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Leading Article

Creating Awareness for Early Identification of Congenital Heart Disease

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Over the past 30 years there has been increasing awareness of the importance of early referral of newborn with heart disease to special center. Continuous advances in technology and training in pediatric cardiology and pediatrics have improved long term outcome and promised better quality of life.[1] Like other developing countries, Bangladesh is facing a multitude of health problems. Pediatric heart diseases are only one of them. Congenital heart disease (CHD) occurs in 5-8 out of every 1000 live births.[2,3] Approximately one quarter of these children have critical congenital heart disease (CCHD), which by definition requires surgery or catheter intervention in the first year of life and responsible for more deaths than any other type of malformation.[4]

Without early recognition, diagnosis and treatment, a majority of infants and children with congenital heart disease die in their first month of life in developing countries.[5] Although, pediatric heart diseases are not explicitly included in the Millennium Development Goals,[6] accomplishing some of the goals will have a direct impact on children with heart disease in developing countries. Improving vaccination and sanitation, increasing the number of skilled health care professionals and awareness for early diagnosis can benefit children with congenital and acquired heart disease.

Status of Cardiac Care

There is a wide gap between the developed and developing countries regarding pediatric cardiac care. Absence of pediatric cardiac centers, presence of cardiac centers only in large cities, unstable political systems affecting social stability, and absence of specific health care policies in various countries are the reasons for this variation.[7]

Four major reasons due to which treatment for congenital heart disease is currently out of reach for a majority of children in Bangladesh are: (1) Scarcity of pediatric cardiac care in public hospitals; (2) Expense of treatment in private hospitals; (3) Lack of resources and trained personnel in this field; and (4) Lack of awareness.[8] Although there has been establishment of tertiary-level pediatric cardiac care services in public hospitals, the time has arrived to further enhancement in the country. Due to a lack of resources cardiac care inadequacy cannot be solved within a short span of time. Giving urgent attention to 2 important reasons for inadequate pediatric cardiac treatment in Bangladesh can change the scenario drastically. These are (1) Lack of awareness about CHD and (2) High rate of unsupervised home deliveries because of which CHD are not detected at birth. Too often referrals are delayed because of ignorance or limited knowledge of CHD or its natural history, inaccurate diagnosis and lack of awareness about available facilities. Moreover many families may seek advice from unqualified doctors, complicating the situation.

In the current era, congenital heart surgery allows for repair or palliation of nearly all types of congenital heart malformations.[9] Congenital heart surgery, together with transcatheter interventions, has resulted in a marked improvement in survival for those with CCHD.[10] With the advent of prostaglandin therapy for ductus arteriosus-dependent lesions, many previously lethal congenital heart conditions that present with severe hypoxemia, shock, and acidosis in the newborn period are now survivable and can be palliated. Intervention is typically performed in the first weeks of life to optimize hemodynamics and prevent end-organ injury associated with delayed diagnosis. Because timely recognition of CCHD could improve outcomes, it is important to identify and evaluate strategies to enhance early detection. In this scenario it is now vital to take basic steps such as increasing awareness of CHD amongst the general population and amongst healthcare providers in order to triage CHD cases for

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early referral and proper management. Only then it will be possible and feasible to improve the diagnosis and treatment of congenital heart disease within a short span of time.

Diagnosis of CHD

Recent studies show that a high proportion of neonate with critical CHD experienced late or no referral to cardiac specialty center accounting for significant number of death.[11] Methods to improve early detection of CCHD appear warranted. Evidence is there that routine pulse oximetry performed on asymptomatic newborns after 24 hours of life, but before hospital discharge, may detect CCHD.[12-13]

In practice, infants and older children with CHD may be diagnosed on the basis of physical examination findings, such as heart murmurs, tachypnea, or overt cyanosis. But, in neonate such findings are not always evident following birth. Skilled physical examination, which is a sensitive and specific screening tool in older children, does not always distinguish between neonates with and without congenital heart disease.[14] A recent study in the United Kingdom suggested that 25% of infants with CCHD are not diagnosed with heart disease until after discharge from the newborn nursery.[15] The median age of diagnosis in these cases was 6 weeks. A recent publication from the United States also suggested that delayed or missed diagnosis occurs in 7 per 100 000 livebirths.[12] Newborns with CCHD are susceptible to profound, sudden worsening in clinical status leading to shock in the first few days and weeks of life. The severity of organ damage is a function of the extent of insult, differential flow to organs as the neonatal circulation responds to the hypoxic/ischemic insult, and the oxygen requirement of each organ.

A number of children with CCHD are so severely compromised at presentation that they die before surgical intervention. Investigators have reported that between 3% and 6% of neonates with dextro-transposition of the great arteries died because of hemodynamic compromise before surgical intervention could be offered.[16] After birth, screening for congenital heart disease by physician or pediatrician is currently not even accomplished by physical examination within the first 24 hours of life and on subsequent nursery visits. Presently in the cities of Bangladesh due to their easy availability there is indiscriminate use of echocardiograms without prior CXR, sometimes even without any indication or pediatric cardiology consultation.

