

Visit Planner

Date of Visit: ____ / ____ / ____

Provider:	Patient: Adda Patient	Gender: Female	Phone:
Visit Provider:	MRN:	DOB: 10/13/2011 (0)	
Conditions: GA-1 Intake	Preferred Language:		
Comorbidities:	First Measure Date:		
Other Allergies:			
Drug Allergies:			
Medications:			

Demographics	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
Unique registry ID (2digFIPS/2digBirthYr/1digCenter/4digAssession)	(2digFIPS/2digBirthYr/1digCenter/4digAssession)				Per Visit	
Was Intake in IBEM-IS refused?	Yes, No				Per Visit	
Was assent obtained	yes, no				Per Visit	
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				Per Visit	
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				Per Visit	
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				Per Visit	
Is patient followed by >1 metabolic center?	Missing/unknown data, yes, no				Per Visit	
If patient is followed by >1 metabolic center note which Metabolic Centers in which States (enter N/A if not applicable)					Per Visit	
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				Per Visit	
If deceased, date of death (if N/A enter 01/01/1901)					Per Visit	
Biological mother's maiden name(enter N/A if unavailable)					Per Visit	
Specify ethnicity if ethnicity is listed as "other", enter N/A if not applicable					Per Visit	
Metabolic 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				Per Visit	
Socioeconomic Status	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
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 Associate degree, college graduate, post-graduate, never attended school				Per Visit	
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 Associate degree, college graduate, post-graduate, never attended school				Per Visit	
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 Associate degree, college graduate, post-graduate, never attended school				Per Visit	

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Socioeconomic Status		Today's Action		Last Value	Date Last	Pt. Goal	Int. Days	Due Date
Parent/guardian considers patient Hispanic?		Missing/unknown data, yes, no					Per Visit	
Is patient/primary caregiver proficient in written English?		Missing/unknown data, yes, no					Per Visit	
Is patient/primary caregiver proficient in spoken English?		Missing/unknown data, yes, no					Per Visit	
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					Per Visit	
Was written/web-based information on this disorder provided in primary caregiver's primary language?		Missing/Unknown data, yes, no					Per Visit	
Family History		Today's Action		Last Value	Date Last	Pt. Goal	Int. Days	Due Date
Consanguinity (defined as any common ancestor)?		Missing/unknown data, known consanguinity, no known consanguinity					Per Visit	
History of sibling death?		Missing/unknown data, yes, no, N/A (only child)					Per Visit	
If sibling death(s): enter sibling #, date (s) of death and cause(s) of death if known (enter 99999 if N/A)							Per Visit	
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					Per Visit	
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					Per Visit	
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					Per Visit	
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					Per Visit	
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					Per Visit	
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					Per Visit	
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					Per Visit	
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					Per Visit	
If sibling(s) affected with this IBEM enter sibling # and sibling unique registry ID number(s) here (enter 99999 if N/A)							Per Visit	
Prenatal History		Today's Action		Last Value	Date Last	Pt. Goal	Int. Days	Due Date
Was prenatal testing for this disorder done during this pregnancy?		Missing/unknown data, yes, no					Per Visit	
Method(s) if prenatal testing for this		Missing/unknown data, N/A, amniocentesis					Per Visit	

