Common Pediatric and Congenital Cardiac Abnormalities – Selected Cases

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Goals
- Review select interesting and overlooked cardiac abnormalities seen in infants
- Outline the thought process involved in managing children with these abnormalities
- Use echo images and other pictures and sounds to keep everyone awake

Case 1
- 1 month old asymptomatic male with murmur, no cyanosis, no respiratory distress, no feeding difficulty, normal growth
- PMHx, Fam Hx, Soc Hx, ROS all negative
- Vitals: HR 140, RR 33, BP 80/46 RUE, 85/48 RLE, O2 sats 100%
- PE significant for palpable thrill, nl S1, split S2, with 4/6 harsh systolic ejection murmur at LUSB, ejection click, strong pulses 4 extremities, no HSM
Pulmonary Valve Stenosis
- Initially described in 1761 by John Baptist Morgagni – thought to be rare
- Now known to be somewhat common, in 8-10% of patients with CHD
- Familial occurrence – 2%
- Cause – Failed cellular migration, cell signaling pathway errors

Pulmonary Stenosis
- Caused by fused or absent commissures. Leaflet fusion with thickening and valve dysplasia, usually with supravalvar attachments called raphe.
- These valvar abnormalities cause poor leaflet excursion, variable stenosis, and can lead to pulmonary insufficiency
- Myxomatous thick dysplastic leaflets seen in the Noonan’s subtype
Normal, trivial, mild, moderate, severe, critical
Moderate, severe, critical require intervention.
Severe is suprasystemic RV pressure
Moderate is 1/2 systemic to systemic RV pressure
Trivial and mild are on the radar with valve abnormalities noted, but simply need to be intermittently followed to monitor for progression
Post-stenotic dilatation of MPA and LPA is common and well tolerated in PS.

RV in Pulmonary Stenosis
- Diffuse RV hypertrophy (infundibulum) - dynamic subvalvar obstruction
- Thickening and dysplasia of tricuspid valve, with regurgitation
- RA dilation / thickening
- PFO vs ASD
PS Murmur link

http://www.wilkes.med.ucla.edu/PulmStenMain.htm

http://www.wilkes.med.ucla.edu/Systolic.htm

UCLA website for murmurs
https://www.med.ucla.edu/wilkes/index.htm

PS Murmur – other factors

- A hemodynamically significant ASD, increases blood flow across pulmonary valve and makes obstruction appear worse
- PS – while PVR is still high – masks the gradient due to downstream obstruction
- Presence of PDA – competing PBF
- PS – agitation during echocardiogram will increase the gradient
Pulmonary Valve Stenosis

- Why do we care if the RV is pressure loaded with mild / moderate PS?
  - We do and we don’t
    - Duration – for a short time is ok
    - Timing – during infancy
    - RV features - causes RVH, and decreased compliance of the RV ensues. Atrial shunting is thus affected.
    - Crowding on the left ventricle – RV is normally crescent shaped and wraps around LV.
- Symptoms – none unless severe or critical PS (cyanosis if an ASD, older pts with chest pain, syncope, sudden death with exercise)
- Natural history of Valvar PS is self-resolution

Continuous Doppler – $4V^2 = \text{Peak Pressure Gradient}$

- Peak to peak pressure gradient in cath lab of ~60 mmHg
- Consistent with moderate to valvar PS
PS Therapy in Evolution

- Balloon pulmonary valvuloplasty or surgical intervention
- SBE prophylaxis historically was given – endocarditis is exceedingly rare, no longer recommended
- Results of cath intervention are excellent – 4% of patients require 2nd intervention
- Post-intervention pulmonary insufficiency is common, mild, and well-tolerated

Balloon pulmonary valvuloplasty

- 20-30% upsize on the balloon size
- Static inflation (as opposed to BAS)
- Serial inflations for thick dysplastic valve
- Suicide RV – fatal RV failure from dynamic obstruction
- PGE can be discontinued at the time of the cath – some require extended PGE therapy or stenting of the ductus while RV remodels
- Results- larger balloon relieves PS but increases severity of PI in long term
PI post BPV

- Moderate PI (PR) post valvuloplasty is reported as 5-24%, and even is higher after BPV in neonates, with up to 40% with moderate PI at 10-12 yrs post.
- PI leads to RV dilation and has been associated with diminished exercise tolerance
- Follow by MRI – mid teenage years
- Pulmonary valve replacement

http://www.utswmedicine.org/Heartvalvesurgery.com

www.miotinternational.com Cardioegypt.com
Pulmonary Stenosis - summary

- Not a zebra
- Common cardiac disease, seen frequently in the NICU, with characteristic diagnostic findings
- Managed either expectantly or by cath intervention
- Aggressive balloon dilation is not benign, with pulmonary regurgitation and RV dilation

Peripheral Pulmonic Stenosis

- Newborn murmur, especially common with premature infants, ejection systolic radiating from the LUSB to axillae and the back
- Cause? – small size of the branch pulmonary arteries following birth and the right angle take-off from the MPA, which becomes less acute with growth.
- LPA PPS after ductal closure very common.
- Murmur should disappear in first 6 months of life.

