

Region 4 Priority 2: IBEM-IS Survey Status

Condition	Intake Survey Status	Interval Survey Status
<i>Amino Acidemias</i>		
Maple syrup urine disease	completed	completed
Homocystinuria (CBS, MTHFR, Cbl D variant 1, Cbl E, Cbl G)	completed	completed
Tyrosinemia	completed	completed
Arginemia		
Argininosuccinate acidemia (ASA)		
Citrullinemia Type I (argininosuccinate synthetase)		
Citrullinemia Type II (citrin deficiency)		
Hypermethioninemia		
Defects of bipterin cofactor biosynthesis/regeneration		
Hyperphenylalaninemia/phenylketonuria		
<i>Fatty Acid Oxidation</i>		
Carnitine uptake deficiency (CUD)	completed	completed
CACT deficiency	completed	completed
CPT-1 deficiency	completed	completed
CPT-2 deficiency	completed	completed
SCAD deficiency	completed	completed
MCAD deficiency	completed	completed
LCHAD deficiency	completed	completed
Trifunctional protein deficiency	completed	completed
VLCAD deficiency	completed	completed
Dienoyl-CoA reductase deficiency		
Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)		

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Glutaric acidemia type II	Revisions pending at Covisint	Revisions pending at Covisint
Medium/Short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)		
<i>Organic Acidemias</i>		
2-methylbutyryl-CoA dehydrogenase deficiency	completed	completed
Isovaleric acidemia	completed	completed
Glutaric acidemia type I	completed	completed
Isobutyryl-CoA dehydrogenase deficiency	completed	completed
3-MCC deficiency	completed	completed
2-methyl 3-hydroxybutyryl CoA dehydrogenase (2M3HBA) deficiency	completed	completed
Holocarboxylase synthetase deficiency	completed	completed
3-methylglutaconic aciduria type I	completed	completed
Beta-ketothiolase	completed	completed
Succinyl CoA-3-keto transferase (SCOT) deficiency	completed	completed
3-hydroxy 3-methylglutaryl (HMG) CoA lyase deficiency	completed	completed
Propionic Acidemia	completed	completed
MMA (Mut-, Mut0, cbl A, cbl B, cbl D variant 2)	completed	completed
MMA + Hcy (Cbl C, Cbl D, Cbl F, Transcobalamin II)	completed	Revisions pending at Covisint
Malonic acidemia	Reviewed by WG,	Reviewed by WG, submitting to Covisint after further revision

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	submitting to Covisint after further revision	
<i>Non-MS/MS Conditions</i>		
Biotinidase deficiency	completed	completed
GALT deficiency	completed	completed
Galactokinase (GALK) deficiency galactosemia	Pending WG review 03/18/11	Pending WG review 03/18/11
UDP-galactose-4-epimerase (GALE) deficiency galactosemia	Pending WG review 03/18/11	Pending WG review 03/18/11
<i>Other Surveys</i>		
Neuropsychological survey		completed
Dialysis survey		completed
Pregnancy survey		completed
Acute cerebral event/imaging survey		
Transplant survey		
Death survey		

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