Familial bicuspid aortic valve

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Familial bicuspid aortic valve. ORPHA:402075

Familial bicuspid aortic valve is a rare, genetic, aortic malformation defined as a presence of abnormal two-leaflet aortic valve in at least 2 first-degree relatives. It is frequently asymptomatic or may be associated with progressive aortic valve disease (aortic regurgitation and/or aortic stenosis, typically due to valve calcification) and a concomitant aortopathy (i.e. aortic dilation, aortic aneurysm and/or dissection).