Appendix D. Summaries of Unpublished Studies

**Appendix Table D1. Unpublished Studies From Manufacturer’s Scientific Information Packet and Current Registered Clinical Trials**

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| **Disease** | **Product**  | **Manufacturer** | **Posters** | **Abstracts** | **Data on File With Manufacturer** | **Ongoing Studies** |
| Fabry’s Disease | Fabrazyme® (agalsidase beta) | Genzyme Corporation | Not reported | Not reported | Not reported | **2010:****NCT01196871:** Drug-Drug Interaction Study Between AT1001 and Agalsidase in Subjects With Fabry Disease**NCT01218659:** Study to Compare the Efficacy and Safety of Oral AT1001 and Enzyme Replacement Therapy in Patients With Fabry Disease**2007:****NCT00455104:** Canadian Fabry Disease Initiative (CFDI) Enzyme Replacement Therapy (ERT) Study*(Status has not been verified in more than two years)* **NCT00487630:** Evaluation of Efficacy and Safety of Agalsidase Beta in Heterozygous Females for Fabry Disease (HEART)**2005:****NCT00196742:** Fabry Disease Registry**NCT00230607:**A Study of the Effects of Fabrazyme (Agalsidase Beta) on Mother's Lactation and on the Growth, Development and Immunologic Response of Their Infants |
| Gaucher Disease Type I | Ceredase® (alglucerase) | Genzyme Corporation | Not reported | Not reported | Not reported | **2006:****NCT00302146:** Positron Emission Tomography (PET) Imaging in People With Gaucher Mutations |
| Cerezyme® (imiglucerase) | Genzyme Corporation | Not reported | Not reported | Not reported | **2011:****NCT01344096:** Thrombocytopathy in Gaucher Disease Patients |
| Velaglucerase® (velaglucerase alfa) | Shire Human Genetic Therapies Inc | **2008:**Zimram A, Altarescu G, Phillips M, Bhirang K, Mensah R, Elstein D. Velaglucerase alfa: a Phase I/II long-term study of enzyme replacement therapy (ERT) in patients with type 1 Gaucher disease [poster]. Presented at: Annual Meeting of the American Society of Human Genetics; November 11-15, 2008: Philadelphia, PA. | **2010:**Zimran A, Gonzalez D, Crombez E, et al. Enzyme replacement therapy with velaglucerase alfa improves key clinical parameters in a pediatric subgroup with type 1 Gaucher disease [abstract]. Presented at: World Symposium 2010; the Annual Meeting the Lysosomal Disease Network; February 10-12, 2010c; Miami, FL.Zimram A, Gonzalez D, Lukina EA, et al. Enzyme replacement therapy with velaglucerase alfa significantly improves clinical parameters in type 1 Gaucher disease: positive results from a randomized, double-blind, global, phase III study [abstract]. Presented at: World Symposium 2010, the Annual Meeting of the Lysosomal Disease Network; February 10-12, 2010b; Miami, FL. | **2009:**A multicenter, randomized, double-blind, parallel-group, two-dose study of gene-activated human glucocerebrosidase (GA-GCB) enzyme replacement therapy in patients with type 1 Gaucher disease. Clinical Study Report: TKT032, Cambridge, MA; Shire Human Genetic Therapies; Jul 2009A multicenter, randomized, double-blind, parallel-group study of gene-activated human glucocerebrosidase (GA-GCB) enzyme replacement therapy compared with imiglucerase in patients with type 1 Gaucher disease. Clinical Study Report: HGT-GCB-039, Cambridge, MA; Shire Human Genetic Therapies; Aug 2009.A multicenter open-label study of gene- activated human glucocerebrosidase (GA-GCB) enzyme replacement therapy in patients with type 1 Gaucher disease previously treated with imiglucerase. Clinical Study Report: TKT034, Cambridge, MA; Shire Human Genetic Therapies; Aug 2009.**2006:**A phase I/II safety study of velaglucerase alfa, a glucocerebrosidase replacement therapy in patients with type 1 Gaucher Disease. Clinical Study Report: TKT025, Final Version 1.0 Cambridge, MA; Shire Human Genetic Therapies; Jun 2006. | **2011:****NCT01356537:** Home Therapy With VPRIV in Gaucher's Disease |
