

**Priority 2: IBEM-IS Pregnancy Survey****Prenatal History**

Patient's age at the time this pregnancy occurred in years (integer field, units "years")

For the current pregnancy, enter Gravida number (enter 99999 if missing/unknown) (integer)

For the current pregnancy, enter number of Term pregnancies (enter 99999 if missing/unknown) (integer)

For the current pregnancy, enter number of Preterm pregnancies (enter 99999 if missing/unknown) (integer)

For the current pregnancy, enter number of pregnancies resulting in Abortion/miscarriage (enter 99999 if missing/unknown) (integer)

For the current pregnancy, enter number of pregnancies resulting in Live births (enter 99999 if missing/unknown) (integer)

First day of Last Menstrual Period (if unknown enter 01/01/1901) (date field)

Estimated Date of Confinement (if unknown enter 01/01/1901) (date field)

At how many weeks gestation was EDC by date and by ultrasound different (enter 99999 if not applicable) (integer field, units "weeks")

How long (in months) did patient try to become pregnant before pregnancy occurred? (pick list – Missing/unknown data, 0-3, 4-6, 7-12, 13-24, >24) (units "months")

History of infertility for patient? (pick list – Missing/unknown data, yes – related to inborn error of metabolism, yes – not known to be related to inborn error of metabolism, no)

Cause(s) of infertility, if known (enter N/A if not applicable, enter unknown if not known) (free text)

Type of fertility treatment(s) received prior to this pregnancy (enter N/A if not applicable or unknown) (free text)

It how many weeks gestation was this pregnancy realized? (enter 99999 if N/A) (integer field, units "weeks")

Was the patient's partner tested for this patient's specific IBEM? (check boxes/check all that apply —Missing/unknown data, yes-biochemical, yes-molecular, no)

Is the patient's partner also affected with this IBEM?  
(pick list – Missing/unknown data, yes, no)

Was preimplantation genetic diagnosis for this IBEM done for this pregnancy?  
(pick list – Missing/unknown data, yes, no)

Was prenatal testing done for this fetus for this IBEM during this pregnancy?  
(pick list —Missing/unknown data, yes, no)

If prenatal testing was performed for this IBEM during this pregnancy, which type of testing was performed?  
(check boxes/check all that apply – Missing/unknown data, biochemical, molecular, chorionic villus sampling, amniocentesis)

Was additional prenatal testing done to assess for fetal effects because of mother's IBEM diagnosis during this pregnancy? Describe testing (example fetal echocardiogram) and results (enter N/A if not applicable) (Free text)

Treatment(s) prescribed for this patient's IBEM during this pregnancy (check boxes/check all that apply – Missing/unknown data, metabolic diet, medications, biochemical lab monitoring, avoidance of fasting, other (specify below)

Other treatments prescribed for this patient's IBEM during this pregnancy (enter N/A if not applicable) (free text)

Was patient in good metabolic control prior to pregnancy? (pick list – Missing/unknown data, yes, no)

Was patient in good metabolic control during the 1<sup>st</sup> trimester of pregnancy? (pick list – Missing/unknown data, yes, no)

Was patient in good metabolic control during the 2<sup>nd</sup> trimester of pregnancy? (pick list – Missing/unknown data, yes, no, N/A – pregnancy not sustained until 2<sup>nd</sup> trimester)

Was patient in good metabolic control during the 3<sup>rd</sup> trimester of pregnancy? (pick list – Missing/unknown data, yes, no, N/A – pregnancy not sustained until 3<sup>rd</sup> trimester)

How many outpatient metabolic visits for this patient occurred during this pregnancy?  
(enter 99999 if missing/unknown data) (integer field)

Number of ED visits for management of IBEM during this pregnancy?  
(pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Number of hospitalizations for management of IBEM during this pregnancy?  
(pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Were there complications during this pregnancy? (pick list – Missing/unknown data, yes-related to IBEM, yes-not known to be related to IBEM, no)

Describe the complications during this pregnancy, if any (enter N/A if not applicable)?  
(Free text)

Number of ED visits for complications during this pregnancy, unrelated to management of this IBEM

(pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Number of hospitalizations for complications during this pregnancy, unrelated to management of this IBEM

(pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Duration of longest inpatient hospitalization (in days) for any reason during this pregnancy

(integer field, units “days”)

Laboratory studies done on patient during pregnancy

(check boxes/check all that apply)

ARE THERE OTHERS, OR DO WE WANT TO ASK THESE QUESTIONS IN ANOTHER WAY?

