

**CMD Children's Metabolic Disorders Parent
Visit Planner**

Date of Visit: ____ / ____ / ____

Provider: _____ **Patient:** Enroll Galactosemia (GALT deficiency) **Gender:** _____ **Phone:** _____
Visit Provider: _____ **MRN:** _____ **DOB:** _____

Conditions: Galactosemia Enrollment **Preferred Language:** _____
Comorbidities: _____ **First Measure Date:** _____

Allergies: _____

Medications: _____

Demographics	Today's Action
Unique registry ID (2digFIPS/2digBirthYr/1digCenter/3digAscension)	
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
Mother's maiden name	
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
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

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Socioeconomic Status
Was written/web-based information on this IBEM provided in primary caregiver's primary language?

Today's Action
Missing/Unknown data, yes, no

Family History Today's Action
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

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)

Mean parental height (enter 99999 if unknown)

(inches)

Prenatal History Today's Action
Was prenatal testing for this IBEM done during this pregnancy?

Last Value
Missing/unknown data, yes, no

Method(s) if prenatal testing for this IBEM 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

Today's Action
Missing/unknown data, none, antibiotics, breastfed, distress, galactose containing formula, non-galactose containing formula, IV fluids, jaundiced, premature (<37 weeks gestation at birth), TPN, transfused

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

Newborn Screening Today's Action
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 (units/g Hb)
(enter 99999 if N/A)

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

Total galactose level on FIRST newborn screen (mg %)
(enter 99999 if N/A)

Methionine level on FIRST newborn screen (umol/L)

Tyrosine level on FIRST newborn screen (umol/L)

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

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

Total galactose level on SECOND newborn screen (mg %)
(enter 99999 if N/A)

Methionine level on SECOND newborn screen (umol/L)

Tyrosine level on SECOND newborn screen (umol/L)

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

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

Total galactose level on THIRD newborn screen (mg %)
(enter 99999 if N/A)

Methionine level on THIRD newborn screen (umol/L)

Tyrosine level on THIRD newborn screen (umol/L)

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MRN:

Gender:
DOB:

Phone:

Diagnostic Testing

Molecular testing: Common or targeted mutation panel

Today's Action

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 985A>G)

Mutation description: Allele 2
(format example 985A>G)

Galactose-1-phosphate uridylyltransferase (GALT) enzyme assay from RBC

Missing/unknown data, Not done, Abnormal < ____, Abnormal >__ < ____, Normal, Non-diagnostic

Galactosemia Isozymes

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

Galactose-1-phosphate level (RBC) prior to Initiation of galactose restricted diet

Missing/unknown data, not done, abnormally high, normal

Urine galactitol prior to initiation of galactose restricted diet

Missing/unknown data, not done, abnormally high, normal, non-diagnostic

Urine reducing substances prior to initiation of galactose restricted diet

Missing/unknown data, positive color change & negative for glucose by dipstick, no color change

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Past Health History**Today's Action**

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, coma, conjunctivitis, dehydration, dermatitis, developmental delay(s), **dysarthria**, **dysphagia**, dystonia, eczema, **failure to thrive**, fatigue, hearing loss, hepatic encephalopathy, hepatomegaly, hypertonia, hypotonia, hypothermia, infection/**sepsis**, irritability, jaundice, lethargy, **macrocephaly**, malignant hyperthermia, myopathy, optic nerve atrophy, pancreatitis, poor feeding, **profuse sweating**, **retinal hemorrhage**, seizure, splenomegaly, stomatitis, stridor, stroke, **subdural hemorrhage**, sudden death, syncope, 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 – coagulopathy, yes -elevated CK, yes - elevated liver function tests, yes - hyperammonemia, yes-hypertriglyceridemia, yes - hyperuricemia, yes - hypoglycemia, yes – ketosis, yes – low/absent ketones, yes - metabolic acidosis, yes -myoglobinuria, yes – plasma total carnitine elevation, 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

0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10, Unknown

Was genetic counseling for this IBEM 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 emergency alert program?

Today's Action

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

Last Visit Comment: **Visit Comment:**