

Priority 2: IBEM-IS Pregnancy Survey

Prenatal History

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

Is patient considered advanced maternal age (older than 35 years)? (pick list – Missing/unknown data, yes, no) - Do we need this question considering #1 above?

What pregnancy number is this for this patient?
(pick list – Missing/unknown data, 1,2,3,4,5,6,7,8,9,10,11+)

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)

Dating by ultrasound—enter date - Rephrase?

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

Was this pregnancy planned? (pick list – Missing/unknown data, yes, no)

If pregnancy was not planned, at how many weeks gestation was this pregnancy realized?
(enter 99999 if N/A) (integer field, units "weeks")

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).

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

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 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 teratogenic effects on the fetus during this pregnancy? Describe testing (example fetal echocardiogram) and results (enter N/A if not applicable) (Free text)

Was this pregnancy terminated due to the results of prenatal testing? (pick list – Missing/unknown data, yes, no)

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 (free text)

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

Was metabolic control achieved during this pregnancy? (pick list – Missing/unknown data, yes, no, intermittently)

Gestational age (in weeks) at which time IBEM was controlled well? (enter 99999 if unknown or N/A) (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)

Duration of longest inpatient hospitalization (in days) during this pregnancy (integer field, units "days")

Laboratory studies done on patient during pregnancy (check boxes/check all that apply)

Missing/unknown data

Ammonia – Abn high

Ammonia – WNL

Blood glucose – Abn low

Blood glucose - WNL

BNP – Abn high

BNP – WNL

C02 – Abn low

C02 – 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)

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)

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)

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

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) during this pregnancy (integer field, units “days”)

Was there spontaneous loss of this pregnancy?

(pick list – Missing/unknown data, yes, no)

Gestational age (in weeks) at time of pregnancy loss (enter 99999 if N/A)

(Integer field), (units “weeks”)

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, other (specify below)

Describe other planned 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/check all that apply — hypoglycemia, hyperammonemia, metabolic decompensation, elevated liver function tests, no labs checked during delivery, normal labs during delivery)

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 death determined to be related to patient’s IBEM? (pick list – Missing/unknown data, yes, no)

Newborn Delivery History

Weeks gestation at time of delivery (enter 99999 if unknown) (integer field, units “weeks”)

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

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)

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 teratogenic 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 related to maternal IBEM (pick list – Missing/unknown data, yes, no)

Maternal IBEM-Specific History (Urea Cycle Disorders)

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)

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)

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)