LATE CLINICAL PRESENTATION OF CONGENITAL HEART DISEASES

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??? LATE PRESENTATION??
“PROPER” presentation:

When the patient presents at a stage which does not necessitate an emergency management, and when treated does not carry a higher risk than international figures for the same anomaly and does not need a different plan of management with worse outcome than if treated earlier.
Always make sure of chromosomal abnormalities
Why Late?

Inavailability of medical service
Improper medical service
No clear clinical findings

“BRIDGING THE GAPS”
Impact of the problem

Prevalence of (CHD) at birth is 4 to 6 per 1000 live births
In preterm infants (gestational age <37 weeks), 12 per 1000 live preterm births.

*Potentially life-threatening lesions in 15 percent of infants with CHD:*
62% diagnosed before discharge from the hospital
8% diagnosed antenatally.
25% diagnosed after discharge from the birthing hospital
5% percent diagnosed after death
1/3 of newborns with congenital heart anomalies are discharged home without being diagnosed

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Pre-Natal Screening

*Sensitivity*: 0 - 80 % detection rates.

- Operator experience
- Gestational age
- Maternal weight
- Fetal position
- Type of defect
Lack of early findings before hospital discharge

1,067 babies with CHD who had a routine newborn examination:
876 (82%) not recognized to have CHD before hospital discharge.
Of these undiagnosed infants, 306 (35%) became symptomatic or died without a diagnosis before six weeks of age.

Abnormalities were detected on the neonatal examination (usually cyanosis or a murmur) in 476 (45%), but only 170 were referred for diagnosis.

Routine examination of babies available at six weeks (the remainder became symptomatic or died) detected abnormalities in only 164 of 252 (65%).

Lack of early findings before hospital discharge

So,

routine examinations of asymptomatic infants in the immediate newborn period and at six weeks of age failed to detect heart disease in more than one-half and one-third of affected infants, respectively. A normal examination did not exclude heart disease, including potentially lethal abnormalities. Good prenatal care should not lull the practitioner into a false sense of security. The clinician must remain vigilant to detect early signs and refer infants promptly to a pediatric cardiologist for definitive evaluation.

Suspicious Signs & Symptoms

Poor feeding: bottle feedings longer than 20 to 30 minutes, taking too little volume, resting frequently during feeds, or otherwise unexplained choking, gagging and/or frequent vomiting with feeds
Breathing too fast or hard, particularly increasing with feeds
Persistent unexplained cough or wheeze
Color changes: central cyanosis, persistent pallor, grey
Excessive sweating, even while sleeping, increasing with feeds and other exertion
Excessive, unexplained irritability
Decreased activity; increased or excessive sleeping
Poor weight gain
Infants with severe lesions

Can deteriorate precipitously

*Retrospective study* - 490 infants with *critical heart disease* (defined as CHD that required invasive intervention or resulted in death in the first 30 days of life)

44% diagnosed prenatally.

Significant physiologic compromise (defined as severe metabolic acidosis, seizures, cardiac arrest, or laboratory evidence of renal or hepatic injury) occurred in 16% of the overall group.

There was no difference in the frequency of these events between infants diagnosed prenatally and those diagnosed postnatally.

The majority of these events occurred after 12 hours of age.

The most common underlying cardiac lesion was aortic arch obstruction.

Cyanosis

? Anemia,
? dark skin
? Mild cyanosis

Non cardiac causes (pulmonary, hematologic, PHT, poor perfusion)
Pulse oxymetry screening

Sensitivity & specificity depend on
Criteria used for an abnormal test -  
- SaO2 <95: 0.75, 0.88
- SaO2 <90: 0.53, 1

Timing of screening ( ?1st hr, 1st day.....)

Probe site- postductal

Quality of the equipment

Signal quality and infant behavior — crying, moving-
not accurate

Health care workers expertise
Oxymetry

Postductal oxygen in 50,008 healthy Norwegian newborns upon admission to the nursery from the delivery unit:

The initial SaO2 was less than 95 percent in 3% of patients. Of these 1360 newborns, 324 cases were classified as pathologic because of symptoms of disease or persistent SaO2 <95 percent.

Of the 324 infants who failed the pulse oximetry screen, 43 had CHD, of whom 27 had critical lesions, and 134 had pulmonary or other disorders. The remaining 147 patients were healthy with transitional circulation.

CHD was diagnosed in 658 patients as follows:
- Prenatal diagnosis: 46 cases
- Pulse oximetry screening: 40 cases
- Routine exam in the nursery: 320 cases
- Post discharge: 74 cases
- Diagnosis while in the neonatal or intensive care unit: 178 cases

Eight newborn infants with critical CHD were not detected by oxymetry screening, four of whom were detected by routine examination.

Significant additional benefit of universal screening by pulse oxymetry to clinical assessment for the detection of CHD in infants admitted to the normal newborn nursery. The cost/benefit ratio of a universal pulse oxymetry screening program remains unknown.

This was illustrated in a 2006 report from the Tennessee Task Force on Screening Newborn Infants for Critical Congenital Heart Disease, which recommended not implementing a mandatory screening program based upon the available literature. Their primary concerns were high rates of false-positive results (specificity), as discussed previously, and unreliability of oxymeter testing for mass screening.

No Murmur

In a study reported in 1963 of 166 infants diagnosed with CHD by 12 months of age at Texas Children's Hospital, no murmur was heard in the first three months in 77 percent.

In a contemporary report, 48 percent of patients diagnosed with CHD before 12 months of age did not have a neonatal murmur.
Shock

A late sign, at whatever age-
HPLHS, Interr arch, critical AS
Severe Cyanosis

When associated with severe distress (gasping): Very late-
TGV, Pulm atresia
Failure to thrive

Shunts
TAPVD
Tissue Changes

(e.g Clubbing)
Indicates tissue hypoxia- late presentation
- Old Tetralogies & pulmonary atresias
- Pulmonary hypertension due to untreated shunts
Pulmonary Hypertension

Truncus arteriosus
Neglected shunts (VSD’s, PDA’s….)
Pulmonary atresias with huge collaterals
Systemic Hypertension

Coarctation (neglected or treated lately)
Strokes

Hemorrhagic
Embolic-Thrombotic
Infections

Infective Endocarditis
Brain abscesses
Arrhythmias

Atrial septal defects
Ebstein’s
Prolonged QT intervals
THANK YOU
Challenges in the management of late presenters
Psychosocioeconomic

In older ages, to treat or not to treat (will treatment improve quality of life)???
Cost of management much more
Outcomes are less glorious
Often longer course treatment with much disturbance of family life
Clinical

Operable or not?
Is there need for medical treatment first?
Is there time for medical treatment?
Can we treat the patient?
- Is the patient fit for surgery
- Can the patient tolerate surgery
- Will the patient be clinically better after surgery?
Patient with Neurologic impairment

Hemiplegia, Quadriplegia, brain atrophy, blindness
Truncus arteriosus presenting after 4 months of age
Univentricular Circulation with NO pulmonary stenosis

? How late can we do banding???
Old Tetralogies

Collaterals
Pulmonary hemorrhage
Intrapulmonary shunting
Large shunts

? How much pulmonary hypertension can we treat?
? Is there role for drugs for PHT before surgery?
? Is there role for partial closure?
? Is there role for banding?