Missing/unknown data

Ammonia – Abn high

Ammonia – WNL

Blood glucose – Abn low

Blood glucose - WNL

BNP – Abn high

BNP – WNL

CO2 – Abn low

CO2 – WNL

CK – Abn high

CK – WNL

Liver function tests – Abn high

Liver function tests - WNL

Plasma acylcarnitine profile – Abn

Plasma acylcarnitine profile – WNL

Plasma amino acids – Abn

Plasma amino acids – WNL

Plasma carnitine levels – Abn low

Plasma carnitine levels – WNL

Urine acylcarnitines – Abn

Urine acylcarnitines – WNL

Urine acylglycines – Abn

Urine acylglycines –WNL

Urine ketones – Abn high

Urine ketones - WNL

Urine organic acids – Abn

Urine organic acids –WNL

Other (specify below)

Add nutrition labs (prealb, transferrin, INR)

CBC

GTT

Other laboratory studies done on patient during this pregnancy: describe test(s) and result(s), enter N/A if not applicable (free text)

Were additional interventions required during this pregnancy due to this metabolic condition? (check boxes/check all that apply – Missing/unknown data, ED visits for hyperemesis/IVF, hospitalizations for hyperemesis/IVF, TPN, tube feedings, additional medications, home lab monitoring, increased frequency of lab monitoring, more intensive fetal monitoring, bedrest, other)

IBEM related complications during this pregnancy move to disease specific sections, (check list – Missing/unknown data, yes-related to IBEM, yes-not known to be related to IBEM, no)

Non-IBEM related complications during this pregnancy, if any (enter N/A if not applicable)? (Free text) SEE ATTACHED PRAMS QUESTIONS – DO WE WANT TO INCLUDE ANY OF THOSE HERE?

[http://www.cdc.gov/PRAMS/References/Phase6MailCore\\_FINAL.doc](http://www.cdc.gov/PRAMS/References/Phase6MailCore_FINAL.doc)

Total maternal weight gain (in kg) during this pregnancy (enter 99999 if unknown) (integer field)

Was this pregnancy terminated? (check boxes/check all that apply – Missing/unknown data, yes – elective due to fetus affected with inborn error of metabolism, yes – elective due to other fetal well being unrelated to IBEM, yes – elective for other reason, yes-elective due to maternal well being, yes –spontaneous, no)

Gestational age (in weeks) at time of pregnancy termination – elective or spontaneous – (round to the nearest week) (enter 99999 if N/A) (Integer field), (units “weeks”)

**Maternal IBEM-Specific History (Urea Cycle Disorders) – If N/A skip to next disorder-specific section**

Number of documented episodes of hyperammonemia during this pregnancy (pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Treatment(s) for hyperammonemic episodes during this pregnancy (check boxes/check all that apply – Missing/unknown data, hospital admission, protein restriction, Ammonul, arginine)

Was maternal protein intake restricted during this pregnancy?  
(pick list – Missing/unknown data, yes, no)

Number of grams of synthetic protein prescribed per day (enter 99999 if N/A or unknown) (integer field)

Number of grams of natural protein prescribed per day (enter 99999 if N/A or unknown) (integer field)

If ammonia scavenger drugs were used during this pregnancy, which drugs were used?  
(check boxes/check all that apply) – Missing/unknown data, biphenyl, sodium benzoate

Other medications/supplements used during this pregnancy (enter N/A if not applicable)  
(free text)

**Maternal IBEM-Specific History (Organic Acidemias, MSUD) – If N/A skip to next disorder-specific section**

Number of documented episodes of ketonuria during this pregnancy  
(pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Number of documented episodes of metabolic acidosis during this pregnancy  
(pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Number of documented episodes of hyperammonemia during this pregnancy  
(pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Was maternal protein intake restricted during this pregnancy?  
(pick list – Missing/unknown data, yes, no)

Number of grams of synthetic protein prescribed per day (enter 99999 if N/A or unknown) (integer field)

Number of grams of natural protein prescribed per day (enter 99999 if N/A or unknown) (integer field)

If ammonia scavenger drugs were used during this pregnancy, which drugs were used?  
(check boxes/check all that apply) – Missing/unknown data, biphenyl, sodium benzoate

Other medications/supplements used during this pregnancy (enter N/A if not applicable)  
(free text)

**Maternal IBEM-Specific History (Fatty Acid Oxidation Disorders) – If N/A skip to next section**

Was this pregnancy complicated by maternal HELLP syndrome?  
(pick list – Missing/unknown data, yes, no)

Was this pregnancy complicated by acute fatty liver of pregnancy (AFLP)?  
(pick list - Missing/unknown data, yes, no)

Number of documented episodes of hypoglycemia during this pregnancy  
(pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Lowest maternal blood glucose value documented during this pregnancy (enter 99999 if N/A or unknown) (integer field)

Number of documented episodes of rhabdomyolysis during this pregnancy  
(pick list – Missing/unknown data, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10)

Highest maternal CK value documented during this pregnancy (enter 99999 if N/A or unknown) (integer field)

Longest permitted fasting interval (in hours) recommended during this pregnancy (enter 99999 if not applicable or unknown) (integer field, units “hours”)

Was L-carnitine supplementation used on a regular basis during this pregnancy?  
(pick list – Missing/unknown data, yes, no, L-carnitine not prescribed)

Did patient adhere to a fat-restricted diet during this pregnancy?  
(pick list – Missing/unknown data, yes, no, fat-restriction not prescribed)

Did patient take metabolic formula regularly during this pregnancy?  
(pick list – Missing/unknown data, yes, no, metabolic formula not prescribed)

