

# Enrollment Tyrosinemia

## Demographics

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

Was enrollment 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

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)

Biological mother's maiden name(enter N/A if unavailable)

Specify ethnicity if ethnicity is listed as "other", enter N/A if not applicable

## 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 primary caregiver proficient in written English? Missing/unknown data, yes, no

Is primary caregiver proficient in spoken English? Missing/unknown data, yes, no

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) Missing/unknown data, not tested, tested -

evaluated for this IBEM?	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)	
Age at time of first abnormal dexa scan (enter 99999 if N/A or unknown)	(years)
<b>Prenatal History</b>	
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
<b>Neonatal History</b>	
Additional information about newborn period	Missing/unknown data, none, antibiotics, breastfed, distress, galactose containing formula, IV fluids, jaundiced, non-galactose containing formula, premature (<37 weeks gestation at birth), TPN, transfused
<b>Measurements</b>	
Birth weight (enter 99999 if unknown)	(kg)
Birth length(enter 99999 if unknown)	(cm)
Birth head circumference (OFC), (enter 99999 if unknown)	(cm)
<b>Newborn Screening</b>	
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)	
Tyrosine level on FIRST newborn screen (enter 99999 if N/A)	(umol/L)
2 <sup>nd</sup> Tier succinylacetone on FIRST newborn screen	Missing/unknown data, not done, positive, negative
Methionine on FIRST newborn screen (enter 99999 if N/A)	(umol/L)
Phenylalanine on FIRST newborn screen (enter 99999 if N/A)	(umol/L)
Tyrosine level on SECOND newborn screen (enter 99999 if N/A)	(umol/L)

2 <sup>nd</sup> Tier succinylacetone on SECOND newborn screen	Missing/unknown data, not done, positive, negative
Methionine on SECOND newborn screen (enter 99999 if N/A)	(umol/L)
Phenylalanine on SECOND newborn screen (enter 99999 if N/A)	(umol/L)
Tyrosine level on THIRD newborn screen (enter 99999 if N/A)	(umol/L)
2 <sup>nd</sup> Tier succinylacetone on THIRD newborn screen	Missing/unknown data, not done, positive, negative
Methionine on THIRD newborn screen (enter 99999 if N/A)	(umol/L)
Phenylalanine on THIRD newborn screen (enter 99999 if N/A)	(umol/L)

**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
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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
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Mutation description: Allele 1 (format example Q188R)

Mutation description: Allele 2 (format example Q188R)

Fumarylacetoacetate hydrolase enzyme assay (enter 99999 if unknown or N/A)	Missing/unknown data, not done, abnormally low, normal
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Tyrosine aminotransferase enzyme assay (enter 99999 if unknown or N/A)	Missing/unknown data, not done, abnormally low, normal
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4-Hydroxyphenylpyruvate dioxygenase enzyme assay (enter 99999 if unknown or N/A)	Missing/unknown data, not done, abnormally low, normal
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Specify source tissue for enzyme assay

Urine succinylacetone at time of diagnosis (enter 99999 if N/A)	Missing/unknown data, not done, detected, not detected
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Urine organic acids	Missing/unknown data, not done, abnormal normal, non-diagnostic
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Plasma tyrosine at time of diagnosis	(umol/L)
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Plasma methionine at time of diagnosis	(umol/L)
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Plasma phenylalanine at time of diagnosis	(umol/L)
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Alpha-fetoprotein at time of diagnosis	(ug/L)
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Initial diagnosis of this IBEM found by: 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

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

Symptom(s) at time of initial metabolic  
 contact

Missing/unknown data, none, acute liver  
 failure, acute renal failure, alopecia, apnea,  
 arrhythmia, ataxia, athetosis, 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, multiorgan failure,  
 myopathy, optic nerve atrophy, pancreatitis, **peripheral neuropathy**, **photophobia**,  
 poor feeding, profuse sweating, retinal  
 hemorrhage, **rickets**, rigidity, seizure, splenomegaly,  
 stomatitis, stridor, stroke, subdural  
 hemorrhage, sudden death, syncope, **tachycardia**,  
 tachypnea, tremors, vision loss, vomiting, other  
 (go to next question to explain)

Other symptom(s) at time of initial  
 metabolic contact (enter N/A if not  
 applicable)

Lab abnormalities at time patient or  
 primary care provider (on behalf of  
 patient) first contacts metabolic  
 specialist.

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-ketonuria,  
 yes - ketosis, yes-lactic acidosis,  
 yes-low/absent ketones, yes - metabolic  
 acidosis, yes -myoglobinuria, yes-plasma total  
 carnitine elevation, yes-proteinuria, yes -renal  
 tubular acidosis, yes - other (go to next  
 question to explain)

Lab tests (other) at time of initial  
 metabolic contact (indicate type and if  
 WNL or Abn), enter N/A if not  
 applicable

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

Number of hospitalizations prior to  
 enrollment in IBEM-IS

Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10,  
 >10

Was genetic counseling for this disorder  
 provided?

Missing/unknown data, yes, no

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

**Emergency Management**  
 Patient was enrolled in a web-based

Missing/unknown data, yes - MEMSCIS, yes -

emergency alert program?

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

History of transplant?

Missing/unknown data, yes (complete Transplant survey now), no