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WOMEN'S HEALTH CARE PHYSICIANS

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Committee on Genetics

This Committee Opinion was developed by the American College of Obstetricians and Gynecologists' Committee on Genetics in collaboration with committee members Britton Rink, MD, and Stephanie Dukhovny, MD.

Newborn Screening and the Role of the Obstetrician–Gynecologist

ABSTRACT: Newborn screening is the largest genetic screening program in the United States, with approximately 4 million infants screened annually. Newborn screening is a mandatory state-based public health program that provides all newborns in the United States with testing and necessary follow-up health care for a variety of medical conditions. The goal of this public health program is to decrease morbidity and mortality by screening for disorders in which early intervention will improve neonatal and long-term health outcomes. The program's functions include the initial screening of all newborns, identifying screen-positive newborns, diagnosing conditions, communicating with families, ensuring that affected children are referred to treatment centers, following up with long-term outcomes, and educating physicians and the public according to individual state or jurisdictional guidelines. All states and the District of Columbia have newborn screening programs with varying screening panels, policies, statutes, and regulations. Most programs have adopted the guidelines suggested by the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children. Obstetrician–gynecologists and other obstetric care providers should make resources about newborn screening available to patients during pregnancy. Providing newborn screening information during prenatal care visits can be accomplished in several ways and should be adapted to individual practice style. Integrating education about newborn screening into prenatal care allows parents to be prepared for having their child undergo screening as well as for receiving newborn screening test results. This document includes updated information on the Recommended Uniform Screening Panel (RUSP) and recommendations for incorporating newborn screening into obstetric practice.

Recommendations and Conclusions

The American College of Obstetricians and Gynecologists makes the following recommendations and conclusions:

- Newborn screening is a mandatory state-based public health program that provides all newborns in the United States with testing and necessary follow-up health care for a variety of medical conditions. However, parents or guardians can opt-out of the program for religious reasons following state guidelines.
- Obstetrician–gynecologists and other obstetric care providers should make resources about newborn screening available to patients during pregnancy. Information can be disseminated through informational brochures and electronic sources and through review or discussion at some time during prenatal care. Integrating education about newborn screening into prenatal care allows parents to be prepared for having their child undergo screening as well as for receiving newborn screening test results.
- Newborn screening does not replace the potential value of carrier screening nor does carrier screening replace newborn screening.

Newborn screening is the largest genetic screening program in the United States, with approximately 4 million infants screened annually (1). Newborn screening is a mandatory state-based public health program that provides all newborns in the United States with

testing and necessary follow-up health care for a variety of medical conditions. However, parents or guardians can opt-out of the program for religious reasons following state guidelines. A treatable condition is diagnosed in 1 out of 300 newborns, and severe disorders are detected in approximately 5,000 newborns per year in the United States (2). The goal of this public health program is to decrease morbidity and mortality by screening for disorders in which early intervention will improve neonatal and long-term health outcomes. This screening is done through blood testing in the newborn within the first 24–48 hours of life in addition to other noninvasive means to screen specifically for hearing loss and critical congenital heart disease.

Most of the disorders screened for have no clinical findings at birth although some may present before the results are available from the newborn screening test. The five most commonly diagnosed conditions by newborn screen in the United States are 1) hearing loss, 2) primary congenital hypothyroidism, 3) cystic fibrosis, 4) sickle cell disease, and 5) medium-chain acyl-CoA dehydrogenase (MCAD) deficiency (3). As of 2018, 35 conditions are listed on the Recommended Uniform Screening Panel (RUSP) as part of the comprehensive preventive health guidelines that have been supported by the Advisory Committee on Heritable Disorders in Newborns and Children and recommended by the Secretary of Health and Human Services (Table 1).

