC-WNL, WBC-Abn, Other
Lab tests liver function collected at this visit	Missing/unknown data, none, Albumin-WNL, Albumin-Abn, AlkPhos-WNL, AlkPhos-Abn, ALT-WNL, ALT-Abn, AST-WNL, AST-Abn, direct bili-WNL, direct bili-Abn, GGT - WNL, GGT - Abn, Globulin - WNL, Globulin - Abn, prealbumin-WNL, prealbumin-Abn, total bili-WNL, total bili-Abn, Total Protein - WNL, Total Protein - Abn, Other
Lab tests renal function collected at this visit	Missing/unknown data, none, BUN-WNL, visitBUN-Abn, Cr-WNL, Cr-Abn, 24-hour creatinine clearance-WNL, 24-hour creatinine clearance-Abn, Other
Lab tests miscellaneous collected at this visit	Missing/unknown data, none, BNP-WNL, BNP-Abn, CK-WNL, CK-Abn, CRP-WNL, CRP-Abn, ESR-WNL, ESR-Abn, ferritin-WNL, ferritin-Abn, IgF1-WNL, IgF1-Abn, IgFBP3-WNL, IgFBP3-Abn, transferrin-WNL, transferrin-Abn, TSH-WNL, TSH-Abn, T4 (free)-WNL, T4 (free)-Abn, UA-WNL, UA-Abn, Zinc-WNL, Zinc-Abn, Other (go to next question and explain)
Lab tests other general (indicate type and if WNL or Abn) collected at this visit, enter N/A if not applicable	
Biochemical testing specific to this IBEM collected at this visit	Missing/unknown data, not done, amylase-WNL, amylase-Abn, lipase-WNL, lipase-Abn, plasma acylcarnitines-WNL, plasma acylcarnitines-Abn, plasma amino acids-WNL, plasma amino acids-Abn, plasma carnitine levels-WNL, plasma carnitine levels-Abn, plasma total hcy-WNL, plasma total hcy-Abn, serum B12-WNL, serum B12-Abn, serum MMA-WNL, serum MMA-Abn, urine ketones-WNL, urine ketones-Abn, urine organic acids-WNL, urine organic acids-Abn, Other (go to next question and explain)
Explain all abnormal results checked above	

Enzyme assay from fibroblasts done at this visit? Missing/unknown data, no, yes - abnormal, yes - normal, yes - non-diagnostic

Other biochemical testing specific to this IBEM (indicate type and if WNL or Abn) collected at this visit, enter N/A if not applicable

Imaging Studies

Today's Action

Abdominal imaging done since last outpatient metabolic visit?

Missing/unknown data, no, yes-abdominal CT-WNL, yes-abdominal CT-Abn, yes-abdominal MRI-WNL, yes-abdominal MRI-Abn, yes-abdominal ultrasound-WNL, yes-abdominal ultrasound-Abn, Yes-Abdominal x-ray-WNL, Yes-Abdominal x-ray-abn

Cardiac imaging done since last outpatient metabolic visit?

Missing/unknown data, no, yes-chest x-ray-WNL, yes-chest x-ray-Abn, yes-echocardiogram-WNL, yes-echocardiogram-Abn, yes-EKG-WNL, yes-EKG-Abn, yes-stress test-WNL, yes-stress test-Abn

Musculoskeletal imaging done since last outpatient metabolic visit?

Missing/unknown data, No, Yes-bone x-rays-WNL, Yes-bone x-rays-Abn, Yes-EMG-WNL, Yes-EMG-Abn

Dexa scan since last outpatient metabolic visit (z-score >-2), specify site

Missing/unknown data, N/A, hip, heel, pelvis, spine, total body, wrist, other

If abnormal dexa scan since last outpatient metabolic visit (z-score = -3 to -2), specify site

Missing/unknown data, N/A, hip, heel, pelvis, spine, total body, wrist, other

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

If abnormal dexa scan since last outpatient metabolic visit (z-score <= -4), specify site

Missing/unknown data, N/A, hip, heel, pelvis, spine, total body, wrist, other

Neurological imaging done since last outpatient metabolic visit?

Missing/unknown data, no, yes-cranial ultrasound-WNL, yes-cranial ultrasound-Abn, yes-EEG-WNL, yes-EEG-Abn, yes-head CT-WNL, yes-head CT-Abn, yes-head MRI-WNL, yes-head MRI-Abn

Is there evidence of delayed myelination on CNS imaging since last outpatient metabolic visit?

Missing/unknown data, no, yes, N/A-imaging not done

Is there evidence of thin corpus callosum on CNS imaging since last outpatient metabolic visit?

Missing/unknown data, no, yes, N/A-imaging not done

Is there evidence of ventriculomegaly on CNS imaging since last outpatient metabolic visit?

Missing/unknown data, no, yes, N/A-imaging not done

Is there evidence of abnormalities in the basal ganglia on CNS imaging since last outpatient metabolic visit?

Missing/unknown data, no, yes, N/A-imaging not done

Renal/pelvic/genital imaging done since last outpatient metabolic visit?

