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Co-occurrence of Glycogen Storag Disease Type 2 and Congenital Myasthenic Syndrome Type 5 in a Pediatric Patient

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Glycogen storage disease type 2 (also known as Pompe disease) is a metabolic disorder characterized by an accumulation of glycogen within lysosomes. Pathophysiologically, this condition is caused by an autosomal recessive deficiency of the lysosomal acid alpha-glucosidase enzyme, resulting in defects in lysosomal metabolism. Glycogen accumulation causes advanced muscle weakness (myopathy) throughout the body, including the heart, skeletal muscles, liver, and the neurological system. Currently, there is no definitive treatment for Pompe disease. However, recent studies have indicated that enzyme replacement therapy (ERT) can be effective. Myasthenia gravis (MG) is an autoimmune illness that affects the postsynaptic acetylcholine receptors and causes fatigue that can be eased by rest. MG is frequently accompanied by a thymoma. Dyspnea and/or bulbar symptoms can indicate an imminent crisis requiring immediate intervention. Here, we present a rare case of a four-year-old female patient who initially presented at the age of one month with the infantile form of Pompe disease and congenital myasthenia syndrome type 5. The patient presented with bradycardia, poor suckling, respiratory distress, and respiratory failure requiring assisted ventilation, subglottic stenosis, and tachypnea. Whole exome sequencing was used for definitive diagnosis. ERT (Myozyme) was administered with good results. We propose that early identification and management of Pompe disease with Myozyme can improve patients' condition and ultimately increase the possibility of survival.

Biography

Raneem Y. Gazzaz is a Faculty of Medicine, and has experience of 4years in teaching from Batterjee Medical College, Jeddah, SAU and wrote, many articles on pediatric medicine