Prevalence of congenital heart disease

Marelli et al Circulation 2007;115:163-172
Heart disease

- In children
  - Mostly congenital
    - Congenital heart disease is the most common single group of structural malformation in infants
    - 6-8 per 1000 live born infants have significant malformation
    - About 1 in 10 stillborn infants have a cardiac anomaly
    - About 10-15% have complex lesions with more than one cardiac anomaly
    - About 10-15% also have a non cardiac abnormality
The most common congenital heart lesions

- **Acyanotic**
  - VSD: 32%
  - PDA: 12%
  - PS: 8%
  - ASD: 6%
  - Coarctation of aorta: 6%
  - AOS: 5%

- **Cyanotic**
  - Fallot IV: 6%
  - TGA: 5%
Fig. 14.4 Changes in the circulation from the fetus to the newborn. When congenital heart lesions rely on blood flow through the duct – duct-dependent circulation – there will be a dramatic deterioration in their clinical condition when the duct closes.
Presentation

• Congenital heart disease may present with:
  – Antenatal cardiac ultrasound diagnosis
    • (18-20 weeks of gestation)
  – Detection of heart murmur
    • Diff: innocent murmur
  – Cyanosis
    • It can be recognised clinically if the concentration of reduced Hb in the blood exceeds 5g/dl (anaemia)!
      • (PH, IRDS, pneumonia, polycythaemia)
  – Heart failure
  – Shock
Diagnostic tools:

- **Chest X-ray:**
  - Baseline for assessing future changes

- **ECG**

- **Echocardiography:**
  - With doppler: to evaluate almost all cases of CHD

- **Cardiac catheterisation**
  - Now reserved for haemodynamic measurements and therapy

A normal chest X-ray and ECG do not exclude congenital heart disease
Echocardiography
Methods used as routine procedures:

• M mode in the 1960s (Edler, Hertz)
• 2D in the 1970s
• Doppler in the 1980s
• TOE in the 1990s
Echocardiography

Ultrasound

>20 000 Hz

- Currently in use: 2 - 7.5 MHz
- The piezoelectric crystal functions as both the transmitter and the receiver
- Real time ECHO can sample as many as 1000/second
Echocardiography

M-Mode recording

- **or time motion recording** is a graph of echo signal depths (down any single scan line) against time

- **M-Mode is used for:**
  - Imaging intracardial structures
  - Imaging the movement of intracardial entities (opening and closing of valves, movement of septum and the wall of the ventricles)
  - Assessing ventricular function
• Christian Johann Doppler
  – Austrian physicist, who noticed the change in the light of the stars as they move related to the Earth
  – In 1842 he described a ”change in frequency of sound, light or other waves caused by the motion of the source or the observer”
  – The degree of change is proportionate to the velocity of motion
Echocardiography

Doppler

- During the course of a Doppler examination, the received frequency shift signals are output both to the display screen and as an audio signal (as Doppler signals fall within the audible range of 400Hz-5kHz).

- Velocities above the zero line represent flow towards the transducer and below the line flow away from the transducer.
Echocardiography

Doppler

- The shift in frequency of the sound waves is dependent upon the velocity and the direction of flow which the transducer is directed at.
- Systemic and pulmonary flow volumes can be calculated from the blood velocities in the aorta and the pulmonary artery respectively.
Echocardiography

Doppler

• ECHO machines calculate pressure gradients through the valves or VSD from velocity, using the modified Bernoulli equation:

\[
\text{Gradient (Hgmm)} = 4V_{\text{max}}^2
\]
Echocardiography

Color flow mapping

• The color coded blood flow data is superimposed, in real time onto the 2D image so that a spatially oriented map of blood flow information is obtained.

