
海外招請講演

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New Insights in the Marfan Syndrome

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Marfan syndrome is a connective tissue disorder, caused by mutations in the FBN1 gene encoding fibrillin-1 protein, a structural component of elastic fibers in the tunica media in large arteries. Mortality and morbidity is mainly determined by the development of an aortic aneurysm and subsequent dissection. Prophylactic aortic surgery has increased survival tremendously. In addition, apart from β -blockers the angiotension blocker losartan has recently been shown to slow down aortic dilatation. However, the beneficial effect of losartan appears to be heterogeneous and is more pronounced in patients with a mutation causing haploinsufficiency (mutations resulting in deficient fibrillin-1 protein) compared to patients with a dominant negative mutation (mutations resulting in abnormal fibrillin-1). Around one third of Marfan patients have a haploinsufficiency mutation. They are at increased risk for cardiovascular death, aortic dissection or prophylactic surgery compared to dominant negative patients. So, especially in these haploinsufficient Marfan patients, losartan therapy should be advised both in unoperated patients and after elective aortic root surgery. After aortic root replacement the distal part of the aorta is at increased risk of type B dissection. Type B dissection has become a major problem in these patients since it may occur when the proximal descending aorta is only slightly dilated. Losartan appears to reduce the incidence of type B dissections. In conclusion, for optimal assessment of prognosis and treatment of Marfan patients, more extensive genetic screening, and evaluation of the FBN1 mutation effect on fibrillin-1 protein is warranted. Treatment with losartan seems beneficial in many Marfan patients, but for assessment of the exact role of losartan in Marfan syndrome, the results of running trials should be awaited.