

CP 1.6 Newborn Screening Program Confirmatory Testing

<i>Disorder</i>	<i>Test/Method</i>	<i>Processing Laboratory</i>	<i>Specimen Requirements</i>	<i>Packaging & Supplies/ Special Handling</i>	<i>Transport Method</i>	<i>Parents' Sample Needed?</i>
Isolated PKU Positive in Amino Acid Panel	Repeat Phenylalanine by MS/MS on Filter Paper	NAPS	Dried Blood Spots on Filter Paper	Sent with regular NBS	Golden State Overnight/Courier	NO
Other Amino Acid Panel Positive (or PKU positive plus other positive analytes) Acylcarnitine Panel Positive	Repeat Amino Acid Panel by MS/MS	Quest, after referral to Metabolic Center	Urine and/or Blood as determined by test and Metabolic Center		Quest Courier	NO, unless ordered by Metabolic Specialist
Biopterin	Urine for Biopterin and Neopterin Dried Blood Spot for dihydropteridine reductase	GDL	Urine 10-20ml Dried Blood Spots (4) on NBS specimen collection filter paper	GDL provides containers for urine, and filter paper for blood. Urine Specimen must be protected from light during collection, handling and shipping. Dried blood packed in plastic bag and shipped on dry ice with urine specimen Intake Form.	Next day delivery or Courier arrangements made by metabolic center and/or hospital	NO
Galactosemia	Galactose-1-phosphate-uridyl transferase by Fluorometric Method Mutational analysis	ARUP	Whole Blood min. 2 ml in green top tube, 2 ml. in purple top tube)	Not provided Intake Form Do Not Freeze	Next day delivery or ARUP Courier Arrangements made by metabolic center or hospital	Only if necessary to clarify baby's results.

3.16 Newborn Screening Program Recall and Confirmatory Testing

Disorder	Test/Method	Processing Laboratory	Specimen Requirements	Packaging & Supplies/ Special Handling	Transport Method	Parents' sample needed?
BD –values of 6.01-10.00 ERU	2 nd NBS test	NAPS Lab	filter paper	n/a	Next day delivery or Courier arrangements made by ASC and/or hospital	NO
BD – values of 6.00 ERU and lower	Biotinidase enzyme activity	Stanford Biochemical Genetics Laboratory after referral to CCS metabolic center	2-3cc whole blood – red (gold) or green (mint) top tube	Spin, freeze serum or plasma within 30” Ship on dry ice	Next day delivery or Courier arrangements made by ASC and/or hospital	YES, if possible
PCH	Ordered by physician; may include TSH, T4, T3, Free T4, Thyroid Antibodies	Accredited lab outside of NBS Program	Serum or whole blood		Should be by overnight courier.	NO
CAH	17 OHP, electrolytes Ratio of Androstendione to Cortisol by MS/MS	Accredited lab outside of NBS Program	Whole Blood		Should be by overnight courier.	NO
Hemoglobin Disorders	May include: CBC, DNA, Thin Layer Isoelectric Focusing (TLIF) electrophoresis	CHRCO Hemoglobin Reference Lab	Whole Blood 1.5-2 ml (EDTA Lavender Tube)	Prepaid envelopes provided by CHO or express/priority mail AND Intake Form	2 day US Priority Mail	YES
Cystic Fibrosis	Sweat Test	CFF- approved lab outside of NBS Program	Sweat	N/A	N/A	NO
SCID	Flow Cytometry	Quest Diagnostics	1.5 cc Whole blood lavender top EDTA tube	Sent at ambient temp.	Quest Courier—1 day	No