

Visit Planner

TYROSINEMIA INTERVAL

Demographics

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

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

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

Is there evidence of corneal opacity/crystals on ophthalmology exam done since last exam outpatient visit? Missing/unknown data, N/A - ophthalmology not done since last outpatient visit, 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? 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
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

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

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

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

If 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
Missing/unknown data, N/A (no Special Ed

are received currently?

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

Patient does home monitoring? Missing/unknown data, no, yes-dried bloodspot phenylalanine and tyrosine

Frequency of home monitoring Missing/unknown data, N/A, only when symptomatic, weekly, every two weeks, monthly, other frequency

Recommended therapeutic Phenylalanine goal (enter 99999 if N/A) (umol/L)

Average phenylalanine on dried blood spots done since the last visit (umol/L)

Recommended therapeutic tyrosine goal (enter 99999 if N/A) (umol/L)

Average tyrosine on dried blood spots done since the last visit (umol/L)

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

Laboratory Studies

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, ABG-Abn, Anion Gap-WNL, Ammonia – WNL, Ammonia – Abn, Anion Gap-Abn, Ca⁺⁺-WNL, Ca⁺⁺-Abn, Cl-WNL, Cl-Abn, CO₂-WNL, CO₂-Abn, Gluc-WNL, Gluc-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, 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

Has patient required blood/blood product
transfusion since the last outpatient
metabolic visit? Missing/unknown data, yes (related to poor treatment compliance), yes-unrelated to
tyrosinemia treatment
compliance, no

Has patient undergone liver transplant
since the last outpatient metabolic visit? Missing/unknown data, yes (complete Transplant Survey now), no

Biochemical testing specific to this
IBEM collected at this visit Missing/unknown data, not done, alpha-fetoprotein – Abn high, alpha-fetoprotein –WNL,
plasma tyrosine –Abn high, plasma tyrosine-WNL, plasma phenylalanine-Abn high, plasma
phenylalanine-WNL, plasma methionine-Abn high, plasma methionine-WNL, filter paper
succinylacetone –detected, filter paper succinylacetone-not detected, urine succinylacetone-
detected, urine succinylacetone- not detected, urine organic acids-Abn, urine organic acids-WNL,
Total 25-hydroxyvitamin D -WNL, Total 25-hydroxyvitamin D -Abn low, other (go to next question
and explain)

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

Is there evidence of
hepatocellular carcinoma on imaging
done since last outpatient visit? Missing/unknown data, no, yes, N/A - imaging not done

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 site

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

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 oral nitisinone prescribed? Missing/unknown data, yes, no

If on nitisinone, dose prescribed (enter 99999 if N/A)? (mg/day)

If on nitisinone, frequency prescribed? Missing/unknown data, N/A – not prescribed, once/day, twice/day, three times/day, other

Patient/Primary caregiver reports nitisinone 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 nitisinone is not taken as prescribed, reason given (enter N/A if not applicable)?

Method of payment for nitisinone, 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

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

Per Visit

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

Per Visit

If on a tyrosine restricted diet, prescribed tyrosine in mg/day from foods (enter 99999 if N/A)

(mg/day)

If on a phenylalanine restricted diet, prescribed phenylalanine in mg/day from foods (enter 99999 if N/A)

(mg/day)

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

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

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.

Patient uses feeding device (NG tube,
G tube, GJ tube)?

Missing/unknown data, yes, no

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