

UDP-galactose 4'-epimerase (GALE) INTAKE SURVEY

Demographics

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

Was Intake 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.

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

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

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

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

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

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Parent/guardian considers patient Hispanic?	Missing/unknown data, yes, no
Is patient/primary caregiver proficient in written English?	Missing/unknown data, yes, no
Is patient/primary caregiver proficient in spoken English?	Missing/unknown data, yes, no
Socioeconomic Status	
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) 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

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Sibling #8 (eighth 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

If sibling(s) affected with this IBEM
enter sibling # and sibling unique
registry ID number(s) here (enter 99999
if N/A)

Mean parental height (enter 99999 if
unknown) (inches)

Prenatal History

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

Was maternal galactose intake
restricted during this pregnancy? Missing/unknown data, yes, no

Neonatal History

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

Measurements

Birth weight (enter 99999 if unknown) (kg)

Birth length(enter 99999 if unknown) (cm)

Birth head circumference (OFC), (enter
99999 if unknown) (cm)

Newborn Screening

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)

GALT enzyme activity on FIRST
newborn screen (enter 99999 if N/A) (units/g Hb)

GALT enzyme (Beutler) on FIRST
newborn screen Positive, Negative, N/A

Total galactose level on FIRST
newborn screen (enter 99999 if NA) (mg %)

GALT enzyme activity on SECOND
newborn screen (enter 99999 if N/A) (units/g Hb)

GALT enzyme (Beutler) on SECOND
newborn screen Positive, Negative, N/A

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Total galactose level on SECOND newborn screen (enter 99999 if NA)	(mg %)
GALT enzyme activity on THIRD newborn screen (enter 99999 if N/A)	(units/g Hb)
GALT enzyme (Beutler) on THIRD newborn screen	Positive, Negative, N/A
Total galactose level on THIRD newborn screen (enter 99999 if NA)	(mg %)
Methionine level on FIRST newborn screen (enter 99999 if N/A)	(umol/L)
Tyrosine level on FIRST newborn Screen (enter 99999 if N/A)	(umol/L)
Methionine level on SECOND newborn screen (enter 99999 if N/A)	(umol/L)
Tyrosine level on SECOND newborn screen (enter 99999 if N/A)	(umol/L)
Methionine level on THIRD newborn screen (enter 99999 if N/A)	(umol/L)
Tyrosine level 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

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

Mutation description: Allele 1 (format example Q188R)

Mutation description: Allele 2 (format example Q188R)

Mother's mutation description: Allele 1 (format example 985A>G)

Mother's mutation description: Allele 2 (format example 985A>G)

Father's mutation description: Allele 1 (format example 985A>G)

Father's mutation description: Allele 2 (format example 985A>G)

Clinical Impression Missing/unknown data, generalized GALE deficiency, peripheral GALE deficiency, intermediate GALE deficiency

Galactose-1-phosphate uridylyltransferase (GALT) enzyme assay from RBC (enter 99999 if unknown or N/A) (U/g Hb)

Galactose-1-phosphate uridylyltransferase (GALT) enzyme assay (umol/hr/g Hb)

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from RBC(enter 99999 if unknown or N/A)

UDP-galactose 4'-epimerase (GALE) (umol/hr/g Hb)
enzyme assay from RBC

UDP-galactose 4'-epimerase (GALE) (umol/hr/g Hb)
enzyme assay from WBC

Galactokinase (GALK) enzyme assay (umol/hr/g Hb)
from RBC

Galactosemia Isozymes

Missing/unknown data, not done, D/G, G/G, N/N, N/G, N/D, D/D

Galactose-1-phosphate level (RBC)
prior to initiation of galactose restricted
diet(enter 99999 if unknown or N/A) (mg % or mg/dL)

Reference range for
Galactose-1-phosphate level (RBC)
obtained prior to initiation of galactose
restricted diet(enter 99999 if unknown
or N/A) (mg % or mg/dL)

Galactose-1-phosphate level (RBC)
prior to initiation of galactose restricted
diet(enter 99999 if unknown or N/A) (umol/g Hb)

Reference range for
Galactose-1-phosphate level (RBC)
obtained prior to initiation of galactose
restricted diet(enter 99999 if unknown
or N/A) (umol/g Hb)

Urine galactitol prior to initiation of
galactose restricted diet(enter 99999 if
unknown or N/A) (mmol/mol Cr)

Reference range for urine galactitol
prior to initiation of galactose restricted
diet(enter 99999 if unknown or N/A) (mmol/mol Cr)

Urine reducing substances prior to
initiation of galactose restricted diet
Missing/unknown data, positive color change &
negative for glucose by dipstick, no color
change, N/A - not done

Past Health History

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, autistic-like
features, 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, microcephaly, 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-aminoaciduria**, 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-low plasma free carnitine, yes - low plasma total carnitine, 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)

Age at time of first abnormal dexta scan (enter 99999 if N/A or unknown) (years)

Emergency Management

Patient was enrolled in a web-based emergency alert program? Missing/unknown data, yes - MEMSCIS, yes - 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

Other
Other Comments