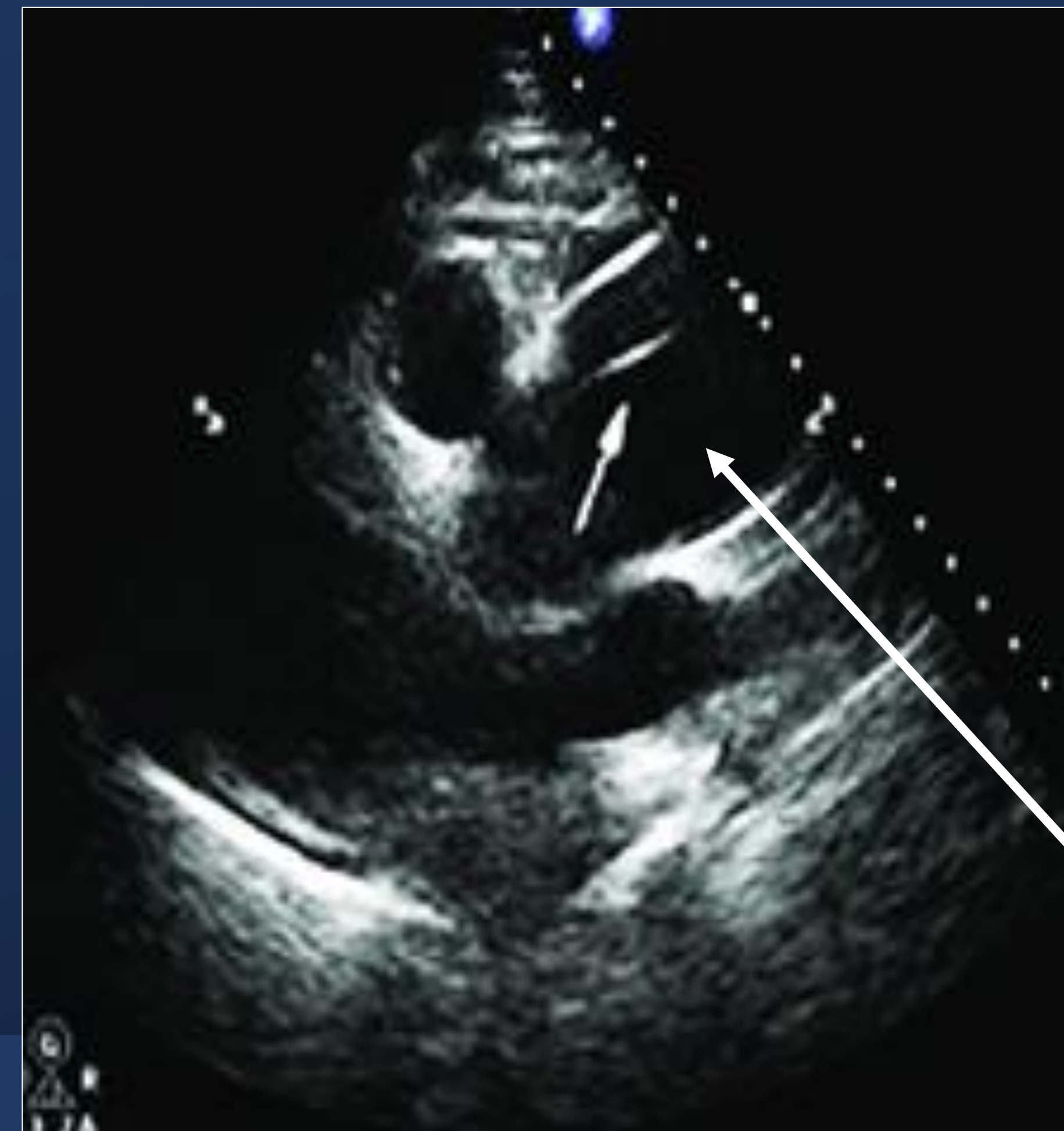


Case study: Marfan Syndrome detected via telephone consultation in the third trimester of pregnancy

Dr Kelsie Sykes, MRCOG

Case discussion

- 28 year old female
- Para 1, previous uncomplicated pregnancy and home birth
- Referred for telephone consultation at 31 weeks gestation due to aortic dissection in her Mother and Uncle
- Patient was 183cm tall and questioning revealed long digits and possible pectus carinatum.
- No cardiovascular, ocular or joint symptoms.



