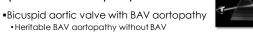


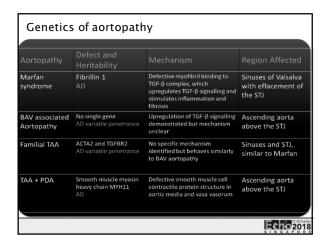
Spectrum of aortopathy



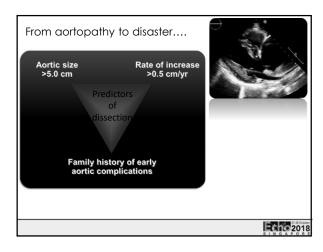


- Coarctation with aortopathy
- ■Connective tissue syndromes
- Marfan Syndrome
- Loeys-Dietz syndrome
- Ehlers-Danlos (vascular)
- •Familial thoracic aortic aneurysm
- •Thoracic aortic aneurysm + PDA
- •MASS phenotype
- Congenital contractural arachnodactyly
- Homocystinuria





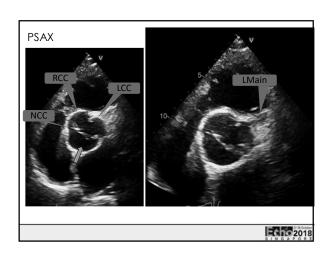
Aortopathy			
MASS phenotype (MVP, Ao ULN, skin, skeletal)	No consistent gene, sporadic Dominant negative mutation	No specific mechanism identified	Aortic root and ascending aorta
Loeys-Dietz syndrome	TGFBR1, TGFBR2, SMAD3 AD	Upregulation of TGF-β signalling stimulates aggressive and widespread arterial aneurysm formation	Aortic root, ascending aorta, arch, descending
Ehlers-Danlos (vascular)	COLSA1, COLSA2, COLSA1	Upregulation of TGF-β signalling stimulates aggressive and widespread arterial aneurysm formation	Aortic root, ascending aorta, arch, descending
Congenital contractural arachnodacyly (Beals-Hecht)	Fibrillin 2 AD	No clear mechanism	Aortic root



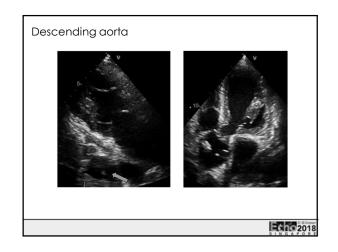
Case -54 year old male -Presents with acute retrosternal chest pain to regional centre -Transferred after CT scan suggests Ao dissection

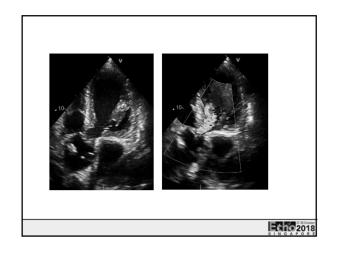


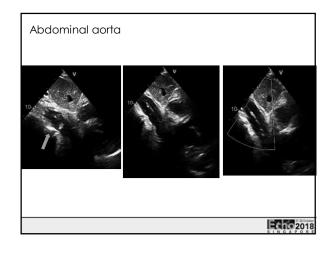


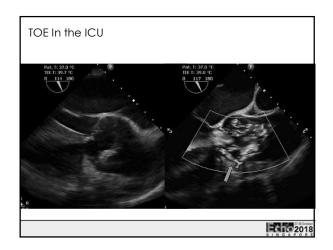


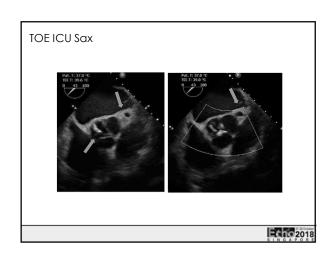


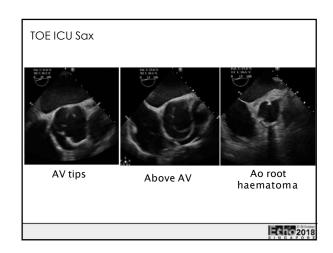








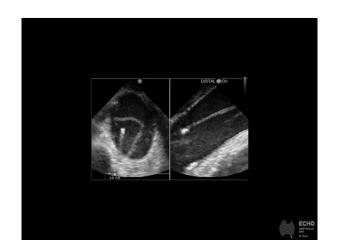














Imaging the aorta 2018 •Aortopathy is a slowly progressive,	
often genetic, completely silent illness •Echo is cornerstone of identification, sequential follow-up and screening	
■ Fastidious attention to detail – 1 mm counts ■ Use of ancillary imaging taken in	-
 Context Always look at previous studies. Acute aortic syndromes can be complex and require careful evaluation 	
complex and require careful evaluation	
2:3 0:00 2018 5 N Q A P O B F	