| Zavesca® (miglustat) | Actelion Pharmaceuticals | Not reported | Not reported | Not reported | *(Status has not been verified in more than two years)* **2007:****NCT00418847:** Pharmacokinetics and Tolerability of Zavesca® (Miglustat) In Patients With Juvenile GM2 Gangliosidosis |
| Glycogen Storage Disease Type II (Pompe disease) | Myozyme® (alglucosidase alfa) | Genzyme Corporation | Not reported | Not reported | Not reported | **2011:****NCT01288027:**Exploratory Muscle Biopsy Assessment Study in Patients With Late-Onset Pompe Disease Treated With Alglucosidase Alfa**NCT01410890 :** Pharmacokinetics of Alglucosidase Alfa in Patients Aged 8-18 Years of Age (PAPAYA)**2008:** **NCT00701701:** Immune Tolerance Induction Study**2007:** **NCT00486889:** Growth and Development Study of Myozyme (Alglucosidase Alfa).**NCT00566878:** Pompe Lactation Sub-Registry**NCT00567073:** Pompe Pregnancy Sub-Registry |
| MPS I (Hurler disease) | Aldurazyme® (laronidase) | Genzyme Corporation | Not reported | Not reported | Not reported | **2009:****NCT00852358:** A Study of Intrathecal Enzyme Therapy for Cognitive Decline in MPS I**2008:****NCT00638547:** Intrathecal Enzyme Replacement for Hurler Syndrome**NCT00741338:** Immune Tolerance Study With Aldurazyme®**2007:****NCT00418821:** A Study of the Effect of Aldurazyme® (Laronidase) Treatment on Lactation in Female Patients With Mucopolysaccharidosis I (MPS I) and Their Breastfed Infants**2005:****NCT00144768:** A Study Investigating the Relationship Between the Development of Laronidase Antibody and Urinary GAG (Glycosaminoglycan) Levels in Aldurazyme® Treated Patients**NCT00144794:** Mucopolysaccharidosis I (MPS I) Registry |
| MPS II (Hunter disease) | Elaprase® (idursulfase) | Shire Human Genetic Therapies Inc | Not reported | Not reported | Not reported | **2011:****NCT01330277:** Biomarker for Hunter Disease (BioHunt)**NCT01506141:** An Extension Study of HGT-HIT-045 Evaluating Long-Term Safety and Clinical Outcomes of Idursulfase (Intrathecal)in Conjunction With Elaprase® in Pediatric Patients With Hunter Syndrome and Cognitive Impairment**2009:****NCT00920647:** A Safety and Dose Ranging Study of Idursulfase (Intrathecal) Administration Via an Intrathecal Drug Delivery Device in Pediatric Patients With Hunter Syndrome Who Have Central Nervous System Involvement and Are Receiving Treatment With Elaprase® **NCT00937794:** A Screening Study to Identify Pediatric Patients With Hunter Syndrome Who Demonstrate Evidence of Central Nervous System (CNS) Involvement and Who Are Currently Receiving Treatment With Elaprase®**NCT01449240:** Collection and Study of Cerebrospinal Fluid in Patients With Hunter Syndrome |
| MPS VI (Maroteaux- Lamy syndrome)  | Naglazyme® (galsulfase) | BioMarin Pharmaceutical Inc | **2011:**Kim KH, Burton BK. Treatment with galsulfase results in improved endurance in a MPS VI patient with history of bone marrow transplantation in early childhood. 61st Annual Meeting of the American Society of Human Genetics (ASHG). Montreal, Canada. 11-15 October 2011. Poster.M. L. Raff. Galsulfase enzyme replacement therapy improves urine GAG excretion and clinical course in Maroteaux-Lamy syndrome (MPS type VI) after donor-engrafted bone marrow transplant. Genomics Institute, MultiCare Health System, Tacoma, WA. 14 October 2011. Poster.