ECHOCARDIOGRAPHIC APPROACH TO CONGENITAL HEART DISEASE: THE UNOPERATED ADULT

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NO DISCLOSURES
CONGENITAL HEART DISEASE

DEFINITION

Heart disease present since birth

Atrial Septal Defect
Ventricular Septal Defect
Atrioventricular Septal Defect
Congenital valvular stenosis
Congenital valvular regurgitation
Coarctation of the aorta
Interrupted aortic arch
Tetralogy of Fallot
Transposition of the great arteries
Truncus arteriosus
Valve atresia
Single ventricle
Anomalous pulmonary venous return
Etc.
INCIDENCE AND SURVIVAL OF CHD

0.8% of live births

- Less Severe Disease
- Survival to 18
- Severe Disease
- Survival to 18
- Total Adults/Yr

Before 1940: <10%

After 1940: 90%
ROLE OF IMAGING

Evaluate anatomy
- Chambers and valves
- Extracardiac vasculature
- Coronary anatomy
Evaluate ventricular function
Evaluate valvular function
Evaluate shunts

ESTABLISH DIAGNOSIS

&

ESTABLISH IMPACT OF ABNORMALITY
TYPICAL CASE

32 year old woman with palpitations and progressive exercise intolerance over the last 6 months

Single S2 and 2/6 systolic murmur at LSB

ECG with incomplete RBBB and right axis
atrial septal defect
ATRIAL SEPTAL DEFECT

anatomy

Secundum - 75%

Primum - 15-20%

Sinus venosus - 5-10%

Webb and Gatzoulis, *Circulation* 2006
ATRIAL SEPTAL DEFECT anatomy

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ATRIAL SEPTAL DEFECT

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ATRIAL SEPTAL DEFECT

HEMODYNAMIC CONSEQUENCES

RA and RV volume overload
Excess volume may result in pulmonary hypertension may develop
More often in women
Generally not severe
May result in paradoxical emboli

Brickner et al  *NEJM* 2000
ECHOCARDIOGRAM

SECUNDUM ASD
ECHOCARDIOGRAM

SECUNDUM ASD
ECHOCARDIOGRAM

SINUS VENOSUS DEFECT
TYPICAL CASE

25 year old woman referred for evaluation of a murmur

Had a hole in her heart as a child, told it closed

Wants to get pregnant

Harsh 5/6 systolic murmur at left sternal border
VENTRICULAR SEPTAL DEFECT
VENTRICULAR SEPTUM

ANATOMY

Inlet (Atrioventricular Canal) From endocardial cushions
Trabecular (Muscular) Septum Invagination of muscle
Outlet (Infundibular) Septum From conotruncal septum
Membranous Septum Small fibrous

Zipes, Braunwald’s Heart Disease, 2005
VENTRICULAR SEPTUM

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- Small, fibrous

Zipes, Braunwald's Heart Disease, 2005
VENTRICULAR SEPTAL DEFECT

PHYSIOLOGY

Systolic volume load

Volume load on pulmonary vasculature and left heart
LA and LV enlargement result

Effect dependent on size of defect and amount of shunting
Amount and direction of shunting dependent on PVR and SVR

Brickner et al  *NEJM* 2000
ECHOCARDIOGRAM

RESTRICTIVE VSD WITH WINDSOCK CLOSURE
ECHOCARDIOGRAM

RESTRICTIVE VSD WITH WINDSOCK CLOSURE
ECHOCARDIOGRAM

MUSCULAR VSD
ECHOCARDIOGRAM

VSD MIMIC?
TYPICAL CASE

40 year old man with Down syndrome

O2 sats 84% on room air

Cyanotic, clubbed

Loud P2, soft murmur at LSB
ATRIOVENTRICULAR SEPTAL DEFECT
ATRIOVENTRICULAR SEPTAL DEFECT

ANATOMY

May be partial or complete
Complete AVSD associated with Down syndrome
May be associated with other anomalies
ATRIOVENTRICULAR SEPTAL DEFECT

COMPONENTS

Primum septal defect

Atrioventricular septal defect

Clefts in AV valves

Left sided AV valve
  “anterior leaflet of mitral valve”

Right sided AV valve
  “septal leaflet of tricuspid valve”
UNREPAIRED COMPLETE AVCD

UNREPAIRED AV SEPTAL DEFECT
ECHOCARDIOGRAM

UNREPAIRED AV SEPTAL DEFECT
ATRIOVENTRICULAR SEPTAL DEFECT
LEFT VENTRICULAR OUTFLOW TRACT

“GOOSENECK” DEFORMITY
ELONGATED, ANTERIOR LVOT
MAY BECOME OBSTRUCTIVE
ECHOCARDIOGRAM

ELONGATED LVOT
COMMON ATRIOVENTRICULAR VALVE

Bridging leaflets

AV valves at same level on imaging

Variable classifications
TYPICAL CASE

30 year old immigrant referred for murmur

Asymptomatic

Continuous murmur under left clavicle
PATENT DUCTUS ARTERIOSUS
PATENT DUCTUS ARTERIOSUS
TYPICAL CASE

23 year old referred for hypertension

BP in right arm 170/70, right leg 100/70

Continuous murmur in back near left scapula
COARCTATION OF THE AORTA
COARCATION OF THE AORTA

ANATOMY

Generally occurs at ductus arteriosus
5-8% of congenital heart disease
Bicuspid aortic valve in 50-80% of patients
Collaterals may provide flow to organs distal to coarctation
ECHOCARDIOGRAM

COARCTATION OF THE AORTA
ECHOCARDIOGRAM

COARCTATION OF THE AORTA
ABDOMINAL PULSE DOPPLER

COARCTATION OF THE AORTA

DIASTOLIC RUNOFF
TYPICAL CASE

50 year old referred for exercise intolerance and pulmonary hypertension

Exam with single S2, 2/6 holosystolic murmur at LSB
CONGENITALLY CORRECTED TRANSPOSITION OF THE GREAT ARTERIES
CONGENITALLY CORRECTED TRANSPOSITION OF THE GREAT ARTERIES

Other terms
L-transposition
“double discordance”
atrioventricular and ventriculoarterial discordance

Other lesions
VSD
pulmonic stenosis
Ebstein abnormality
“inverted coronaries”
ECHOCARDIOGRAM

SIDE BY SIDE SEMILUNAR VALVES
ECHOCARDIOGRAM

APICAL FOUR CHAMBER VIEW
ECHOCARDIOGRAM

SYSTEMIC AV VALVE REGURGITATION
CONCLUSIONS

CHD presenting in adulthood is uncommon

Lesions may be simple or complex, all likely have significant consequences

Echocardiography is mainstay of diagnosis

Referral to ACHD centers is most often needed