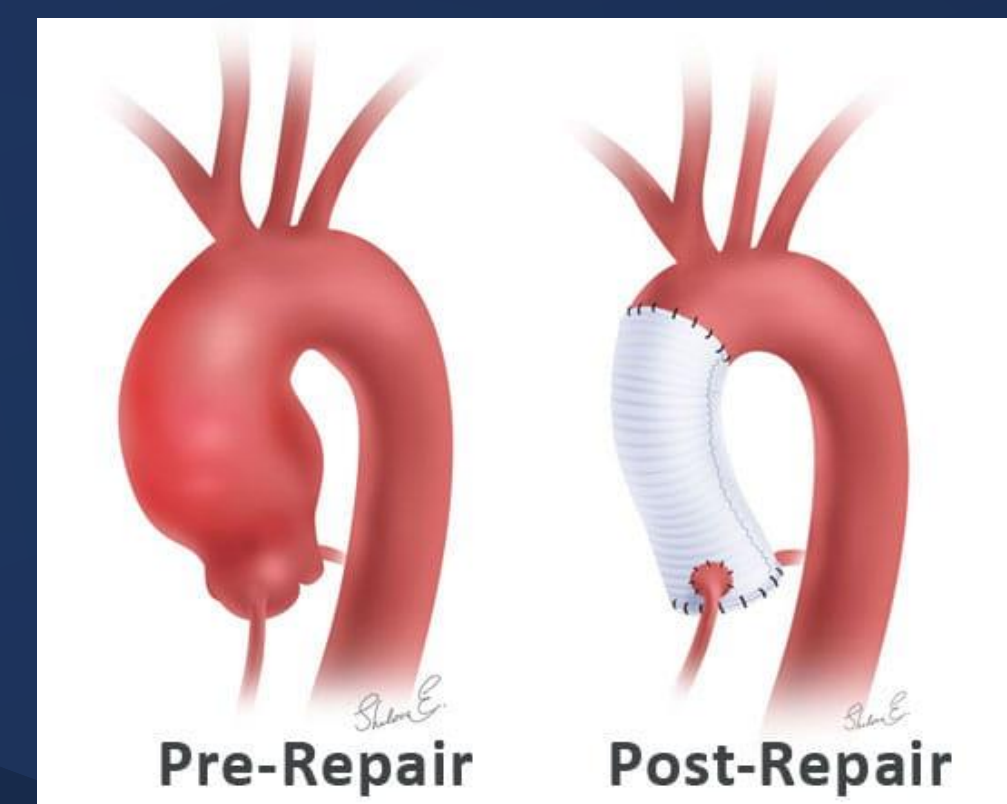
Investigations

Echocardiogram was performed at 32+6 weeks and revealed good biventricular function, but severe dilatation of the aortic root of 50mm.

Dilated aortic root, measuring 50mm

Management

- Referral to a tertiary unit
- Commenced on beta-blockers
- Delivery by uncomplicated Caesarean Section at 33+6 weeks
- Two weeks post delivery underwent valve-sparing aortic root replacement and coronary artery bypass graft



