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# A Systematic Literature Review of Enzyme Replacement Therapy in Early and Late-Onset Pompe Disease

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**Introduction:** Pompe Disease (PD) is characterized by a deficiency of acid alpha-glucosidase, leading to progressive glycogen accumulation in the tissues. Previous systematic review (SR) on early-onset PD (EOPD) and on late-onset PD (LOPD) haven't evaluated important endpoints for enzyme replacement therapy (ERT) such as safety, thus creating the need for reassessing clinical outcomes. **Objective:** To evaluate efficacy and safety of Alglucosidase alfa for EOPD and LOPD. **Methods:** We systematically searched PubMed and Embase for prospective clinical studies published until September 2018 evaluating ERT for EOPD and LOPD. Outcomes of interest were defined a priori. Assessment of quality of evidence (QOE) was performed according to the GRADEpro criteria. **Results:** In EOPD, a total of 1470 articles were identified and, after a selection, 13 articles were included in our analysis. Only 2/9 analyzed endpoints had high GRADEpro scores favoring ERT. Survival was evaluated in 6/13 studies and increased in all of them (GRADEpro moderate). Time to Onset Ventilation (TOV) was evaluated in 7/13 studies and 4/7 showed its increase by ERT (GRADEpro moderate). Cardiomyopathy was evaluated in 10/13 by left ventricular mass, reduced in 10/13 studies after ERT (GRADEpro very low). Safety data were described in 6/13 studies (GRADEpro low) and all analyzed antibody formation, present in at least 85% of patients evaluated. In LOPD, a total of 1172 articles were identified and only 23 of these were included in our analysis. Only 4/10 endpoints analyzed had moderate or high GRADEpro scores. Quality of Life (QOL) was evaluated in 6/23 studies and 5/6 used the SF-36 questionnaire. Despite heterogeneous results across studies, GRADEpro was moderate and favors ERT. Patients under ERT showed improvement in all 12 studies evaluating 6MWT, with high QOE. Muscle strength (MS) was evaluated in 8/23 included studies and only 2/8 were unable to show improvement after ERT, also with high QOE. In 4/5 studies antibody formation was analyzed and all patients developed antibodies. In both, antibody titers were not correlated with severe adverse events (AEs) or infusion-associated reactions (IARs) nor were associated with treatment efficacy and clinical outcomes. Most IARs were mild to moderate in severity in both cases. **Discussion:** Our results add information over previous published SR on ERT, as include data from observational prospective studies, showing benefit for survival, TOV and cardiomyopathy in EOPD, whereas in LOPD the results corroborate previously published SR on ERT impact on 6MWT in LOPD and show positive effect of ERT on QOL and MS in LOPD. Our findings also suggest that ERT is safe in EOPD and in LOPD, once most AEs were mild to moderate and antibody formation did not seem to interfere with any outcome evaluated.