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disorder done	(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					
Neonatal History	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
Additional information about newborn (defined as 0-28 days of life) period	Missing/unknown data, none, antibiotics, breastfed, distress, galactose containing formula, IV fluids, jaundiced, non-galactose containing formula, premature (<37 weeks gestation at birth), TPN, transfused				Per Visit	
Measurements	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
Birth weight (enter 99999 if unknown)	(kg)				Per Visit	
Birth length(enter 99999 if unknown)	(cm)				Per Visit	
Birth head circumference (OFC), (enter 99999 if unknown)	(cm)				Per Visit	
Newborn Screening	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
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					Per Visit	
State newborn screen serial number (enter 99999 if N/A or unknown)					Per Visit	
C5-DC on FIRST newborn screen (enter 99999 if N/A)	(umol/L)				Per Visit	
C5-DC/C5OH ratio on FIRST newborn screen (enter 99999 if N/A)					Per Visit	
C5-DC on SECOND newborn screen (enter 99999 if N/A)	(umol/L)				Per Visit	
C5-DC/C5OH ratio on SECOND newborn screen (enter 99999 if N/A)					Per Visit	
C5-DC on THIRD newborn screen (enter 99999 if N/A)	(umol/L)				Per Visit	
C5-DC/C5OH ratio on THIRD newborn screen (enter 99999 if N/A)					Per Visit	
Diagnostic Testing	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
Molecular testing: Common or targeted mutation panel	Missing/unknown data, not done, abnormal - compound heterozygote, abnormal - homozygote, abnormal - simple heterozygote, alteration(s) of unknown significance detected, no mutations detected				Per Visit	
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				Per Visit	
Mutation description: Allele 1 (format example 985A>G)					Per Visit	
Mutation description: Allele 2 (format example 985A>G)					Per Visit	
Mother's mutation description: Allele 1 (format example 985A>G)					Per Visit	
Mother's mutation description: Allele 2 (format example 985A>G)					Per Visit	
Father's mutation description: Allele 1 (format example 985A>G)					Per Visit	
Father's mutation description: Allele 2 (format example 985A>G)					Per Visit	
Plasma acylcarnitine profile	Missing/unknown data, not done, abnormal, normal, non-diagnostic				Per Visit	
Urine acylcarnitines	Missing/unknown data, not done, abnormal, normal, non-diagnostic				Per Visit	
Urine organic acids	Missing/unknown data, not done, abnormal, normal, non-diagnostic				Per Visit	

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Plasma carnitine levels	Missing/unknown data, not done, abnormally low, normal				Per Visit	
Glutaryl-CoA Dehydrogenase enzyme assay from fibroblasts	Missing/unknown data, not done, abnormal, normal, non-diagnostic				Per Visit	
Glutaryl-CoA Dehydrogenase enzyme assay from leukocytes	Missing/unknown data, not done, abnormal, normal, non-diagnostic				Per Visit	
Glutaryl-CoA Dehydrogenase enzyme assay from other tissue	Missing/unknown data, not done, abnormal, normal, non-diagnostic				Per Visit	
Past Health History	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
Number of hospitalizations prior to Intake in IBEM-IS	0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, >10, Unknown				Per Visit	
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				Per Visit	
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					Per Visit	
Symptom(s) at time of initial metabolic contact	Missing/unknown data, none, acute liver failure, acute renal failure, alopecia, apnea, arrhythmia, ataxia, athetosis, autistic-like features, body odor, brain abnormalities, brain malformation, candidiasis, cardiomyopathy, cataract(s), cerebral edema, chorea, cirrhosis, clonus, cognitive impairment, coma, confusion, conjunctivitis, contracture(s), corneal erosion, dehydration, dermatitis, developmental delay(s), disorientation, drooling/hypersalivation, dysarthria, dysmetria, dysmorphism, dysphagia, dysplastic kidney(s), dystonia, eczema, edema, failure to thrive, fatigue, flapping tremor, fluctuating level of alertness, gait abnormality, genital abnormalities, headache, hearing loss, hepatic encephalopathy, hepatomegaly, hyperreflexia, hypertension, hyporeflexia, hypertonia, hypotonia, hypothermia, increased intracranial pressure, infection/sepsis, irritability, jaundice, keratosis, learning disability, lethargy, loss of consciousness, loss of developmental milestone(s), macrocephaly, malignant hyperthermia, microcephaly, multiorgan failure, myopathy, optic nerve atrophy, pancreatitis, peripheral neuropathy, photophobia, polycystic kidney(s), poor feeding, poor growth, profuse sweating, renal dysplasia, retinal hemorrhage, rickets, rigidity, scotomas, seizure, slurred speech, spasticity, splenomegaly, stomatitis, stridor, stroke, subdural hemorrhage, sudden death, syncope, tachycardia, tachypnea, tremors, trichorrhexis nodosa, vision loss, vomiting, other (go to next question to explain)				Per Visit	
Other symptom(s) at time of initial metabolic contact (enter N/A if not applicable)					Per Visit	
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-aminoaciduria, yes-anemia, yes-bone marrow suppression, yes-elevated amylase, yes -elevated CK, yes-elevated lipase, yes -elevated liver function tests, yes-coagulopathy, yes-hematuria,				Per Visit	

