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Review

U.S. and international efforts on critical congenital heart disease screening: Can we have a uniform recommendation for Europe?

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SUMMARY

An estimated 90% of births or more in the United States will be screened for critical congenital heart disease (CCHD) by the end of 2014. Europe has made less progress despite providing the population-based studies that were critical in driving support for efforts within the United States. Congenital Heart Disease (CHD) advocacy groups, investigators in screening for CCHD and international health organizations have been meeting with health care providers and government officials on a country by country basis. Countries that are implementing or have pilot projects have been identified to track global implementation.

The Nordic countries, the United States, Switzerland and the United Arab Emirates are closest to universal screening for CCHD in newborns. Significant pilot projects tailored to unique care delivery systems screen through the use of midwives in the Netherlands, on maternity wards in the United Kingdom and while developing newborn care infrastructure in China. In Africa, South and Central America, individual countries are in the early stages of organization.

Screening for CCHD is spreading across the globe. Early recognition has the ability to improve care in countries providing CHD treatment and prepare parents for adverse events in countries where care is not accessible. Impact of screening in regions with less access to intervention will be important to track.

In September of 2011, the United States Secretary of Health and Human Services added critical congenital heart disease (CCHD) screening to the Recommended Uniform Screening Panel (RUSP). Within the same year, CCHD screening received the endorsements of major professional organizations including the American Academy of Pediatrics (AAP) [1], the American College of Cardiology (ACC), the American Heart Association (AHA) and the March of Dimes. Much of the foundational evidence supporting this recommendation was provided by population studies conducted in Europe [2,3]. In the three short years since its addition to the RUSP, CCHD screening has quickly become the standard of care in the United States. It is estimated that by the end of 2014 over 90% of the babies born in the US will be covered by CCHD screening (Fig. 1). Interestingly, implementation in many European countries has not occurred at the same speed even though many of the challenges of implementation are the same throughout the developed world.

Congenital heart disease (CHD) is the most common birth defect impacting one in 100 births. The most critical forms of congenital heart disease occur at a rate of roughly three per 1,000. Infants born with CCHD require intervention within the first few weeks or months of life to prevent significant morbidity or mortality. Infants who are identified as having CCHD and receive appropriate interventions have excellent outcomes and in most cases survive well into adulthood. Survival rates for even the most serious forms of CCHD are now excellent. However, studies have shown that as many as 50% of babies born with CCHD are not detected prior to leaving the newborn nursery [4] and death due to CCHD accounts for approximately 40% of all newborn deaths from congenital anomalies.

Pre-natal detection through fetal ultrasound (or fetal echocardiogram) only detects between 23% and 60% of cases of CCHD; even when combined with those identified through clinical assessment, a significant diagnostic gap remains [5]. Detection during newborn assessment is challenging – the human eye is not able to see visible cyanosis until oxygen saturation levels are near 80% or lower. Also, many of the critical forms of CCHD may not have a murmur and infants may not become symptomatic until closure of the ductus arteriosus and foramen ovale and transition between fetal and newborn circulation is complete. Variation in rates around detection depend both on type of lesion and on facility [6,7]. Most frequently missed lesions included hypoplastic left heart syndrome and coarctation

