Adult Congenital Heart Disease Workshop

Doreen DeFaria Yeh, MD
Cardiology Division/Echocardiography section
Adult Congenital Heart Disease Program
Massachusetts General Hospital, Boston, MA

American Society of Echocardiography, EchoFlorida
October 15, 2013

• No disclosures

Natural History of unrepaired CHD

J. Hoffman, 1965
Growing Adult CHD population

- 1.2M Adults in the US with Congenital Heart Disease
  - Pediatric patients
  - Adult patients

1965:
- 75% Pediatric
- 25% Adult

1985:
- 50% Pediatric
- 50% Adult

2005:
- 40% Pediatric
- 60% Adult

ACHD Growth Implications

Hospital admissions for ACHD in the U.S. more than doubled between 1998 and 2005.

Total national hospital charges increased from $696 million in 1998 to $3.16 billion in 2005.

The Trend Continues….

O’Leary, JAMA 2013
ACHD: Challenges

• Complex CHD is not “cured”

• Patients require long term monitoring for sequelae of early surgeries

• Are we prepared for this growing population?
  – Do our fellows feel sufficiently trained?
    • 74% of fellowships < 6 lectures/year
  – Are we prepared for imaging needs?

National Average Scores by Exam Category: 2012 Exam

Bethesda Group I: Simple lesions

These patients can usually be cared for in the general medical community.

Unrepaired conditions:
- isolated small atrial septal defect (ASD)
- isolated small ventricular septal defect (VSD)
- mild pulmonary stenosis
- isolated ductus arteriosus—no other heart problems

Repaired conditions:
- Patent ductus arteriosus (PDA)
- Secundum atrial septal defect (ASD)

Repaired or unrepaired conditions:
- isolated atrio-ventricular valve disease
- isolated patent foramen ovale (PFO)
- isolated aortic valve disease
- isolated mitral valve disease

Group I—Simple Congenital Heart Disease
Bethesda Group II: Moderate Complexity

Overview
- Complex congenital disease: Complex to scan
- Sequential segmental analysis
  - Determination of cardiac location and situs
  - Segments and connections
Sequential segmental analysis

1.) gather data: what do we already know? OPERATIVE REPORTS! Prior imaging

2.) determination of cardiac location

3.) abdominal and cardiac situs

4.) Segments and connections

5.) evaluate coronary origins

Location/Orientation

Cardiac Location
- Levoposition
- Dextroposition
- Ambiguous

Cardiac Orientation
- Levorotation
- Dextrorotation
- Ambiguous

Eidem, Cetta, O’Leary, 2010
Visceral and Cardiac Situs ( sidedness)

Abdominal Situs

Atrial Situs/ventricular looping

Abdominal situs:

- Position of the liver, stomach, spleen and abdominal great vessels, aorta and IVC

Cardiac/Atrial situs

- Atrial situs:
  - Atrial situs solitus
  - Atrial situs inversus
  - Atrial situs ambiguous: rare
Segments and Connections:

- Great veins
- Atrial segments
- Ventricular segments
- Great Arteries

Venous Segment

- Systemic veins:
  - IVC/SVC
  - Coronary sinus
  - Hepatic veins
  - * LSVC

Venous Segment

- Pulmonary veins
  - Anomalous drainage
    - Associated ASD
Atrial Segments

- Thicker muscular limbus is to the right
- RA is more muscular
- RA appendage usually more broad

Eidem, Cetta, O’Leary, 2010

Atrioventricular Connection

- L transposition of the great vessel (L TGA, CC TGA)
  - RA to LV to PA
  - LA to RV to Aorta
- Valvular abnormalities

Eidem, Cetta, O’Leary, 2010
Ventricular Segment

- Morphologic RV
  - Apical septal insertion of AV valve
  - Course apical trabeculations
  - Moderator band
  - Multiple small papillary muscles
  - RVOT always most anterior ventricular structure
  - Muscular infundibulum
- Morphologic LV
  - Smooth endocardial surface
  - Large discrete papillary muscle
  - Fibrous continuity between the AV valve and semilunar valve

- Univentricular heart
  - Determine type of rudimentary ventricle

Ventricular – Great Arterial Connection

- Concordant:
  - Morphologic RV to PA
  - Morphologic LV to Aorta
- Discordant: (L TGA)
  - Morphologic RV to Aorta
  - Morphologic LV to PA
- No anatomic features that distinguish semilunar valves
- Rule of 50%

Transposition of the Great Vessels (D-TGA)

