

Timely diagnosis of congenital heart disease - did we improve?

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Background: 12 years ago, a study was published showing that in our referral population, cardiac diagnosis was made late in 10% of all patients with congenital heart disease (CHD) requiring therapy, resulting in too late initiation of therapy according to accepted standards of treatment timing. The rate of late diagnosis was the same in cyanotic and acyanotic CHD with 10% in each.

Methods: An identical study was performed as 12 years ago with a prospective evaluation of the time of diagnosis of CHD during a 3-year period ending in June 2011. Only patients with newly diagnosed CHD that required either catheter-interventional or surgical therapy were included. Of note that in between the two studies came the nationwide recommendation for neonatal pulse oximetry (POX) screening of all newborns starting in 2006.

Results: A total of 209 patients were included. 41% of these had cyanotic, 59% acyanotic CHD. According to the study criteria, late diagnosis was observed in 21 patients (10%), 6% (5 of 85) of cyanotic and 13% (16 of 124 patients) of acyanotic CHD. The 2 most frequent heart defects with delayed diagnosis were atrial septal defect and coarctation (7 and 6 patients, respectively). Delayed diagnosis resulted in one patient dead (undiagnosed interrupted aortic arch).

Compared to the historical study in our referral population, the striking finding was that still 10% of all CHD diagnosis was made late. The main difference was that late diagnosis in cyanotic CHD decreased from 10 to 6%, whereas in acyanotic CHD an increase from 10 to 13% was seen. Moreover an increase in fetal diagnosis was not surprisingly seen to currently 26% of all children with relevant CHD included in the study.

Conclusion: After 12 years of referring physician education and introduction of a nationwide POX screening, the rate of late diagnosis of CHD in our referral population remained stable at 10% of all CHD, only the rate of delayed recognition of cyanotic CHD showed a decline.