

## R125 Thoracic aortic aneurysm or dissection

### Testing Criteria

1. Thoracic aortic aneurysm\* or dissection with onset before age 50, OR
2. Thoracic aortic aneurysm\* or dissection with onset before age 60 with a first degree relative with thoracic aortic aneurysm or dissection, OR
3. Thoracic aortic aneurysm\* or dissection before age 60 with no classical cardiovascular risk factors, OR
4. Thoracic aortic aneurysm\* or dissection before age 60 with features suggestive of aortopathy, e.g. arterial tortuosity, OR
5. Clinical features suggestive of Loeys-Dietz syndrome, OR
6. Features of Marfan syndrome giving a systemic Ghent score of  $\geq 7$ , following assessment by a clinical geneticist or specialist with expertise in aortopathy, OR
7. High clinical suspicion of a condition predisposing to aortic/arterial disease AND diagnostic testing for other conditions such as Ehlers Danlos syndrome (where indicated) has not identified a causative mutation
8. Any deceased individual with a thoracic aortic aneurysm\* or dissection detected at autopsy meeting one of the above criteria and who have relatives who will benefit from cascade testing using a genetic diagnosis will be suitable for post-mortem genetic testing.

\*Thoracic aortic aneurysm defined as:

- In children: z score  $>2$  for body surface area
- In adults: dilatation  $>38$  mm

Testing should be carried out following assessment in a clinical service specialising in management of patients with aortopathy, including support from clinical genetics; testing may occasionally be appropriate outside these criteria following discussion in an aortic genetics MDT

### Overlapping Indications

- R27 Congenital malformation and dysmorphism syndromes – likely monogenic or R89 Ultra-rare and atypical monogenic disorders tests should be used in individuals with congenital malformations, dysmorphism or other complex or syndromic presentations

Referrals for testing will be triaged by the Genomic Laboratory; testing should be targeted at those where a genetic or genomic diagnosis will guide management for the proband or family.

### Where in Pathway

At presentation

### Requesting Specialties

- Cardiology
- Clinical Genetics

### Specialist Service Group

- Cardiology

### Associated Tests

Please note all the tests below will be undertaken for R125 Clinical Indication requests

Code	Name	Optional Family Structure	Scope(s)	Target Type	Target Name	Method
R125.1	Thoracic aortic aneurysm or dissection WES or medium panel	Singleton	Small variants	Panel of genes or loci	Thoracic aortic aneurysm or dissection (700)	WES or Medium Panel
R125.2	Thoracic aortic aneurysm or dissection MLPA or equivalent	Singleton	Exon level CNVs	Panel of genes or loci	Thoracic aortic aneurysm or dissection (700)	MLPA or equivalent