Grown –Up Congenital Heart Disease
What’s Important?

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GUCH/ACHD

• **Grown Up Congenital Heart** disease or
• **Adult Congenital Heart Disease**
  = any structural abnormality of the heart or intrathoracic great vessels that is present from birth

• 60% diagnosed in infants < 1yr, 30% in children, **10% in adults**.
• Age 16 years onwards
• ~ 85% survival to 16 years (better medical and surgical therapies over the past 20 years)
• **there are now more adults than children living with congenital heart disease**
So why is it important?

- Prevalence 3.2 per 1000 population
- The ACHD population in Wales is currently estimated at 8,500 (~185,000 in the U.K)
- The ACHD population is expected to expand at a rate of ~ 10% per year
- So GPs and secondary care physicians will start to see more pts with ACHD.
- Plus, many ACHD patients wish to start a family...
ACHD lesions

**Simple:**
e.g. ASD/VSD/PDA/anomalous pulmonary venous drainage

**Moderate:**
e.g. Tetralogy of Fallot, coarctation of the aorta, Ebstein’s anomaly, atrioventricular septal defect (AVSD)

**Complex:**
e.g. Single ventricle e.g. with Fontan-type palliation,

Systemic ventricle = morphological right ventricle
e.g. transposition of the great arteries (TGA),
corrected transposition of the great arteries ccTGA.
Initial Management of Congenital Heart Disease

- Aim = to restore cardiac anatomy to as near normal as possible at as early an age as possible (i.e. as neonates/infants).
- Many patients undergo correction of the defect(s) e.g. ASD closure.
- Some disease is so complex e.g. a single ventricle that palliative surgery is the only option, which leaves the patient with gross anatomical distortions.
- Occasionally defects are not operable due to complexity or irreversible pulmonary hypertension at time of diagnosis.
ACHD management

• **All patients should be assessed at least once** by a specialist in ACHD and a management plan formulated

• Currently Dr Dirk Wilson, Dr Nav Masani, myself (Dr Helen Wallis) aim to provide a ACHD service across SW but as yet there is no full time ACHD specialist in Wales.

• A few patients will be discharged with advice, **most will require lifelong FU**.

• Correction of a defect does not equate to a ‘cure’.
CASE 1

- 30 year old, 20/40 pregnant (1\textsuperscript{st} pregnancy)
- Slight increased SOBOE and palpitations
- PMH= mild asthma
- Soft ESM murmur noted in ANC
- ECHO requested
Secundum ASD
Secundum ASD

• The commonest new diagnosis in adulthood
• Often patients labelled with mild asthma, or c/o palpitations or exertional chest discomfort
• Classically a partial RBBB on ECG, fixed split second heart sound and pulmonary ejection systolic murmur
• All patients should be considered for ASD closure unless defect is ‘tiny’
Secundum ASD- closure

• Percutaneous closure is the method of choice, commonly using an Amplatzer® septal occluder device.

• Made from Nitinol, a wire made from an alloy of nickel and titanium which has shape memory.
Secundum ASD- closure
CASE 1 - outcome

• Normal pulmonary pressures on ECHO assessment, good biventricular function
• Normal vaginal delivery with air filter for any IV lines
• Post partum TOE to assess suitability for percutaneous closure and exclude anomalous pulmonary venous connections
CASE 1- learning points

consider the diagnosis of ASD in any patient with breathlessness and palpitations (esp. if young)

often diagnosed incidentally when admitted for another reason

Small risk of paradoxical embolism (c.f. PFO)
CASE 2

• 32 year old man referred from A&E team with headache and newly diagnosed hypertension.
• PMH of ‘heart operation as a child’
• FU until 16 years then ‘discharged’

o/e
• BP 190/110 right brachial, 100/60 left brachial.
• weak femoral pulses bilaterally,
• left thoracotomy scar
• DIAGNOSIS = ?
CASE 2: Coarctation of the aorta

- Coarctation of the aorta is a discrete, ridge-like narrowing in the descending aorta at the site of the aortic ductal attachment, near the left subclavian artery.
Coarctation repair

A = end to end anastomosis, B = subclavian flap repair, C = patch aortoplasty
coarctation of the aorta

- **In older children and adults** with native CoA, **angioplasty** (with or without stenting) is an accepted treatment.
CASE 2 = coarctation of the aorta

CoA is a **diffuse** arteriopathy characterised by widespread changes in vascular structure and function which manifests as:

- hypertension (occ. re-coarctation)
- premature cardiovascular disease,
- aneurysmal disease (ascending aorta and site of repair).

Associated bicuspid aortic valve (fragile aorta), berry aneurysms

Surgical/ catheter intervention is **palliative** not curative, hence lifelong FU
Coarctation- aneurysm at site of repair
CASE 2 - outcome

• ECHO and then MRI aorta confirmed significant re-coarctation at repair site.
• MRA head- no berry aneurysm
• Underwent angioplasty and stenting
• AT II blocker still necessary post procedure

Follow-up
• needs BP checks RIGHT brachial, BP < 130/80
• Fasting cholesterol
CASE 2 – learning points

• Any patient who is not under active FU who has undergone heart surgery as a child or who has a heart defect which has not been operated on needs to be referred to the ACHD service for assessment.

