

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

Patient's age at the time this pregnancy occurred (enter 99999 if missing/unknown) (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 Estimated Date of Confinement 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, N/A – pregnancy was not planned, 0-3, 4-6, 7-12, 13-24, >24) (units "months")

Does the patient have a history of infertility? (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, enter unknown if not known) (free text)

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

How many weeks pregnant was patient when first visit for prenatal care was completed? (enter 99999 if missing/unknown data, enter weeks at time delivery if no prenatal care was received) (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, unknown – partner has not been tested, 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 – 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 1st trimester of pregnancy? (pick list – Missing/unknown data, yes, no)

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

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

Highest value of primary metabolite of concern during this pregnancy (specify
metabolite, value, and units of measurement) (free text)

Lowest value of primary metabolite of concern during this pregnancy (specify metabolite,
value, and units of measurement) (free text)

Laboratory studies done on patient during pregnancy
(check boxes)

Missing/unknown data

Ammonia – Abn high

Ammonia – WNL

Blood glucose – Abn low

Blood glucose – WNL

Blood glucose – Abn high

BNP – Abn high

BNP – WNL
 CBC – Abn
 CBC - WNL
 CO2 – Abn low
 CO2 – WNL
 CK – Abn high
 CK – WNL
 Glucose Tolerance Test (oral) – Abn
 Glucose Tolerance Test (oral) - WNL
 INR – Abn
 INR - 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
 Prealbumin – Abn low
 Prealbumin – WNL
 Transferrin – Abn
 Transferrin - 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)

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/IV fluids, hospitalizations for hyperemesis/IV fluids, 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, if any (enter N/A if not applicable, enter unknown if not known) (free text)

Non-IBEM related complications during this pregnancy, if any (enter N/A if not applicable, enter unknown if not known) (Free text)

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, MSUD, Organic Acidemias) – 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)

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)

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) (units = gm/day)

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

If ammonia scavenger drugs were used during this pregnancy, which drugs were used? (check boxes/check all that apply) – Missing/unknown data, buphenyl, 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) (units = u/l)

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 delivery 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 – 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 actual interventions for labor/delivery (enter N/A if not applicable) (free text)

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 —hypoglycemia, hyperammonemia, metabolic decompensation, elevated liver function tests, no labs checked during delivery, normal labs during delivery

Were there additional maternal interventions during 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) (units = days)

*****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, enter unknown if not known) (free text)

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

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

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)

How long did the baby stay in the hospital after birth? (pick list – Missing/unknown data, <24 hours, 24-48 hours, 3-5 days, 6-14 days, >14 days, N/A – baby was not born in a hospital, baby is still in the hospital)

Routine newborn screening done? (pick list – Missing/unknown data, yes-normal for all screened disorders, yes – abnormal (specify below), yes – results pending, 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 for the baby after birth 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, enter unknown if not known) (free text)

Newborn examined by a genetics professional 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, microcephaly, congenital anomalies, other), if any (enter N/A if not applicable, enter unknown if not known) (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)

Is the baby alive currently? (pick list – Missing/unknown data, yes, no)