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PREVENTION OPPORTUNITIES UNDER THE BIG SKY

NEWBORN BLOODSPOT SCREENING: THE FIRST 2½ YEARS OF EXPANDED SCREENING

Newborn screening is the practice of testing blood from newborns to identify developmental, genetic and metabolic disorders so that important action steps can be taken. Although the disorders detected are rare, early diagnosis and proper treatment can support healthy development and prevent lifelong impairment. Since 1973 screening of Montana newborns has been required for phenylketonuria (PKU) and congenital hypothyroidism (CH). When detected early these disorders can be treated with special diet (PKU) or hormone replacement (CH) to prevent the severe sequelae of the untreated conditions. In 2003 tests for galactosemia and hemoglobinopathies were required. In January 2008 newborn screening in Montana was expanded to include 28 metabolic endocrine, hematologic and genetic conditions recommended by the American College of Medical Genetics and the American Academy of Pediatrics.[1,2] More than 99% of babies born in Montana have been screened. This issue of *Montana Public Health* describes the results of the first 2 ½ years of the expanded newborn bloodspot screening (NBS) program.

The Montana NBS Program In Montana the NBS Program is a public health system that includes several important components: laboratory testing to identify the conditions listed in Table 1; follow-up laboratory testing to confirm initial identifications; clinical consultation and management (including special diets) in cooperation with a baby's private physician; and education for families of newborns, health care providers and the general population.

Table 1. Conditions included in Montana NBS Program

Phenylketonuria
Congenital hypothyroidism
Galactosemia
Hemoglobinopathies
Biotinidase deficiency
Congentical adrenal hyperplasia
Cystic fibrosis

Metabolic disorders
- Fatty acid oxidation (5 disorders)
- Organic acidemia (9 disorders)
- Amino acid (5 disorders)

Results of NBS 2008-2010 From January 2008 through June 2010, the blood from 30,550 newborns (about 1000 each month) was screened and from 701 rescreened to verify an initial finding.(Table 2) This testing led to 36 confirmed diagnoses.(Table 3) The infants and families with confirmed diagnoses received follow-up care and treatment from metabolic and genetic specialists. In addition, specialized nutritional counseling, physical therapy, occupational therapy, and other specialized services are provided.

Table 2. Number of newborns screened and test results, Montana NBS Program, 2008-June 2010.

	Year			Total
	2008	2009	2010	
Screened	12455	12118	5977	30550
Initial positive*	286	257	158	701 (2.3%)
Repeat positive	67	45	18	130 (0.4%)
Confirmed diagnosis	17	15	4	36 (0.1%)

*includes low T4/TSH in low birth weight babies

Table 3. Confirmed diagnoses identified by the NBS Program, Montana, 2008-June 2010

Conditions diagnosed	Year		
	2008	2009	2010
Congenital hypothyroidism	9	5	1
Cystic fibrosis	3	3	2
Organic acidemia disorders	1	4	1
Fatty acid oxidation disorders	1	2	0
Galactosemia (Duarte variant)	1	1	0
Phenylketonuria	1	0	0
Sickle cell disease	1	0	0

Assessing and Improving the NBS Program The criteria for acceptability of newborn bloodspot specimens became more stringent when screening was expanded in 2008. In 2009 approximately 10% of initial specimens submitted did not meet the stringent criteria. The Montana Public Health Laboratory launched an intensive educational program to submitters at birthing hospitals. As a result of renewed attention to proper collection and submission procedures, the rate of unsatisfactory specimens submitted decreased markedly to less than 1%.

Clinical Expertise Clinical consultation is an important component of the NBS Program. Initial laboratory results may be abnormal for a variety of reasons including: supplemental nutrition for very sick or low birth weight babies, or medications a breast-feeding mother is taking. Consultation is readily available from experts at Shodair Medical Genetics Department through contract with the NBS Program to assist with patient evaluation, patient management, and decisions for further diagnostic testing.

Confidentiality of Information Considerable discussion and news media attention has been directed to length of storage and potential uses of dried blood spots (DBS) after initial newborn screening has been completed. It is the policy of the Montana Public Health Laboratory to store DBS specimens for one year to allow retesting (validation) of original test findings or testing in studies to develop new screening tests. After one year DBS specimens are destroyed.

Recommendations for health care providers

- Screen all newborns: collect a bloodspot specimen from all newborns. If you are not certain that NBS was performed in the nursery for your patient, call the MT Public Health Laboratory, 800-821-7284, or simply collect and submit another specimen.
- If initial test is out-of-range: if you are notified that an initial test result was out-of-range or the specimen was unsatisfactory, collect and submit another specimen as soon as possible.
- Resources available: visit www.newborn.hhs.mt.gov or www.lab.hhs.mt.gov .

For more information, contact Denise Higgins, Newborn Screening Manager, dehiggins@mt.gov, 406-444-3040; or Linda Beischel, Newborn Screening Coordinator, lbeischel@mt.gov, 406-444-0984.

References:
1. Watson MS, et al. Newborn screening: toward a uniform screening panel and system. Genetics in Medicine 2006; 8: 1S-11S http://www.acmg.net/resources/policies/NBS
2. PHSD/DPHHS. Expanded newborn bloodspot screening in Montana, 2008. Montana Public Health 2008; 3(6):1-2.

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