



Region 4 Priority 2 Workgroup
Telemeeting Agenda - Friday, December 19, 2008
1/866/489-0573; *4545164*

I. Welcome

II. Introductions and IBEM-IS Participation Updates – by State Roll Call

Please announce yourself. Your state, and, if applicable, the clinic you are representing and provide a brief update of where your center is in the IBEM-IS process (waiting for approval, finalizing IRB application, enrolling, consenting, etc.)

III. IBEM-IS Update

A. Building the Information System

1. Review set up of surveys for C3 elements
2. GA – 1 Interval Survey (Handout 1)
3. Galactosemia Enrollment Survey (Handout 2)
4. Galactosemia Interval Survey (Handout 3)

B. Implementation

1. Issues and Concerns
 - a. Assignment of UPIN numbers
 - b. Other?
2. SOPs for Implementation
 - a. IRB Renewals – must be submitted to MPH
Updated surveys posted to website
 - b. Invoicing for period ending 12/13/08
 1. Clinical State Lead Activities
 2. Case Enrollment

C. Clinical State Lead Activities

1. Updates on involving other sites within state

IV. Request from DocSite – integrating lab results into DocSite Applications

V. Family Functioning and Chronic Disease Project

VI. Comments / Announcements

- A. Next Meeting – January 16th 1 pm, CT / 2 pm ET
- B. Please enter/update your member information in the member registry
www.region4genetics.org
- C. Other

VII. Adjourn



Region 4 Priority 2 Workgroup
Telemeeting Notes - Friday, November 21, 2008

Participating: Carolyn Anderson - Co-Lead, Susan Berry Co-Lead, Kristi Bentler, Darin Erickson, Anne Jurek, MN; Barb DeLuka, IL; Susie Romie, IN; Jan Askren, KY; Ayesha Ahmad, Jerry Feldman, MI; Nancy, Leslie, Shawn McCandless, Kim Regis, OH; Bill Rhead, WI; Jill Skrabel, NE; Sara Copeland, IA; Jill Shuger, HRSA; Sally Hiner, Region 4 Genetics Collaborative Coordinator.

I. IBEM-IS Participation Updates:

Sue updated on total enrollment to date: 47 subjects enrolled, 43 were MCADD

State/Lead	Center	Status
IL/Burton	Children's Memorial Hospital	No Report
	University of IL	No Report
	Rush University Medical Center	No Report
IN/Hainline	Riley Hospital for Children	Consented 14, none enrolled yet
KY/Gowans	University of KY	No Report
	University of Louisville (Askren)	Application development in process
MI/Feldman	Wayne State	Consenting, none entered to date
	University of MI	Trained
MN/Berry	University of MN	Enrolling all disorders for which data elements are in process - so have consented more than have entered; Done enrollment data for several, starting to add interval surveys
	Mayo	No Report
OH/ Leslie	Cincinnati Children's Hospital	Enrolling, none entered-to-date
	Case Western Reserve	IRB in process
	Nationwide Children's, Columbus	7 Consented, entering data on six of the 7; amended IRB to include phone consent and phone script to be able to contact and enter "in-actives"
	Akron	No Report
	Dayton	No Report
WI/Rhead	Waisman Center	No Report
	Medical Center of WI	IRB Approval, staff change,
NE/Skrabel	Nebraska	Developing IRB Application
IA/Copeland	University of Iowa	Developing IRB Application

Resources for IRB

- Kim Regis will forward her phone consent and script to Sally to post to the Region 4 Website.
- Anyone desiring IRB assistance should contact Sally (shiner@mphi.org)

Resources for Data Entry

- MN has discovered that using the paper visit planner to collect the data and then add it to the Registry later is quickest. We will continue to post the visit planners in pdf form on the Region 4 website so they can be accessed from the Region 4 Website

II. IBEM-IS Update - Building the Information System

A. No disorders have been added this month.

B. Disorder Pending:

a. Disorders which we have ready to add include:

- i. C5 Hydroxy
- ii. Biotinidase
- iii. C3

They will be added in the above order

b. Next to be finalized into final form for submitting to DocSite

- i. SCADD
- ii. Carnitine update
- iii. IBD

c. Disorders needing further input

- i. Glutaric Acidemia Type 1 – we are waiting on input on the neuro elements

d. Disorders needing finalizing by the group

- i. Tyrosynemia
- ii. Galactosemia

This will be on the December Agenda

III. IBEM-IS Implementation

A. Issues and concerns

- 1 Question about MTC Oil – this question on the survey will be reworded to make sure it reflects our intent – want to differentiate when MTC oil is prescribed separately from the metabolic formula.
- 2 As questions/concerns arise or ambiguity is noted, please forward to Sally. (shiner@mphi.org)
- 3 Consents – permission for re-contact. A large number of individuals have not given consent to be re-contacted. It seems the forms may be from one institution. It may be that individuals really did want the “no” box checked, it may mean that the neither the yes or no box was checked. People are reminded to ask the question. Lack of response cannot be taken as consent, so by default must be assumed to be a “no” response. Being able to re-contact is essential for future cohort studies.

