

Disorders	Abnormal 1st screen	Abnormal 2nd screen	Contact
Biotinidase deficiency	repeat screen requested	metabolic specialist recommends diagnostic testing	Metabolic Clinic RN: 303-724-2352
Congenital Adrenal Hyperplasia	borderline: repeat screen requested	presumptive positive: endocrinologist recommends diagnostic testing	Children's Hospital Endocrine: 720.777.6128 Pediatric Endocrine Associates: 303.783.3883 Rocky Mountain Pediatric Endocrinology: 720-420-3636
	presumptive positive: endocrinologist recommends diagnostic testing		
Congenital Hypothyroidism	borderline: repeat screen requested	presumptive positive: CDPHE recommends PCP consults with endocrinology for diagnostic testing	
	presumptive positive: CDPHE recommends PCP consults with endocrinology for diagnostic testing		
Cystic Fibrosis	repeat screen requested	Only performed if 1st NBS was abnormal. If IRT is elevated, DNA mutational analysis is performed.	On-Call CF Provider: 720-777-6181 Children's Hospital Laboratory Colorado (accredited lab for sweat test): 720-777-8173
		zero mutation: no additional testing	
		1 mutation: sweat test within 2 weeks	
		2 mutations: CF provider will contact PCP and coordinate care	
Galactosemia	metabolic specialist recommends diagnostic testing	Only performed if 1st NBS was abnormal	Metabolic Clinic RN: 303-724-2352 On-Call Metabolic Provider: 720-777-3999
Hemoglobinopathies	repeat screen requested	Only performed if 1st NBS was abnormal	Hematology RN: 303-724-9075
		If abnormal, confirmatory testing performed, hematology RN contacts PCP with results	
Amino Acidemia	NICU: CDPHE contacts NICU for repeat specimen and/or diagnostic testing	only performed if 1st NBS was abnormal	Metabolic Clinic RN: 303-724-2352 On-Call Metabolic Provider: 720-777-3999
	Time Critical result: metabolic specialist recommends diagnostic testing		
Organic Acidemia	NICU: CDPHE contacts NICU for repeat specimen and/or diagnostic testing	only performed if 1st NBS was abnormal	Metabolic Clinic RN: 303-724-2352 On-Call Metabolic Provider: 720-777-3999
	Time Critical result: metabolic specialist recommends diagnostic testing		
Fatty Acid Oxidation	NICU: CDPHE contacts NICU for repeat specimen and/or diagnostic testing	only performed if 1st NBS was abnormal	Metabolic Clinic RN: 303-724-2352 On-Call Metabolic Provider: 720-777-3999
	Time Critical result: metabolic specialist recommends diagnostic testing		
SCID	Immunologist recommends diagnostic testing	only performed if 1st NBS was abnormal	Immunology: 303-398-1196