



Effects of enzyme replacement therapy on the respiratory & motor functions among patients with late-onset Pompe disease for long-term

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Background: Pompe is a congenital metabolic disorder and muscles involvement is the most common presentation of it. The late-onset Pompe disease is a type of Pompe according to the age of clinical presentation. In LOPD, the muscles of respiratory system, trunk and proximal limbs involved. Enzyme replacement therapy is a new method for treatment of these patients, but the efficacy and safety of this method are challenging for long-term so that in this pilot study efficacy and safety were evaluated for two years.

Methods: In this prospective study, the patients with LOPD included this study, and they received rhGAA every two weeks for two years, and the motor and respiratory function of them were evaluated. Also, CPK and LDH were measured every six months. This study was registered as a trial and approved by the ethics committee of Tehran medical sciences.

Results: Two adults who were suffering LOPD for 17 and 12 years were investigated in this study. Both of them did not show any side effects, also the motor function got slightly better, and one of them could walk for more distance, on the other hand, the respiratory function was slightly improved and the CPK levels dramatically reduced.

Conclusion: The results of this study showed the ERT was safe and effective for control the disease activity of patients with LOPD, but this method is so expensive special for long-term. This is a small pilot study and needs more.