Extended Abstract
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Extent: a completely new method of treatment of patients with Marfan syndrome

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Abstract

Marfan syndrome is the most common genetic disorder of connective tissue. One complication that threatens the lives of patients is progressive dilatation of the ascending aorta with development of aortic valve regurgitation or the emergence of dissection, often leading to sudden death. Until now, these patients were operated only after dilatation of the ascending aorta causing thermodynamically significant regurgitation of the aortic valve. The surgery consisted of the replacement of the ascending aorta and aortic valve or valve sparing procedure. This new method is a preventive operation. The method involves creating a custom made external support of the root and the ascending portion of the aorta. Based on the CT examination, prosthesis Extent is created. The surgery is performed from the longitudinal median sternotomy without cardiopulmonary bypass. Entire aortic root is dissected to its origin from the left ventricle, ostia of the coronary arteries are encircled, the prosthesis is pulled underneath and fixed to the root and then sutured longitudinally. It is interesting that the prosthesis was developed and as the world's first has it sewn on himself (Mr Tal Golesworthy), 13 years ago in Oxford. Neither him nor the other 100 patients operated in this department with this disease had dilation or dissection throughout the study. It is because the prosthesis grows over time into the aortic wall thereby enforces it while maintaining the elastic properties of the wall. At our institute, we have so far experience with operations of 20 patients, with good results. This operation moves the care of patients with Marfan syndrome to qualitatively higher level.

By immunhistochemical studies of Marfan patients it was shown, that most of the patients with MFS display an abnormal pattern of microfibrils in the extracellular matrix. Further studies revealed a 350 kD protein named Fibrillin which was characterised to be the main component of the extracellular microfibrils, to be responsible for these abnormalities. Subsequently the fibrillin gene could be mapped to the long arm of chromosome 15 and in situ hybridisation assigned the gene locus to 15q15-21. However, genetic testing proved to be less reliable than expected, with only 66% mutation detection rate of patients diagnosed with MFS by the Ghent nosology. The reason for these fair results are based on the fact that there are a variable number of mutations, which precludes broad spectrum genetic screening. Furthermore Fibrillin mutations have also been detected in Marfan related disorders.

Further refinements were included in the Gent criteria and recently revised by De Paepe et al in 1996. The criteria are subdivided into the following organ systems: skeletal, ocular, cardiovascular, pulmonary, skin, dura and family respectively genetic history. For a clinical diagnosis of MS, in the absence of a positive family history, a person suspected to have MS should at least display major criteria in two organ systems and involvement of a third organ system. When patients have a positive family history, major criteria in one and minor criteria in another organ system is required.

Particular emphasis should be placed on the predominance of skeletal features in childhood, as this can lead to earlier diagnosis and more appropriate surveillance, of cardiovascular status. In younger patients with a family history of MFS who do no fulfil the diagnostic criteria as well as young patients who fail to meet criteria in one organ system are required to repeat evaluations on a regular basis until the age of 18.

Cardiovascular complications usually do not occur before the age of 16 years. A this point the aortic root dimensions approached the adult size so that one can refer to the recommendations for the adult population regarding aortic root dimensions which should be always measured in relation to body surface area.

Infrequently, the infantile form of Marfan syndrome occurs, which is characterised by mitral valve regurgitation and aortic root dilatation which become apparent during the first three months of life. Mortality is substantial with 14% of affected children dying during the first year of life.