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Studies on aortic and aortic valve diseases

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Abstract

Bicuspid aortic valve (BAV) is the most common of the congenital cardiac malformations with an estimated incidence of 1-2% in the general population. Patients with BAV are predisposed to early and frequent endocarditis, stenosis, regurgitation, which often accompanied by aneurysm and dissection. A family-based genome-wide analysis found that BAV was linked to chromosomal regions 5q, 13q, and 18q with autosomal dominant inheritance, reduced penetrance, and a non-Mendelian pattern. Mutations have been detected in the transmembrane receptor *NOTCH1* (gene map locus 9q) in familial and sporadic BAV cases. Mutations in the *vascular smooth muscle cell alpha actin* gene (mapped to chromosome 10q) have also been identified in BAV patients. Downregulated expression of *ubiquitin fusion degradation 1-like*, a gene that is highly expressed in the outflow tract during embryogenesis, was observed in the cusps of BAV patients as compared with those of controls. Heterozygous *Nkx2-5*-deficient, *Fgf8*, *Nos3*, and *Gata5* mice are at higher risks for developing BAV. BAV can also manifest as different types of left ventricular outflow tract abnormalities, including aortic coarctation, arch hypoplasia, and supravalvular and mitral valve stenosis. A male predominance of more than 3:1 has been reported for BAV, and this anomaly is very frequent in X0 Turner's syndrome. This talk will give an overview on our present understanding of aortic and aortic valve pathogenesis, particularly in patients with BAV and other connective tissue disorders. We discuss the genetic basis and the basic pathology underlying these disorders. In addition, we discuss recent insights into pathophysiology and treatment.

Biography

Salah A. Mohamed, Laboratory and Group Leader in Department of Cardio and Thoracic Vascular Surgery, University Clinic of Schleswig-Holstein Campus Luebeck, the group dedicated research interests to aging, biomarker, aortic and aortic valve diseases. He has published more than 20 papers in reputed journals and serving as an editorial board member of repute.