Fellow's Column: A Case of Hypoplastic Left Heart Missed on Congenital Heart Defect Screening

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Introduction:

In September 2010, the US Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) recommended that screening for critical congenital cyanotic heart defects be added to the uniform newborn screening panel. The goal was to identify infants with structural heart disease that develop morbidity and mortality due to normal physiologic changes in infancy, such as the closure of the ductus arteriosus. They initially recommended a list of seven cardiac lesions to be primary targets of such screening: hypoplastic left heart syndrome, pulmonary atresia, tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriousus. All of these lesions are traditionally associated with hypoxia. (1)

The SACHDNC assembled a workgroup composed of the AAP, pediatric generalists, pediatric specialists, and other relevant parties in 2011 and developed the screening recommendations for congenital heart defects, which were published in Pediatrics in November 2011. In that article, an additional list of secondary screening targets were added: coarctation of the aorta, interrupted aortic arch, double outlet right ventricle, Ebstein anomaly, and single ventricle complex. (1)

Since that time, congenital heart defect screening has become mandated and has undergone implementation across the nation. The CDC has tracked outcomes since these recommendations have been published. According to CDC data, pulse oximetry screening has reduced early infant death related to critical congenital heart defects by 120 instances per year (a 33% reduction). Additional research has postulated that there is a cost of $12,000 for every year of life gained for babies detected by congenital heart defect screening. This would indicate that pulse oximetry screening for congenital heart disease is, in fact, quite effective. (2)

We will discuss the case of an infant impacted by congenital cardiac defect screening.

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Case Presentation:

Our patient is a full term Caucasian male born via SVD to a 32-year old G3P3 woman. Pregnancy was uncomplicated with normal prenatal labs and normal prenatal ultrasound anatomy scan. Delivery was uncomplicated, and baby remained with mother throughout the nursery course. Baby breastfed well and did not have excessive weight loss or hyperbilirubinemia. He was discharged home with mother on day of life two after normal congenital heart defect screening.

After discharge, the baby was noted to develop rhinorrhea on day of life three with subsequent development of left eye discharge. Siblings were noted to have viral URI symptoms as well. PCP prescribed a seven-day course of ocular erythromycin for presumed lacrimal duct obstruction. On day of life 12, mother noted that the baby developed increased work of breathing with subcostal retractions but continued to feed well with normal elimination habits.

On day of life 19, the mother again took the baby to PCP's office. In PCP's office, the baby was noted to have retractions with SpO_2 as low as 86% and so was directed to ED. While in the ED, the baby was noted to have tachycardia, tachypnea, nasal flaring, and retractions. He was started on high-flow nasal cannula (HFNC) 10L with noted improvement. The patient was admitted to NICU for further management.

At time of admission to the NICU, the initial exam was notable for yellow discharge from the left eye, bilateral subcostal retractions, upper airway sounds transmitted throughout all lung fields, and mildly diminished breath sounds in the right lung fields. Labs on admission were notable for a metabolic acidosis thought to be secondary to dehydration and were otherwise unremarkable, including normal lactate. Initial CXR showed pulmonary vascular congestion with the suggestion of bilateral

