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Poster Abstract: Therapeutic

Two Cases of Danon Disease – A 'Cousin' of Pompe Disease

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Danon disease is an extremely rare X-linked cardiac 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 dehydrogenase (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 diagnosed 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 examinations 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 pseudohypertrophy. The 7-year-old demonstrated aggressive, deviant 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 excluded 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 patients were referred to a cardiologist and were cured. Only cardiotrophic and symptomatic therapy were used. Limited physical activity was recommended.

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