Aortic root replacement

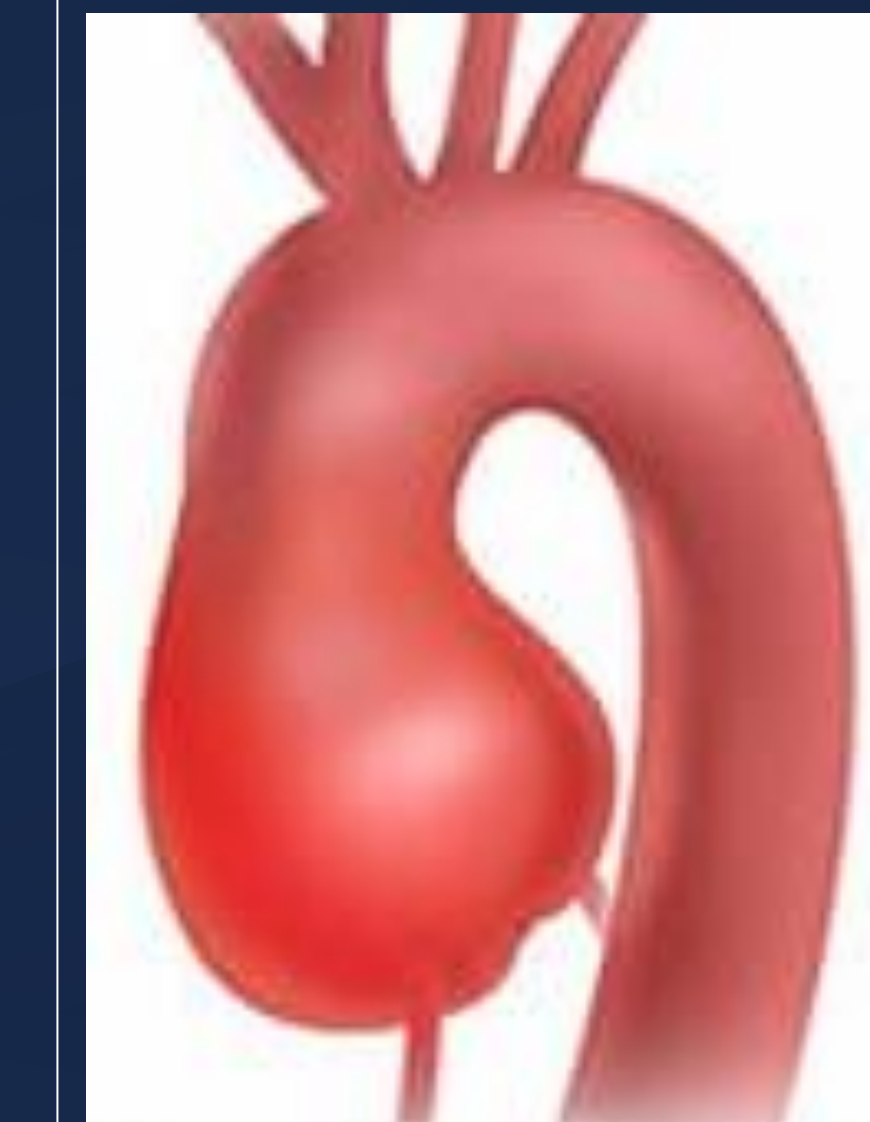
- Dilated portion of the aorta is removed and replaced with a graft
- Aortic valve is removed and then replaced within the graft