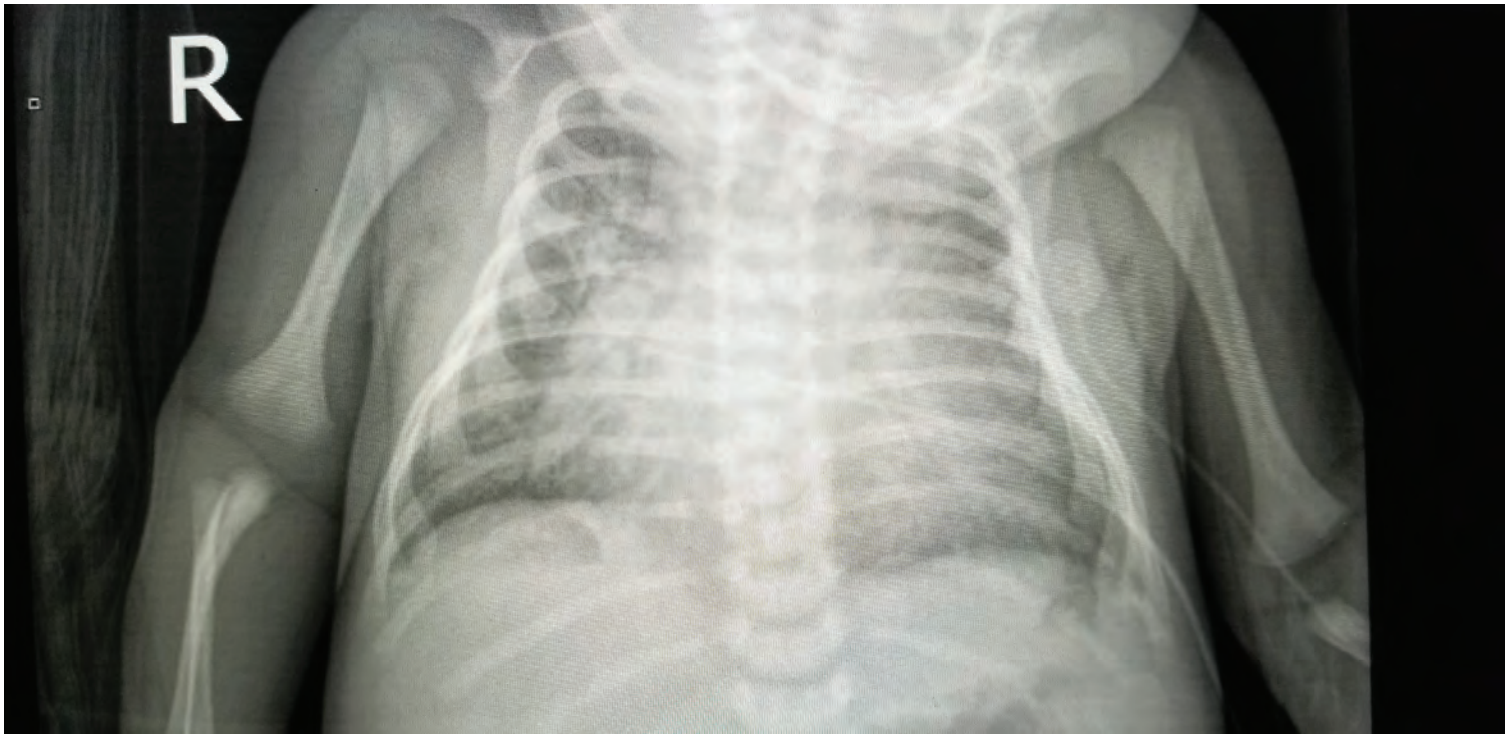

Fig 1. Initial CXR showed pulmonary vascular congestion with the suggestion of bilateral perihilar airspace disease, which obscured the cardiothymic silhouette.

perihilar airspace disease, which obscured the cardiothymic silhouette. Baby was diagnosed with acute respiratory failure likely secondary to viral vs bacterial infectious etiology and was started on ampicillin and gentamicin.

During the first few days of treatment, only minimal improvement in clinical status was noted. By day five of admission, the baby had been unable to wean fully from HFNC support and had frequent episodic tachypnea and desaturations requiring tactile stimulation. Additionally, a new mid systolic murmur was heard.

Echocardiogram was subsequently performed and demonstrated hypoplastic left heart syndrome, long-segment coarctation, moderate PDA with bidirectional shunting and signs of impaired RV function. The patient was started on milrinone and alprostadil. The baby underwent Norwood procedure on day of life 32. After recovery from surgery, the patient was noted to be improved clinically and was discharged to home to await further palliative surgery and probable eventual cardiac transplant.

Discussion:

While current data may indicate the cost-effective nature of pulse oximetry for congenital heart defect screening at the population level, cases like the one above may call into question the sensitivity of the screen at the patient level.

A study published in Pediatrics in 2015 evaluated the number of infants detected and missed on neonatal congenital heart defect screening. Data analyzed from metropolitan Atlanta, GA from 2000 to 2005 was used to create a simulation model of detection rates of both primary and secondary screening targets. (3)

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It was determined that ~25% of all congenital heart defects would fall under the critical disease heading, which constitutes the primary and secondary screening targets for congenital heart defect screening. Using data previously gathered by the National Birth Defects Prevention Study, it was determined that

30% of critical congenital heart defects were detected prenatally on ultrasound. An additional 40% were detected within the first three days of life due to neonatal symptoms. These two categories were labeled "timely detection," in that they would lead to initial echocardiogram by the third day of life and prior to birth hospital discharge. The remaining 30% of congenital heart defect cases would, therefore, fall into the "late detection" category and represent the infants that we would hope to detect on neonatal cardiac screening. (3)

The simulation model predicted that over the course of a year, half of the infants who would fall into the "late detection" category would be caught by newborn congenital heart defect screening; ~800-1100 cases per year. However, the remaining half of late detection cases were predicted to be false negatives on the congenital heart defect screen and therefore remain undetected. This represents ~800-1000 cases per year of critical congenital heart defects that remain undetected past the third day of life in spite of congenital heart defect screening. (3)

It is worth noting that the rates of detection vary based upon which critical congenital heart defect is being screened. For example, pulmonary atresia is highly likely to present clinically while still admitted to the birth hospital, and essentially zero cases are predicted to be missed on congenital cardiac defect screening. In contrast, 16% of cases of tetralogy of Fallot and 38% of cases of coarctation of the aorta are predicted to slip past screening. Looking at the data, we see that an anticipated two percent of hypoplastic left heart syndrome cases are predicted to be missed through initial discharge, similar to the patient discussed above. (3)

These infants are at substantial risk of morbidity and mortality related to their critical congenital heart defects, which are often exacerbated by normal newborn physiologic changes such as PDA closure. The above case likely represents one such infant whose condition was potentially exacerbated by physiologic alterations of blood flow across the PDA. It is conceivable that other missed cases of congenital heart defects may present similarly.

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We would like to discuss one possible method for improved detection of congenital heart defects that has been proposed, called peripheral perfusion index (PPI). PPI is calculated by taking the ratio of pulsatile to non-pulsatile blood flow through tissue and is measurable by certain types of pulse oximeters. PPI is affected by stroke volume and therefore, may be an indicator of circulatory or cardiac disease.

A case-control study was conducted in Sweden, where data from 10,000 newborns without CHD were tested to define a normal PPI range. During data collection, nine infants were diagnosed with ductal-dependent systemic circulation related to congenital heart disease; four cases of coarctation of the aorta, two of interrupted aortic arch, two of hypoplastic left heart syndrome, and one of critical aortic stenosis. All nine of these infants were noted to have measurement of PPI below the interquartile ranges of the normal controls and five of the nine tested had a PPI below the fifth percentile of the calculated normal range. Additionally, it was noted that three of these nine infants were not detected on standard pulse oximetry screening. This data may suggest that PPI merits further evaluation as a congenital heart defect screening tool. (4)

While this study does not provide definitive data on rates of detection for various cardiac defects or delve into important population health topics like rates of false positive screening and cost-effectiveness, it does open the door to further discussion on new or adjunctive congenital heart defect screening practices.

References:

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