Brickner, NEJM 2000
Great Arterial Segment

- PA
  - Supravalvular stenosis
  - Branch pulmonic stenosis
  - PA hypoplasia
  - Absent L PA
- Aortic
  - Supravalvular stenosis (Williams)
  - Aortic coarctation
  - Associated ascending aneurysm

Complex Congenital Step by Step Summary

1.) Gather data about the patient—operative report!
2.) Determine location
3.) Visceral and cardiac situs
4.) Sequential approach to segments and connections
   - Great Veins, atrial, AV connection, ventricular, VA connection, great vessels
5.) Ask for help when needed

Case review
72 year old male with new atrial fibrillation

- History of hypothyroidism and hypertension
- Exertional fatigue
- Multiple normal TSHs
- Two prior normal echocardiograms

Cardiology visit:
- Low atrial rhythm, iRBBB
- Wide splitting of S2
- Review of echo imaging:
  - RV enlargement

Right Ventricular Quantification:
JASE 2010

GUIDELINES AND STANDARDS

Guidelines for the Echocardiographic Assessment of the Right Heart in Adults: A Report from the American Society of Echocardiography
Endorsed by the European Association of Echocardiography, a registered branch of the European Society of Cardiology, and the Canadian Society of Echocardiography

Lawrence D. Bandt, MD, FAAC; Chao, Wenyue M. Jin, MD, FACC; and the Joint Guidelines Committee for Right Heart Assessment in Adults

© 2010 American Society of Echocardiography
RV linear dimensions

RVD1 < 42mm
RVD 2 <35mm
RVD 3 < 86mm

Quantification of RV function by echo

• Newer echo modalities better approximate MR RVEF
  – Tricuspid annular plane systolic excursion (TAPSE)
  – Fractional area change (FAC)
  – RV Tissue Doppler
  • E', S'
  • Myocardial Performance Index or Tei index
  – 3D volumetrics
  – Strain, strain rate
  – RV diastolic parameters

Agitated saline study
RV dilation must be explained…

Atrial septal defects in adults

- **Ostium Secundum:**
  - MVP (10-20%)
  - ECG: RBB morphology, RAD

- **Ostium Primum:**
  - MR, cleft MV leaflet, VSD
  - ECG: RBB morphology, LAD
  - 1st degree AVB (75%)

- **Sinus Venosus:** (rare)
  - ECG: Junctional or low atrial rhythm
  - Anomalous pulmonary venous drainage into RA or vena cavae
Complications related to ASDs in adults

- Paradoxical embolization
- Atrial arrhythmias
- Right heart enlargement; Exertional fatigue
- Pulmonary hypertension

ACHD Guidelines

CLASS I
- RA or RV enlargement
  - With or without symptoms
- Percutaneous closure for secundum defects
- Surgical closure: sinus venosus, coronary sinus, or primum ASD

CLASS IIa
- Surgical closure of secundum ASD is reasonable:
  - If concomitant surgical correction of tricuspid valve is needed
  - when the anatomy of the defect precludes the use of a percutaneous device. (C)
- paradoxical embolism (C)
- orthodeoxia-platypnea (B)

Sinus Venosus Defects
Our patient: Follow up

- TEE consistent with sinus venous defect with RUPV draining to the SVC/RA junction
- Underwent surgical correction
- Significant improvement in exercise capacity
- Continues to have intermittent atrial fibrillation

Take Home Points:

- RV enlargement must be explained (consider TEE)
- Congenital Heart Defects may first present later in life
- Surgical correction and improvement in RV size/function will improve symptoms of exercise tolerance

Case II

46M h/o restrictive VSD new diastolic murmur
Ventricular Septal Defect

- **Inlet**: AV septal defect, may be associated with ASD
- **Supracristal/Outlet**: can lead to Ao RCC prolapse
- **Membranous**: Commonly closes spontaneously
- **Muscular**: May be multiple

Associated lesions:
- Pulmonic stenosis, BAV, coarctation, subaortic membranes

Complications of VSD:

- Left atrial or left ventricular enlargement
- Atrial arrhythmias
- Endocarditis
- Aortic insufficiency
- Sinus of Valsalva aneurysm→fistula (continuous murmur)
- Pulmonary hypertension/ Eisenmenger physiology
VSD: echocardiographic findings

ACHD Guidelines: VSD

CLASS I
- Catheterization to assess operability of adults with VSD and PAH
- Closure for Qp/Qs of > 2.0 and clinical evidence of LV volume overload (B)
- History of endocarditis

CLASS IIa
- Closure is reasonable:
  - Net L>R shunt with Qp/Qs > 1.5 and PASP < 2/3 systemic, PVR < 2/3 SVR (B)
  - Net L>R shunt with Qp/Qs > 1.5 in the presence of LV systolic or diastolic failure (B)

