What’s known:
In 2011, screening for critical congenital heart defects (CCHD) became the second point-of-care newborn screening test added to the Recommended Uniform Screening Panel, and it has since been widely adopted. Heart defects are the primary targets for CCHD screening, which often require evaluation by echocardiogram. An original list of seven conditions represented the most common critical lesions which routinely present with hypoxemia for newborns. Endorsed by the American Academy of Pediatrics and four other professional medical societies, the CCHD screening using pulse oximetry is required by law in all but two states.

Remaining challenges include national data collection and outcomes analyses at the population level.

What’s new:
An expert panel including Gerard R. Martin, MD, a cardiologist at the Center for Translational Science at Children’s National Health System, reviewed current practices in newborn screening for CCHD and identified opportunities for improvement. The panel’s study expanded the list of core conditions to 12 to emphasize the importance of other potentially critical, yet treatable secondary conditions. Roughly 79 percent of “positive” screens for CCHD identify secondary conditions, such as sepsis and pulmonary diseases. The study found algorithm misinterpretation was common in states collecting outcomes data, emphasizing needs for proper training and quality-assurance feedback mechanisms. Public health surveillance varied dramatically, with nearly one-fifth of states neither actively collecting data nor planning to do so. Additional CCHD screening research in special settings like the NICU, out-of-hospital settings, and areas with high altitude may result in adaptations to screening protocol. Future improvements to the current screening algorithm and analyses of the impact on CCHD outcomes will rely on further investment in a national data repository.

Questions for future research:
Q: What is the optimal algorithm for CCHD based on screening and testing ease of use, costs, resource utilization, and sensitivity for different treatment settings?
Q: What will be the impact on present screening for CCHD on outcomes of non-CCHD secondary conditions?