Missing/unknown data, no, yes-genitogram-WNL, yes-genitogram-Abn, yes-pelvic ultrasound-WNL, yes-pelvic ultrasound-Abn, yes-renal ultrasound-WNL, yes-renal ultrasound-Abn, yes-testicular ultrasound-WNL, yes-testicular ultrasound-Abn, yes-VCUG-WNL, yes-VCUG-Abn

Other imaging (indicate type of imaging and if WNL or Abn) done since last outpatient metabolic visit?, enter N/A if not applicable

PharmacoTherapy	Today's Action
Is L-carnitine prescribed?	Missing/unknown data, yes, no
If on L-carnitine, dose prescribed(enter 99999 if N/A)?	(mg/kg/day)
If on L-carnitine, route prescribed?	Missing/unknown data, N/A - not prescribed, feeding tube, oral, IV
If on L-carnitine, frequency prescribed?	Missing/unknown data, N/A – not prescribed, once/day, twice/day, three times/day, four times/day, only when ill, other
Patient/primary caregiver reports L-carnitine is taken as prescribed?	Missing/unknown data, N/A – not prescribed, yes - 4-7 days/week, yes - 1 to 3 days/week, no (0 days/week)
If L-carnitine is not taken as prescribed, reason given (enter N/A if not applicable)?	
Method(s) of payment for L-carnitine, if prescribed	Missing/unknown data, N/A - not prescribed, Commercial/private insurance, Military insurance, None - can't afford to fill medication prescription, Self-pay, State program (newborn screening funds/special needs program/MCH block grant), State/Federal insurance (Medicaid/Medicare), Other
Method of payment for L-carnitine, if prescribed	Missing/unknown data, N/A - Not prescribed, commercial/private insurance, military insurance, none - can't afford to fill medication prescription, self-pay, State program (newborn screening funds, special needs program, MCH block grant), State/Federal insurance (Medicaid/Medicare), Other
Is cyanocobalamin (B12) prescribed?	Missing/unknown data, yes, no
If on cyanocobalamin (B12), dose prescribed(enter 99999 if N/A)?	(mg/day)
If on cyanocobalamin (B12), route prescribed?	Missing/unknown data, N/A - not prescribed, FeedingTube, Oral
If on cyanocobalamin (B12),, frequency prescribed?	Missing/unknown data, N/A - not prescribed, Once/day, Twice/day, Three times/day, Four times/day, only when ill, Other
Patient/primary caregiver reports cyanocobalamin (B12), is taken as prescribed?	Missing/unknown data, N/A - not prescribed, Yes - 4-7 days/week, Yes - 1-3 days/week, No (0 days/week)
If cyanocobalamin (B12), is not taken as prescribed, reason given(enter N/A if not applicable)?	
Method of payment for cyanocobalamin (B12), if prescribed	Missing/unknown data, N/A - not prescribed, Commercial/private insurance, State/Federal insurance (Medicaid/Medicare), Military insurance, self-pay, None - can't afford to fill medication prescription, State program (newborn screening funds, special needs program, MCH block grant), Other
Is hydroxocobalamin (B12) prescribed?	Missing/unknown, Yes, No
If on hydroxocobalamin (B12) , dose prescribed(enter 99999 if N/A)?	(mg/kg/day)
If on hydroxocobalamin (B12) , route prescribed?	Missing/unknown data, N/A - not prescribed, FeedingTube, SQ, IM
If on hydroxocobalamin (B12) , frequency prescribed?	Missing/unknown data, N/A - not prescribed, Once/day, Twice/day, Three times/day, Four times/day, only when ill, Other
Patient/primary caregiver reports hydroxocobalamin (B12) is taken as prescribed?	Missing/unknown data, N/A - not prescribed, Yes - 4-7 days/week, Yes - 1-3 days/week, No (0 days/week)
If hydroxocobalamin (B12) is not taken as prescribed, reason given(enter N/A if not applicable)?	
Method of payment for hydroxocobalamin (B12), if prescribed	Missing/unknown data, N/A - not prescribed, commercial/private insurance, military insurance, none - can't afford to fill medication prescription, self-pay, State program (newborn screening funds, special needs program, MCH block grant), State/Federal insurance (Medicaid/Medicare), Other
Is cystadane prescribed?	Missing/unknown data, yes, no
If on cystadane, dose prescribed(enter 99999 if N/A)?	(mg/kg/day)
If on cystadane, route prescribed?	Missing/unknown data, N/A - not prescribed, feeding tube, oral
If on cystadane, frequency prescribed?	Missing/unknown data, N/A - not prescribed, once/day, twice/day, three times/day, four times/day, only when ill, other
Patient/primary caregiver reports	Missing/unknown data, N/A - not prescribed,

