Danon disease is an extremely rare X-linked cardi-ac and skeletal myopathy that leads to early mortality. We describe the cases of two boys aged 5 and 7 years. Both complained of weakness and fatigue. They were admitted to the Hepatology Department with increased transaminases (>10 N), lactate dehy-drogenase (about 2000 IU/L), creatine kinase (10 N). The preliminary diagnosis of the 5-year-old boy was drug-induced hepatitis (giardiasis had been treated with albendazole), and the 7-year-old boy was diag-nosed with cryptogenic hepatitis.

Examination revealed arrhythmia, WPW syndrome in both. The first signs had manifested 2 years previously. Both cases were misdiagnosed due to increased transaminases. Virologic and parasitologic examina-tions were performed. The phenotype was not specific, with multiple stigmas (low hair growth epikant, palpebral, gothic palate, short neck, wing-folds of the neck), both had equine foot and muscle pseudohyper-trophy. The 7-year-old demonstrated aggressive, devi-ant behavior which had been apparent since the age of 4 years.

Both cases had mild hepatomegaly and hypertrophy of the right atrium and left and right ventricles. The results of KT, US-elastography revealed fibrosis F3 in both. Liver biopsy and electromyography were performed in the 5-year-old. PAS positive reaction – a glycogen like substance was found in hepatocytes, which looked like plant cells with fat vacuolization. Electromyography was subnormal. Creatine kinase-MB was 10–20 N, NT-proBNP was >25,000 IU/L.

The mother of the younger boy had a myopy. Within 1 year, she had born a second boy and had refused any investigations.

Pompe disease was excluded by the normal activity of acid maltase. Normal enzyme activity also exclud-ed Gaucher and Fabry diseases.

Danon disease was confirmed by the presence of X-linked mutations in LAMP-2 in the 5-year-old. The 7-year-old was diagnosed with Danon disease only by normal acid maltase and phenotypically. Both pa-tients were referred to a cardiologist and were cured. Only cardiotrophic and symptomatic therapy were used. Limited physical activity was recommended.

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