**Pulse Oximetry Screening For Congenital Heart Disease in Newborns**

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Congenital malformations are one of the leading causes of infant death in the United States, and critical congenital heart defects (cCHD) are responsible for more deaths than any other type of malformation\(^1\). With the increased use of prenatal ultrasound\(^2\) and a better understanding of the postnatal clinical diagnosis of cCHD in the past decade\(^3\), the overall risk of death from cCHD has declined\(^4\). Congenital heart disease occurs in about 1% of live births, which is estimated to be approximately 26,000 per year in the United States. Approximately 25% of these infants will require expert cardiac care during the immediate newborn period or in early infancy\(^5\). Over the last 30 years, advances in surgical and interventional cardiology have greatly improved, and survivability into adulthood is much more common. Currently, there are approximately 800,000 adult survivors of congenital heart disease in the U.S., and the number continues to increase on an annual basis\(^6\). This has created a new patient population with a number of different challenges such as the impact of adult onset diseases on their underlying cardiac pathology, cognitive, physical, and psychosocial development, as well as issues related to reproduction, employment and health insurance. With this dramatic improvement in overall survival, minimizing morbidity takes on a greater urgency, starting with the newborn period.

Despite the magnitude of improved survival, undetected cCHD continues to remain a significant health problem. Up to 30% of infant deaths from cCHD occur before diagnosis\(^7\). Current methods of cCHD detection rely largely on newborn physical examination but fail to identify approximately 50% of cCHD\(^8\)

**“I hope he doesn’t inherit your ears”**

Dr. Catherine Karimov, MD

While we can easily accept imperfections in ourselves, we want no less than perfection for our children. Since the 1980’s, parents have cut their children’s leisure time in half to enroll them in activities that expand their minds or give them an edge in college admissions. Ensuring children’s success means pushing the envelope—even if it entails manipulating their genetic potential.

In one study, at least 10% of parents said they would approve of genetic testing to guarantee their child was athletic or intelligent, 10% would even test for height. Most would consider such selection of traits unethical, but what about less clearly vain choices? Not brawn or eye color but something more significant, like poor eyesight.

Just as we are finding worrisome trends in parents' preferences, genetic testing is becoming more attainable. Technology has now progressed to the point where comparative genomic array (array-CGH) testing may become the standard of care over the traditional labor intensive and expensive karyotype. As obstetricians, we have to adapt to the changing landscape and be ready for the pressure from patients who may want to know everything about their future offspring.

Current cytogenetic techniques were first discovered in the early 1950s. Stains for chromosomes were developed and perfected by the mid-1970s and allowed us to observe chromosomal deletions, duplications, inversions, and translocations. However, microscopy then could only show us a minimum of about 4 megabases. In the 1980’s, fluorescence in situ hybridization (FISH) was developed to increase the “resolution” and find deletions of DNA as small as 1 megabase. Still, we were limited by our clinical suspicions of an abnormality in specific area of the genome. It was not until 1990s that the array-CGH provided a breakthrough in genetic diagnosis for non-specific clinical disorders.

Arrays were originally developed for quick comparisons of gene expression in multiple cell lines. They were adapted to whole genome hybridization or “multiple FISH testing” by the year 2000. Initial array-CGH platforms were composed of multiple bacterial artificial clones (BACs) that contained reference cDNA up to 200 bases long. These have now been largely replaced by arrays of much shorter oligonucleotides, each between 25 and 80 nucleotides in length. Alternative platforms to those “traditional” approaches have already been developed and...
cases that were not previously detected on prenatal ultrasound 8. In 2010, the US Health and Human Services (HHS) Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) recommended that cCHD be added to the recommended universal screening panel. A workgroup was convened in January 2011. The SACHDNC, in collaboration with the American Academy of Pediatrics (AAP), the American College of Cardiology Foundation (ACCF) and the American Heart Association (AHA) worked together to outline cCHD screening implementation strategies that included pulse oximetry screening (POS). The SACHDNC considered seven specific lesions as primary targets for screening: hypoplastic left heart syndrome, pulmonary atresia, tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriosus. The workgroup also recognized the importance of developing electronic health information exchange systems to allow health care providers to easily track pulse oximetry monitoring and diagnostic outcomes 9.