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Past Health History	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
	yes-hemolytic anemia, yes - hyperammonemia, yes-hyperglycemia, yes-hyperglycinemia, yes-hypertriglyceridemia, yes - hyperuricemia, yes - hypoglycemia, yes-hypokalemia, yes-hypoproteinemia, yes-immunologic abnormalities, yes-ketonuria, yes - ketosis, yes-lactic acidosis, yes-low/absent ketones, yes - metabolic acidosis, yes -myoglobinuria, yes-plasma total carnitine elevation, yes-low plasma free carnitine, yes - low plasma total carnitine, yes-proteinuria, yes -renal tubular acidosis, yes-respiratory alkalosis, 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					Per Visit	
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					Per Visit	
Was genetic counseling for this disorder provided?	Missing/unknown data, yes, no				Per Visit	
Date of last outpatient metabolic visit (if unknown enter 01/01/1901)					Per Visit	
Identify specific birth defects	Missing/unknown data, none, Patent Ductus Arteriosus (P.D.A.), Atrial Septal Defect (A.S.D.), Ventricular Septal Defect (V.S.D.), Atrioventricular Septal Defect, Transposition of the great arteries, Tetralogy of Fallot, Tricuspid Valve Atresia and Stenosis, Pulmonary Valve Atresia and Stenosis, Truncus arteriosus, Endocardial Cushion Defect, Coarctation of Aorta, Ebstein's Anomaly, Hypoplastic Left Heart Syndrome, Aortic Valve Stenosis, Common Truncus, Bronchopulmonary Dysplasia, Esophageal Atresia/Tracheoesophageal Fistula, Rectal and Large Intestinal Atresia/Stenosis, Pyloric Stenosis, Hirshsprung's Disease, Biliary Atresia, Renal Agenesis/Hypoplasia, Congenital Hypothyroidism, Congenital Adrenal Hyperplasia, Epilepsy, Hydrocephalus, Microcephalus, Severe Combined Immunodeficiency, Club Foot, Missing or reduction of limb, Gastroschisis, Omphalocele, Congenital Hip Dislocation, Diaphragmatic Hernia, Spina Bifida, Hearing Loss, Retinopathy of prematurity, Blind, Congenital Cataract, Hemoglobinopathies, Hemophilia, Hypospadias and Epispadias, Bladder Exstrophy, Obstructive Genitourinary Defect, Down Syndrome, Klinefelter Syndrome, Fetal Alcohol Syndrome, Trisomy 13 (Patau syndrome), Trisomy 18, Cleft lip and/or palate, Choanal Atresia, Other (explain below)				Per Visit	
List specific comorbidities (enter N/A if not applicable, enter unknown if missing information)					Per Visit	
Emergency Management	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
Patient was enrolled in a web-based emergency alert program?	Missing/unknown data, yes - MEMSCIS, yes - other web-based program, no				Per Visit	
Patient/primary caregiver was given the 24 hour on-call contact information for a metabolic provider	Missing/unknown data, yes, no				Per Visit	
Patient/primary caregiver was given a written emergency letter for this IBEM?	Missing/unknown data, yes, no				Per Visit	
Patient/primary caregiver was given a	Missing/unknown data, yes, no				Per Visit	

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Emergency Management	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date	
Patient/primary caregiver was given a sick day plan specific to this IBEM?	missing/unknown data, yes, no				Per Visit		
Other	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date	
Other Comments					Per Visit		

Last Visit Comment:

Visit Comment: