Medical Condition Testing Platform / Analyte  Initiations and Interference Normal Range Propionic acidemia (PA)  Tandem mass spectrometry (MS/MS)  Tandem mass spectrometry (MS/MS)  Tandem mass spectrometry (MS/MS)  Tandem mass spectrometry (MS/MS)  C3, C3/C2  Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Methylmaionic acid (MMA)  Tandem mass spectrometry (MS/MS)  C3, C3/C2  Unsatisfactory specimens or speci	II I - II:	_	rders (Those with these disorders a			•	
Methylmalonic acid (MMA) Tandem mass spectrometry (MS/MS) C3, C3/C2  Tandem mass spectrometry (MS/MS) C4  Tandem mass spectrometry (MS/MS) C5  Tandem mass spectrometry (MS/MS) C6  Tandem mass spectrometry (MS/MS) C7  Tandem mass spectrometry (MS/MS) C8  Tandem mass spectrometry (MS/MS) C9  Tandem mass spectrometry	Medical Condition	Testing Platform/ Analyte	Limitations and Interference	Normal Range	Symptoms if not treated	Common Medical Treatment	Confirmatory Test
Unsatisfactory specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Methylmalonic acid (MMA)  Methylmalonic acid (MMA)  Tandem mass spectrometry (MS/MS) C3, C3/C2  Methylmalonic acid emia (IVA)  Tandem mass spectrometry (MS/MS) C3, C3/C2  Tandem mass spectrometry (MS/MS) C3 (Tandem mass spectrometry (MS/MS) C4 (Tandem mass spectrometry (MS/MS) C5 (Tandem mass spectrometry (MS/MS) C5 (Tandem mass spectrometry (MS/MS) C5 (Tandem mass spectrometry (MS/MS) C6 (Tandem mass spectrometry (MS/MS) C7 (Tandem mass spectrometry (MS/MS) C8 (Tandem mass spectrometry (MS/MS) C9	Propionic acidemia (PA)	Tandem mass spectrometry	Total parenteral nutrition (TPN	Normal	Breathing problems,	Protein-restricted diet;	Referral to metabolic
specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Methylmalonic acid (MMA)  Tandem mass spectrometry (MS/MS) C3, C3/C2  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Sovaleric acidemia (IVA)  Tandem mass spectrometry (MS/MS) C5  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens or or intellectual disabilities  Total parenteral nutrition (TPN and blood transfusions) Unsatisfactory specimens or specimens or or specimens or or intellectual disabilities  Total parenteral nutrition (TPN and blood transfusions) Unsatisfactory specimens or specimens or or specimens or or the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions) Unsatisfactory specimens or specimens or specimens or specimens or or the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions) Unsatisfactory specimens or specime		(MS/MS)	and blood transfusions		seizures, swelling of the	medical formula; no fasting;	center for urine
hours of age or received after seven days may limit the ability of the assay to detect disease states  Methylmalonic acid (MMA)  Tandem mass spectrometry (MS/MS)  C3, C3/C2  Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  S-methylcrotonyl CoA carboxylase deficiency (MS/MS)  Tandem mass spectrometry (MS/MS)  Tandem mass spectrometry (MS/MS)  C5OH  Tandem mass spectrometry (MS/MS)  Tandem mass spectrometry (MS/MS)  C5 Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimen		C3, C3/C2	Unsatisfactory specimens or		brain, stroke, coma/death.	carnitine and biotin therapy	organic acids analysis,
seven days may limit the ability of the assay to detect disease states  Methylmalonic acid (MMA)  Tandem mass spectrometry (MS/MS)  C3, C3/C2  Unsattifactory specimens or specimens or specimens or seven days may limit the ability of the assay to detect disease states  Isovaleric acidemia (IVA)  Tandem mass spectrometry (MS/MS)  C5  Tandem mass spectrometry (MS/MS)  Tandem mass spectrometry (MS/MS)  C5  Tandem mass spectrometry (MS/MS)  Tandem			_ ·				· ·
Methylmalonic acid (MMA)  Mothylmalonic acid (Mishins)  Mothylmalo			hours of age or received after				analysis, and/or
Methylmalonic acid (MMA)  Tandem mass spectrometry (MS/MS) (A3, C3/C2  Total parenteral nutrition (TPN and blood transfusions C3, C3/C2  Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Sovaleric acidemia (IVA)  Tandem mass spectrometry (MS/MS) C5  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions) Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions) Unsatisfactory specimens or specimens or specimens or specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions) Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Total parenteral nutrition (TPN and blood transfusions) Unsatisfactory specimens or speci			· · ·				mutation analysis
Methylmalonic acid (MMA)  Tandem mass spectrometry (MS/MS) C3, C3/C2  Unsatisfactory specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  3-methylcrotonyl CoA  Tandem mass spectrometry (MS/MS)  Tandem mass spectrometry (MS/MS)  Tandem mass spectrometry  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Total parenteral nutrition (TPN and blood transfusions under the parenteral nutrition (TPN and blood transfusions)  Unsatisfactory specimens or specimen			of the assay to detect disease				
(MS/MS) C3, C3/C2 Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions can be provided after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions can be provided after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions can be provided after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions can be provided after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions can be provided after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions can be provided after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions can be provided at the provided and transfusions can be provided and transfusion analysis and/or mutation analysis can be provided and transfusion analysis and/or mutation analysis can			states				
C3, C3/C2 Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens or specimens or specimens or days may limit the ability of the assay to detect disease states  3-methylcrotonyl CoA carrboxylase deficiency (MS/MS)  C5OH  Tandem mass spectrometry (MS/MS)  C5OH  Unsatisfactory specimens or spec	Methylmalonic acid (MMA)	Tandem mass spectrometry	Total parenteral nutrition (TPN	Normal	Breathing problems,	Protein/fat-restricted diet;	Referral to metabolic
Specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Tandem mass spectrometry (MS/MS)  C5  Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions or intellectual disabilities of the assay to detect disease states  Total parenteral nutrition (TPN and blood transfusions or intellectual disabilities or intellectual disabilities or intellectual disabilities  Protein-restricted diet; no fasting; carnitine and glycine therapy organic acids analysis, specific enzyme analysis, and/or mutation analysis or intellectual disabilities  Protein-restricted diet; no fasting; carnitine and glycine therapy organic acids analysis, specific enzyme analysis, and/or mutation analysis or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease or the damage resulting in life-long learning problems or life-l					-	medical formula; no fasting;	center for urine
hours of age or received after seven days may limit the ability of the assay to detect disease states  Isovaleric acidemia (IVA)  Tandem mass spectrometry (MS/MS)  C5  Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  3-methylcrotonyl CoA carboxylase deficiency (3MCC)  C5OH  Unsatisfactory specimens or specimens or specimens or specimens or states  Total parenteral nutrition (TPN and blood transfusions)  Unsatisfactory specimens or specimens or specimens or states  Normal  Brain damage resulting in life-long learning problems or intellectual disabilities  or intellectual disabilities  or intellectual disabilities  brain, stroke, coma/death.  (MS/MS)  C5OH  Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  1. Ver failure and brain damage resulting in life-long learning problems, specific enzyme analysis, and/or mutation analysis  analysis, and/or metabolic center for urine or intellectual disabilities  Protein-restricted diet; no fasting; carnitine and glycine therapy or ganic acids analysis, specific enzyme analysis, and/or mutation analysis  All of the assay to detect disease states  Normal  Breathing problems, seizures, swelling of the brain, stroke, coma/death.  Liver failure and brain damage resulting in life-long learning problems or intellectual disabilities  All of the assay to detect disease analysis, and/or mutation analysis and/or mutation analysis and/or mutation analysis and/or mutation analysis.		C3, C3/C2			brain, stroke, coma/death.	, , , , , , , , , , , , , , , , , , , ,	
seven days may limit the ability of the assay to detect disease states    Sovaleric acidemia (IVA)   Tandem mass spectrometry (MS/MS)   Total parenteral nutrition (TPN and blood transfusions (MS/MS)   Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states    3-methylcrotonyl CoA (arboxylase deficiency (MS/MS)   Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states    3-methylcrotonyl CoA (arboxylase deficiency (MS/MS)   Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease (International Control of the days may limit the ability of the assay to detect disease (International Control of the days may limit the ability of the assay to detect disease (International Control of the days may limit the ability of the assay to detect disease (International Control of the assay to detect disease)			'			B-12 therapy	· ·
of the assay to detect disease states  Tandem mass spectrometry (MS/MS) C5 Unsatisfactory specimens or specimens collected before 24 hours of age or received after serven days may limit the ability of the assay to detect disease  3-methylcrotonyl CoA carboxylase deficiency (3MCC)  C5OH  Tandem mass spectrometry (MS/MS)  Total parenteral nutrition (TPN and bility of the assay to detect disease states  Total parenteral nutrition (TPN and bilox)  Total parenteral nutrition (TPN and bilox analysis, and/or mutation analysis  Total parenteral nutrition (TPN analysis, and/or mutation analysis  Total parenteral nutrition (TPN analysis, and/or mutation analysis)  Total parenteral nutrition (TPN analysis, and/or mutation analysis)  Total parenteral nutrition (TPN analysis, and/or mutation analysis, and/or mutation analysis  Total parenteral nutrition (TPN analysis, and/or mutation analysis  Total parenteral nutrition (TPN analysis, and/or mutation analysis  Total parenteral nutrition (TPN an			_				•
Isovaleric acidemia (IVA)  Tandem mass spectrometry (MS/MS)  C5  Unsatisfactory specimens or specimens collected before 24 hours of age or received after carboxylase deficiency (3MCC)  Tandem mass spectrometry  (MS/MS)  Tandem mass spectrometry  (MS/MS)  Tandem mass spectrometry  (MS/MS)  Tandem mass spectrometry  Total parenteral nutrition (TPN and blood transfusions or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  Tandem mass spectrometry  Total parenteral nutrition (TPN and blood transfusions or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease of the distance of the specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease of the distance of the							mutation analysis
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(MS/MS) C5 Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  3-methylcrotonyl CoA carboxylase deficiency (3MCC)  Tandem mass spectrometry (MS/MS)  C5OH  Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease    Iife-long learning problems or intellectual disabilities   therapy   center for urine organic acids analysis, specific enzyme analysis, specific enzyme analysis, specific enzyme analysis, and/or mutation analysis							
C5 Unsatisfactory specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  3-methylcrotonyl CoA carboxylase deficiency (MS/MS)  (3MCC)  Tandem mass spectrometry (MS/MS)  C5H  Tandem mass spectrometry (MS/MS)  C5OH  Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Or intellectual disabilities  therapy  organic acids analysis, specific enzyme analysis, specific enzyme analysis, specific enzyme analysis, specific enzyme analysis, and/or mutation analysis	Isovaleric acidemia (IVA)	· · · · · ·		Normal			Referral to metabolic
specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states  3-methylcrotonyl CoA carboxylase deficiency (MS/MS)  CSOH  Tandem mass spectrometry (MS/MS)  CSOH  Total parenteral nutrition (TPN and blood transfusions (MS/MS)  CSOH  Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease							
hours of age or received after seven days may limit the ability of the assay to detect disease states  3-methylcrotonyl CoA carboxylase deficiency (MS/MS) (3MCC)  CSOH		C5			or intellectual disabilities	therapy	-
seven days may limit the ability of the assay to detect disease states  3-methylcrotonyl CoA carboxylase deficiency (MS/MS)  CSOH  C			'				·
of the assay to detect disease states  3-methylcrotonyl CoA carboxylase deficiency (MS/MS)  C5OH  C6DH  C6DH			_				
states  3-methylcrotonyl CoA carboxylase deficiency (3MCC)  CSOH  Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  States  Total parenteral nutrition (TPN and blood transfusions (MS/MS)  Seizures, swelling of the brain, stroke, coma/death.  Liver failure and brain damage resulting in lifelong learning problems or intallectical disabilities.  States  Referral to metabolic center for urine organic acids analysis, specific enzyme analysis, and/or mutation analysis							mutation analysis
Tandem mass spectrometry (MS/MS) (3MCC)  Tandem mass spectrometry (MS/MS)  C5OH  Total parenteral nutrition (TPN and blood transfusions (Disabilities)  Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Total parenteral nutrition (TPN and blood transfusions (Disabilities)  Breathing problems, seizures, swelling of the brain, stroke, coma/death.  Liver failure and brain damage resulting in life-long learning problems or intellected disabilities.							
carboxylase deficiency (3MCC)  (MS/MS)  C5OH  Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  (MS/MS)  Seizures, swelling of the brain, stroke, coma/death.  Liver failure and brain damage resulting in lifelong learning problems or intellectual disabilities.							
Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Unsatisfactory specimens or specimens or specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  Drain, stroke, coma/death.  Liver failure and brain damage resulting in life-long learning problems or intellectual disabilities.	·	· · · · · ·		Normal			
specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease  specific enzyme analysis, and/or damage resulting in life- long learning problems or intellectual disabilities	•						
hours of age or received after seven days may limit the ability of the assay to detect disease  Liver failure and brain damage resulting in life-long learning problems or intellectual disabilities.	(3MCC)	С5ОН			brain, stroke, coma/death.	carnitine therapy if deficient	-
seven days may limit the ability of the assay to detect disease  damage resulting in life- long learning problems or					Liver failure and brain		·
of the assay to detect disease long learning problems or			_				
intellectual disphilities							mutation analysis
			, and the second				

3-hydroxy-3- methylglutaryl CoA lyase deficiency (HMG)	Tandem mass spectrometry (MS/MS)	Total parenteral nutrition (TPN and blood transfusions	Normal	Breathing problems, seizures, coma/death	Protein restriction medical formula, no fasting	Referral to metabolic center for urine
, , , ,	C5OH, C6DC	Unsatisfactory specimens or		, ,	, ,	organic acids analysis,
		specimens collected before 24				specific enzyme
		hours of age or received after				analysis, and/or
		seven days may limit the ability				mutation analysis
		of the assay to detect disease				
		states				
Multiple carboxylase	Tandem mass spectrometry	Total parenteral nutrition (TPN	Normal	Difficulty breathing, motor	Biotin therapy	Referral to metabolic
deficiency (MCD)	(MS/MS)	and blood transfusions		skill delay, hearing loss,		center for urine
	C50H	Unsatisfactory specimens or		speech loss, lack of		organic acids analysis,
		specimens collected before 24		coordination, seizures,		specific enzyme
		hours of age or received after		brain damage, death		analysis, and/or
		seven days may limit the ability				mutation analysis
		of the assay to detect disease				
		states				
Beta-ketothiolase	Tandem mass spectrometry	Total parenteral nutrition (TPN	Normal	Intellectual disability,	Protein restricted diet, no	Referral to metabolic
deficiency (BKT)	(MS/MS)	and blood transfusions		enlarged heart, poor	fasting, carnitine therapy,	center for urine
	C5:1, C5OH	Unsatisfactory specimens or		growth, abnormal muscle	bicitra supplement	organic acids analysis,
		specimens collected before 24		tone, jerky movement, low		specific enzyme
		hours of age or received after		white blood cells and		analysis, and/or
		seven days may limit the ability		platelets.		mutation analysis
		of the assay to detect disease				
		states				
Glutaric acidemia, type 1	Tandem mass spectrometry	Total parenteral nutrition (TPN	Normal	Muscle contractions and	Protein restriction; carnitine	Referral to metabolic
(GA-1)	(MS/MS) C5DC	and blood transfusions		spasms, poor coordination	therapy, riboflavin	center for urine
		Unsatisfactory specimens or		and balance, metabolic	supplement	organic acids analysis,
		specimens collected before 24		acidosis, seizures, swelling		specific enzyme
		hours of age or received after		of the brain, coma/death		analysis, and/or
		seven days may limit the ability				mutation analysis
		of the assay to detect disease				
	   Fatty Δcid Ων	states  kidation Disorders (People affecte	d with these disorder	   sare difficulty hurning fat fo	or energy)	
	-	1 1			2.1.	
Medical Condition	Testing Platform/ Analyte	Limitations and Interference	Normal Range	Symptoms if not treated	Common Medical Treatment	Confirmatory Test

Carnitine uptake defect	Tandem mass spectrometry	Total parenteral nutrition (TPN	Normal	Breathing problems,	Avoid fasting; low fat diet;	Plasma and urine
(CUD)	(MS/MS)	and blood transfusions		swelling of the brain,	Carnitine therapy	carnitine analysis
		Unsatisfactory specimens or		seizures, coma/death,		
	Free Carnitine	specimens collected before 24		enlarged heart or liver,		Referral to metabolic
		hours of age or received after		muscle weakness, anemia		center for specific
		seven days may limit the ability				enzyme analysis,
		of the assay to detect disease				metabolite analysis,
		states				and/or mutation
						analysis
Medium chain acyl- CoA	Tandem mass spectrometry	Total parenteral nutrition (TPN	Normal	Breathing problems,	Avoid fasting; low fat diet;	Quantitative plasma
dehydrogenase deficiency	(MS/MS)	and blood transfusions		seizures, coma/death	Carnitine therapy	acetylcarnitine profile,
(MCAD)	C6, C8, C10, C8/C10	Unsatisfactory specimens or				urine acylglycine,
		specimens collected before 24				urine organic acids,
		hours of age or received after				DNA analysis
		seven days may limit the ability				
		of the assay to detect disease				Referral to metabolic
		states				center for specific
						enzyme analysis,
						metabolite analysis,
						and/or mutation
						analysis
Very long chain acyl- CoA	Tandem mass spectrometry	Total parenteral nutrition (TPN	Normal	Breathing problems,	Avoid fasting; low fat diet	Quantitative plasma
dehydrogenase deficiency	(MS/MS)	and blood transfusions		seizures, coma/death	with medium chain	acetylcarnitine profile,
(VLCAD)	C14, C14:1, C14:2, C14:1/C16	Unsatisfactory specimens or			triglyceride (MCT) oil	urine organic acids,
		specimens collected before 24			supplement; carnitine therapy	mutation analysis of
		hours of age or received after				the VLCAD gene,
		seven days may limit the ability				genetic testing
		of the assay to detect disease				
		states				Referral to metabolic
						center for specific
						enzyme analysis,
						metabolite analysis,
						and/or mutation
						analysis

Long chain 3 hydroxyacyl- CoA dehydrogenase deficiency {LCHAD} Trifunctional protein deficiency (TFP)	Tandem mass spectrometry (MS/MS) C16:10H, C160H, C18:10H, C18:20H	Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states	Normal	Breathing problems, swelling of the brain, seizures, coma/death,	Avoid fasting; low fat diet; Long chain fatty acid restriction; medium chain triglycerides (MCT) oil supplement; carnitine therapy	Quantitative plasma acylcarnitine profile, urine organic acid analysis, free e-OH-fatty acids, biochemical and molecular genetic testing to differentiate LCHAD from TFP  Referral to metabolic center for specific enzyme analysis, metabolite analysis, and/or mutation analysis
		Ami	ino Acid Disorders			unarysis
Medical Condition	Testing Platform/ Analyte	Limitations and Interference	Normal Range	Symptoms if not treated	Common Medical Treatment	Confirmatory Test
Argininosuccinic aciduria (ASA)  Characterized by ammonia build-up in the blood	Tandem mass spectrometry (MS/MS) Citrulline	Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states	<55 umol/L	Muscle weakness and decreased tone, breathing problems, seizures, swelling of the brain, coma/death  Poor growth, enlarged liver, learning/intellectual disability	High-calorie, protein- restricted diet; arginine supplementation	Quantitative plasma ammonia and amino acid analysis, urine orotic acid.  Immediate consult with a metabolic specialist at a metabolic treatment center
Citrullinemia, type I, (classic form) (CIT)  Characterized by ammonia build-up in the blood	Tandem mass spectrometry (MS/MS) Citrulline	Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states	<55 umol/L	Muscle weakness and decreased tone, breathing problems, seizures, swelling of the brain, coma/death  Poor growth, enlarged liver, learning/intellectual disability	High-calorie, protein- restricted diet; arginine supplementation, sometimes dialysis	Quantitative plasma ammonia and amino acid analysis; urine for orotic acid.  Immediate consult with metabolic specialist at a metabolic treatment center

Maple syrup urine disorder (MSUD)  Characterized by the inability to digest leuicine, isoleucine, and valine	Tandem mass spectrometry (MS/MS) Leucine	Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states	<305 umol/L	Rigid/floppy muscles, brain swelling, seizures, metabolic acidosis, coma	Protein-restricted diet; medical formula	Immediate consult with metabolic specialist at a metabolic treatment center
Homocystinuria (HCY)  Characterized by the inability to digest methionine in food	Tandem mass spectrometry (MS/MS) Methionine, C3, and/or C3/C2	Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states	<75 umol/L	Intellectual and developmental disability; delays in crawling/walking/talking; poor growth; poor vision; behavior and emotional problems	Protein- restricted diet; medical formula; vitamin B6, betaine, and folic acid supplements; vitamin b12 injections or L-cysteine supplements	Quantitative plasma amino acid analysis, total plasma homocysteine
Phenylketonuria (PKU)  Characterized by the inability to digest phenylalanine in food.	Fluorescence assay Phenylalanine	Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states	<188 umol	Intellectual disabilities, behavior problems, hyperactivity, restlessness or irritability, seizures, eczema	Protein-restricted diet; medical formula	Repeat newborn screen; consult metabolic clinic; Quantitative plasma amino acid analysis, red blood cell DHPR assay, urine neopterin, DNA testing for PAH gene mutation
Tyrosinemia, type I  Characterized by the build- up of tyrosine in the blood	Tandem mass spectrometry (MS/MS) Succinylacetone (SUAC)	Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after seven days may limit the ability of the assay to detect disease states	<5.42 umol/L	Rickets, delays in walking, severe liver and kidney problems, death	Protein-restricted diet; medical formula; medication and vitamin D supplement	Immediate consult with metabolic specialist at a metabolic treatment center
Tyrosemia, type II and III  Characterized by the build- up of tyrosine in the blood	Tandem mass spectrometry (MS/MS) Tyrosine	Total parenteral nutrition (TPN and blood transfusions Unsatisfactory specimens or specimens collected before 24 hours of age or received after	<360 umol/L	Intellectual disabilities	Protein-restricted diet; medical formula; medication	Immediate consult with metabolic specialist at a metabolic treatment center

		seven days may limit the ability of the assay to detect disease states						
		Endo	ocrine Disor	ders				
Medical Condition	Testing Platform/ Analyte	Limitations and Interference	Normal Ran	ige	Symptoms if not treated	Common Medical Treatment	Confirmatory Test	
Primary congenital hypothyroidism  Characterized by an Insufficient thyroid hormone	Time resolved fluorescence assay Thyroid hormone T4 and Second tier TSH	Maternal medications, EDTA or sodium citrate anticoagulant interference, iodine exposure, cardiac medications	>6.0 ug/dL  Breathing problems, delays in sitting/crawling/walking/tal king, poor weight gain and growth, goiter, anemia, slow heart rate, hearing loss		sodium citrate anticoagulant interference, iodine exposure, cardiac medications in sitting/crawlin king, poor wei growth, goiter		Oral intake of throxine upon consultation of pediatric endocrinologist	Thyroid stimulating hormone (TSH)
Congenital adrenal	Solid phase, time-released	Treatment with	Weight dependent:		Confusion, irritability, rapid heart rate, coma	Hydrocortisone pills, treatment for salt-wasting	Repeat newborn screen; Serum 17-OHP; sodium, potassium and plasma renin if salt wasting is suspected.	
Characterized by a lack of 21-hydroxylase  17-hydroxyprogesterone (17-  hydrocortison may result in fresults  Preterm or low newborns, and collected beformay result in fresults.  Some congeni defects of the biosynthesis, wido not cause in the control of the con	dexamethasone, hydrocortisone, or prednisone may result in false-negative results  Preterm or low weight newborns, and samples	Birth weight (grams)	17-OHP (ng/mL)					
		collected before 24 hours of age may result in false positive results.						
	Some congenital enzyme defects of the steroid biosynthesis, which cause CAH do not cause in increase in 17 $\alpha$ OHP, resulting in false negative	1500- 2499	<63					
		results.  Anomalous results can also be caused by uneven saturation, poorly collected or improperly dried specimens, or contamination of the blood spot with fecal material	>2500	<32				

		Puln	nonary Disorders			
Medical Condition	Testing Platform/ Analyte	Limitations and Interference	Normal Range	Symptoms if not treated	Common Medical Treatment	Confirmatory Test
Cystic fibrosis (CF)  Characterized by a buildup of thick mucus in the lungs	Fluorescence assay Immunoreactive Trypsinogen (IRT) Second tier genotyping	Meconium ileus, heat degradation, contamination with EDTA or heparain	<80.1 ng/mL	Bronchitis/pneumonia, poor growth, chronic diarrhea, impaired learning ability	Pulmonary therapy; prevent infection; replace digestive enzymes	Sweat chloride (gold standard) and/or DNA mutation analysis
		Other I	Metabolic Disorders			
Medical Condition	Testing Platform/ Analyte	Limitations and Interference	Normal Range	Symptoms if not treated	Common Medical Treatment	Confirmatory Test
Biotinidase deficiency  Characterized by insufficient biotin	Colorimetric assay Biotinidase	Sulfonamides sulfisoxazole or trimethoprim and sulfamethoxazole can cause a false negative Enzyme destruction by heat, humidity, or delay between collection and testing (false positive); transfusion (false negative)	Enzyme Present	Intellectual and developmental disability; seizures; skin rash; alopecia; hearing loss; death	Biotin therapy	Repeat newborn screen; Quantitative enzyme assay; consultation with a metabolic clinic
Classic galactosemia (GALT)  Characterized by the inability to digest galactose	Fluorescence assay Galactose-1 Phosphate Uridyl-1- Transferase (GALT)	Enzyme destruction by heat, humidity, or delay between collection and testing (false positive); transfusion (false negative)	>3.00 u/gHb	Hypoglycemia, seizures, enlarged liver, jaundice, bleeding, blood infections, cataracts	Galactose and lactose- restricted diet, Vitamin C and K supplement	Galactose-1 phosphate uridyltransferase enzyme and galactosemia DNA panel
Medical Condition	Tocting Platform / Analyta	Limitations and Interference	Normal Range	Sumptoms if not trantad	Common Medical Treatment	Confirmatory Tost
Hemoglobinopathy Hemoglobin disease or trait	Isoelectric focusing (IEF) with high-performance liquid chromatography (HPLC) reflex Hemoglobin fractions: Fetal (F), Adult (A), Sickle (S), C-Hemoglobin (C), E-Hemoglobin (E), D-Hemoglobin (D) Hemoglobin patterns:	Red blood cell transfusion— repeat screen 90-120 days post transfusion	Normal (F+A)	In sickle cell disease: death by sepsis or splenic sequestration anemia; sickling crisis	Fluids, pain killers, blood transfusion Hemoglobin disease: prophylactic penicillin until age five	High performance liquid chromatography (HPLC) reflex, DNA testing

Characterized by pathogenic variants in the SMN1 gene					rehabilitation	number assessment
Spinal Muscular Atrophy	Real-Time qPCR SMN1 gene		Presence of <i>SMN1</i> Exon 7	Increasingly lilted mobility	Injections, physical and occupational therapy,	SMN1 Exon 7 absence and SMN2 copy
Medical Condition	Testing Platform/ Analyte	Limitations and Interference	Normal Range	Symptoms if not treated	Common Medical Treatment	Confirmatory Test
			Other Disorders			
Medical Condition  Severe combined immunodeficiency (SCID)  Characterized by the baby's lack of T-cells and severely reduced B-cell function	Beta Thalassemia FSC=Hemoglobin SC disease FSA=Sickle Beta Thalassemia FE=Hemoglobin E disease/Hemoglobin E-Beta Thalassemia FSD=Hemoglobin SD disease FSE=Hemoglobin SE disease F only=Beta Thalassemia Major  Testing Platform/ Analyte  Real-Time qPCR T-cell receptor excision circles (TRECs)	Limitations and Interference  Specimen contamination can cause false negative	nunology Disorders  Normal Range  <=1.079 MoM	Symptoms if not treated  Death in infancy or early childhood	Common Medical Treatment  Bone marrow transplant.  Prophylactic antibiotics, antiviral and antifungal agents, and intravenous gamma globulin are given until bone marrow transplant is performed.	Confirmatory Test  Complete Blood Count (CBC) with differential and flow cytometry to determine the extent of cell lymphopenia
	FA=Normal FS-Hemoglobin S disease/Sickle					

<sup>\*</sup>Information included in this table was taken from the Medical Home Portal (Medical Home Portal, n.d.) and the Wisconsin Newborn Screening Program (Wisconsin State Laboratory Newborn Screening Program, 2020) (Association of Public Health Laboratories); (Clinical and Laboratory Standards Institute, 2019); (Wisconsin State Laboratory Newborn Screening Program, 2020)