A routine neonatal physical examination includes observation of skin color and peripheral perfusion, respiratory pattern and frequency, auscultation of heart and lungs, and palpation of the abdomen and femoral pulses. Findings that elicit suspicion for cCHD may include unusual heart murmurs, tachypnea, or decreased pulses. Visible cyanosis may occur as the acute physiologic changes correspond with ductal closure and decreasing pulmonary resistance within the first hours of life. However, subtle findings may not be evident prior to early discharge, frequently before 48 hours of life, and may be missed 10. Approximately half of all newborns with congenital heart disease are asymptomatic in the first few days of life 11. Whereas cyanotic heart disease is more likely to be detected with visual examination, left-sided obstructive heart disease is more likely to present with shock and death at the time of ductal closure, making nursery detection of subtle changes just as critical 11. It is important to identify and evaluate strategies which enhance early detection of cCHD in addition to routine physical examination.

Pulse oximetry can detect mild hypoxemia which may not be detectable by the human eye 12. Pulse oximetry screening (POS) has been supported as a simple, economical, and useful complimentary tool in addition to the established methods of prenatal ultrasound and postnatal clinical observation 13. Legislation has been proposed in some states to mandate routine use of POS for cCHD screening of newborns before hospital discharge 14. To date, only New Jersey and Maryland have actually passed the legislation.

Arterial saturation varies within the first 24 hours of life, with many healthy newborns having values less than 95% due to shunting at the ductal level. To achieve an acceptable specificity, testing >24 hours after birth would be optimal; however, with increasing early hospital discharges, this may not be possible. The SACHDNC workgroup (2011) recommended that screening not begin until 24 hours of life, or as late as possible if earlier discharge is planned. This can be done when routine screenings are completed and final weight assessment is obtained. Readings from both the right hand (pre-ductal) and either foot (post-ductal) should be obtained, either in parallel or direct sequence. If there is a >3% absolute difference in oxygen saturation between the right hand and foot, the screen is considered positive. The range of readings from 91% - 94% requires additional evaluation and/or rescreeing 15. An oximeter reading below 90% is considered an indicator of hypoxemia that warrants immediate intervention 16. Although readings that are > 95% in right hand or foot with < 3% difference is with < 3%

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difference is considered a negative screen, it is important that parents understand that this type of screening test does not detect all cCHDs, so it is possible to still have a critical or other heart defect with a negative screening result.

Screening based on SACHDNC recommendations utilizes a screening algorithm to provide consistent guidelines. It needs to be performed by qualified personnel who have been educated in the use of the algorithm and trained in motion-tolerant pulse oximeters. Congenital heart disease is a health issue that can affect any child, any race, any socioeconomic status, in any community. Universal POS can lead to early diagnosis and treatment for serious cCHD, potentially saving lives. There is a need for continued advocacy regarding cCHD and the importance of POS legislation in New York State.

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References:

*Please note: This topic is being presented at each of our upcoming Regional Perinatal Center Affiliate Quadrant QA meetings.*

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A new site has been recently created by the March of Dimes for partners to access a large bank of resources. The Prematurity Prevention Resource Center (PPRC), is a comprehensive source of information on prematurity and prematurity prevention. The site is targeted to professionals and includes the most current information on interventions, research, advocacy, professional education, global initiatives, teaching tools and resources to use with patients.

It’s also home for Healthy Babies are Worth the Wait® Program, the Prematurity Prevention Network and the 39+ Week Fee for Service Package, including the 39-Week Toolkit.

To register for the PPRC and join in the effort to reduce the incidence of preterm birth and improve birth outcomes go to: www.prematurityprevention.org

In the News

WASHINGTON, DC, FEB. 8, 2012 – The March of Dimes campaign to reduce medically unnecessary early deliveries is being elevated to the forefront of the nation’s maternal and child health agenda.

U.S. Department of Health and Human Services (HHS) Secretary Kathleen Sebelius announced the launch of Strong Start, a multi-faceted perinatal health campaign. This public-private partnership includes expansion of “Healthy Babies are Worth the Wait™” – the March of Dimes public awareness campaign to let women and health care providers know that if a pregnancy is healthy, it is best to wait for labor to begin on its own, rather than scheduling an induction of labor or a cesarean section.

To help reduce the increasing number of preterm births in America and ensure more babies are born healthy HHS Secretary Kathleen Sebelius announced more than $40 million in grants to test ways to reverse that trend, as well as a public campaign to reduce early elective deliveries.

HHS will also work with ACOG and a variety of other professional organizations.

For more information on the grants announced today and to learn more about efforts to reduce preterm births and early elective deliveries, please visit: www.innovation.cms.gov/initiatives/strong-start

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put into clinical practice in the past 2 years. These platforms are based on single sample hybridization to arrays that were initially designed for single nucleotide polymorphism (SNP) detection. This plethora of technologies may seem confusing, so it is helpful to think of them in terms of what they can do.