B. SOPs for Implementation

- 1 Reminder – IRB Renewals must be submitted to MPHI.
- 2 Data and information required for IRB Renewals – please submit requests for information to Sally. Information of interest to the broader group, such as number enrolled in entire project, will be posted on the Region 4 website so that in the future you will be able to access it without requesting.
- 3 Update on number/case entry for each participating site
 1. This is a reminder that we will be asking each site to report this data at a minimum of every six months.

C. Clinical State Lead Activities

1. Updates on involving other sites within state
 - i. No activity was reported
 - ii. Reminder – this is part of the Clinical State Lead Role and activity will need to be reported when invoicing for the honoraria.

IV. Region 4 Carryforward Request

Jill announced this has been approved and we should be receiving our Notice of Grant Award any day. Of interest to this workgroup, the following activities are funded:

- Support for the IRB process, including completing required forms. As the IBEM-IS expands (EHDI, additional IBEM D/O; and CAH/DSDs) more and more clinicians will be working through the IRB process, perhaps for the first time. MPHI will provide staff support to facilitate the process for all partners.
- Family Functioning and Chronic Disease Project - focus groups of families in 3-7 Region 4 states to gather qualitative data to be used to develop a survey tool
- Expanding our evaluation activities to measure change in practice resulting from Regional activities

Other activities to be supported with carryforward include:

- Case enrollment in the EHDI Registry
- Addition of CAH & DSD to the IBEM-IS
- State meetings on site with the Region 4 Staff Team
- Expanding 3 year follow-up of CH study to additional states
- Expansions of the modified second tier CAH screening protocol
- Adapt MEMSCIS for Sickle Cell, expand beyond IL
- Develop and conduct a survey of families of children with heritable disorders to assess families knowledge of and perceived need for genetic services
- Market and disseminate Region 4 products and best practice guidelines
- MS/MS training of one additional rep from each Region 4 NBS Lab at Mayo Clinic

V. Family Functioning and Chronic Disease Project

Darin will begin to develop an action plan.

VI. Expanding to include other Regions

Sally had a telemeeting with representatives from the Heartland Region and a series of follow up phone calls. The Heartland RC has requested use of carryforward to support clinician participation in the IBEM-IS. Out of the conversations with interested persons from this region, Sally developed and provided a document of FAQs. We have received contact information for 7 individuals in this region who will be participating. Several Heartland sites have already initiated the IRB application process.

VII. December meeting 12/19 –

Sally will poll the workgroup for RSVPs as requested by Sue.

VIII. Comments

A. NBS Symposium - Sue presented on the IBEM-IS at the APHL NBS Symposium in early November. The audience was primarily public health, not as many clinicians. There was a bit of interest in long term follow up, especially as it pertains to public health outcomes since these kids are identified through NBS. It

was a good opportunity for our work to be showcased. Especially since the workgroup has been struggling to foster the partnership between clinicians and public health – we both have the same goal of improved outcomes.

B. Meeting of National data group – The national data Group which ACMG has received funding to support held their first meeting in late October. The group will be working to facilitate national evaluations that will support long term follow up data. We are positioned well to participate because we already have a data collection mechanism

C. Jill Shuger reported that the Secretary's Advisory Committee on Heritable Disorders will be hosting a session on long-term follow-up during a regularly scheduled session this winter. CA, UT and the New England Region will be presenting. UT and NEW England have public health labs collecting the information from clinicians. They are CDC grantees whose goal is to expand the already in place birth – 3 registries to see if they can expand into long term follow up.

D. Jill Shuger noted that regions who are not yet participating in the IBEM-IS may consider writing the activity into their reapplication.

Notes by Hiner

Provider:
Visit Provider:

Patient: Interval GA-1
MRN:

Gender:
DOB:

Phone:

Demographics

Follow up status

Today's Action

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

Today's Action

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)

Today's Action

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?

(free text)

If not applicable enter N/A.

Complications associated with surgical procedure(s) done since the last outpatient metabolic visit?

(free text)

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

Today's Action

Missing/unknown data, yes, no

Patient/primary caregiver currently has a written emergency letter for this IBEM?

Missing/unknown data, yes, no

Patient/primary caregiver currently has a sick day plan specific to this IBEM?