CLASS IIb
- Pulmonary vasodilators for VSDs with progressive/severe pulmonary vascular disease (B)

CLASS III
- VSD closure is not recommended in patients with severe irreversible PAH (B)

38F history of PDA closure in infancy, subaortic membrane resection age 19, bicuspid aortic valve, presented with exertional headaches and head bobbing
38F history of PDA closure, subaortic membrane resection, BAV presented with exertional headaches and head bobbing

- Bicuspid systolic ejection click
- Bruit L scapula
- Bilateral bounding carotid pulsation
- Weak and delayed peripheral pulses diffusely, BP difficult to auscultate
- No radial-femoral delay

38F history of PDA closure in infancy, subaortic membrane resection age 19, presented with weak peripheral pulses and prominent carotid pulsation
Aortic Coarctation

- 6-8% of all congenital heart disease
- 4/10,000 live births
- Male: Female 2:1
- Diffuse arteriopathy
- **Hypertension**
- Berry aneurysm screening

**Associated abnormalities**
- Bicuspid aortic valve (50-60%)
- Mitral valve abnormalities
- Subaortic membrane
- VSD, PDA
- Aortic arch anomalies/ aberrant subclavians

Guidelines for intervention

**CLASS I:**
- Peak to peak gradient > 20 mmHg
- Gradient <20mmHg with evidence of collaterals
- Persistent hypertension

Role for Functional testing? Exercise ABIs?
Percutaneous Interventional approaches

- Coarctation Stenting:

Images Courtesy Ignacio Inglessis, MD

Coarctation of the Aorta
Extra-anatomic Bypass

9 years post s/p ascending to descending aortic graft progressive fatigue and exertional dyspnea

Mean gradient 56mmHg
PASP 60mmHg
Case: 50 year old male history of tetralogy of Fallot, s/p repair age 7, CAD s/p stenting, Aflutter ablation new patient evaluation
RV measures 5.2cm at the base:
MRI with RVEDVI 206ml/m2
Tetralogy of Fallot

- Rightward deviation of the aortic valve with overriding of the ventricular septum
- Ventricular septal defect
- Subpulmonary infundibular stenosis
- Right Ventricular Hypertrophy

**anterior cephalad deviation of the outflow septum**

Bricker NEJM 2000
Associated abnormalities:

- Varying degrees of RVOT obstruction/PA hypoplasia
  - Mild obstruction: pink tetralogy
  - Most severe form: pulmonary atresia, cyanosis
- ASD (pentalogy)
- Right sided aortic arch (25%)
- Absent left pulmonary artery
- Coronary anomaly:
  - LAD from RCA, courses anteriorly across RVOT
  - Anomalous circumflex

Adult with Tetralogy of Fallot

- Unrepaired (rare)
  - Degree of RVOT obstruction will determine pulmonary hypertension
- Palliative Repair
  - Systemic arterial to pulmonary arterial shunt
- Definitive Repair
  - VSD patch
  - Subinfundibular resection
  - Transannular Patch or RV to PA conduit

Palliative Systemic-Pulmonary Shunts

- (BT Shunt) Alfred Blalock - Helen Taussig
  - 1944: left subclavian artery to the left pulmonary artery
- Willis Potts
  - 1946: descending thoracic aorta and LPA
- David Waterston
  - 1962: ascending aorta to the RPA
Definitive Surgical Correction

• Complete Repair
  – 1954 C. Walton Lillehei
  – 1955 John Kirklin (Mayo Clinic)

• Takedown of prior palliative shunt
• Open RVOT/PA
• VSD closure with patch
• Resection of subpulmonic obstruction
• Transannular patch developed in 1959
• Conduit between RV to PA in 1965 for pulmonary atresia

**REPAIRED: NOT CURED**
Sequelae post TOF repair

• Residual lesions (VSD, sub-PS, branch PA stenosis)
• **Pulmonary regurgitation**
• Progressive RV dilation and dysfunction: RHF
• Exercise intolerance/fatigue
• Ventricular arrhythmia and sudden death
• Secondary LV dysfunction
• Aortic dilation, insufficiency (dissection is quite rare)

Pulmonic Regurgitation:

• Often underappreciated
• May be progressive over decades
• Volume load on the RV
• Color Doppler
  – Widejet occupying RVOT
  – Reversal of flow in the main PA
• CW Doppler:
  – Rapid deceleration time
Timing of pulmonic valve replacement

- Class I recommendation:
  - Pulmonic valve replacement is indicated for severe PR with any of the following:
    - 1.) symptoms of decreased exercise tolerance
    - 2.) moderate to severe RV dysfunction or enlargement
    - 3.) sustained atrial or ventricular arrhythmias
    - 4.) moderate to severe TR

Warnes et al. Circ 2008

Case: GT

- 52 M h/o TOF s/p right classic BT shunt age 4, complete repair age 9, St. Jude PVR 2007, PV endocarditis 2008, ICD for NSVT, cirrhosis due to long standing right heart failure, GI bleeding...

