

Region 4 IBEM-IS Survey Status

<u>Enrollment</u>	<u>Interval</u>	<u>Disorders (30)</u>	<u>Posted R4 Website</u>
X	X	Maple syrup urine disease	Both
X	X	Glutaric Acidemia Type I	Enrollment only
X	X	Isobutyryl-CoA dehydrogenase deficiency	Both
X	X	Carnitine uptake disorder	Both
X	X	CACT deficiency	Both
X	X	CPT-1 deficiency	Both
X	X	CPT-2 deficiency	Both
X	X	SCAD deficiency	Both
X	X	MCAD deficiency	Both
X	X	LCHAD deficiency	Both
X	X	Trifunctional protein deficiency	Both
X	X	VLCAD deficiency	Both
X	**	3-MCC deficiency	Enrollment only
X	!	2-methyl 3-hydroxybutyryl CoA dehydrogenase deficiency	Enrollment only
X	***	Holocarboxylase synthetase deficiency	Enrollment only
X	***	3-methylglutaconic aciduria type I	Enrollment only
X	!	Beta-ketothiolase deficiency	Enrollment only
X	***	3-hydroxy 3-methylglutaryl (HMG) CoA lyase deficiency	Enrollment only
X	!	Succinyl CoA-3-keto transferase (SCOT) deficiency	Enrollment only
X	!	Propionic Acidemia	Enrollment only
X	!	MMA (includes Mut-, Mut0, cobalamin A, cobalamin B, Cobalamin D variant 2)	No
X	!	MMA + Hcy (includes Cobalamin C, Cobalamin F, Transcobalamin II deficiency)	No
X	X	Biotinidase deficiency	Both
X	X	GALT deficiency	No
***	***	Tyrosinemia	No
		?Isovaleric acidemia	
		?2-methylbutyryl-CoA dehydrogenase deficiency	
		?Homocystinuria (includes Cobalamin G, Cobalamin E, CBS, MTHFR)	

Prepared by: Kristi Bentler  
Date: 03/09/09  
Rev: 07/14/09

## Region 4 IBEM-IS Survey Status

X=completed and available in IBEM-IS

\*= in process at DocSite

\*\*= reviewed by group preparing to send to DocSite

!= scheduled for P2 workgroup review/discussion 07/17/09

\*\*\* = needs to be reviewed by P2 workgroup

Prepared by: Kristi Bentler

Date: 03/09/09

Rev: 07/14/09