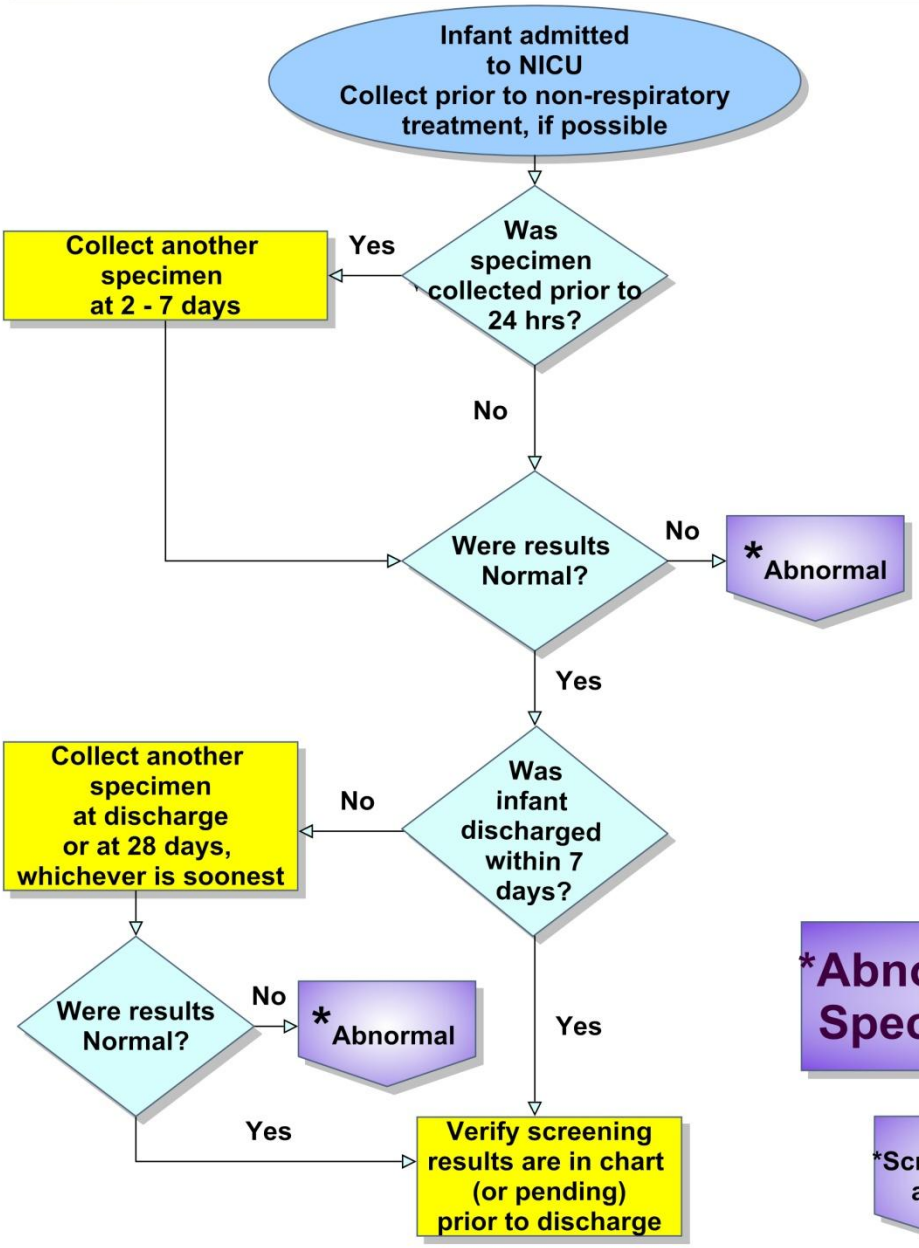
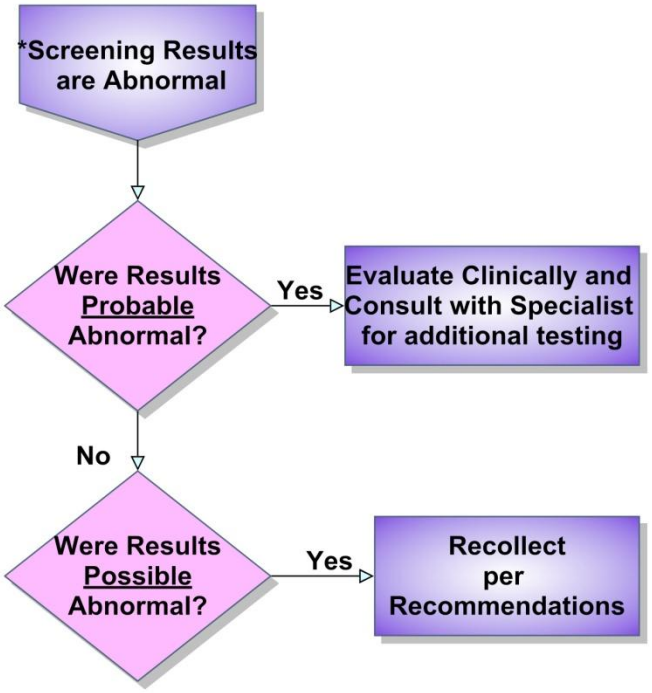
MT Newborn Screening NICU Recommended Protocol



