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“Cardiac disorders in young patients with beta thalassemia major”

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Beta thalassemia is an inherited disorder, associated with decreased production or absence of the beta chain of the hemoglobin. Regular hemotransfusions lead to temporarily improvement of the clinical condition. Over time, complications of chronic hemolysis and iron overload, such as hemochromatosis, that affects primarily the heart, occur. Survival rates have improved significantly since the introduction of the new iron chelators, but cardiac complications in these patients remain the leading cause of mortality in up to 70% of the patients.

The purpose of the present study is to determine if there are signs of early myocardial involvement, as well as the time of their presentation, in children and young adults with beta thalassemia major. The study will include new echocardiographic techniques and investigation of laboratory expression of certain, selected microRNAs.

MicroRNAs are small, non-coding molecules that take part in normal development of the cardiovascular system, as well as in many pathologic processes. They are stable, resistant and widely distributed in the body fluids that makes them particularly suitable for isolation, identification and research. There are many evidence on the involvement of certain micro-RNAs (miR-1, miR-21, miR-29, miR-150) in the pathogenesis and progression of heart failure, myocytic hypertrophy and apoptosis, interstitial fibrosis, and cardiac remodeling.

Forty young patients with beta thalassemia major will be included in the study. Major tasks include echocardiographic examination of cardiac function, evaluation of laboratory markers for iron overload (ferritin, non-transferrin bound iron), analysis of expression of specific microRNAs, as well as a correlation between the results obtained, transfusion regimens and chelation therapy performed.

The study results will give information about the current cardiovascular status of the youngest patients with beta thalassemia major. This will give an opportunity for identification of higher risk patients for future cardiovascular complications, together with the possibility of therapy optimization. The presence or absence of expression of certain microRNAs, associated with cardiac failure, remodeling and fibrosis, will be study. Given the potential of microRNAs for future biomarkers, we believe that examining their expression in children and young adults with beta thalassemia major will contribute to a better understanding of their involvement in pathologic processes, as well as to a better care for patients starting from early childhood.