

Immunoadsorption therapy for dilated cardiomyopathy: a case report

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Nephronophthisis is the most common monogenic cause of end-stage kidney disease (ESKD) in children. Mutation of the NPHP1 gene can cause several extrarenal manifestations, but it is not known to cause dilated cardiomyopathy (DCM). Also, DCM is not a classic complication of chronic kidney disease (CKD).

In September 2022, a 22-year-old male patient was referred to the North Estonia Medical Centre due to ESKD. Anaemia and secondary hyperparathyroidism had developed as complications of CKD. The patient had previously been examined in Ukraine, but the aetiology of the CKD was unknown. A genetic testing was performed, which revealed a homozygous deletion of the NPHP1 gene, which is disease-related with nephronophthisis. On the basis of echocardiography, DCM was suspected, cardiac ejection fraction (EF) was preserved. Magnetic resonance imaging (MRI) did not reveal the cause of DCM. The patient was referred to the kidney transplant waiting list.

In October 2023, the patient was hospitalized due to pulmonary oedema and acute respiratory failure in the cardiac intensive care unit. Echocardiography demonstrated severe dilation of the left ventricle, EF was 27%. MRI showed no evidence of myocarditis. Since the cardiac function did not improve with conservative treatment, cardiac autoantibodies were measured and five immunoadsorption procedures with intravenous immunoglobulin infusion were performed in March 2024. After a month, the patient's heart function had improved significantly, EF was 49%.

Since studies have proven the effectiveness of immunoadsorption in the treatment of idiopathic DCM, this treatment option should not be overlooked in unclear cases of DCM.