



Pediatric Clips

Newborn Screening Program (NBS)

By Mukund Dole, MD

Pediatric Clips from The Children's Medical Center of Dayton are quick reviews of common pediatric conditions.

The Children's Medical Center of Dayton is the region's pediatric referral center for a 20-county area. As the only facility in the region with a full-time commitment to pediatrics, Dayton Children's offers a wide range of services in general pediatrics as well as in 35 subspecialty areas for infants, children and teens. We welcome your inquiries about services available – call 937-641-3666 or e-mail marketing @childrensdayton.org.



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Newborn screening (NBS) is a population-based preventative public health program that is being carried out in the United States and other countries. The goal of NBS is to help diagnose certain medical illnesses by early detection, institute prompt intervention and prevent:

- development impairment (including neurologic deficits and mental retardation)
- delayed/poor physical growth
- severe medical illness and organ dysfunction, and sometimes
- death.

In 2005, the American Academy of Pediatrics (AAP) endorsed a report from the American College of Medical Genetics (ACMG) that all states screen newborns for a core panel of 29 congenital conditions and an additional 25 conditions that may be detected by screening. The program screens for illnesses that have treatments available, have public health benefits, have available diagnostic technology for mass screening and prove to have clear cost benefit analysis.

LIMITATIONS

Limitations of the NBS program are:

- NBS panels cannot test for all congenital conditions/ illnesses.
- Results of NBS are tools for more specific testing and evaluation of children with congenital illnesses.
- Normal test results (in range) for a given condition do not necessarily rule out its diagnosis in a child with the characteristic clinical illness and these tests may need to be repeated.
- Primary care providers (PCPs) should be aware that infants may present with clinical symptoms or deterioration before the test results become available, therefore clinical knowledge and suspicion of these conditions is important.

COLLECTION OF SPECIMENS

Specimens are collected after 24 hours of birth but no later than 5 days of age. The specimen is sent to Ohio Department of Health (ODH) laboratory within two days of collection. Specimens should be collected prior to a blood transfusion whenever possible. All birthing centers should designate a specific NBS coordinator and

physician to oversee the process and keep the ODH informed of any changes.

RESPONSIBILITIES OF PUBLIC HEALTH LABORATORIES

1. Make all necessary arrangements for specimen collection.
2. Once the specimen is submitted, complete the screening procedure within seven days.
3. Promptly transmit all results to the PCP.
4. Keep all records for at least 21 years.
5. Provide all relevant reports to the necessary programs/entities for appropriate action.
6. Provide ongoing education to families and PCPs about their condition.

ABNORMAL SPECIMENS

If the initial screening results are abnormal and the public health laboratory (ODH) determines the baby is at risk, the following actions are taken immediately:

1. Results are communicated to the appropriate physician or provider.
2. The provider then communicates the results to the child's parent or guardian and should arrange for a second blood specimen to be submitted for screening or further diagnostic testing.
 - If results are abnormal for a hemoglobin disease or trait, diagnostic testing should be done before the baby reaches 2 months.
 - If results indicate other genetic or metabolic disorders, a repeat specimen should be obtained immediately, no later than 10 days of receiving the notice from the ODH laboratory.
3. All diagnostic specimens obtained for confirmatory testing should be submitted to an approved laboratory and results transmitted to the appropriate provider.
4. If the provider is unable to locate the child, he/she should notify the health commissioner of the district in which the child and family resides. The health commissioner will then undertake a good faith effort to locate the child and the family.

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ROLE OF THE PRIMARY PHYSICIAN

The important roles PCPs fulfill in order to make the NBS program successful are:

1. Check early in the child's care that NBS has been successfully completed.
2. If NBS is not done, provide the family with the necessary education and reorder screening as soon as possible.
3. If the parents refuse NBS, obtain and document a waiver in the medical record, flag the charts and monitor them closely for warning signs of illnesses.
4. If the results of NBS are not available at the two to four week visit, make every attempt to contact the birthing facility or state laboratory (ODH) for results.
5. Action based on screening results:
 - If the NBS results are in range (normal), the results should be shared with the family. False negatives should be kept in mind.
 - If the specimen is invalid, the NBS must be reordered immediately.
 - If the results are out of range (abnormal), rapid rescreening or obtaining confirmatory testing is necessary. Most state programs have specific action (ACT) sheets which delineate the steps to be taken in such cases. If ACT sheets for a given medical condition are not available from the state, these can be downloaded at www.acmg.net/resources/policies/ACT/conditionanalyte-links.htm.
6. Actions needed after rescreening/confirmatory testing:
 - If the confirmatory tests indicate that the child is normal, parents should be reassured and the child watched carefully.
 - Appropriate consultations with subspecialists is recommended to establish a final diagnosis when necessary.
 - Children with positive confirmatory tests should be identified as special needs and chronic management plans specific for that condition should be initiated.

Continued from the front.

- The PCP should provide a medical home and a central source of family centered, accessible, coordinated, comprehensive, compassionate and continually effective care. The complex nature of some of these illnesses may require multidisciplinary care involving appropriate subspecialist therapies, nutritionists and others.

ROLE OF SUBSPECIALISTS

The major roles subspecialists play in the success of the NBS program are:

1. Work with the PCP to provide care both during acute illness and helping manage chronic medical issues such as neurodevelopmental disabilities, behavioral issues and late effects secondary to these conditions.
2. Coordinate genetic counseling and evaluation, and ensure appropriate referral occurs.
3. Be a source for patient education and links to resources.
4. Work with the PCPs and assist in transitioning of patients with chronic illnesses to adult care.

NEWBORN SCREENING PROGRAM IN OHIO

The NBS program in Ohio has been in place since 1990 and all newborns are screened for the following genetic, metabolic and endocrine disorders:

argininemia	argininosuccinic acidemia	biotinidase deficiency	carnitine / acylcarnitine translocase deficiency	carnitine palmitoyl transferase deficiency type II
carnitine uptake defect	citrullinemia	congenital adrenal hyperplasia	congenital hypothyroidism	cystic fibrosis
galactosemia	glutaric academia, type I and II	homocystinuria	hypermethioninemia	isobutyryl-CoA dyhydrogenase deficiency
isovaleric acidemia	long chain hydroxyacyl-CoA dehydrogenase deficiency	maple syrup urine disease	medium chainacyl-CoA dehydrogenase deficiency	methylmalonic acidemia
multiple CoA carboxylase deficiency	phenylketonuria	propionic acidemia	trifunctional protein deficiency	very long chain acyl-CoA dehydrogenase deficiency
2-methylbutyryl-CoA dehydrogenase deficiency	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	3-ketothiolase deficiency	3-methylcrotonyl-CoA carboxylase deficiency	sickle cell and other hemoglobinopathies

FEATURED SPECIALIST



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Center and also an associate professor of pediatrics at Wright State University Boonshoft School of Medicine. He is board certified in pediatrics and pediatric hematology/oncology. He has special interests in hemoglobinopathies, sickle cell disease, resistant sarcomas and general oncology.

HEMATOLOGY/ONCOLOGY AT DAYTON CHILDREN'S

The Comprehensive Care Center for Cancer and Blood Disorders provides advanced

hematology and oncology care for patients up to age 21. The department is one of the few pediatric programs in the nation to be accredited by the American College of Surgeons Commission on Cancer. The hematology/oncology team provides a multidisciplinary approach to ensure that every child's medical and psychosocial needs are met. To make a referral or speak with someone in hematology/oncology, call 937-641-3111.



For further information about The Children's Medical Center of Dayton or its specialists contact us at 937-641-3666 or marketing@childrensdayton.org.



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