*Abnormal Newborn Screening Specimen Collection Protocol



Montana Dept. of Public Health and Human Services
Public Health Laboratory
Newborn Screening Program
800-821-7284

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NEWBORN SCREENING FOR PRETERM, LOW BIRTH WEIGHT, AND SICK NEWBORNS: UPDATED RECOMMENDATIONS FOR 2012

Each Neonatal Intensive Care Unit (NICU) should have a standard policy for newborn screening to optimize the timing and minimize the number of bloodspot specimen collections. The neonate's immaturity and the necessary therapeutic interventions can interfere with both the collection of samples and the interpretation of the newborn screening results. Based on a survey of Montana NICU's, most have already implemented updated protocols. The recommendations below combine elements of 2009 Approved Guideline I/LA31-A from the Clinical and Laboratory Standards Institute (CLSI) and current Montana Administrative Rules. These recommendations apply to all neonates receiving intensive care.

- **Collect the newborn screen by heelstick whenever possible.**
  Umbilical catheter may be used if cleared of IV fluids, amino acids, heparin, etc.

- **Collect the initial newborn screen at 0 to 48 hours of age. Collect before treatment** (antibiotics, steroids, parenteral nutrition) is started or transfusion given, even if less than 24 hours, **unless medically contraindicated**. Otherwise, collect at 24-48 hours.

- **If the initial screen was collected at less than 24 hours, order another newborn screen collection at 48 hours to 7 days at the latest.** Out of range results on the less than 24 hour screen will usually normalize. Some conditions manifest in the first week, but may not be detected in blood collected at less than 24 hours.

- **Screen transferred infants**.
  Collect a newborn screen when infant is admitted to the NICU after transfer.

- **Screen all infants at least twice if hospitalized more than 7 days**.
  Order another screen prior to discharge or at 28 days, whichever comes first.
  At least two screens are required for infants less than 1500g birth weight.

- **Check for normal newborn screen results at 7 days and prior to discharge.**
  Make sure screening was done, report(s) received, and no repeats are needed.

- **Follow up out of range newborn screen results.**
  The faxed screening report will recommend another screen or consultation with a specialist for diagnostic testing. Check if another screen has been ordered or collected.

- **Arrange for additional collection if infant was transfused prior to initial screen.**
  Screen for hemoglobin disorders and galactosemia 120 days after transfusion.

- **Remember the hearing screen.** Infants admitted for more than 5 days should have auditory brainstem response (ABR) to detect neural hearing loss.

- **Remain vigilant for signs of conditions**, even if screening results are normal.

- **Questions? Need a newborn screen report?** Call the Montana Newborn Screening Program (Public Health Lab) at 406-444-0984 or 800-821-7284 (24/7). Fax 406-444-1802. For more information see www.newborn.hhs.mt.gov .