- Progressive decline in exercise capacity with chronic right heart failure...

RV dysfunction
Repaired TOF: When to plan PVR?

MRI RV volumes may help predict outcomes?

- RVEDVI < 120 ml/m² (all normalized)
- RVEDVI ≥ 160 ml/m²
  - Sensitivity 55%
  - Specificity 92%
- RVEDVI > 190 ml/m² (none normalized)

Sudden Cardiac Death

- Risk factors:
  - Older age at time of correction (>12 years)
  - Severe PR
  - RV dilation
  - RV dysfunction
  - RV mass/volume ratio?
  - NSVT, inducible on EPS
  - history of syncope or sudden cardiac death
  - QRS >180ms
- When to consider ICD??

Case: GT

50 M h/o TOF s/p right BT shunt age 4, complete repair age 9, St. Jude PVR 2007, PV endocarditis 2008, ICD for NSVT, cirrhosis due to long standing right heart failure, GI bleeding...

Progressive decline in exercise capacity with chronic right heart failure...

- LHC: anomalous left main
- RHC: could not cross his mechanical PVR
  - Invasive RVSP (60mmHg), LVEDP (20mmHg)
  - Echo trans RVOT gradients (mean 20)
  - Extrapolate PASP pressures 40mmHg
  - Cardiac output measured by echo and Fick (RV) 4.2L/min
  - → PVR < 2Wu
Don’t wait too long….

- PVR done at RVEDV of 195ml/m2
- Severe residual RV dysfunction → secondary LV dysfunction and cirrhosis
- Recently underwent Heart/Lung transplant

Transcatheter Pulmonary Valve

Summary: tetralogy of Fallot

- Think about associated anomalies (R aortic arch, coronary anomalies, ASD)
- Watch carefully for late sequela ("PR", RV dysfunction, VT)
- PVR timing: Don’t wait too long….
  - symptoms, exercise capacity
  - RV function/size
- SCD risk assessment, ICDs when indicated
- RV/PA conduits or failed bioprosthetic PVR: percutaneous pulmonary placement may be considered
Transposition of the Great Arteries:

37 M h/o D-TGA s/p Mustard atrial switch in infancy, presents with cirrhosis, marked ascites
→ Transferred to MGH for heart and liver transplant evaluation

D-Transposition of the Great Arteries

• Normal ventricular situs
• Great arteries:
  – Failure of spiral septation of the truncus arteriosus
• Atrioventricular concordance
• Ventriculoarterial discordance
• 2 separate parallel circulations

Need for a shunt: atrial septostomy

Creation of an Atrial Septal Defect Without Thoracotomy

1965
Atrial Switch Procedure

- 1958: Ake Senning
  - atrial tissue
- 1964: William Mustard
  - Excised atrial septum
  - Synthetic material

Bricker NEJM 2000

Atrial Switch: The Systemic Right Ventricle

- D TGA: The systemic RV is dilated and severely hypertrophied.
- Atrial switch pulmonary venous pathway: LLPV drains into RA through the baffle.

Issues after Atrial Switch

- Arrhythmia
  - Only 40% NSR @ 20 years post op, 11 % need pacers
  - Sudden death risk
- Systemic RV failure
  - RV systolic dysfunction — ? ACE/ARB inhibitors
    - Progressive TR (systemic AV valve)
- Venous baffle obstruction
  - Mustard: SVC>IVC (SVC syndrome, hepatic congestion → ascites)
  - Senning: Pulm vein—Systemic Veins
- Baffle leaks (25%)
  - Risk of paradoxical embolism
37M D-TGA s/p Mustard admitted with ascites, no orthopnea

- Lost to follow up for years
- Superior> inferior limb baffle obstruction
- Cirrhosis
- Post baffle superior and inferior limb stenting
  - Autodiuresis
  - Improved ascites
- Regression of cirrhosis

Ebstein Anomaly

- Failure of delamination of the tricuspid valve
- Associated: ASD/PFO, WPW
- Tricuspid regurg
- Right heart failure
- Cyanosis

