Postnatal screening for congenital heart disease: Clinical findings, Pulsoximetry, Echocardiography

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Introduction: Congenital heart defects (CHD) are the most common congenital malformations. Echocardiography performed by a pediatric cardiologist is regarded as the gold standard, detecting even small cardiovascular defects. The primary objective of newborn screening is the pre-symptomatic identification of life-threatening CHD in order to achieve a timely diagnosis before collapse or death occurs.

Methods: Supported by the government of Hessen, and in written informed consent of the parents, screening echocardiograms (ECHO) were offered for a period of eleven years for all babies who were born between August 1, 2007 and November 30, 2018 at a large maternity hospital managed by obstetricians. The population of neonates comprised those who were unremarkable during pregnancy and who were born at term. The pulse oximetry readings (OXI) were taken between 2nd and 4th hour of life. An experienced pediatric cardiologist performed the echocardiograms using a 10 MHz transducer within the first days (median: 27 hours) of neonatal life after the clinical examination (CLIN). In this study, the results of ECHO were compared with those obtained from CLIN and OXI (Sp02 < 95%).

Results: 13050 neonates (86%) could be included. 599 cases (45.9/1000) of CHD were detected by ECHO: 18 critical, life-threatening CHD (1.4/1000), 155 hemodynamically significant CHD (11.9/1000), 415 minor VSD (31.8/1000) and 11 other defects. Only 16% of the congenital heart defects were detectable by CLIN, 3% by OXI. The sensitivities with regard to the critical CHD were 100% (ECHO), 39% (CLIN), 33% (OXI), 56% (CLIN+OXI). In 16% of the neonates, a fetal echocardiography was performed as a result of the patient history or family history. No congenital heart defect was detected antenatally which was a prerequisite of this maternity hospital.

Conclusions: OXI is an additional tool to detect missed diagnoses of CHD in early life. ECHO is expensive and personnel-intensive, but the only strategy to rule out missed diagnoses of CHD.