Newborn screening programs are implemented and managed on the state level and operate through collaborations between public health programs, laboratories, hospitals, pediatricians, subspecialists, and specialty diagnostic centers. The programs' functions include the initial screening of all newborns, identifying screen-positive newborns, diagnosing conditions, communicating with families, ensuring that affected children are referred to treatment centers, following up with long-term outcomes, and educating physicians and the public according to individual state or jurisdictional guidelines. Screening programs test newborns primarily through blood samples obtained by a heel blood sample shortly after birth that are placed on a special filter paper and sent to a designated state newborn screening laboratory at 24–48 hours of life. In patients for whom a blood sample is unable to be obtained shortly after birth—such as newborns who require a transfusion or total parenteral nutrition, are sick, born preterm, or born out of the hospital setting—the screening protocol may be modified and, in some cases, repeat testing is warranted.

Creation of the Recommended Uniform Screening Panel

Conditions that are included or considered for inclusion on the Recommended Uniform Screening Panel are chosen based on evidence that supports the benefit of screening, feasibility of screening at the state level, and the availability of treatment. The uniform panel of core conditions was intended to be flexible, and criteria have been established to perform evidence-based reviews to expand the panel over time. This panel consists of five main categories of disorders: 1) hemoglobinopathies, 2) organic acid disorders, 3) amino acid disorders, 4) fatty acid oxidation disorders, and 5) miscellaneous disorders, such as cystic fibrosis, hypothyroidism, and hearing loss (Table 1). In the process of screening, it is possible to unintentionally identify "secondary conditions" or conditions in the differential diagnosis of the core list that may have clinical significance (4). This document includes updated information on the Recommended Uniform Screening Panel and recommendations for incorporating newborn screening into obstetric practice.

Core conditions may be screened by enzyme analysis, DNA-based testing, and immunoassay techniques. Beyond blood testing, pulse oximetry screening with skin sensors is used to detect the lower oxygen saturation levels in the blood that are associated with some forms of critical congenital heart defects (5, 6). Screening for hearing loss in the newborn is performed by either otoacoustic emissions or automated auditory brainstem response (7).

State Guidelines

All states and the District of Columbia have newborn screening programs with varying screening panels, policies, statutes, and regulations. Most programs have adopted the guidelines suggested by the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children. The selection of disorders screened for is affected by the disease prevalence within the state or jurisdiction, detection rates, treatment availability, and cost considerations (8). A current list of conditions screened for in each state is maintained online by Baby's First Test (9). States also vary in their policies for parental or legal guardian consent for the use of residual blood samples for secondary purposes, such as research or program quality evaluation (10) or for refusal of newborn screening in rare circumstances.

The Department of Health and Human Services and 15 other agencies have issued a body of regulations that protect human participants involved in research. This is known as the "common rule," which is an attempt to balance individual autonomy over biospecimens versus the beneficence of secondary research use in the public health context. A great deal of variability exists among states relative to blood spot retention and how the blood spot cards are made available for research. Whether or not the use of blood spot cards is considered human research and the nature of institutional review board consent should be clarified relative to state and federal guidelines for any patient with questions or for any investigator who is considering blood spot card use. Families with questions on informed consent and

Table 1. Recommended Uniform Newborn Screening Panel of Core Conditions*

Disease Categories	Diseases
Inborn errors of organic acid metabolism	Isovaleric acidemia
	Glutaric acidemia type I
	3-Hydroxy-3-methylglutaric aciduria
	Holocarboxylase synthetase deficiency
	Methylmalonic acidemia (methylmalonyl-CoA mutase)
	3-Methylcrotonyl-CoA carboxylase deficiency
	Methylmalonic acidemia (cobalamin disorders)
	Propionic acidemia
	β-ketothiolase deficiency
Inborn errors of fatty acid metabolism	Medium-chain acyl-CoA dehydrogenase deficiency
	Very long-chain acyl-CoA dehydrogenase deficiency
	Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency
	Trifunctional protein deficiency
	Carnitine uptake defect/transport defect
Inborn errors of amino acid metabolism	Classic phenylketonuria
	Maple syrup urine disease
	Homocystinuria
	Citrullinemia, type I
	Argininosuccinic aciduria
	Tyrosinemia, type I
Hemoglobinopathies	S,β-thalassemia
	S,C disease
	S,S disease (sickle cell anemia)
Miscellaneous multisystem diseases	Biotinidase deficiency
	Primary congenital hypothyroidism
	Congenital adrenal hyperplasia
	Classic galactosemia
	Cystic fibrosis
	Glycogen Storage Disease Type II (Pompe)
	Mucopolysaccharidosis type 1
	Severe combined immunodeficiency
	Spinal muscular atrophy
	X-linked adrenoleukodystrophy
Newborn screening by methods other than by heel stick	Critical congenital heart disease
	Hearing loss