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 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

Date of Visit: ____ / ____ / ____

Provider:
Visit Provider:

Patient: Interval GA-1
MRN:

Gender:
DOB:

Phone:

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

Emergency Management

Today's Action

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

Provider:
Visit Provider:

Patient: Interval GA-1
MRN:

Gender:
DOB:

Phone:

Care Coordination

Other health services received currently

Today's Action

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?

Today's Action

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 ones 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

Date of Visit: ____ / ____ / _____

Provider: _____ **Patient:** Interval GA-1 **Gender:** _____ **Phone:** _____
Visit Provider: _____ **MRN:** _____ **DOB:** _____

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
 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 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

Laboratory Studies **Today's Action**
 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, 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 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**, Plat-WNL, Plt-Abn, RBC-WNL, RBC-Abn, WBC-WNL, WBC-Abn, Other
 Lab tests chemistry collected at this visit Missing/unknown data, none, ABG-WNL, ABG-Abn, Ammonia – WNL, Ammonia – Abn, Anion Gap-WNL, 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 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, Other
 Lab tests miscellaneous collected at this visit Missing/unknown data, none, CK-WNL, CK-Abn, CRP-WNL, CRP-Abn, ESR-WNL, ESR-Abn, ferritin-WNL, ferritin-Abn, transferrin-WNL, transferrin-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, plasma acylcarnitines – WNL, plasma acylcarnitines – Abn, plasma amino acids-WNL, plasma amino acids-Abn, plasma carnitine levels-WNL, plasma carnitine levels-Abn, urine acylcarnitines-WNL, urine acylcarnitines-Abn, urine ketones-WNL, urine ketones-Abn, urine organic acids-WNL, urine organic acids-Abn, Other (go to next question and explain)**

Explain all abnormal results checked above (free text)

Other biochemical testing specific to this IBEM (indicate type and if WNL or Abn) collected at this visit, enter N/A if not applicable

Glutaryl-CoA Dehydrogenase enzyme assay from fibroblasts done at this visit **Missing/unknown data, Not done, Abnormal, Normal, Non-diagnostic**

Glutaryl-CoA Dehydrogenase enzyme assay from leukocytes done at this visit **Missing/unknown data, Not done, Abnormal, Normal, Non-diagnostic**

Provider: **Patient:** Interval GA-1 **Gender:** **Phone:**
Visit Provider: **MRN:** **DOB:**

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 age-WNL, yes-bone age-Abn, yes-bone x-rays-WNL, yes-bone x-rays-Abn, yes-dexa scan-WNL, yes-dexa scan-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
Is there evidence of reduced brain volume due to increased fronto-operculo-temporal extra-axial fluid on central nervous system imaging?	Missing/unknown data, N/A – imaging not done, no, yes
How has the finding of reduced brain volume due to increased fronto-operculo-temporal extra-axial fluid changed from previous imaging?	Missing/unknown data, N/A – no known history of increased fronto-operculo-temporal extra-axial fluid, N/A - repeat imaging not done, greater than previous imaging findings, less than previous imaging findings, same as previous imaging findings
Is there evidence of reduced brain volume due to increased ventricular size on central nervous system imaging?	Missing/unknown data, N/A – imaging not done, no, yes
How has the finding of reduced brain volume due to increased ventricular size changed from previous imaging?	Missing/unknown data, N/A – no known history of increased ventricular size, N/A - repeat imaging not done, greater than previous imaging findings, less than previous imaging findings, same as previous imaging findings
Is there evidence of reduced brain volume due to generalized increased extra-axial fluid on central nervous system imaging?	Missing/unknown data, N/A – imaging not done, no, yes
How has the finding of reduced brain volume due to generalized increased extra-axial fluid changed from previous imaging?	Missing/unknown data, N/A – no known history of generalized increased extra-axial fluid, N/A - repeat imaging not done, greater than previous imaging findings, less than previous imaging findings, same as previous imaging findings
Is there evidence of basal ganglia injury on imaging done since last outpatient metabolic visit?	Missing/unknown data, N/A -imaging not done, no, yes
Is there evidence of subdural hemorrhage on imaging done since last outpatient metabolic visit?	Missing/unknown data, N/A -imaging not done, no, yes
Is there evidence of acute striatal necrosis on imaging done since last outpatient metabolic visit?	Missing/unknown data, N/A -imaging not done, no, yes
Is there evidence of leukodystrophy on imaging done since last outpatient metabolic visit?	Missing/unknown data, N/A -imaging not done, no, yes
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

Is L-carnitine prescribed?