NEWBORN SCREENING FOR PRETERM, LOW BIRTH WEIGHT, AND SICK NEWBORNS: FACTORS AFFECTING DETECTION OF SCREENED CONDITIONS

Adapted from CLSI Approved Guideline I/LA31-A

Condition	Inci-dence	Best age to screen	Abnormal analyte	Factors affecting NBS (False positive)	Factors affecting NBS (False negative)
Congenital Hypothyroidism (CH)	One in 3,000 births	12 to 72 hours and 2-6 weeks	Low T4 and/ or high TSH	Low T4/ normal TSH with prematurity or acute illness; physiologic high TSH at 12-24 hours; transient low T4 from topical iodine	Delayed TSH rise in premature infants with CH; **steroid** treatment may depress TSH
Congenital Adrenal Hyperplasia (CAH)	One in 12,000 births	12 to 48 hours and 2-4 weeks	High 17-OHP	Preterm, sick and stressed infants; maternal CAH; incorrect birth weight (determines normal range)	Maternal **steroid** treatment; dexa-methasone in infant; incorrect birth weight
Cystic Fibrosis (CF)	One in 2,500 births	24 hours to 7 days	High IRT	Hypoxic organ damage, neonatal stress, hypo-glycemia; preterm; trisomies; early collection	If clinical CF symptoms, should have confirmatory (sweat/ mutation) testing regardless of NBS result
Hemoglobin-opathies (Sickle Cell SS, SC, Sβthal)	One in 400 to 1 in 10,000 births	Birth to 72 hours	HbS present and no HbA	May need further evaluation after fetal hemoglobin disappears	RBC **transfusion** prior to collection; other serious hemoglobinopathies may not be detected by NBS
Classic Galactosemia (GALT)	One in 60,000 births	Birth to 48 hours	Low Gal-1-PUT	Enzyme may be destroyed by heat, humidity, or delay in specimen transit	RBC t**ransfusion** prior to collection; no milk feeding (soy or **parenteral** only)
Biotinidase Deficiency (BIOT)	One in 60,000 births	Birth to 72 hours	Absent biotinidase	Premature or jaundiced infant	Transient effect from plasma **transfusion**
Phenylketonuria (PKU)	One in 10,000 births	24 to 48 hours	High phenyl-alanine	**Parenteral nutrition**; liver disease; maternal PKU	Specimen collected at less than 24 hours
Maple Syrup Urine Disease (MSUD)	One in 185,000 births	24 to 48 hours	High leucine and valine	**Parenteral nutrition**	Specimen collected at less than 24 hours
Tyrosinemia (TYR)	One in 100,000 births	24 to 48 hours	High tyrosine/ high succinyl-acetone	Liver disease and immature liver enzymes; **parenteral nutrition**	Specimen collected at less than 24 hours
Citrullinemia/ Arginosuccinic Acidemia (CIT/ASA)	One in 70,000 to 100,000 births	24 to 48 hours	High citrulline	**Parenteral nutrition** (if enough arginine is added, it can cause upstream buildup of citrulline)	Specimen collected at less than 24 hours
Homocystinuria (HCY)	One in 300,000 births	3 to 7 days	High methionine	Liver dysfunction and/ or prematurity; **parenteral nutrition**; cholestasis	Specimen collected at less than 72 hours
Organic acidemia disorders (Nine)	One 50,000 births overall	24 to 48 hours	High acyl-carnitines C5, C5OH, C3, C5DC, etc)	**Parenteral nutrition** and/ or **antibiotics** can elevate C5; Vitamin B12 deficiency in mother may elevate C3	Unknown
Fatty Acid Oxidation Disorders (Five)	One in 10,000 births overall	Birth to 48 hours	High acyl-carnitines C8, C14:1,C16:OH, low C0, etc	**Carnitine supplementation** or MCT oil	Conditions may be masked by **carnitine supplementation** or well-fed state