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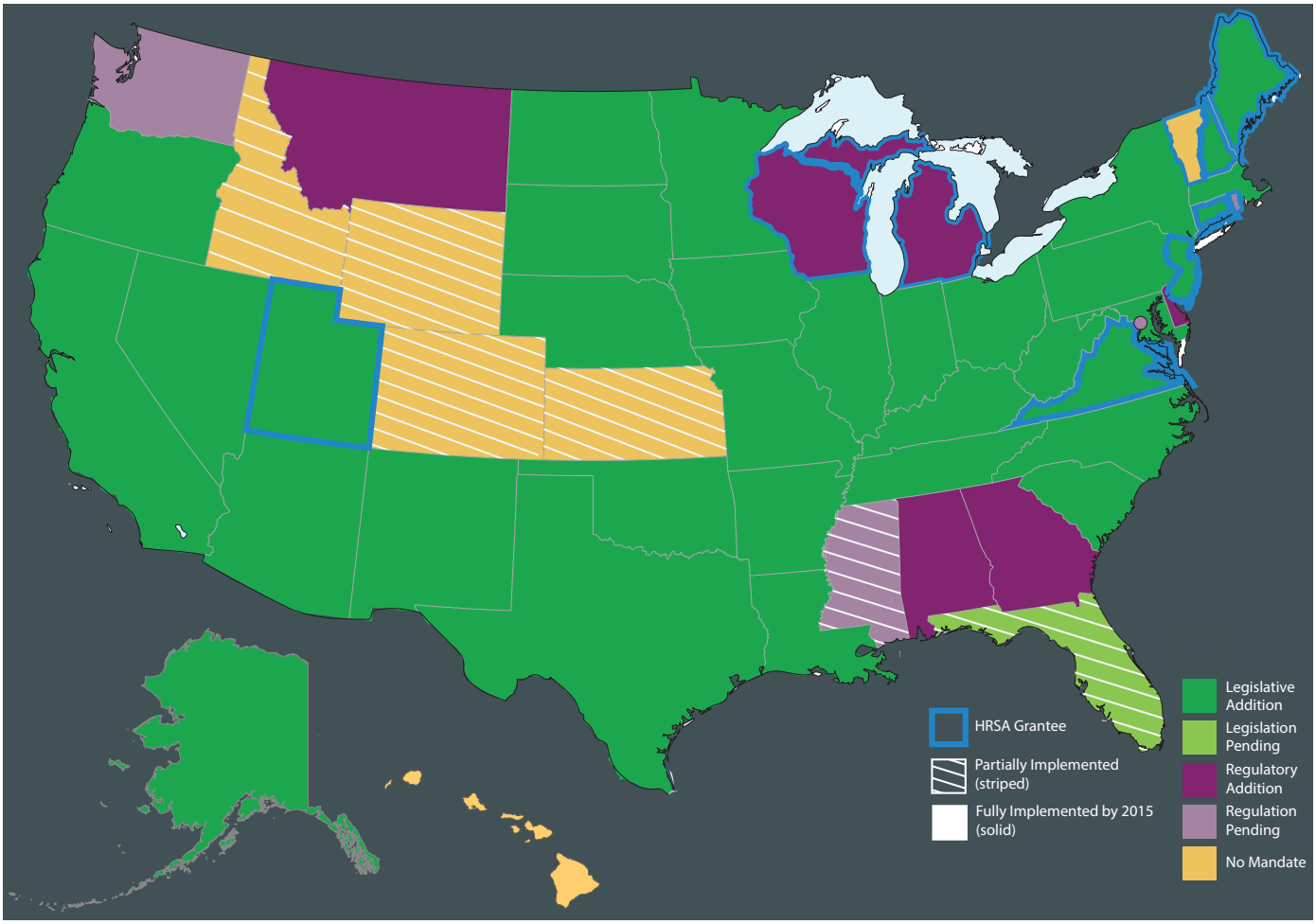

Fig. 1. United States implementation map – states with CCHD screening mandates (as of July 2014). The United States Health Resources and Services Administration (HRSA) awarded three year (2012–2015) education and implementation grants specifically for CCHD screening using pulse oximetry.

of the aorta; cyanotic lesions such as pulmonary atresia were also important.

Using pulse oximetry to assist in the detection of CCHD has met the criteria for universal newborn screening. Analysis conducted by the Centers for Disease Control and Prevention (CDC) suggests that CCHD screening can be cost effective, and the incremental cost using a reusable sensor is less than one U.S. dollar ($1) [8]. Implementation can be achieved smoothly without the hiring of additional nursery staff to perform the screen [9]. Pulse oximetry screening has a lower false positive rate than pre-natal ultrasound or clinical assessment alone, and in combination with traditional detection methods is well documented to significantly improve detection rates [2,3,10].

The algorithm recommended at the national level in the U.S. involves screening measurements on two limbs, the infant's right hand (pre-ductal measurement) and either foot (post-ductal measurement) (ref. [11] and Fig. 2). It is recommended for use in detecting asymptomatic newborns in a normal newborn nursery prior to discharge from the hospital of birth. The screen is performed between 24 and 48 hours of age or as close to discharge as possible if the infant is scheduled to be discharged prior to reaching 24 hours of age. Earlier screening is associated with a higher number of false positives, later screening increases the risk of the neonate suffering circulatory collapse on the newborn unit prior to detection [3]. For a failed screen, infants receive a complete assessment and echocardiogram to determine the cause for an abnormal reading. When combined with newborn physical examination, sensitivity using this algorithm is 82.7% and specificity is over 97.8% [2].

Although an infant may still have CCHD and pass the screen, de-Wahl Granelli et al. [2] demonstrated that CCHD screening using pulse oximetry also identifies infants with other serious causes for hypoxia such as sepsis, respiratory infection, congenital diaphragmatic hernia and persistent pulmonary hypertension. The implications of identifying these illnesses is particularly important in the developing world where complex open heart surgery or cardiac catheterization may not be available but fluid boluses, oxygen and antibiotics may be feasible life-saving interventions. In fact, 45% of false positives are positives for secondary targets (other serious situations that require identification prior to discharge) [2]. Therefore, it is crucial that infants who fail the screen be ruled out for other causes of hypoxia in addition to CCHD. Because of the low number of false positives and the detection of secondary targets, screening has not led to a significantly higher number of referrals to pediatric cardiologists or echocardiograms. In a recent Australian study where 18,801 infants were screened over a 42-month period, only nine additional echocardiograms were needed as a result of implementation [12].

In the United States, once CCHD screening was added to the RUSP at the national level it was then left to the states to determine whether they would require implementation of the screening recommendation within each state. The first states