

# INTERVAL: 3-MGA TYPE 1

## Demographics

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)

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

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

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

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

Is there evidence of optic nerve atrophy on ophthalmology exam done since last outpatient metabolic visit? Missing/unknown data, N/A –ophthamology exam not done since last outpatient metabolic visit, yes, no

Is there evidence of dystonia on physical exam done today? Missing/unknown data, yes, no

Is there evidence of spasticity on physical exam done today? Missing/unknown data, yes, no

Is there evidence of hypotonia on physical exam done today? Missing/unknown data, yes, no

Is there evidence of ataxia on physical exam done today? Missing/unknown data, yes, no

Is there evidence of speech-language problems on exam done today? 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**

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
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
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 CoordinationToday'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(complete Transplant survey now if not previously done), Transplant received - heart(complete Transplant survey now if not previously done), Transplant received-liver(complete Transplant survey now if not previously done), 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**

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

Has there been a progressive loss of developmental milestones since the last outpatient metabolic visit? 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)

**Education**

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 qualified for services? Missing/unknown data, N/A - no Special Ed services are currently received, N/A - patient is >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

**Laboratory Studies**

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, ABG-Abn, Anion Gap-WNL, Ammonia – WNL, Ammonia – Abn, Anion Gap-Abn, Ca<sup>++</sup>-WNL, Ca<sup>++</sup>-Abn, Cl-WNL, Cl-Abn, CO<sub>2</sub>-WNL, CO<sub>2</sub>-Abn, Gluc-WNL, Gluc-Abn, K<sup>+</sup>-WNL, K<sup>+</sup>-Abn, LDH - WNL, LDH - Abn, Magnesium - WNL, Magnesium - Abn, Na<sup>+</sup>-WNL, Na<sup>+</sup>-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, BUN-Abn, Cr-WNL, Cr-Abn, 24-hour creatinine clearance-WNL, 24-hour creatinine clearance-Abn, Nuclear Medicine GFR-WNL, Nuclear Medicine GFR-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, transferrin-WNL, transferrin-Abn, TSH-WNL, TSH-Abn, T4 (free)-WNL, T4 (free)-Abn, T4(total)-WNL, T4(total)-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, plasma acylcarnitines-WNL, plasma acylcarnitines-Abn, plasma amino acids-WNL, plasma amino acids-Abn, plasma carnitine levels-WNL, plasma carnitine levels-Abn, urine organic acids-WNL, urine organic acids-Abn, urine C5OH acylcarnitine -WNL, urine C5OH acylcarnitine -Abn, Other (go to next question and explain)

Quantitative plasma 3-methylglutconate level (nmol/L)

Quantitative urine 3-hydroxyisovaleric acid level (mmol/mol Cr)

Quantitative urine 3-methylglutaric acid level (mmol/mol Cr)

Quantitative urine 3-methylglutaconic acid level (mmol/mol Cr)

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

Enzyme assay from leukocytes 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

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 abnormal myelination on CNS imaging since last outpatient 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-nuclear medicine DMSA renogram-WNL, yes-nuclear medicine DMSA renogram-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

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 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

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**

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 (grams/day) restricted diet, prescribed protein grams/day from foods - not including metabolic formula (enter 99999 if N/A)

**Nutrition**

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

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

If prescribed metabolic formula, amount of protein prescribed from metabolic formula per day (enter 99999 if N/A): (grams/day)

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), WIC, Other

If other nutritional supplementation is taken (explain), enter N/A if not applicable.

Additional nutritional comments? (enter N/A if not applicable)

**Other**

Other Comments