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SHORT REPORT

Failed detection of complex congenital heart disease (including double outlet right ventricle and total anomalous pulmonary venous return) by neonatal pulse oximetry screening

Patrick G. Hetzel · René Glanzmann · Joëlle Günthard · Elisabeth Bruder · Esther Godi · Christoph Bührer

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Abstract We report on a newborn infant with complex congenital heart disease (CHD) featuring double outlet right ventricle and hypoplastic left ventricle who had postductal oxygen saturation well above 95% and thus eluded pulse oximetry screening for CHD.

Keywords Complex heart defect · Double outlet right ventricle · Hypoplastic left ventricle · Neonatal pulse oximetry screening

After an uneventful pregnancy with unsuspicious first and second trimester out-of-hospital ultrasound scans, this girl was born at term from meconium-stained amniotic fluid with Apgar scores of 9, 10 and 10. She was transferred to the neonatology ward at 15 h of age because of tachypnea. There was no heart murmur, pulses were equally palpable, and upper and lower extremities blood pressure was normal. Chest X-rays showed some minor pulmonary opacities, heart was normal by size and shape. She was transiently given supplemental oxygen (maximum 40%) during the first 9 h. Thereafter, pre-and postductal oxygen saturations (Hewlett-Packard HP66S) were consistently above 95% also with room air. Blood gas analysis was always normal (minimum pH 7.34, worst base deficit-5 mmol/l). Tachypnea persisted until day 9 of life when the infant suddenly had progressive cyanosis (minimum oxygen saturation 84%) responsive to supplemental oxygen, apneas, absence of femoral pulses, hypotension of the lower extremities, oliguria and metabolic acidosis. Echocardiography revealed complex CHD, including a double outlet right ventricle, total anomalous pulmonary venous return into the superior vena cava, primum-type atrial septal defect, mitral valve atresia, hypoplastic left ventricle, coarctation, and patent ductus arteriosus. After discussions with the cardiologists and neonatologists involved, the parents decided against surgery, and the girl died soon afterwards. Autopsy confirmed the echocardiographic findings.

A recent report in this journal demonstrated that postductal pulse oximetry screening had 100% sensitivity and 99.7% specifity for detection of critical CHD [2]. Out of 3,262 asymptomatic newborn infants, pulse oximetry screening identified 17 infants with critical CHD, while there were no false negative cases. Based on this study, the Swiss Societies of Pediatric Cardiology and Neonatology have recommended general screening by pulse oximetry [1].

Some critical CHD (such as coarctation with large ventricular septal defect or critical aortic stenosis) may go undetected if there is abundant central right-to-left shunt [3–6].

P. G. Hetzel · R. Glanzmann · C. Bührer
Department of Neonatology, University Children's Hospital, Basel, Switzerland

J. Günthard
Department of Cardiology, University Children's Hospital, Basel, Switzerland

E. Bruder
Department of Pathology, University Hospital, Basel, Switzerland

E. Godi
Department of Obstetrics, Women's Hospital Bruderholz, Basel, Switzerland

P. G. Hetzel (✉)
Universitäts-Kinderspital beider Basel, CH-4005 Basel, Switzerland
e-mail: patrick.hetzel@ukbb.ch

Moreover, CHD with common outlet, such as double outlet ventricle (left or right) may show equal and even normal oxygen saturation in both great arteries if pulmonary blood flow greatly exceeds systemic blood flow. Thus, pulmonary overcirculation may mask CHD by normal saturations and tachypnea may remain the only clinical sign.

In our case, two hemodynamic peculiarities contributed to regular postductal saturations: 1. Pre-atrial mixing of oxygenated and desoxygenated blood (total anomalous pulmonary venous return) with high saturation in the right atrium and ventricle. 2. Origin of both great arteries from the right ventricle. Clinical deterioration ensued following narrowing of the duct and the aortic isthmus with diminished perfusion of the lower body half.

Our case emphasises that pulse oximetry screening, even when used in conjunction with clinical examination, may miss critical CHD. Persistent unexplained tachypnea in newborn infants should prompt echocardiographic examination.

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