Protocol

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Document Statement

This protocol aims to ensure identification of patients with potential genetic mutations associated with thoracic aortic aneurysms (TAA) and Aortic dissections, and appropriate referral for genetic testing.

TAAs or Aortic Dissections in young patients with no cardiovascular risk factors are likely to have a genetic cause. These can be identified in patients with classic “syndromic” features of known connective tissue disorders such as Marfan Syndrome, Loeys-Dietz Syndrome, Vascular Ehler Danlos Syndrome and Turner Syndrome. There is growing appreciation for a group of patients who suffer aortopathies at a young age without known evidence of connective tissue disorders (non-syndromic) (1). These aortopathies may run in families, have a higher rate of growth and may dissect at near normal dimensions when associated with specific gene lesions such as ACTA2 and MYLK.

Some patients with specific gene mutations might be offered surgery at near normal aortic sizes based on what is known from the natural history. Also, screening family members may reveal a genetic mutation that necessitates surveillance. Non clinical implications affect choice for careers, reproductive health, disease-labelling, insurance and others. Therefore it is important to identify patients with potential genetic mutation, and offer them genetic testing. (1)

1. Roles and Responsibilities

- The clinical lead for aortic surgery is responsible for the overall implementation of this protocol.
- All clinicians reviewing patients in Aortic clinic are responsible for following the guidance in this protocol.
- Clinicians should ensure adequate correspondence to involved geneticist as well as communication with the lead Aortic surgeon in clinic on the day.
- Adequate patient communication regarding the reasons behind advised referral is the responsibility of the reviewing clinician.
- Involved geneticist should ensure the aortic team is aware of significant results either via correspondence or via Aortopathy MDT.
- Outpatient clinic manager is responsible for ensuring a copy of this guidance is available in the surgical folder available in OPD.

2. Procedure

Potential candidates for genetic testing:

- Patients referred for incidental detection or under surveillance for a TAA or Type B Aortic dissection

- Post-operative surveillance for patients who underwent surgery or endovascular intervention for TAA or Aortic Dissection

Consultation:

- Detailed family history to detect affected family members.
- Clinical examinations to detect significant features of connective tissue disorder (below).
- Discussion with the patient regarding a possible inherited component.
- Genetic testing to be offered if the patient meets the criteria for referral (below).

Criteria for Referral:

- Thoracic aortic aneurysm (z score >2 for body surface area) or previous acute or chronic aortic dissection treated surgically or conservatively in patients younger than 50.

- Thoracic aortic aneurysm (z score >2 for body surface area) or previous acute or chronic aortic dissection treated surgically or conservatively in patients younger than 60 with a first degree relative with thoracic aortic aneurysm or dissection.

- Thoracic aortic aneurysm (z score >2 for body surface area) or previous acute or chronic aortic dissection treated surgically or conservatively in patients younger than 60 with no classical cardiovascular risk factors.

- Thoracic aortic aneurysm (z score >2 for body surface area) or previous acute or chronic aortic dissection treated surgically or conservatively in patients younger than 60 with features suggestive of aortopathy, e.g. arterial tortuosity on imaging, cerebral aneurysms.

- Features suggestive of connective tissue disorder at any age:

List of most characteristic or easy recognizable clinical features associated with syndromic forms of thoracic aortic disease.

Craniofacial features	Craniosynostosis
	Widely spaced eyes (hypertelorism)
	Cleft palate or bifid uvula
Ocular features	Lens subluxation/dislocation (ectopia lentis)
	Retinal detachment
	High myopia (−6.00 diopters or higher)
	Iris hypoplasia or flocculi
Cardiovascular features	Mitral valve prolapse
	Arterial tortuosity
	Multiple aneurysms or dissections
	Left-sided congenital heart defect or patent ductus arteriosus
Musculoskeletal features	Pectus excavatum or carinatum
	Joint hypermobility or contractures
	Recurrent joint subluxations/dislocations
	Severe, early-onset osteoarthritis
	Severe kyphosis or scoliosis
Cutaneous features	Thin, translucent skin with easily visible veins
	Hyperelastic skin
	Livedo reticularis
	Striae at unusual sites/not related to weight gain
	Atrophic or wide scars
Other features	Short or tall stature
	Disproportionately long limbs (dolichostenomelia)
	Abnormal long and slender fingers (arachnodactyly)
	Spontaneous pneumothorax
	Recurrent abdominal wall hernias
	Spontaneous rupture of internal organs

Expert consensus recommendations on the cardiogenetic care for patients with thoracic aortic disease and their first-degree relatives (2)

Correspondence:

- Details of the patient consultation including discussions regarding genetic testing should be included in the clinic letter and copied to the patients GP.
- A separate referral letter should be addressed to the clinical geneticist. This should include patients history, the rationale behind the referral as per this guidance and a summary of the discussion with the patient.
- The consultant aortic surgeon in clinic on the day should be informed.

3. Policy Implementation Plan

- The clinical lead for aortic surgery will be responsible for implementing this protocol.
- All junior doctors as well as the Aortic team will be made aware of the content of this guidance via email and departmental meeting.
- A copy of this guidance will be available in the surgical folder in outpatient clinic.

4. Monitoring of Compliance

Clinic lists and patient referrals will be reviewed by the members of the Aortic Team and recommendations for genetic testing will be supervised and prospectively monitored.

6. References

1-2014 ESC Guidelines on the diagnosis and treatment of aortic diseases. ***European Heart Journal (2014) 35, 2873–2926 doi:10.1093/eurheartj/ehu281***

2- *Expert consensus recommendations on the cardiogenetic care for patients with thoracic aortic disease and their first-degree relatives Judith M.A. Verhagen a, Marlies Kempers b, Luc Cozijnsen c, Berto J. Bouma d, Anthonie L. Duijnhouwer e, Jan G. Post f, Yvonne Hilhorst-Hofstee g, Sebastiaan C.A.M. Bekkers h,Wilhelmina S. Kerstjens-Frederikse i, Thomas J. van Brakel j, Eric Lambermon k, Marja W. Wessels a, Bart L. Loeys b,l, Jolien W. Roos-Hesselink m, Ingrid M.B.H. van de Laar a,□, on behalf of the NationalWorking Group on BAV & TAA -* ***International Journal of Cardiology 258 (2018) 243–248***

8. Endorsed By:

Name of Lead Clinician / Manager or Committee Chair	Position of Endorser or Name of Endorsing Committee	Date
Mr Omar Nawaytou		

9. Record of Changes

Section No	Version No	Date of Change	Description of Amendment	Description of Deletion	Description of Addition	Reason