Providing anaesthesia for caesarean section surgery to a pompe disease patient

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Background. Pompe disease, alternatively recognized as an acid maltase deficiency or glycogen storage disease type II, is a genetic disease transmitted in autosomal-recessive way. It emerges due to a shortage of the enzyme acid-A-glucosidase within lysosomes. Consequently, PD leads to the buildup of glycogen in multiple body tissues, particularly in skeletal, cardiac, and smooth muscles. The morbidity of PD varies depending on patient's age when the symptoms first appear, the degree of complexity of harm to skeletal, cardiac, and respiratory muscles, and the speed of disease progression.

Case presentation. A 29-year-old pregnant woman, known to have juvenile pompe disease, was admitted for a scheduled caesarean section. Her symptoms first emerged when she was 20 years old. She commenced enzyme replacement therapy with alglucosidase alpha, administered intravenously at a dose of 20 mg/kg over a 5-hour infusion every 2 weeks. The pregnancy was not planned, yet the patient consistently received enzyme replacement therapy throughout the gestation period. However, pulmonary function tests conducted while standing and sitting indicated significant restrictions.

Conclusions. In 1963, the connection between the inherited shortage of the lysosomal enzyme acid A-glucosidase and PD was initially established. This particular enzyme plays a vital role in breaking down glycogen into glucose. Insufficient GAA results in glycogen buildup within lysosomes, primarily within muscle cells, triggering a gradual decline in muscle function.