*Recommendations as of September 2018. For updated information, see Health Resources and Services Administration. Advisory Committee on Heritable Disorders in Newborns and Children. Rockville (MD): HRSA; 2018. Available at: https://www.hrsa.gov/advisory-committees/heritable-disorders/index.html. Retrieved October 3, 2018.

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research are directed to the U.S. Department of Health and Human Services (www.hhs.gov).

Parent Education and the Role of Obstetric Care Providers

The results of surveys and focus groups of expectant parents demonstrate that women and their families would like to receive information about newborn screening during their prenatal care visits (11, 12). Obstetrician–gynecologists and other obstetric care providers should make resources about newborn screening available to patients during pregnancy. Information can be disseminated through informational brochures and electronic sources and through review or discussion at some time during prenatal care. Integrating education about newborn screening into prenatal care allows parents to be prepared for having their child undergo screening as well as for receiving newborn screening test results. Newborn screening does not replace the potential value of carrier screening nor does carrier screening replace newborn screening. Box 1 includes important information about newborn screening to guide obstetric care providers when counseling patients. There are also many available resources on newborn screening for patients and health care professionals, including videos, printable brochures, and websites (see www.acog.org/More-Info/NewbornScreening).

Providing newborn screening information during prenatal care visits can be accomplished in several ways and should be adapted to individual practice style. For example, newborn screening information could be provided during the first-trimester obstetric visit and could include written or website information along with other patient education materials. Newborn screening information could be given to patients later in pregnancy when other educational information is routinely distributed, such as at the time of glucose screening or group B streptococcal screening in the third trimester. Information on newborn screening also could be reviewed during a discussion of past adverse pregnancy outcomes related to a positive newborn screening test result or birth defect, while options for prenatal or preimplantation genetic screening or diagnostic testing are considered.

Box 1. What Obstetric Care Providers Need to Know About Newborn Screening

Patients should be offered information regarding newborn screening at some time during prenatal care including informational brochures, electronic sources, and review or discussion. This can be accomplished at different moments in prenatal care:

- During the first-trimester new obstetric visit and should include written or website information along with other patient education materials
- Later in pregnancy when other educational information is routinely distributed, such as at the time of glucola or group B streptococcal screening in the third trimester
- During a discussion of past adverse pregnancy outcomes related to a positive newborn screening test result or birth defect, while options for prenatal or preimplantation genetic screening or diagnostic testing are considered

Newborn screening programs are state-based public health programs that provide all newborns in the United States with presymptomatic testing and necessary follow-up health care for a variety of medical conditions for which early intervention will improve neonatal and long-term health outcomes.

- All states and the District of Columbia have individual newborn screening programs with varying policies, statutes, and regulations.
- Newborn screening consists of blood spot screening for metabolic and genetic conditions, hearing screening, and pulse oximetry screening for critical congenital heart defects.
- State health department's newborn screening programs vary as to the long-term disposition of samples for storage and research. For further information, please check with your state health department.
- Newborn screening does not replace the potential value of carrier screening, nor does carrier screening replace newborn screening.

For More Information

The American College of Obstetricians and Gynecologists has identified additional resources on topics related to this document that may be helpful for obgyns, other health care providers, and patients. You may view these resources at www.acog.org/More-Info/NewbornScreening.

These resources are for information only and are not meant to be comprehensive. Referral to these resources does not imply the American College of Obstetricians and Gynecologists' endorsement of the organization, the organization's website, or the content of the resource. The resources may change without notice.

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