cystadane is taken as prescribed?	yes - 4-7 days/week, yes - 1-3 days/week, no (0 days/week)
If cystadane is not taken as prescribed, reason given(enter N/A if not applicable)?	
Method of payment for cystadane, if prescribed	Missing/unknown data, N/A - Not prescribed, commercial/private insurance, military insurance, none - can't afford to fill medication prescription, self-pay, State program (newborn screening funds, special needs program, MCH block grant), State/Federal insurance (Medicaid/Medicare), Other
If on folic acid, dose prescribed(enter 99999 if N/A)?	(mg/kg/day)
If on folic acid, route prescribed?	Missing/unknown data, N/A - not prescribed, feeding tube, oral
If on folic acid, frequency prescribed?	Missing/unknown data, N/A - not prescribed, once/day, twice/day, three times/day, four times/day, only when ill, other
Patient/primary caregiver reports folic acid are taken as prescribed?	Missing/unknown data, N/A - not prescribed, yes - 4-7 days/week, yes - 1-3 days/week, no (0 days/week)
If folic acid is not taken as prescribed, reason given(enter N/A if not applicable)?	
Method of payment for folic acid, if prescribed	Missing/unknown data, N/A - not prescribed, commercial/private insurance, military insurance, none - can't afford to fill medication prescription, self-pay, State program (newborn screening funds, special needs program, MCH block grant), State/Federal insurance (Medicaid/Medicare), Other
If on metronidazole (Flagyl), dose prescribed(enter 99999 if N/A)?	(mg/kg/day)
If on metronidazole (Flagyl), route prescribed?	Missing/unknown data, N/A - not prescribed, feeding tube, oral
If on metronidazole (Flagyl), frequency prescribed?	Missing/unknown data, N/A - not prescribed, once/day, twice/day, three times/day, four times/day, only when ill, other
Patient/primary caregiver reports metronidazole (Flagyl) are taken as prescribed?	Missing/unknown data, N/A - not prescribed, yes - 4-7 days/week, yes - 1-3 days/week, no (0 days/week)
If metronidazole (Flagyl) is not taken as prescribed, reason given(enter N/A if not applicable)?	
Method of payment for metronidazole (Flagyl), if prescribed	Missing/unknown data, N/A - not prescribed, commercial/private insurance, military insurance, none - can't afford to fill medication prescription, self-pay, State program (newborn screening funds, special needs program, MCH block grant), State/Federal insurance (Medicaid/Medicare), Other

Other medications (see optional DocSite detailed medication survey also)

Missing/unknown data, none, Ammonul, analgesics, antacids, antianxiety, antibiotics, anticoagulants/thrombolytics, anticonvulsants, antidepressants, antiemetics, antifungals, antivirals, antihistamines, antihypertensives, antiinflammatories, antipsychotics, antipyretics, aromatase inhibitor, biophosphonates, bronchodilators, contraceptives - oral, contraceptives - injections, corticosteroids, diuretics, estrogen, GnRH analog, growth hormone, immunosuppressives, insulin, insulin sensitizers, iron, laxatives, manitol, progesterone, sleeping medications, testosterone, vitamins, other

Nutrition

Today's Action

What type(s) of milk/formula is patient taking?	Missing/unknown data, None, Baby formula (regular), Baby formula (soy), Breastmilk, Rice milk, Skim milk, 1% milk, 2% milk, Soy milk, Special metabolic formula, Toddler formula (regular), Toddler formula (soy), Whole milk, Other
Is patient prescribed a protein restricted diet?	Missing/unknown data, yes, no
If patient is prescribed a protein restricted diet, prescribed protein grams/day from foods - not including metabolic formula (enter 99999 if N/A)	(grams/day)
If protein restricted diet (from foods, not including metabolic formula) is	

prescribed and followed 0 or 1-3 days/week, reason given (enter N/A if not applicable)	
Is patient prescribed low protein foods?	Missing/unknown data, yes, no
Method of payment for low protein foods, if prescribed	Missing/unknown data, N/A - not prescribed, commercial/private insurance, military insurance, none - can't afford to fill medication prescription, self-pay, State program (newborn screening funds/special needs program/MCH block grant), State/Federal insurance (Medicaid/Medicare), Other (grams/day)
If prescribed metabolic formula, amount of protein prescribed from metabolic formula per day (enter 99999 if N/A):	
If on special metabolic formula, name(s) of formula(s), enter N/A if not applicable	
Patient/primary caregiver reports metabolic formula is taken as prescribed?	Missing/unknown data, N/A-not prescribed, yes - 4-7 days/week, yes - 1-3 days/week, no (0 days/week)
If metabolic formula is prescribed and not taken as prescribed, reason given? (enter N/A if not applicable)	
Method of payment for metabolic formula, if prescribed	Missing/unknown data, N/A - not prescribed, commercial/private insurance, military insurance, none - can't afford to fill prescription, self-pay, State program (newborn screening funds/special needs program/MCH block grant), State/Federal insurance (Medicaid/Medicare), Other
If other nutritional supplementation is taken (explain), enter N/A if not applicable.	
Patient uses feeding device (NG tube, G tube, GJ tube)?	Missing/unknown data, yes, no
Additional nutritional comments? (enter N/A if not applicable)	