• By convention, blood flow away from the transducer is colored blue, flow towards the transducer is colored red, with an increase in brightness for the higher velocities.
Echocardiography

Color flow mapping

- Turbulent flow is coded green
- Small or multiple shunt jets otherwise unrecognised are easily detectable, and the direction of flow may be determined
- Exact diagnosis of valvular regurgitation is possible
Echocardiography

Doppler tissue imaging

• Allows the Doppler imaging of the myocardium. Movements of the myocardium are color coded according to velocity.
Echocardiography

Transesophageal echocardiography

- A transducer is attached to a probe and driven down the esophagus behind the heart
- Gives clear image of the atrial septum, left atrium, aorta, mitral valve
Congenital cardiac defects

- Lesions with left-to-right shunt
- Lesions with obstruction
- Lesions with cyanosis
I. Lesions with left to right shunt

- PDA
- VSD
- ASD
- AVSD
- PAPVR
Patent ductus arteriosus (PDA)

Ductus arteriosus:
Connects the pulmonary artery to the descending aorta

PDA:
The failure of the DA to close shortly after birth
frequently occurs in preterm or sick neonates.
In other children it is due to a defect in the heart muscle

Flow:
The flow of the blood across the PDA is left to right
(from the aorta to the arteria pulmonalis, following the
fall in pulmonary vascular resistance after birth)
Clinical features

- In preterm infants:
  - collapsing pulse
  - Systolic murmur at the LSE
  when it is severe: the resulting heart failure may make it difficult to wean the infant from the artificial ventilation.

- Other children present a continuous murmur beneath the left clavicle
  - The pulse is bounding
  when the ductus is large: increased pulmonary blood flow with heart failure (pulmonary hypertension)
PDA III.

- **Investigations**
  - Chest X-ray:
    - Normal - enlarged heart. Enlarged pulmonary arteries, increased pulmonary vasculature
  - ECG:
    - Normal
    - LVH with large left-to-right shunt
    - RVH with pulmonary hypertension
  - Echocardiography:
    - Ductus can be identified
    - Doppler: continuous flow
PDA IV.

- **Management**
  - If preterm infant is symptomatic:
    - Fluid restriction
    - Indomethacin (prostaglandin synthetase inhibitor)
    - Surgical ligation
  - Young child is asymptomatic:
    - PDA closure recommended because of lifelong risk of bacterial endocarditis
    - Surgical ligation
    - Transvenous umbrella occlusion
Patent ductus arteriosus

ECG
- Usually normal
- Left ventricular hypertrophy with large left to right shunt
- Right ventricular hypertrophy with pulmonary hypertension

Fig. 14.15 Patent ductus arteriosus. (a) Murmur. (b) Chest X-ray. (c) ECG. (d) A patent ductus arteriosus visualised on echocardiography. (e) A small (12 or 17 mm diameter) double umbrella device which is passed through a catheter via the femoral vein. (f) The distal and proximal umbrellas are deployed in the aortic and pulmonary arterial end of the duct respectively.

PT = Pulmonary trunk, AO = Aorta.
VSD accounting for 32% of all cases of CHD

- Main types:
  - Perimembranosus
  - Muscular

- Clinical features:
  - Asymptomatic
  - Heart failure
  - Failure to thrive
  - Recurrent chest infections
  - Cyanosis (due to pulmonary vascular disease, now rare)
  - Endocarditis (late)
VSD II.

- **Physical signs**
  - Parasternal thrill
  - Heart murmur (LLSE)
    - Loud pansystolic murmur (small VSD)
    - Unimpressive ejection murmur (large VSD)
  - Variable pulmonary component of second heart sound
    - Normal when small defect
    - Loud, when large with PH
  - Tachypnoe, tachycardia, enlarged liver because of heart failure

- Tachypnoe, tachycardia, enlarged liver because of heart failure
VSD III.

- **Investigations**
  - **Chest X-ray:**
    - Normal cardiomegaly, enlarged pulmonary arteries and increased pulmonary vascular markings from pulmonary oedema
  - **ECG:**
    - Wide range, from normal to grossly abnormal
    - The most important abnormality: RVH
  - **Echocardiography:**
    - Precise anatomy of the defect
    - Doppler: can assess the haemodynamic effect
VSD IV.

