

Cardiac phenotype and genetic variabilities? An example from beta-thalassemia patients

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Introduction:

New Echocardiographic modalities such as speckle tracking echocardiography (STE) have not only allowed the early detection of myocardial dysfunction in systemic diseases but have allowed as well to distinguish the pattern cardiac affection. Several mutations are characterized within the beta-thalassemia patients.

Methods:

40 beta thalassemia patients were studied pertaining to two groups, Group 1 with thalassemia causing mutation IVSI (Intervening Sequence)-110 and Group 2 with predominant IVSI-6 mutation. Both groups were subjected to cardiac MRI for measurement of cardiac iron load, 2D speckle tracking echocardiography for measurement of global longitudinal strain (GLS) and its endocardial vs. epicardial component

Results:

Group 1 had more cardiac iron load compared to Group 2 (G1:14.1±1.4 vs. G2: 26±2.2, P<0.01), Group 2 had more endocardial longitudinal strain involvement than Group 1 (G1: 20.1±3.2 vs. G2: 12±1.1, P<0.01). Epicardial longitudinal strain was more affected in Group 1 compared to Group 2 (G1:15.2±0.9 vs. G2: 17±3.2, P=0.04). Epicardial GLS was well correlated with myocardial iron load (r=78 %; P<0.001). Epicardial GLS proved 80% sensitivity in predicting IVSI-110 mutation, seemingly Endocardial GLS has recorded 82 % sensitivity in predicting IVSI-6 mutation.

Conclusions:

The aforementioned findings point towards different mechanisms of myocardial injury through different genotypes of Thalassemia, Group 1 develops mainly superficial subepicardial dysfunction due to iron load in the more vascularized layers while Group 2 seem to have more involvement of ischemic Subendocardium by unknown mechanisms. This study underlies as well the possibility of using STE in indirect genetic diagnosis of as displayed in this study.

