

The Need of Screening for Relatives of Patients Affected by Thoracic Aortic Diseases

Daniele Maselli¹, Saverio Nardella¹, Aamer Ahmed² and Giovanni Mariscalco^{2*}

¹Department of Cardiovascular Surgery, Cardiac Surgery Unit, S. Anna Hospital Catanzaro, Italy

²Department of Cardiovascular Sciences, University of Leicester, Glenfield Hospital, Leicester, UK

*Corresponding author: Giovanni Mariscalco, Department of Cardiovascular Sciences, University of Leicester, Glenfield Hospital, Leicester, UK, Tel: 4401162583019; E-mail: giovannimariscalco@yahoo.it

Received date: November 21, 2016, 2016; Accepted date: November 23, 2016; Published date: November 30, 2016

Copyright: © 2016 Maselli D et al. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

Citation: Maselli D, Nardella S, Ahmed A, Mariscalco G (2016) The Need of Screening for Relatives of Patients Affected by Thoracic Aortic Diseases. Angiol 4: e116. doi:10.4172/2329-9495.1000e116

Thoracic Aortic Diseases

The Global Burden Disease 2010 project demonstrated that the overall global death rate from aortic aneurysms and aortic dissection increased from 2.49 per 100000 to 2.78 per 100000 inhabitants between 1990 and 2010, with higher rates for men [1]. Aortic size currently remains the main criterion for prophylactic surgical intervention in patients affected by thoracic aortic disease (TAD) [2]. However, there is evidence to suggest that patients with specific genetic mutations and familial traits require earlier interventions at smaller aortic sizes. The guidelines for management of TAD already include specific recommendations regarding treatment of patients with Marfan syndrome and other syndromic forms of TAD [3,4]. Recent studies on the genetics of TAD have also shown that patients with certain specific genetic abnormalities have a different natural history course [5].

Modern information suggests that genetic testing of patients with TAD may be important for determining the appropriate personalized treatment strategy for individual patients. However, no clear screening indications exist for relatives of patients already diagnosed with TAD [3,4]. Guidelines recommend screening of relatives of patients affected by syndromic TAD forms (i.e. Marfan syndrome, Loeys-Dietz syndrome, Ehlers-Danlos syndrome) and BAV aortopathies only [3,4]. A recent Australian cohort study demonstrated that 37% of family members of TAD patients can be easily identified through common screening tests, including transthoracic echocardiogram, CT scan and/or MRI of the thoracic aorta [6]. In light of low risk mortality associated with elective surgical procedures, prospective studies, both

randomized and observational, with the aid of possible genetic screening tests should be routinely offered to the relatives of patients already affected by TAD.

References

1. Sampson UKA, Norman PE, Fowkes GR, Aboyans V, Yanna Song, et al. (2014) Global and regional burden of aortic dissection and aneurysms. *Global Heart* 8: 171-180.
2. Davies RR, Gallo A, Coady MA, Tellides G, Botta DM, et al. (2006) Novel measurement of relative aortic size predicts rupture of thoracic aortic aneurysms. *Ann Thorac Surg* 81: 169-177.
3. Hiratzka LE, Bakris GL, Beckman JA, Bersin RM, Carr VE, et al. (2010) 2010 ACCF/AHA/AATS/ACR/ASA/SCA/SCAI/SIR/STS/SVM Guidelines for the diagnosis and management of patients with thoracic aortic disease. *J Am Coll Cardiol* 55: e27-129.
4. Erbel R, Aboyans V, Boileau C, Bossone E, Bartolomeo RD, et al. (2014) 2014 ESC Guidelines on the diagnosis and treatment of aortic diseases: document covering acute and chronic aortic diseases of the thoracic and abdominal aorta of the adult. The Task Force for the Diagnosis and Treatment of Aortic Diseases of the European Society of Cardiology (ESC). *Eur Heart J* 35: 2873-2926.
5. Milewicz DM, Guo DC, Tran-Fadulu V, Lafont AL, Papke CL, et al. (2008) Genetic basis of thoracic aortic aneurysms and dissections: focus on smooth muscle cell contractile dysfunction. *Annu Rev Genomics Hum Genet* 9: 283-302.
6. Robertson EN, van der Linde D, Sherrah AG, Vallely MP, Wilson M, et al. (2016) Familial non-syndromal thoracic aortic aneurysms and dissections: Incidence and family screening outcomes. *Int J Cardiol* 220: 43-51.