P-52

A case of early diagnosis the mucopolysaccharidosis type I (Hurler's syndrome) in 2-month-old girl with acute myocarditis.

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Mucopolysaccharidosis I-Hurler is the most severe form of a metabolic genetic disease caused by mutations of the IDUA gene on chromosome 4p16.3 encoding the lysosomal α L-iduronidase. Acute cardiomyopathy has been found in some infants younger than 1 y.o.
Objective To present a rare clinical case.
Methods Complex inpatient examination.
History of the disease: 2 months old patient was hospitalized to ED with severe cardiac and respiratory insufficiency symptoms, convulsions.
Laboratory findings: high cardiospecific enzymes level (CK 271 U/l, CK-MB 122,5 U/l, LDH 546 U/l, Troponin I 0,2013) and NT-proBNP (82681pg/ml).
ECG and Holter monitoring A multiply atrial tachycardia 195 b.p.m. was recorded.
Echocardiography: LVEDD 39-40 mm. LVEF 20%.
Anomaly of the coronary arteries was excluded by CTA.
Chest X-ray Cardiomegaly (CTR 63.2%).

The management included ALV, tube feeding, intravenous inotropes and loop diuretics, complex HF and antiarrhythmic therapy, intravenous immunoglobulinG.
Positive dynamics was noted in the form of arresting the multi-organ failure syndrome, reducing signs of heart failure.
According to laboratory data-lack of viremia, decrease cardiospecific enzymes' level, NT pro-BNP (9356 pg/ml), normalization level of troponin. By ECHO LVEF was increase to 33-34% and there was't heart chambers size dynamic.
Due to early age of debut, the polysystemic lesion, the absence of positive myocardial remodeling dynamics on the optimal drug therapy lysosomal diseases were included to differential diagnosis.
Results. Low α-L-iduronidase by tandem mass spectrometry were detected. We researched IDUA gene by non-radioactive direct sequencing and found homozygous mutation Gln63Term, that associated with Hurler's syndrome. External manifestations disease were absent until 5 months. There was no family history of cardiac or mitochondrial disease. Specific therapy (Laronidase) was prescribed. Allogenic bone marrow transplantation was performed at 8 month. After transplantation, normalization NT-proBNP (550 pg/ml), increase LVEF to 48%, a decrease LVEDD to 30 mm were registered.
Conclusions. This patient had early clinical debut of the Hurler's syndrome with severe heart failure. The highlights of the case is that we did't see typical clinical manifestations for mucopolysaccharidosis typeI. The key point for effective diagnostics was atypical clinical features of acute myocarditis and genetic testing. Dynamic monitoring of the patient was continued.