• Management:
  – Drug therapy for heart failure
    • Diuretics: furosemide, thiazide, spironolactone
    • ACE inhibitor
    • Digoxin?
  – Surgical therapy:
    • Palliation: pulmonary banding
    • Correction: closure of the VSD/ Goretex
Indication for performing surgery within the first year of life:

- Severe symptoms with failure to thrive
- Pulmonary hypertension with possible progression in pulmonary vascular disease

In general: a child with a long, loud pansystolic murmur and a normal component in the second heart sound will not require surgery, even if symptomatic early in life.

Even if asymptomatic, surgery will be required for any child with an unimpressive murmur, but a loud pulmonary component in the second heart sound, implying a raised pulmonary arterial diastolic pressure.
II. Lesions with obstruction

- AOS
- PS
- Coarctation of the aorta
Coarctation of the aorta

- Clinical features
  - In neonatal period severe coarctation presents with ductus dependent circulation and circulatory collapse
  - If less severe, it may present with heart failure and murmur between the shoulder blades
  - In older children it may present with hypertension
  - The key: to the clinical diagnosis the recognition of the weak or absent femoral pulse
  - The BP in the arms will be higher than in the legs
COA II.

Palpation of the femoral pulse must be performed routinely during the cardiovascular examination of any child!
COA III.

- **Investigations**
  - Chest X-ray:
    - Usually normal
    - Rib notching from aortic to aortic collateral arteries in teenagers and adults
  - ECG:
    - In neonatal period: RVH
    - In older children: LVH
Coarctation of the aorta

(a) Coarctation of the aorta. There is narrowing of the aorta distal to the left subclavian artery adjacent to the insertion of the arterial duct.

(b) Murmur.

(c) Chest X-ray.

(d) ECG.

Ejection systolic murmur between shoulder blades or normal

Rib notching from aortic-to-aortic collateral arteries in teenagers and adults

Deep S wave in $V_2$ and tall R wave in $V_6$ (>45mm total) and upright T wave indicates left ventricular hypertrophy

Downgoing T wave suggests left ventricular strain and severe coarctation and/or hypertension.
COA IV.

• Management:
  – Surgery:
    • The subclavian flap procedure is most widely used though left arm pulse is lost, the arm develops normally
  – Balloon dilatation:
    • Is performed in some centres, but the exact role remains uncertain
    • If restenosis occurs after operation, balloon dilatation is the treatment of choice
III. Lesions with cyanosis

- TGA
- Fallot IV
- TAPVR
- Pulmonary atresia with intact ventricular septum
- Pulmonary atresia with VSD
- Truncus arteriosus communis
- Ebstein’s anomaly
- DORV
- Tricuspid atresia
- Single ventricle
Tetralogy of Fallot

• Clinical features:
  – Large inlet ventricular septal defect
  – Overriding aorta with respect to ventricular septum
  – Right ventricular outflow tract obstruction
  – Right ventricular hypertrophy
Tetralogy of Fallot II.

• Symptoms:
  – Severe cyanosis may be present in the first few days of life with a ductus dependent pulmonary circulation but:
    most are diagnosed in the first few months of life following the identification of the
  – murmur
    (nowdays the classical combination of severe cyanosis, hypercyanotic spells and squatting on exercise developing in late infancy is rare.)
Tetralogy of Fallot III.

- **Hypercyanotic spells**: are characterised by a rapid increase in cyanosis usually associated with irritability or inconsolable crying because of severe hypoxia and breathlessness and pallor because of tissue acidosis.

  - **Signs**:
    - Loud ejection murmur in the third left intercostal space with a single second heart sound.
    - Murmur will shorten and cyanosis will increase as the right ventricule obstruction increases.
    - During hypercyanotic spells the murmur will be very short or inaudible as the pressure in the two ventricles become similar.
    - **Clubbing** if the fingers and toes may develop.
Tetralogy of Fallot

Fig. 14.21  (a) Tetralogy of Fallot. The right ventricular outflow tract obstruction results in blood flowing from right to left across the ventricular septal defect. (b) Murmur. (c) Chest X-ray. (d) ECG.