Today's Action

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

Provider:
Visit Provider:

Patient: Interval GA-1
MRN:

Gender:
DOB:

Phone:

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)?

Pharmacotherapy

Method(s) of payment for L-carnitine,
if prescribed

Today's Action

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

Method 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

Is Riboflavin prescribed?

Missing/unknown data, yes, no

If on Riboflavin, dose prescribed
(enter 99999 if N/A)?

(mg/kg/day)

If on Riboflavin, route prescribed?

Missing/unknown data, N/A - not prescribed, feeding tube, oral, IV

If on Riboflavin, 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
Riboflavin 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 Riboflavin is not taken as prescribed,
reason given (enter N/A if not applicable)?

Method(s) of payment for Riboflavin ,
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

Method of payment for Riboflavin,
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

Are other components of GA-1 powder (glutamine,
creatine, lipoic acid, coenzyme Q10, pantothenate)
prescribed (if so, document dose, route, frequency;
if not applicable enter N/A)

(free text)

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,
bronchodilators, contraceptives - injection, contraceptives - oral, corticosteroids, diuretics,
hormones, immunosuppressives, insulin, insulin sensitizers, iron, laxatives, mannitol, sleeping
medications, vitamins, other

Nutrition

Is patient prescribed a protein restricted diet?

Today's Action

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

Provider: _____ **Visit Provider:** _____ **Patient:** Interval GA-1 **Gender:** _____ **Phone:** _____
MRN: _____

What type(s) of milk/formula is patient taking? Missing/unknown data, None, Baby formula (regular), Breastmilk, Rice milk, Skim milk, 1% milk, 2% milk, Soy milk, Special metabolic formula, 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

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)

**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

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

Date of Visit: ____ / ____ / ____

Provider: _____ **Patient:** Enroll Galactosemia **Gender:** _____ **Phone:** _____
Visit Provider: _____ **MRN:** _____ **DOB:** _____

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

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

Date of Visit: ____ / ____ / ____

Provider: **Patient:** Enroll Galactosemia **Gender:**
Visit Provider: **MRN:** **DOB:** **Phone:**

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)

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

Date of Visit: ____ / ____ / ____

Print Date: 9/13/2008

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Provider:
Visit Provider:

Patient: Enroll Galactosemia
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

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

Date of Visit: ____ / ____ / _____

Provider:
Visit Provider:

Patient: Enroll Galactosemia
MRN:

Gender:
DOB:

Phone:

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:**

CMD Children's Metabolic Disorders Parent
Visit Planner

Date of Visit: ____ / ____ / ____

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

Conditions: Galactosemia (GALT deficiency) Interval Update **Preferred Language:**
Comorbidities: **First Measure Date:**

Allergies:

Medications:

Demographics

Follow up status

Today's Action

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

Today's Action

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)

Today's Action

Is there evidence of cataract(s) on ophthalmology exam done since last outpatient metabolic visit?

Missing/unknown data, N/A - ophthalmology exam not done since last outpatient metabolic visit, yes, no

Is there evidence of tremor on physical exam done today?

Missing/unknown data, yes, no

Is there evidence of ataxia on physical exam done today?

Missing/unknown data, yes, no

Is there evidence of speech-language problems on exam done today?

Missing/unknown data, yes, no

Tanner Stage of breast development at this time

Missing/unknown data, I, II, III, IV, V

Tanner Stage of pubic hair development at this time

Missing/unknown data, I, II, III, IV, V

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?

(free text)

If not applicable enter N/A.

Complications associated with surgical procedure(s) (free text)
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

Today's Action

Missing/unknown data, yes, no

Patient/primary caregiver currently has a written emergency letter for this IBEM?

Missing/unknown data, yes, no

Patient/primary caregiver currently has a sick day plan specific to this IBEM?

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

CMD Children's Metabolic Disorders Parent Visit Planner

Date of Visit: ____ / ____ / _____

Print Date: 9/13/2008
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Provider: **Patient:** Interval Galactosemia (GALT deficiency) **Gender:** **Phone:**
Visit Provider: **MRN:** **DOB:**
Conditions: Galactosemia (GALT deficiency) Interval Update **Preferred Language:**
Comorbidities: **First Measure Date:**
Allergies:
Medications:

Has patient accessed their web-based emergency plan since the last 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

Emergency Management

Total number of hospital admissions with NO metabolic decompensation since last outpatient metabolic visit

Today's Action

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

CMD Children's Metabolic Disorders Parent Visit Planner

Date of Visit: ____ / ____ / ____

Provider:
Visit Provider:

Patient: Interval Galactosemia (GALT def)
MRN:

Gender:
DOB:

Phone:

Care Coordination

Other health services received currently

Today's Action

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?

