

## References – Myozyme

1. Kishnani P.S. et al. Recombinant human acid-glucosidase: Major clinical benefits in infantile-onset Pompe disease. *Neurology* 2007;68:1-1.
2. Kishnani PS, Hwu W, Mandel H, et al. A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. *J Pediatr* 2006;148:671-6.
3. Kishnani PS, Howell RR. Pompe disease in infants and children. *J Pediatr* 2004;144:S35-43.
4. Kishnani PS, Steiner RD, Bali D, et al. Pompe disease diagnosis and management guideline. *Genet Med* 2006;8:267-88.
5. Kishnani P, Byrne B, Nicolino M, et al. Enzyme replacement therapy (ERT) with recombinant human acid alpha glucosidase (rhGAA) in infantile-onset Pompe disease: interim results from a pivotal trial. 55th Annual Meeting of the American Society of Human Genetics, Salt Lake City, Utah, October 25-29, 2005. (abstract no.199)
6. Kishnani P, Byrne B, Nicolino M, et al. Enzyme replacement therapy (ERT) with recombinant human acid alpha glucosidase (RHGAA) in infantile onset Pompe disease (IOPD). SSIEM 42st Annual Symposium, Paris, France, September 6-9, 2005. (abstract no. 387-O)
7. Nicolino M, Kishnani P, Spencer C, et al. Safety and efficacy of CHO-cell derived recombinant human acid alpha glucosidase (rhGAA) in patients with infantile-onset Pompe disease (IOPD) treated after 6 months of age. Second Annual Symposium on Lysosomal Storage Disorders, Athens, Greece, March 2-4, 2005. (abstract)
8. Kishnani P, Spencer C, Nicolino M, et al. Safety and efficacy of CHO-cell derived recombinant human acid alpha glucosidase (rhGAA) in patients with infantile-onset Pompe disease (IOPD) treated after 6 months of age. ACMG Annual Clinical Genetic Meeting, Dallas, Texas, March 17-20, 2005. (abstract 55)
9. Kishnani P, Spencer C, Byrne B, et al. Long term efficacy of enzyme replacement therapy (ERT) in children with Pompe disease. 2006 Annual Clinical Genetics Meeting, American College of Medical Genetics, San Diego, California, March 23-26, 2006. (abstract)
10. Kishnani PS, Nicolino M, Voit T, et al. Chinese hamster ovary cell-derived recombinant human acid -glucosidase in infantile-onset Pompe disease. *J Pediatr* 2006;149:89-97.

11. Cook AL, Kishnani PS, Carboni MP, et al. Ambulatory electrocardiogram analysis in infants treated with recombinant human acid  $\alpha$ -glucosidase enzyme replacement therapy for Pompe disease. *Genet Med* 2006;8:313-7.
12. Ansong AK, Li JS, Nozik-Grayck E, et al. Electrocardiographic response to enzyme replacement therapy for Pompe disease. *Genet Med* 2006;8:297-301.
13. Chien Y, Lee N, Peng S, et al. Brain development in infantile-onset Pompe disease treated by enzyme replacement therapy. *Pediatr Res* 2006;60:349-52.
14. Amalfitano A, Bengur AR, Morse RP, et al. Recombinant human acid  $\alpha$ -glucosidase enzyme therapy for infantile glycogen storage disease type II: Results of a phase I/II clinical trial. *Genet Med* 2001;3:132-8.