Right ventricular hypertrophy
Upright T wave in $V_1$ with 'pure' R wave (no S wave)
TGA

- There are two parallel circulations
- It is incompatible with life if there is no mixing between the two circulations
- Naturally occurring associated anomalies: VSD, ASD, PDA
- Therapeutic intervention: Rashkind septostomy
TGA II.

- **Symptoms:**
  - Cyanosis
  - The children present when the DA is closing

- **Physical signs:**
  - Cyanosis and clubbing (the other symptoms vary depending on the associated malformations)
  - The second heart sound is usually single
  - Systolic murmur due to increased flow or stenosis within the left (pulmonary) outflow tract
TGA III.

• **Investigations:**
  – Chest X-ray:
    • Narrow upper mediastinum with “egg on side” appearance of cardiac shadow
    • Increased pulmonary vascular marks are common
  – ECG:
    • Is rarely helpful in establishing diagnosis
  – Echocardiography:
    • Essential in demonstration of abnormal arterial connection
Transposition of the great arteries

(a) Complete transposition of the great arteries

(b) Coincident $A_2P_2$ single

Variable systolic murmur

(c) Narrow pedicle

'Egg on side'
cardiac contour
Increased pulmonary vascular markings

(d) ECG

Usually normal neonatal pattern
TGA IV.

- **Management:**
  - Maintaining the patency of DA is essential (prostaglandin E inf.)
  - Ballon atrioseptostomy is life-saving procedure
  - All patients with TGA will require surgery
    - Mustard, Senning operation
    - Arterial switch (Jatene) operation
Hypoplastic left heart syndrome

Definition:

• A rare, heterogenous and severe deformation of the left heart.

• It can mean the hypoplasia, stenosis or atresy of the...
  
  • aorta
  • aortic valve
  • musculature of the left heart
  • mitral valve
  • left atrium

• **Incidence:** 0,2 % (USA: 2000 per annum)
Causes:

- SECONDARY: the blood flow to the developing embryonic left ventricle is restricted → abnormal left ventricle development?
- there is strong evidence pointing to GENETIC etiology → BUT: the genetic mechanism is yet unknown
- P. Grossfeld; Circulation Research; 2007, 100: the FIRST research to show the hereditary nature and the probable genetic background of the HLHS
The genealogy of 38 study groups:

- in 55% of the families there was at least one member with HLHS or other heart disease
- 193 relatives → 21.4% suffering from heart disease (age: 3-74 years)
- many chromosome abnormalities concomitant with HLHS (eg. 11% occurrence of terminal 11q deletion– Jacobsen syndrome)
- at least one genetic mutation present in HLHS patients: one transcription factor, the NKX2.5 has been identified
- the role of genes TBX 5 and IRX is being studied
- assumption: HLHS is caused by the defectiveness of muscular tissue
Treatment

Initiation of the family!

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Changes in left heart hemodynamics after technically successful in-utero aortic valvuloplasty


*Department of Cardiology, Children’s Hospital Boston and Departments of †Radiology and ‡Obstetrics and Gynecology, Brigham and Women’s Hospital, Harvard Medical School, Boston, MA, USA

stabilization for surgical intervention

• multiple-step, palliative surgical interventions...
1. **Norwood procedure:**
   - the right heart is the maintainer of the systemic circulation
   - Blalock-Taussig shunt / Sano shunt
   - age: neonate

2. **Bidirectional Glenn procedure:**
   - SVC → art. pulm.
   - closure of BT/Sano
   - age: 3-6 months
3. Fontan procedure:

- age: 18 months – 4 years

4. Hybrid approach:

- maintenance of the stent ductus
- band on the pulm. branches

(Gutgesell HP; Curr. Opin. Cardiol.; 2007, 22)
Prognosis:

• chronic health problems

• no survivors till the 1980’s, the first survivors are in their early 20’s, long-term prognosis is unknown

• endocarditis

• close controll is necessary