Marfan Syndrome

What is it?

- It is an autosomal dominant disorder of the body's connective tissue due to gene mutations.
- Can affect men and women of any race or ethnic group.
- It can be inherited or occur spontaneously with no family history.
- It affects the eyes, skeleton and heart.
- Common features include excessive height with long limbs, fingers and toes.

How is it Diagnosed?

- Blood test demonstrating the abnormal gene.
- Physical exam.
- Cardiac MRI.

How does it affect the heart?

- Dilation of the aortic root is found in 60-80% of patients.
- This can lead to aortic dissection or rupture.
- Can have MV prolapse and/or calcification.
- Dilation of the pulmonary artery.
- Dilatation or dissection of the descending aorta.

What is the long term management?

- Beta blocker and Losartan.
- Avoiding moderate dynamic exercise and sports with risk of bodily collision.
- Cardiac surgery if aortic root dimension is increased.
- Pregnancy – high risk.

What is the follow up?

- Annual cardiac follow up is mandatory in all Marfan patients.
- Echocardiography to monitor aortic root and valve, as well look at LV and MV and serial MRI

Reference: The European Society of Cardiology (2012) *Guidelines for the Management of Grown-Up Congenital Heart Disease.* Oxford University Press: Oxford.