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Editorial OMICS International

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

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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 affcted 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 thorugh common screening tests, including transthorcaic 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 randomzied and observational, with the aid of possible genetic screening tests should be routinely offered to the relatives of patients already affected by TAD.

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