

Patient: Interval BKT Deficiency

Interval SCOT Deficiency

Demographics

Today's Action

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

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)
 Missing/unknown data, yes, no

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?

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

Emergency Management	Today's Action
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
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 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 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	Missing/unknown data, Yes, No

visit? (If yes, complete Neuropsych Survey)
 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
 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, yes-uses glucometer to check glucose, yes-has ketostix but doesn't use them, yes-has glucometer but doesn't use it**

Frequency of home monitoring Missing/unknown data, N/A, only when symptomatic, routinely

Patient has a prescribed glucometer for this IBEM for home glucose monitoring? Missing/unknown data, yes, no

Lowest reported glucose on home monitoring samples since last outpatient metabolic visit (if N/A enter 99999) (mg/dL)

Number of home monitoring samples reported since last outpatient metabolic visit (enter 99999 if N/A) (integer)

Laboratory Studies

Today's Action

Enzyme assay (specific to this IBEM) done at this visit? Missing/unknown data, no, yes - abnormal, yes - normal, yes - non-diagnostic
 Fatty acid oxidation probe assay done at this visit? Missing/unknown data, no, yes - abnormal, yes - normal, yes - non-diagnostic
 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
 Documented hypoglycemia (blood glucose <60 mg/dL) since the last outpatient metabolic visit? Missing/unknown data, glucose not measured, yes (answer next question), no
 Lowest blood glucose value since last outpatient metabolic visit (enter 99999 if not measured) (mg/dL)

DISCUSS: Does hyperglycemia (during acute episode) need to be added as a data element here? (format similar to the above 2 questions)

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, BUN-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, , bloodspot

acylcarnitines-WNL, bloodspot acylcarnitines-Abn, plasma acylcarnitines-WNL, plasma acylcarnitines-Abn, plasma amino acids-WNL, plasma amino acids-Abn, **plasma carnitine levels -WNL, plasma carnitine levels-Abn low, urine C5OH acylcarnitine-WNL, urine C5OH acylcarnitine-Abn, urine acylglycines-WNL, urine acylglycines-Abn, urine organic acids-WNL, urine organic acids-Abn, other (go to next question and explain)**

Explain all abnormal results checked above

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

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

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
Have glucose paste/gel/tablets been prescribed to this patient for this IBEM?	Missing/unknown data, yes, no
If glucose paste/gel/tablets are prescribed for this IBEM, number of times patient has used them since the last outpatient metabolic visit	N/A - not prescribed, 0,1,2,3,4,5,6,7,8,9,10, >10, Missing/unknown data
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
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
Specific information given to avoid fasting?	Missing/unknown information, yes, no
Longest interval permitted between feedings in well state during DAY (in hours)	Missing/unknown information, no fasting restriction given, 1,2,3,4,5,6,7,8,9,10,11,12
Longest interval permitted between feedings in well state during NIGHT (in hours)	Missing/unknown information, no fasting restriction given, 1,2,3,4,5,6,7,8,9,10,11,12
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

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), 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)	