Case # 2

- 7 year old twin with years of frequent URIs, worsening stridor, loud breathing, decreased exercise tolerance in comparison to his twin.
- Vitals normal, exam normal
- No respiratory distress noted
- Recent ER and pulmonary visits led to CT
Vascular Ring

- Anomaly of aortic arch formation leading to complete encirclement of trachea and esophagus by vascular structures
- Two common types consist of 90% of surgical cases
  - Double aortic arch
  - Right aortic arch with aberrant left subclavian artery and diverticulum of Komerell
Right aortic arch with Aberrant Left Subclavian Artery

- 2nd most common vascular ring – includes a retroesophageal vessel from which the left subclavian artery and left ductus or ligamentum arises.
- Regression of left fourth embryonic arch
- Symptoms – swallowing or respiratory problems. Most patients are asymptomatic.
- CXR – shows right arch.
- Barium esophagogram – posterior indentation on esophagus
- MRI – ideal for imaging both airway and vasculature.
Double Aortic Arch

- Persistence of embryonic right and left aortic arches with complete encirclement of trachea and esophagus
- Right aortic arch is dominant – 75% of cases. Left arch is either hypoplastic or atretic.
- Rarely associated with congenital heart disease – tetralogy of fallot, transposition of the great arteries, or coarctation of one or both arches
- Symptoms – stridor, respiratory problems, or swallowing difficulty
- CXR – can show tracheal indentation
- Barium studies are more diagnostic
- Confirmation by Echo (+/-), but please proceed with MRI/CTA

Vascular rings

- Symptoms due to tracheal and esophageal compression – stridor, dyspnea, barking cough – all worse with feeding or exertion
- Older children have chronic cough, wheezing, “asthma”.
- Recurrent respiratory infections
- Vomiting, choking, dysphagia, poor feeding

Vascular ring imaging

- CXR – right aortic arch, tracheal deviation
- Barium esophagograms – doesn’t help to describe the arch anatomy
- MRI and CT
- Treatment is always surgical ligation – left posterolateral thoracotomy. Video-assisted thoracoscopic surgery for certain cases
Case # 3

- 6 yo female with referral for murmur – subtle cardiac complaints of intermittent chest pain, fatigue, and palpitations at low level exercise. No syncope or cyanosis
- PMHx – asthma. No prior surgeries
- Family / Social Hx unremarkable
- Vitals HR-82 RR-24 BP-110/63 RA 139/70 LL
- Exam significant for late systolic click in supine position
- Click moves to mid-systole in standing position, with late systolic regurgitant murmur trailing the click
- High right clavicular continuous hum also present while seated or standing
Mitral valve prolapse

- A very confusing disease
- Diagnosis made by exam and confirmation by echocardiogram
- Constant mid-to-late systolic ejection click
- Variable regurgitant murmur of MR
- Echo shows – non planar Mitral valve with anterior and/or posterior leaflet prolapse with systolic displacement and leaflet thickening
- Echo with valsalva or upright posture should accentuate the prolapse
Mitral valve prolapse

- Common in connective tissue disorders
  - Marfan syndrome
  - Ehlers-Danlos syndrome
  - Osteogenesis imperfecta
  - Pseudoxanthoma elasticum
  - Stickler syndrome
- Other skeletal abnormalities include scoliosis, pectus excavatum, asthenic body habitus, and straight back syndrome
- Also consider hyperthyroidism, sickle cell disease, muscular dystrophy, hypomastia, and von Willebrand disease
Mitral valve prolapse

- Systolic click moves closer to S1 with decreases in LV volume (standing or valsala)
- Systolic click moves later toward S2 with increases in LV volume (squatting)
MVP – Clinical presentation

- Atypical chest pain, palpitations, fatigue, dizziness, syncope, dyspnea, anxiety attacks
- No basis of association of symptoms with MVP – no difference noted vs controls on exercise testing
- Murmur and click as described above
- Nonspecific EKG – but atrial and ventricular arrhythmias are more common than in the normal population
- Mitral regurgitation is usually trivial to mild – but can progress with age and necessitate valve repair
- Infective Endocarditis – routine dental prophylaxis is no longer recommended.

