Fabry Cardiomyopathy: Early Detection and Intervention

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Fabry (or Anderson-Fabry) disease is a rare x-linked lysosomal storage disorder leading to an accumulation of globotriaosylceramides in all tissues and organs including the heart. Cardiac involvement is of major prognostic importance in Fabry disease. Left ventricular hypertrophy represents the dominant cardiac feature. The clinical diagnosis must be confirmed by assay of α-galactosidase A activity in leukocytes or plasma and should be followed by molecular genetic analysis. In addition, new biomarkers like LysoGb3 might be helpful for the clinical understanding of the found genetic abnormalities. Specific treatment with enzyme replacement therapy with agalsidase beta (Fabrazyme®, Genzyme, USA) became available since 2001. In early randomized trials using Fabrazyme® a clearance of globotriaosylceramides from the myocardium (mainly the vascular endothelium) could be shown by sequential myocardial biopsies. Furthermore clinical studies showed a reduction of left ventricular wall thickness/mass and improvement of regional myocardial function during enzyme replacement therapy. Lately it became obvious that early therapy at least in male patients might be appropriate. In advanced cardiomyopathies with typically located replacement fibrosis an additional therapy is necessary. This presentation will focus on typically signs of the cardiomyopathy which might be helpful for an early intervention.