

Table 1: Disorders Screened

Grouping	Metabolites Measured	Disorder	Abbrev	New or Existing Screen
Metabolic Disorders				
Amino Acid Disorders	Amino Acids	Phenylketonuria	PKU	Existing
		Maple Syrup Urine Disease	MSUD	New
		Citrullinemia	CIT	New
		Argininosuccinic Acidemia	ASA	New
		Homocystinuria	Hcy	New
		Tyrosinemia, Type I	Tyr I	New
Fatty Acid Oxidation Disorders	Acylcarnitines	Medium-chain Acyl-CoA Dehydrogenase Deficiency	MCAD	Existing
		Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD	Existing
		Very-long Chain AcylCoA Dehydrogenase Deficiency	VLCAD	New
Organic Acid Disorders	Acylcarnitines	Propionic Acidemia	PROP	New
		Methylmalonic Acidemia	MUT	New
		Cobalamin Disorders	Cbl A, B	New
		Glutaric Aciduria, Type I	GA I	Existing
		Isovaleric Acidemia	IVA	New
Galacto-semia	GALT enzyme activity		GALT	Existing
Endocrine Disorders	Thyroid stimulating hormone (TSH)	Congenital Hypothyroidism	CH	Existing
	17OH-progesterone (1 st tier) Steroid panel (2 nd tier)	Congenital Adrenal Hyperplasia	CAH	New (starting in the fall 2010)
Hemoglobinopathies	Hemoglobin HPLC	Sickle Cell Disease & related disorders	SCD	New
Cystic Fibrosis	Immunoreactive trypsinogen (IRT) (1 st tier) CFTR mutation panel (2 nd tier)		CF	New