УДК 61:615.1(062)(476-25)
ББК 52я73
А 43
ISBN 978-985-21-1008-2

Jadczak K., Pieńkowska A., Franaszek K.

WHY EARLY ONSET FORM OF GLYCOGEN STORAGE DISEASE TYPE II IS UNPREDICTABLE? - CASE REPORT

Tutor MD Kadłubiska A., MD, PhD Bossowski A.
Department of Pediatrics, Endocrinology, Diabetology with Cardiology Division
Medical University of Bialystok, Bialystok

Glycogen-storage disease type II, also known as Pompe's disease is a genetically determined disorder, inherited in an autosomal recessive manner with a prevalence of 1:40,000-1:50,000 live births. The disease is characterised by the deficiency of the enzyme - acid alpha-glucosidase, that degradate glycogen. Its storage contributes to cell damage, mainly muscle. Based on age, an early onset form and a delayed onset form are distinguished.

11-year-old female patient was diagnosed with early-onset Pompe disease at 3 months old. The girl is on a treatment program for Pompe disease - every 2 weeks Myozyme infusions, started in the 4th month of her life. The patient is under the care of the Department of Pediatrics, Endocrinology, Diabetology and Cardiology, Medical University of Bialystok. She was first hospitalized on the 15th day of life due to tachycardia. Physical examination revealed decreased muscle tone and liver enlargement. Electrocardiogram showed paroxysmal ventricular tachycardia and features of left ventricular hypertrophy. Additional tests revealed elevated transaminases levels. Echocardiogram showed features of hypertrophy of both heart chambers. Based on an overall assesment a metabolic background was suggested. During 4-week diagnostics in Children's Health Centre in Warsaw Pompe disease was diagnosed. Since the beginning of the therapy with Myozyme, a slowdown of the disease progression has been observed. Due to numerous infections along with the progression of the disease required the placement of transcutaneous endoskopic gastrostomy and tracheostomy with respiratory support.

Early diagnosis with subsequent enzyme replacement therapy, despite its beneficial effect in slowing the disease progression and prolonging patients' lives, will not completely stop the disease progression. For this reason, it is indicated to extend the diagnostics in neonates with features of generalized flaccidity and cardiomyopathy by dry blood drop testing and performing genetic tests. Specific emphasis should be placed on the prevention of infectious diseases through vaccination of patiens and close contacts.