Hypoxemia is difficult to detect in newborns, and the transitional circulation masks important clinical findings such as absent femoral pulses while the ductus arteriosus remains patent. Earlier, though, it was found that clinical assessment is as good as investigational workup in differentiating innocent and pathological murmurs in children.[17] Perhaps most importantly, physical examination skills are on the decline in current trainees.[18] Practicing pediatricians currently have limited experience in discriminating innocent from pathological murmurs even in older children. In a contemporary series in which echocardiography was performed to evaluate for possible heart disease based on suspicious physical examination, fewer than 15% of subjects were found to have significant congenital heart disease.[19]

When used as a screening tool, echocardiography has a high frequency of either false-positive results (usually related to the transitional circulation) or recognition of clinically benign diagnoses (eg. PFO, small muscular ventricular septal defects). In addition, there may be an inadequate supply of trained personnel who could perform this screening with a reasonable degree of accuracy.

Therefore, to improve timely detection of CCHD a number of investigators have proposed that pulse oximetry can be considered as a complementary modality to the newborn physical examination.[20]

Pulse Oximetry and CHD

Pulse oximetry has the potential to identify hypoxemia that might not otherwise produce visible cyanosis. Pulse oximetry is used routinely in the assessment of young children in neonatal intensive care units and emergency departments. It has been proposed as an adjunct to the assessment of the newborn in the delivery room.[21] As such, some have proposed that pulse oximetry be considered as a vital sign equivalent in importance to pulse, respirations, and blood pressure.[22]

The investigators observed that pulse oximetry is much more effective in identifying infants with CCHD and is more accurate and much less expensive than screening all newborns with echocardiography. Using a cutoff of 95% in lower-extremity saturation, Hoke et al,[23] suggested that 81% of neonates with CCHD could be identified.

However, arterial oxygen saturation varies considerably in the first 24 hours, with many healthy newborns

having arterial saturations of less than 95%. As such, oximetry screening before 24 hours of life can result in a significant number of false-positive results due to transition from fetal to neonatal circulation. A study from the United Kingdom reported that the false-positive rate was as high as 5% when oximetry screening was performed in the first 24 hours compared with 1% at the time of hospital discharge.[24] Later screening can miss an opportunity for intervention for defects that are impacted by closing PDA.

Therefore, to achieve an acceptable specificity, testing after 24 hours of birth would appear to be the most reasonable strategy.

A screen is considered positive if (1) any oxygen saturation measure is <90% (in the initial screen or in repeat screens); (2) oxygen saturation is <95% in the right hand and foot on three measures, each separated by one hour; or (3) a >3% absolute difference exists in oxygen saturation between the right hand and foot on three measures, each separated by one hour. Any screening that is 95% in the right hand or foot with a 3% absolute difference in oxygen saturation between the right hand or foot is considered a negative screen and screening would end.[3]

Pulse oxymetry is highly specific for detection of CCHD with moderate sensitivity, thereby meeting criteria for universal screening. Thus screening all newborn babies with pulse oxymeter in addition to the usual routine physical examination is essential to identify CHD and can be used as universal screening for CHD.[26]

Prenatal diagnosis of CHD

Over the past two decades, imaging has become the principle diagnostic tool in prenatal detection of fetal malformations. Cardiac abnormalities are among the major malformations that are most frequently missed in prenatal ultrasound examinations which is a cause for concern because undetected CHD increases the risk of early neonatal mortality. Prenatal diagnosis allows full investigation of affected fetuses for coexisting abnormalities and improved counseling of families and offer a proper perinatal and neonatal management.[27] Studies show that if CHD is detected before birth, there are significant benefits for babies, their families and for medical services around the time of birth and in the first year of life.[28] Prenatal diagnosis and appropriate treatment may prevent the devastating consequences of early circulatory collapse, such as brain damage. But access to and availability of this type of prenatal screening may be limited in rural or low socioeconomic status areas and within certain racial/ethnic groups. Generally, detailed fetal echocardiography is not performed routinely for prenatal screening but is reserved for cases which are at high-risk for CHD like family history of CHD; coexisting maternal disease; exposure to teratogens in early pregnancy; infections such as parvovirus B[19], rubella, coxsackie; abnormal karyotype and extracardiac foetal anomalies such as diaphragmatic hernia, exomphalos noted on a general foetal sonogram. Only 10 percent of the fetuses with cardiac anomalies have identified risk factors.[29] Hence, basing referral for foetal echocardiography on the presence of risk factors only excludes about 85% of foetuses with severe detectable heart defects from screening.[30] In such cases, detailed foetal echocardiography is commonly done between 18 and 22 weeks of gestation.[31] Prenatal diagnosis will improve the chances of survival of a baby with a critical congenital defect by ensuring that the necessary prenatal and postnatal care is provided.

Appropriate detection of cardiac abnormalities can only be achieved by carrying out routine fetal echocardiography in all fetuses in the second trimester, irrespective of the presence or absence of risk factors for the development of congenital cardiac disease. This is not yet possible in a developing country like Bangladesh.

Recommendation

With identifiable risk factors fetal echocardiography at 18-22 weeks of gestation and pulse oxymetry in addition to routine clinical examination of newborn as screening for CHD at 24-48 hours of life should be performed.

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