

<u>Enrollment</u>	<u>Interval</u>	<u>Disorders</u>	<u>Posted R4 Website</u>
X	X	Maple syrup urine disease	Both
X	X	Glutaric Acidemia Type I	Both
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
X	**	2-methyl 3-hydroxybutyryl CoA dehydrogenase deficiency	Enrollment
X	*	Holocarboxylase synthetase deficiency	Enrollment
X	***	3-methylglutaconic aciduria type I	Enrollment
X	X	Beta-ketothiolase deficiency	Both
X	***	3-hydroxy 3-methylglutaryl (HMG) CoA lyase deficiency	Enrollment
X	X	Succinyl CoA-3-keto transferase (SCOT) deficiency	Both
X	X	Propionic Acidemia	Both
X	X	MMA (Mut-, Mut0,cobalamin A,cobalamin B,Cobalamin D variant 2)	Both
X	***	MMA + Hcy (Cobalamin C, Cobalamin F, Transcobalamin II)	Enrollment
X	X	Biotinidase deficiency	Both
X	X	GALT deficiency	Both
***	***	Tyrosinemia	No
***	***	Isovaleric acidemia	No
***	***	2-methylbutyryl-CoA dehydrogenase deficiency	No
***	***	Homocystinuria (Cobalamin G,Cobalamin E,CBS,MTHFR)	No
***	***	?Phenylketonuria	No
***	***	?Urea cycle disorders	No

Prepared by: Kristi Bentler

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X	X	Neuropsychological survey	Yes
***	***	Dialysis survey	No
***	***	Pregnancy survey	No
***	***	Imaging survey	No
***	***	Transplant survey	No

X=completed and available in IBEM-IS

*= in process at DocSite

**= reviewed by group preparing to send to DocSite

*** = needs to be reviewed by P2 workgroup