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Title: Improving respiratory muscular strength in Pompe disease late-onset with muscles respiratory training

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Body: BACKGROUND: Glicogenosis type II late onset or Pompe disease is a genetic disorder due to lysosomial enzyme deficiency: acid α-glucosidase. This deficiency leads to the accumulation of glycogen in muscle tissue that causes a progressive debilitation of the muscles until a severe hypoventilation. The weakness of respiratory muscles is diagnosed by functional respiratory tests (FVC in clino and orthostatism, MIP and MEP), sleep disorders study and blood gas analysis.(1) The main therapeutic option is the administration of surrogate enzyme. Despite a single report in literature, we don't know if respiratory muscle rehabilitation or training could bring any real benefits.(2) OBJECTIVE: We want to evaluate effectiveness of respiratory muscles training using a specific appliance with adjustable resistance (Threshold). First endpoint is muscular strength of respiratory brawn by MIP and MEP measurements. METHODS: We studied 7 patients, aged between 13 and 58 years, affected by Pompe desease late onset with respiratory muscles deficiency on functional respiratory tests. All patients were taking sostitutive enzyme therapy and underwent training with Threshold at specified pressures. During follow up they performed the measurement of MIP and MEP. RESULTS: The values of MIP and MEP increased on average of 16.2% ($p<0.05$) after 3 months and of 21% ($p<0.05$) after 20 months of treatment in our group of patients. CONCLUSION: Our study suggests that the training with Threshold, used in addition to enzyme therapy, could improve respiratory muscular strength in Pompe disease.