Karyotype and array will both detect aneuploidy, unbalanced translocations, large deletions, and duplications. However, the array has much higher resolution and potentially faster turnover rate since culture is not required. Array-CGH does not have the ability to diagnose balanced structural chromosomal abnormalities, such as balanced translocations, inversions, or polyploidy. Array-CGH can act as a defining tool to detect mosaicism for structural genomic abnormalities or marker chromosomes, although very low levels of mosaicism found on karyotype may remain undetected by array-CGH.

Like the karyotype, the array-CGH will not detect point mutations in such Mendelian disorders as cystic fibrosis or sickle cell disease. However, SNP-based platforms may turn this into a clinical reality in the near future. SNP-based array provides information about alleles of single bases so homozygosity can also be detected. Thus, uniparental isodisomy, which is the genetic basis of many disorders such as Russel-Silver or Angelman syndrome, can now be detected. An important caveat is that private information can be discovered from the SNP-array that a patient may not want us to know, such as consanguinity or non-paternity.

True costs of array-CGH have decreased to about $400. Karyotyping cannot be done for much less than that. Array-CGH can detect nearly everything that karyotypes do and even give more information for syndromes that have no detectable ultrasound changes or screening tests. One study in Korea found that 1 in 200 of the women who had amniocentesis for age, abnormal screening, or maternal anxiety, had an array-CGH finding undetectable by karyotype.

Array-CGH may lead to the diagnosis of more and more subtle syndromes and changes that likely lead to not-so-severe abnormalities. For example, a rare form of early severe obesity has been linked to a deletion on the small arm of chromosome 16. In another example, a deletion on the Y chromosome could potentially render a male offspring infertile.

Thus, there are both upsides and downsides to this increasingly affordable new technology. What is the best way to balance them? I do think we should switch to array-CGH once routine karyotyping is too expensive. However, the array should be limited as much as possible to the syndromes that carry the most morbidity for the routine analysis. Higher-resolution or SNP-arrays could be used for fetuses with known congenital anomalies or to aid in diagnosis in at-risk fetuses. Of course, the perfect balance may always be just out of our reach. But like it or not, this future is coming.

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1. http://www.stylist.co.uk/life/how-to-make-the-perfect-baby
OTIS — Organization of Teratology Information Specialists

**Mission**

The Organization of Teratology Information Specialists (OTIS) is dedicated to providing accurate evidence-based, clinical information to patients and health care professionals about exposures during pregnancy and lactation. The organization serves to provide education, to conduct relevant research and to support teratology information services throughout North America.

**Goals**

- Provide counseling to patients and health care professionals about exposures related to pregnancy and breastfeeding, so they can make informed treatment choices.
- Reduce risks and decrease the number of preventable birth defects by educating the public and health care professionals about specific agents of concern.
- Empower patients with accurate information about the true risks of an exposure, thereby preventing unnecessary terminations of wanted pregnancies.
- Facilitate communication and collaboration between teratology information services.
- Contribute collectively to research and the worldwide literature in the field of teratology.

**Brief History**

- OTIS was first established as the Organization of Teratology Information Services in 1990 by individual teratology information services throughout the United States and Canada. It was incorporated in April 1999 as a not-for-profit organization. While OTIS members primarily represent Teratology Information Services (TIS) located in the United States and Canada, some members hail from services outside North America and many members are not directly affiliated with a TIS. To accurately reflect this diverse membership, OTIS changed its name in 2005 from the Organization of Teratology Information Services to the Organization of Teratology Information Specialists. Members of OTIS have a wide range of backgrounds and experience in the field of teratology and include medical doctors, genetic counselors, nurses, researchers, and educators.
- A critical function of OTIS is to educate patients, health care professionals, and the general public about the field of teratology. OTIS has developed many fact sheets about various exposures during pregnancy and lactation in the general categories of medications, infections and vaccines, illicit substances, herbal products, maternal medical conditions, and other common exposures. The OTIS fact sheets are reader friendly and include information about breastfeeding and paternal exposures as well as exposure risks prior to and during pregnancy. OTIS fact sheets, as well as other types of educational resources for patients and health care providers, are available on the OTIS website at www.otispregnancy.org.
- With its diverse membership composed of experts in the fields of teratology and women’s health, its unique approach to evidence-based research, and its dedication to the evaluation and dissemination of current and accurate information regarding exposures during pregnancy and lactation, OTIS has established itself as one of the world’s primary teratology research organizations and a critical resource for patients and health care providers throughout North America.

**For Further Information:** [http://www.otispregnancy.org](http://www.otispregnancy.org) or call 866-626-6847

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