Follow-up

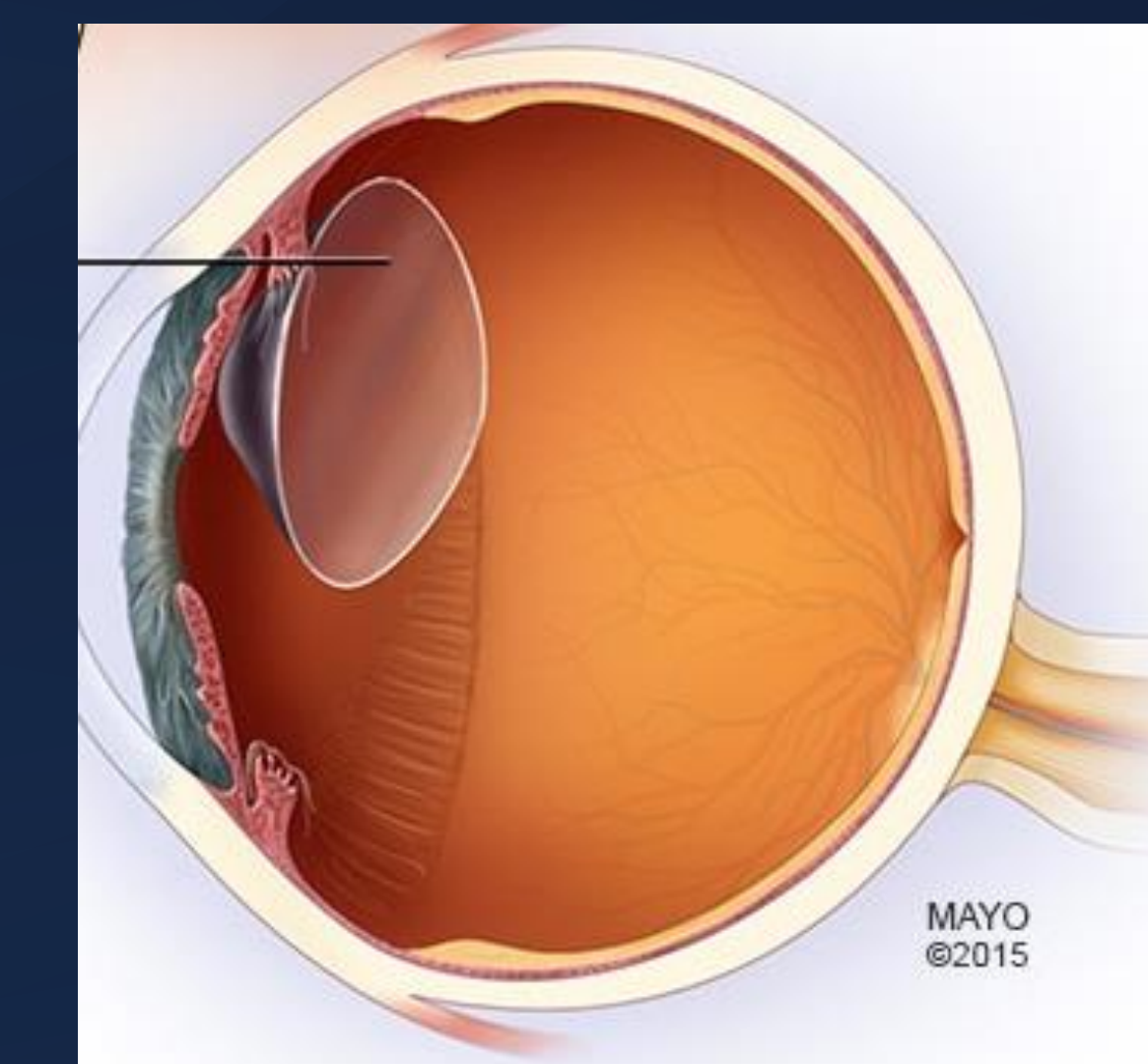
- Post-operative course was turbulent
- MCA territory stroke
- Severe mitral regurgitation requiring a redo sternotomy for valve repair
- Aortic root abscess
- Atrial flutter resulting in pulmonary oedema
- Perforation of anterior mitral valve leaflet with significant mitral regurgitation
- Third surgery for mitral valve replacement at 6 months postnatal

Discussion

- Marfan syndrome is a heritable disorder of connective tissue
- 75% of cases are inherited in an autosomal dominant pattern (remainder due to de novo mutation)
- Caused by mutations in the FBN1 gene which codes for the extracellular matrix fibrillin, a major component of connective tissue
- 3 main manifestations:



Proximal aortic aneurysm



Ocular lens dislocation



Overgrowth of long bones

- Leading cause of mortality and morbidity is aortic dissection
- Aortic root diameter >40mm carries a 10% risk of dissection in pregnancy
- Management is with beta-blockers and lifestyle modifications, aimed at minimising aortic root dilatation