**Conclusion**

Clinical presentation of this anomaly shows great variability and it is difficult to estimate the anatomical progression. Therefore, the treatment of the disease should be personalized and risk score should be formulated for the objective treatment decision but further studies are needed for this.

**Video 1.** Echocardiography showed that excessive right atrial dilation without any tricuspid valve and cardiac anomalies

**Video 2.** During the injection of opaque matter to innominate vein, we detected that enlarged right atrium had became larger than right ventricle as a stomach shape

**References**

The patient presented with fatigue, respiratory distress, chest pain, and 10 kg weight loss in the last 20 days. Vomiting developed five days before admission. His weight was 35 kg (<3rd percentile), height 175 cm (50–75th percentile), head circumference 57.5 cm (>98th percentile), body mass index 9.7 kg/m² (<5th percentile), heart rate 148/min, arterial blood pressure 70/40 mm Hg, and respiratory rate 32/min, and he had a cachectic appearance. Echocardiography revealed severe dilatation of the left ventricle, widespread decrease in contractility, and mild mitral insufficiency (Figs. 1, 2). The left ventricle end-diastolic diameter was 69 mm, ejection fraction 31%, and ventricular shortening fraction 15%. Neurological examination showed cerebellar dysfunction and mild mental retardation.

Complete blood count, sedimentation rate, electrolytes, liver function tests, creatinine kinase, troponin T, thyroid functions, arterial blood gases, serum thiamine, B12, folate, total-free carnitine and acylcarnitine were normal. Viral serologic tests (Coxsackie, adenovirus, Epstein-Barr, cytomegalovirus, and parvovirus B19) were negative. MRI revealed bilateral hyperintense lesions in the frontal cerebral white matter, globus pallidus, and dentate nuclei (Fig. 3).

He was diagnosed with heart failure secondary to DCM. Dobutamine, dopamine, furosemide, and captopril therapy was initiated. L-carnitine (100 mg/kg/day) was instituted. During the follow-up, an increase in urine output and decrease in respiratory distress were observed. In the second week, dobutamine and dopamine were discontinued, and digoxin was added. Urine organic acid analysis demonstrated increased levels of 2-hydroxyglutaric acid (1039 mg/g creatinine, reference: <10 mg/g) and 3-hydroxyglutaric acid (35.6 mg/g creatinine, reference: <5 mg/g). Oral riboflavin (200 mg/day) was prescribed. He showed significant improvement with a good clinical response. In the third week, his clinical status was stable, and echocardiography revealed a 42% ejection fraction. Sequence analysis of the L-2 hydroxyglutarate dehydrogenase (L2HGDH) gene revealed the p.P302L (c.905C>T) mutation. He was diagnosed with heart failure secondary to DCM. Dobutamine, dopamine, furosemide, and captopril therapy was initiated. L-carnitine (100 mg/kg/day) was instituted. During the follow-up, an increase in urine output and decrease in respiratory distress were observed. In the second week, dobutamine and dopamine were discontinued, and digoxin was added. Urine organic acid analysis demonstrated increased levels of 2-hydroxyglutaric acid (1039 mg/g creatinine, reference: <10 mg/g) and 3-hydroxyglutaric acid (35.6 mg/g creatinine, reference: <5 mg/g). Oral riboflavin (200 mg/day) was prescribed. He showed significant improvement with a good clinical response. In the third week, his clinical status was stable, and echocardiography revealed a 42% ejection fraction. Sequence analysis of the L-2 hydroxyglutarate dehydrogenase (L2HGDH) gene revealed the p.P302L (c.905C>T) mutation.

**Discussion**

Dilated cardiomyopathy, a myocardial disorder characterized by a dilated left ventricular chamber and systolic dysfunction that commonly results in congestive heart failure is the most common form of cardiomyopathy. However, understanding the cause of DCM remains difficult, with only 34% of pediatric patients having an identifiable cause. The secondary causes of dilated cardiomyopathy can result from infections, endocrine disorders, neuromuscular diseases and metabolic diseases (1).

L-2 hydroxyglutaric aciduria is an autosomal recessive metabolic disorder characterized by psychomotor delay and cerebellar signs, often associated with macrocephaly. Characteristic MRI findings include subcortical leukencephalopathy, and bilateral nucleus dentatus lesions.

**References**


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**Figure 3. T2-weighted axial slices (A-C), T2-FLAIR coronal (D) sagittal (E) slices in subkortikal deep white matter (A-D), globus pallidus (C, D) dentat nucleus (B, E) hyperintense lesions on MRI**