

UKGTN Testing Criteria

Approved name and symbol of disease/condition(s): Supravalvular Aortic Stenosis; SVAS Cutis Laxa, Autosomal Dominant 1; ADCL1	OMIM number(s): 185500 123700
Approved name and symbol of gene(s): elastin; ELN	OMIM number(s): 130160

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

Referrals will only be accepted from one of the following:	
Referrer	Tick if this refers to you.
Consultant Cardiologist (Grown Up Congenital Heart or Inherited Cardiac Clinician specialists only)	
Consultant Paediatric Cardiologist	
Consultant Clinical Geneticist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	Tick if this patient meets criteria
Supravalvular Aortic Stenosis OR	
Cutis laxa with or without cardiac or other features	
OR At risk family members where familial mutation is known	

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