

**Cardiac functional assessment in mucopolysaccharidosis**

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**Introduction:** In mucopolysaccharidosis (MPS) cardiovascular involvement includes valvular dysfunction, coronary artery disease and conduction abnormalities, with significant morbimortality. The impact of enzyme replacement therapy (ERT) on cardiac lesions is yet not well defined and, additionally, the decision to undergo surgical procedures may be challenging. There are no reliable parameters to monitor the clinical and therapeutic approach in MPS, nor has the role of tissue Doppler (TD) evaluation in this clinical setting been described. Our aim was to characterize the morphological and functional echocardiographic abnormalities in patients with different types of MPS undertaking ERT.

**Methods:** Cardiovascular studies were performed in eight patients with MPS, including biochemical markers of ventricular dysfunction (BNP, serum sodium, C reactive protein), EKG and morphological and functional echocardiographic cardiac evaluation.

**Results:** Patients aged ranged between four and 23 years old (five males). Six had MPS type VI and two type II, all undergoing ERT. The assessed biochemical parameters were all normal and EKG evaluation showed first-degree atrioventricular block in two patients. Valvular thickening with dysfunction was present in all cases, affecting mainly left heart structures (mitral regurgitation: 8; mitral stenosis: 7; aortic regurgitation: 7). Although only one patient had systolic dysfunction by M-mode evaluation (median LVEF 34%; median LVSF 34.5%), the systolic velocities of the lateral and septal walls of the mitral annulus were reduced (four and five cases, respectively). Regarding LV diastolic function, both Doppler velocities (LV inflow) and TD evaluation (early diastolic velocity of the lateral and septal mitral valve annulus) were suggestive of diastolic dysfunction in six cases. Those with normal function corresponded to the youngest patients, with an earlier beginning of the ERT. With respect to the RV function, all cases had normal systolic function (evaluated by TAPSE and TD systolic velocities of the lateral tricuspid valve annulus). In three cases the RV inflow velocities profile was suggestive of RV diastolic dysfunction and TD abnormalities were found in six patients.

**Conclusions:** MPS patients show a significant rate of cardiac involvement and TD abnormalities seem to be an early finding. Taking this into account, a regular follow up of the affected patients is mandatory.