• Monitor BP at the RIGHT brachial
CASE 3

• 24 year old patient with repaired Tetralogy of Fallot
• p/w pre-syncopal episodes
• 6/12 post partum (uncomplicated NVD with planned epidural delivered in tertiary centre)
• o/e ESM and EDM pulmonary area
• ECG SR, RBBB, QRSd = 184 ms
• ECHO, then cardiac MRI
Tetralogy of Fallot
Repaired tetralogy of Fallot
Tetralogy of Fallot

- Post op:
- most pts have a RBBB post ventriculotomy
- Most patients have significant pulmonary regurgitation (PR) from enlarging the right ventricular outflow tract (RVOT)
- PR results in volume loading of the RV with progressive enlargement and then dysfunction
- Pts develop breathlessness and are at risk of arrhythmias (atrial and ventricular)
- QRSd > 180 ms is a risk factor for SCD
CASE 3 - outcome

• pre pregnancy severe pulmonary regurgitation with moderate RV enlargement but good RV function (QRSd= 143 ms)
• post partum, severe RV enlargement/dysfunction (QRSd reflects this)
• event monitor captured VT
• referred for urgent PVR +/- ICD
CASE 3 – learning points

- Pts with repaired Tetralogy will require a pulmonary valve replacement at some point
- RV size and function, TR and symptoms will determine the timing
- Regular clinic and ECHO FU is vital
- Pregnancy can precipitate a decline in RV function
- Development of arrhythmias may indicate a decline in RV function
CASE 4

• 32 year old man with Ebstein anomaly, works as a builder, physically very fit
• seen in A&E with palpitations.
• diagnosed with new onset AF (assumed > 48 hours duration, not compromised)
• admitting team aware that he had a heart condition but had not undergone any surgery),
• commenced on Bisoprolol and Warfarin, not admitted and referred for elective DC cardioversion.
• seen in ACHD OPD 2/52 later with severe SOB at rest with abdominal and ankle swelling.
Ebstein Anomaly
CASE 4 - Outcome

• o/e severe right heart failure, low cardiac output, AF
• Admitted, treatment with IV diuretics, urgent TOE guided cardioversion after amiodarone loading
• Severe TR – assessment for TVR once in SR
CASE 4- learning points

• Just because a patient hasn’t undergone cardiac surgery it doesn’t mean that the heart condition is minor

• Any arrhythmia in a pt with ACHD is potentially serious and may be life threatening

• Development of arrhythmias may be the marker of worsening ventricular function +/- or valve disease
CASE 5

• A 25 year old patient with Down’s syndrome
• Unrepaired atrioventricular septal defect (AVSD)
• Resulting in Eisenmengers syndrome
• i.e. shunt reversal with central cyanosis
• p/w progressive breathlessness and functional decline, with headache and intermittent blurred vision
• o/e finger clubbing, O2 sats 79%, BP 100/60
• No murmur audible, chest clear
CASE 5- AVSD Eisenmengers
CASE 5- Eisenmenger’s syndrome

• Hb 15.2 g/dl (14-18 g/dl)
• Haematocrit 0.52 (0.42-0.54)
• U&Es normal
• TFTs normal
• Good biventricular function on ECHO
• CXR clear

• ? Consider disease modifying therapy for pulmonary hypertension i.e Sildenafil +/- or Bosentan
CASE 5- outcome

• This patient had a relative anaemia (previous Hb 21g/dl)
• Hb > 18 g/dl is expected in such patients
• Pts with Eisenmengers have abnormal clotting and are at risk of thrombotic events and increased bleeding
• Therefore check serum ferritin, TIBC
• Patient was iron deficient resulting in less deformable RBCs causing ‘hyperviscosity type’ Sxs
• **Cautious** oral iron replacement i.e. 200 mg od for 1 week then recheck FBC/ haematocrit
• Venesection is rarely indicated and often results in iron deficiency
CASE 5 - Outcome

- Good response to 2 months of intermittent oral Fe,
- H. Pylori +ve serology treated with triple therapy
- However, even when Hb > 20 g/dl still NYHA III, therefore, referred to PHT centre and commenced on Bosentan with marked improvement to NYHA class II
CASE 5 – learning points

• Don’t be caught out by a Hb level which is within normal range as being normal.....

• Regularly look for Fe deficiency in Eisenmengers patient esp. in young women who are menstruating

• NYHA class III/IV and probably class II need to be considered for pulmonary hypertension therapy as latest studies demonstrate increased survival as well as increased quality of life
CASE 6

- 35 year old man presented with a 3/12 history of increasing breathlessness on exertion, orthopnea, PND, ankle oedema
- No previous history of cardiac disease
- Never particularly sporty but previously fit and well
- o/e clinically in heart failure
- Commenced on diuretics and referred for urgent cardiology assessment
CASE 6- ECHO
CASE 6- congenitally corrected transposition of the great arteries (ccTGA)
CASE 6- ccTGA

- The right ventricle is the systemic ventricle i.e. has to pump blood around the body
- The RV is not designed to cope with high pressure, therefore, over time the RV dilates and then function declines
- Patients with ccTGA and no other associated lesions may not present with symptoms until their 30’s
- A rare cause of heart failure and can be missed
CASE 6- outcome

• Treated with standard heart failure therapy i.e. ACEi, betablockers, spironolactone, biventricular pacing but minimal evidence base for use in RV systemic ventricles

• May need to be considered for heart transplantation
Summary

• If you have any patients with ACHD who have not been recently assessed (within the past 3 years) and/or do not have a clear longterm management plan please refer to the ACHD service

• Any patient with ACHD who wishes to become pregnant (or who is pregnant) needs ACHD review (or alternatively needs appropriate contraceptive advice)

• Atrial as well as ventricular arrhythmias need to be taken seriously in pts with ACHD

• Heart failure is the second most common reason for admission
FINALLY
The Sarah Boulton Grown Up Congenital Heart Disease Memorial Fund

- Trust fund in memory of Sarah, one of my ACHD patients who sadly died at the age of 30 years in November 2011
- Currently more than £1000 raised with a sponsored charity walk up Snowdon planned for 15\textsuperscript{th} September
- All monies raised will be used to improve services/ facilities for GUCH patients within ABM
THANK YOU....
ANY QUESTIONS?