MVP – Complications

- Complications – extremely rare
  - Infective endocarditis
  - Thromboembolic phenomena
  - Cardiac arrhythmias
  - Sudden death
  - Progressive mitral regurgitation – 30-40 yo
  - Ruptured chordae tendineae
  - Congestive heart failure
MVP – Sports Restriction

- No data exists to demonstrate that strenuous exercise in patients with MVP predisposes to death.
- Common sense and good judgment
  - Self limit exercise for symptoms (chest pain, fatigue)
  - Evaluate left ventricular and left atrial function, mitral valve function, and existing symptoms.

Venous Hum

- Best auscultated over the right or left upper chest to low neck area – continuous
- Disappear with changes in head position or compression of the jugular vein
- Vary with respiration
- Best heard with patient upright
- Turbulence of venous flow IJ or EJ

Case # 4

- 6 week old female with a “murmur”
- No cyanosis, no sweating, increasing fatigue, sleeps a lot, seems pale and mottled with cold extremities
- Febrile illness 2 weeks ago with diarrhea
- Hyperemesis and preeclampsia during pregnancy
- Family history – paternal hypothyroidism
Physical Exam

- HR 153, RR 52, BP 76/49 LL
- Nl resp effort, clear lungs, no cyanosis
- PMI displaced laterally, increased precordial activity, gallop present. 1/6 systolic regurgitant murmur @ apex, pulses normal
- Obvious pallor, liver edge down 2 cm
Dilated Cardiomyopathy

- Dilation and impaired systolic function of left or both ventricles – “Hefty bag”
- Most common of the cardiomyopathies (CM) – 50%. Incidence is 0.58 cases per 100,000 kids
- Causes – Infectious, Familial, Mitochondrial, Metabolic, Arrhythmic, Toxic, Inflammatory, and Idiopathic

Inflammatory

- Viral myocarditis leads to abnormal immunologic response (autoimmune), causing tissue destruction. “Burnt-out Myocarditis” – 30-40% of cases
- Coxsackie B (enteroviruses), adenovirus, herpes simplex, cytomegalovirus

Clinical Presentation

- Older kids – exercise intolerance, DOE, tachycardia, palpitations, chest pain, syncope or near syncope, occasionally with cardiovascular collapse and sudden death
- Younger kids – rarely present with adult findings of peripheral edema and postural nocturnal dyspnea
- Infants – respiratory distress, abdominal distension, poor feeding, failure to thrive
Physical Findings - DCM

- Tachypnea, tachycardia, weak pulses, narrow pulse pressure, hypotension, cool extremities, poor capillary refill, wheezing, hepatomegaly
- Laterally and widely displaced cardiac impulse, gallop (S3), Mitral Regurgitation murmur
Diagnostic Tests in DCM

- CXR – cardiomegaly, pulmonary venous congestion, pulmonary edema, effusions, and atelectasis
- EKG – Sinus Tachy, ST-T wave changes, LVH
- Holter - Afib / Aflutter, PVCs, Ventricular tachycardia – 50% of patients at diagnosis
- Echo – LV dilation, decreased systolic performance, pericardial effusions, thrombi, Mitral regurgitation
- Cath – biopsy and pre-transplant hemodynamics
Questions??

IE Prophylaxis

- New recommendations include prophylaxis only for dental prophylaxis on patients with
  - Prosthetic cardiac valve
  - Previous infective endocarditis
  - Congenital heart disease **
  - Cardiac transplant recipients who develop valvulopathy
IE in Congenital Heart Disease

- IE prophylaxis recommended in CHD with
  - Unrepaired cyanotic heart disease, including palliative shunts and conduits
  - Completely repaired congenital heart defect with prosthetic material or device during first 6 months following procedure (while endothelialization occurs)
  - Repaired CHD with residual defects at site (VSD patch leak – prevents endothelialization)

IE

- Dental procedures cause transient bacteremia
  - Tooth extraction (10-100%)
  - Periodontal surgery (36-88%)
  - Scaling and root planing (8-80%)
  - Teeth cleaning (up to 40%)
- Routine daily dental procedures also cause transient bacteremia
  - Tooth brushing and Flossing (20-68%)
  - Wooden toothpicks (20-40%)
  - Chewing food (7-51%)

IE

- The frequency of bacteremia and therefore risk of endocarditis is far greater due to routine daily activities
- Vast majority of IE cases had no recent dental procedure 2 weeks before onset of symptoms
- Current emphasis is for maintenance of good oral hygiene and eradication of dental disease in effort to decrease incidence of IE.
- Risk of antibiotic adverse events may exceed benefit of prophylaxis in many patients
- Additional research is encouraged to substantiate future recommendations.