Case I: 57F feels well quite active, Ebstein diagnosed after abnl ECG. No medications
Case II:

- 78 year old female with a murmur as a child
  - Noted to have a murmur at age 8
  - Catheterization in 1956 nondiagnostic, presumed VSD
  - 3 successful pregnancies in the 1960s
  - Repeat catheterization in 1965: right heart angiogram
    - Intermittent atrial arrhythmias
      - managed with digoxin, no prior ablation
    - Sat 95% on RA, normal HCT, euvoicmic
  - Awaiting ovarian surgery:
    - ? Pre-operative recommendations
Case III: chest pain

- 62F history of hypertension, prior breast cancer
- Murmur all her life
- Generally very well, daily exercise, lives in FL
- Recent 6 months: exertional chest heaviness, relieved with rest
- No dyspnea or cough, no dysphagia
TTE Summary

- RV enlargement with preserved RV systolic function
- Congenitally abnormal and thickened pulmonic valve with stenosis and regurgitation
- Massive PA aneurysm (approximately 8cm), dilation of the proximal R and L PAs
- No evidence of ASD, VSD or PDA
- Normal LV size and systolic function
- Trileaflet AV, mild ascending aortic dilation with mild AR
- PASP estimated 30mmHg
Pulmonic Stenosis

- Valvular, Subvalvular, Supravalvular
- JVP: Prominent A waves
- Systolic ejection click
- Mild PS generally does not progress
- Severe valvular PS usually amenable to balloon valvuloplasty
- RVSP/ps PASP
Syndrome of Absent Pulmonary Valve (APV)

- Pulmonary valve: Dysmorphic, hypoplastic, rudimentary
- Free pulmonic insufficiency
- Associated arteriopathy: Main PA and branch dilation
- Symptoms: bronchial compression by enlarged PA
- Isolated anomaly or associated with genetic syndrome
- Chromosome 22q11.2 deletion

Eisenmenger physiology

32 M h/o double outlet RV, large VSD, progressive cyanosis, HCT 78 and exercise intolerance
→ Referral to MGH for heart/lung transplant
Eisenmenger is a multisystem disease

- advanced pulmonary hypertension, R>L shunting
  - ASD, VSD, AVSD, PDA, unrepaired truncus arteriosus
- Unique to Eisenmenger:
  - Hypoxemia related erythrocytosis: Hyperviscosity clotting risk (stroke, renal infarct)
  - Paradoxical embolism
  - Cyanotic hemostatic abnormalities: bleeding risk
  - Hemoptysis (bronchial artery rupture, pulmonary infarct)
  - Renal impairment
  - Hyperuricemia → gout
  - Marked RVH, chronic RV ischemia, NSVT
  - Linus coronary artery compression from enlarged, hypertensive PA
  - Endocarditis, brain abscess

Treatment options for Eisenmenger

- Oxygen ?, warfarin ?, digoxin +
- Advanced PAH therapy
  - Dual endothelin-receptor antagonist: bosentan
    - Improve exercise capacity, hemodynamics (BREATHE-5)
  - Single endothelin-receptor antagonist: sitaxsentan
  - Improvement in hemodynamics
  - Phosphodiesterase type V inhibitor: sildenafil
    - Improve functional class, oxygen saturation, hemodynamics
  - Combination therapy
- Transplant
  - Increased peri-operative mortality
Gatzoulis Circ 2010: mortality and Eisenmenger

- 229 patients
  - Mean age 34, 35% M
  - 53% NYHA III or IV
  - Mean resting O2 sat 84%
- 64 pts advanced therapies (AT)
  - 74% bosentan
  - 25% sildenafil
  - 1% epoprostenol
- Mean follow up 4 years
  - 52 died, only 2 in AT group

So many ways to go wrong with Eisenmenger...

- Phlebotomy...
  - Only indicated if very high HCT (>70) and symptoms and no evidence of dehydration (with isovolemic replacement)
  - Otherwise keep iron replete, iron deficiency common
- Bringing them into the hospital...
  - Minor procedures/anesthesia are life-threatening
  - Diuretics, may worsen hyperviscosity
  - IV lines, paradoxical embolism, line infection, intracardial pacing
- Intubation
- Missed thromboembolic prevention
- Pregnancy
- Hot tubs/strenuous exercise/high altitude

Summary

- ACHD: growing and aging population
- Repaired, not “cured”
- Obtain prior operative reports, understand anatomy
- Sequential segmental analysis
Thank You

Doreen DeFaria Yeh, M.D.
Phone 617-643-7024, 617-726-8510
ddefariayeh@partners.org