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- Aims and Scope
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- Author Information
- Abstracting/Indexing
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- Pricing Information

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◀ previous 4 of 17 next ▶

Eight years experience with enzyme replacement therapy in two children and one adult with Pompe disease

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Abstract

Pompe disease (type 2 glycogenosis, acid maltase deficiency) is a disorder affecting skeletal and cardiac muscle, caused by deficiency of acid α -glucosidase. In 2006 enzyme therapy with recombinant human α -glucosidase received marketing approval based on studies in infants. Results in older children and adults are awaited. Earlier we reported on the 3-year follow-up data of enzyme therapy in two adolescents and one adult. In the present study these patients were followed for another 5 years.

Two severely affected patients, wheelchair and ventilator dependent, who had shown stabilization of pulmonary and muscle function in the first 3 years, maintained this stabilization over the 5-year extension period. In addition patients became more independent in daily life activities and quality of life improved.

The third moderately affected patient had shown a remarkable improvement in muscle strength and regained the ability to walk over the first period. He showed further improvement of strength and reached normal values for age during the extension phase.

The results indicate that both long-term follow-up and timing of treatment are important topics for future studies.

Keywords: [Pompe disease](#), [Acid maltase deficiency](#), [Glycogenosis type 2](#), [Enzyme replacement therapy](#)

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