

## UKGTN Testing Criteria

<b>Approved name and symbol of disease/condition(s):</b> Familial Thoracic Aortic Aneurysms Dissections (TAAD)	<b>OMIM number(s):</b> 132900, 611788,
<b>Approved name and symbol of gene(s):</b> <i>MYH11, ACTA2, TGFBR1, TGFRB2</i>	<b>OMIM number(s):</b> 160745, 102620, 190181, 190182

<b>Patient name:</b>	<b>Date of birth:</b>
<b>Patient postcode:</b>	<b>NHS number:</b>
<b>Name of referrer:</b>	
<b>Title/Position:</b>	<b>Lab ID:</b>

<b>Referrals will only be accepted from one of the following:</b>	
<b>Referrer</b>	<b>Tick if this refers to you.</b>
<b>Consultant Clinical Geneticist</b>	
<b>Consultant Cardiologist in liaison with clinical geneticist</b>	

<b>Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:</b>	
<b>Criteria</b>	<b>Tick if this patient meets criteria</b>
Dilation and/or dissection of the ascending thoracic aorta, <b>OR</b> dissection of the descending aorta just distal to the subclavian artery	
<b>AND 2.</b> Family history of TAAD or unusually early presentation of dissection	
<b>AND</b> exclusion of Marfan syndrome, Loeys-Dietz aortic syndrome and other connective tissue abnormalities	
<b>OR</b> Family history of known mutation in ACTA2/MYH11/TGFBR1/TGFBR2	

**If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.**