Was there evidence of cardiac hypertrophy during this pregnancy?  
(pick list – Missing/unknown data, yes, no, echocardiogram not performed during pregnancy)

Was there evidence of any cardiac dysfunction during this pregnancy?  
(pick list – Missing/unknown data, yes, no, cardiac testing not performed during pregnancy)

**\*\*\*Stop here if pregnancy was not continued until labor/delivery\*\*\***

#### Maternal Delivery History

Were there additional interventions planned for labor/delivery related to patient’s IBEM?  
(check boxes/check all that apply – Missing/unknown data, no, letter to OB/MFM specialist, IV fluids, planned c-section, additional maternal lab monitoring, change in delivering site, referral for high risk OB management, altered anesthesia plan, no, other (specify below))

Describe other planned interventions for labor/delivery (enter N/A if not applicable) (free text)

Actual interventions for labor/delivery related to patient's IBEM? (check boxes/check all that apply – Missing/unknown data, no, letter to OB/MFM specialist, IV fluids, planned c-section, additional maternal lab monitoring, change in delivering site, referral for high risk OB management, altered anesthesia plan, no, other (specify below)

Acute health concerns experienced by the patient during delivery (enter N/A if not applicable, enter unknown if missing/unknown data) (free text)

Abnormal patient lab results during delivery (check boxes/check all that apply — hypoglycemia, hyperammonemia, metabolic decompensation, elevated liver function tests, no labs checked during delivery, normal labs during delivery **PRAMS**)

Were there additional maternal interventions at or after delivery related to this IBEM (other than labs)? (check boxes/check all that apply—resuscitation, ICU monitoring, medications, infusions, blood/blood product transfusion, TPN, Ammonul, dialysis, other (specify below)

Other specified maternal interventions at or after delivery related to this IBEM (enter N/A if not applicable) (free text)

Did patient die during or shortly after delivery? (pick list – Missing/unknown data, yes, no)

If patient died during or shortly after delivery, was IBEM thought to contribute to death of mother (pick list – Missing/unknown data, yes, no)

**Weeks gestation at time of delivery – round to nearest week (enter 99999 if unknown) (integer field, units “weeks”)**

Method of delivery (pick list—Missing/unknown data, vaginal, scheduled c-section, emergency c-section)

Maternal inpatient days post-delivery (enter 99999 if missing/unknown data) (integer)

**\*\*\*Stop here if delivery of live newborn did not occur\*\*\***

#### Newborn Delivery History

Acute health concerns for newborn related to maternal IBEM (example: fetal distress secondary to maternal acute metabolic decompensation), enter N/A if not applicable (free text)

Additional lab tests and results done on newborn specifically due to maternal IBEM history (enter N/A if not applicable) (free text)

Health concerns for newborn not known to be related to maternal IBEM (enter N/A if not applicable) (free text) – LIST??

Additional interventions for newborn during or shortly after delivery (other than labs) (check boxes/check all that apply—Missing/unknown data, resuscitation, NICU/special care nursery monitoring, medications, glucose infusion, blood/blood product transfusion, TPN, other (specify below)

Other specified neonatal interventions at or after delivery (enter N/A if not applicable) (free text)

Birth Weight (enter 99999 if unknown) (decimal field, units “kg”)

Birth Length (enter 99999 if unknown) (decimal field, units “cm”)

Birth Head Circumference (enter 99999 if unknown) (decimal field, units “cm”)

Apgar score at 1 minute (enter 99999 if unknown) (integer field)

If Apgar score at 1 minute was <8, what were the health concern(s)? (enter N/A if not applicable, enter unknown if not known) (free text)

Apgar score at 5 minutes (enter 99999 if unknown) (integer field)

If Apgar score at 5 minutes was <8, what were the health concern(s)? (enter N/A if not applicable, enter unknown if not known) (free text)

Routine newborn screening done? (pick list – Missing/unknown data, yes-normal for all screened disorders, yes – abnormal (specify below), no (specify reason below)

Describe abnormal newborn screen result (enter N/A if not applicable) (free text)

Explain why routine newborn screening was not done (enter N/A if not applicable) (free text)

Was additional testing (beyond newborn screening) done to rule out the IBEM mother is affected with? (check boxes/check all that apply – Missing/unknown data, yes – biochemical normal, yes – biochemical abnormal, yes – molecular – normal, yes – molecular abnormal affected, yes – molecular abnormal unaffected carrier, no)

Additional studies and results (examples: brain MRI, echocardiogram) done on newborn to assess for effects of maternal IBEM (enter N/A if not applicable) (free text)

Newborn examined by geneticist prior to or shortly after discharge from the birth hospital? (pick list – Missing/unknown data, yes, no)

Abnormal newborn exam findings (examples: dysmorphism, growth retardation, congenital anomalies), if any (enter N/A if not applicable) (free text)

Did newborn die at or shortly after delivery? (pick list – Missing/unknown data, yes, no)

Was newborn's death thought to be related to maternal IBEM (pick list – Missing/unknown data, yes, no)