Today's Action

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 ones 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

CMD Children's Metabolic Disorders Parent
Visit Planner

Date of Visit: ____ / ____ / ____

Provider:
Visit Provider:

Patient: Interval Galactosemia (GALT def)
MRN:

Gender:
DOB:

Phone:

Education

Was patient referred for Special Education evaluation at this time?

Today's Action

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

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 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

Laboratory Studies

Today's Action

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, 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 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, Plat-WNL, Plt-Abn, RBC-WNL, RBC-Abn, WBC-WNL, WBC-Abn, Other

Lab tests chemistry collected at this visit

Missing/unknown data, none, ABG-WNL, ABG-Abn, Ammonia - WNL, Ammonia - Abn, Anion Gap-WNL, 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 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, Other

Lab tests miscellaneous collected at this visit

Missing/unknown data, none, CK-WNL, CK-Abn, CRP-WNL, CRP-Abn, ESR-WNL, ESR-Abn, ferritin-WNL, ferritin-Abn, transferrin-WNL, transferrin-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, GALT enzyme activity - Abn < ____, GALT enzyme activity - Abn > ____, galactose-1-phosphate level - WNL, galactose-1-phosphate level elevated but within acceptable treatment range for classic galactosemia, galactose-1-phosphate level above acceptable treatment range for classic galactosemia, urine galactitol - WNL, urine galactitol - Abn high, urine reducing substances - WNL, urine reducing substances - Abn, other (go to next question and explain)

?NTX, osteocalcin, IgF-1, IgFBP-3, 25-OH vitamin D total, PTH, LH, FSH, bone specific alk phos

Explain all abnormal results checked above

(free text)

Other biochemical testing specific to this IBEM (indicate type and if WNL or Abn) collected at this visit, enter N/A if not applicable

Visit Planner

CMD Children's Metabolic Disorders Parent
Date of Visit: ____ / ____ / ____

Provider: **Patient:** Interval Galactosemia (GALT def) **Gender:** **Phone:**
Visit Provider: **MRN:** **DOB:**

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 age-WNL, yes-bone age-Abn, yes-bone x-rays-WNL, yes-bone x-rays-Abn, yes-dexa scan-WNL, yes-dexa scan-Abn, yes-EMG-WNL, yes-EMG-Abn

Age at time of first abnormal dexa scan (enter 99999 if N/A)

(years)

If abnormal dexa scan since last outpatient metabolic visit (z-score ≤ - 2 to 3), specify site

Missing/unknown, N/A, hip, heel, pelvis, spine, wrist

If abnormal dexa scan since last outpatient metabolic visit (z-score ≤ - 3 to 4), specify site

Missing/unknown, N/A, hip, heel, pelvis, spine, wrist

If abnormal dexa scan since last outpatient metabolic visit (z-score ≤ - 4), specify site

Missing/unknown, N/A, hip, heel, pelvis, spine, wrist

Chronological age at time of most recent bone age done since last outpatient metabolic visit

(years, months)

Results of bone age done since last outpatient metabolic visit

(years, months)

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

Is there evidence of cerebral edema on imaging done since last outpatient metabolic visit?

Missing/unknown data, N/A -imaging not done, no, yes

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
Is calcium prescribed?

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

If on calcium, dose prescribed (enter 99999 if N/A)?

(mg/kg/day)

If on calcium, route prescribed?

Missing/unknown data, N/A - not prescribed, feeding tube, oral

If on calcium, frequency prescribed?

Missing/unknown data, N/A – not prescribed, once/day, twice/day, three times/day, other

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

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

Print Date: 9/13/2008
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Date of Visit: ____ / ____ / ____

Provider:
Visit Provider:

Patient: Interval Galactosemia (GALT def)
MRN:

Gender:
DOB:

Phone:

Pharmacotherapy

Method(s) of payment for calcium,
if prescribed

Today's Action

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

?Vitamin D supplement

Is Estrogen (excluding oral contraceptive) prescribed?

Are Biphosphanates prescribed?

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,
bronchodilators, contraceptives - injection, contraceptives - oral, corticosteroids, diuretics,
hormones, immunosuppressives, insulin, insulin sensitizers, iron, laxatives, mannitol, sleeping
medications, vitamins, other

Nutrition

Is patient prescribed a galactose restricted diet?

Today's Action

Missing/unknown data, yes, no

Other diet questions....

What type(s) of milk/formula is patient taking?

Missing/unknown data, None, Baby formula (regular), Breastmilk, Rice milk, Skim milk, 1%
milk, 2% milk, Soy milk, Special metabolic formula, Whole milk, Other

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)