**2010:**\*Acosta A, Giuliani L, Horovitz D, et. al. Experience with enzyme replacement therapy on very young mucopolysaccharide and Related Diseases. Adelaide, Australia: 23-27 June 2010. Poster.Ribeiro EM, Bezerra KRF, Giovannetti D, et al. Enzyme replacement therapy in mucopolysaccharidosis VI: early treatment with galsulfase in three siblings. 11th International Symposium on Mucopolysaccharide and Related Diseases. Adelaide, Australia: 23-27 June 2010. Poster.**2008:**Lampe C, Miebach E, Arash L, et al. Therapeutic response after two years of Galsulfase enzyme replacement therapy (ERT) in five adult patients with Maroteaux-Lamy syndrome. Poster, ASHG 58th Annual meeting. Philadelphia, Pennsylvania, 11-15 November 2008 | **2010:** Braunlin E, Howard R, Christoph K, et al. Long term cardiac effects of Naglazyme(galsulfase) therapy (NRx). 11th International Symposium on Mucopolysaccharide and Related Diseases. Adelaide, Australia: 23-27 June 2010. Abstract. Decker C, Devereaux D, Kim S, et al. Analysis of the clinical impact of immune response to enzyme replacement therapy with naglazyme. 11th International Symposium on Mucopolysaccharide and Related Diseases. Adelaide, Australia: 23-27 June 2010. Abstract.\*Harmatz P, Guffon N, Garcia P, Cheng S, Lagan K, Decker C. A Phase 4 two dose level study of galsulfase in Mucopolysaccharidoses IV infants. J Inherit Metab Dis (2010) 33 (Suppl 1):S1–S197. Abstract.Horovitz DDG, Magalhaes T, Acosta A, et. al. Enzyme replacement therapy in 25 mucopolysaccharidosis type VI Brazilian children under age five. 11th International Symposium on Mucopolysaccharide and Related Diseases. Adelaide, Australia: 23-27 June 2010. Abstract 103.**2009:**Horovitz DDG, Ribeiro EM, Acosta A, et al. Enzyme replacement therapy in eight mucopolysaccharidosis type VI Brazilian children under age three: preliminary data. 11th International Congress on Inborn Errors of metabolism. San Diego, CA: 29 August - 02 September 2009. Abstract.Ospina S, Benavidez R, Giovannetti D, et al. Maroteaux lamy syndrome enzyme replacement therapy: outcome in a severe form. 11th International Congress on Inborn Errors of Metabolism. San Diego, CA: 29 August - 02 September 2009. Abstract.Sohn YB, Park SW, Kim SJ, et al. Enzyme replacement therapy in a mucopolysaccharidosis type VI patient who was previously treated with bone marrow transplantation. 11th International Congress on Inborn Errors of metabolism. San Diego, CA: 29 August-02 September 2009. Abstract 348.Solano M, Marquez W, Ospina S, et al. Post anesthetic recovery and surgical procedure in 4 patients with MPS VI under ERT. 11th International Congress on Inborn Errors of Metabolism. San Diego, CA: 29 August -02 September 2009. Abstract.Solano ML, Nunez LC, Villamizar I. Severe cardiomyopathy is reverted in patient with advanced MPS VI under ERT. 11th International Congress on Inborn Errors of Metabolism. San Diego, CA: 29 August-02 September 2009. Abstract 426.**2008:**Harmatz P, Giugliani R, Schwartz I, et. al. Enzyme replacement therapy for mucopolysaccharidosis vi: improvement of pulmonary function relative to growth in patients treated with recombinant human N-acetylgalactosamine 4-sulfatase. 10th International Symposium on MPS and Related Diseases. Vancouver, Canada: 26-29 June 2008. AbstractHarmatz P, Wen A, et al. Tracheostomy reversal in an MPS VI patient due to improved pulmonary function while on enzyme replacement therapy : a case study. 15th Annual Meeting of the American College of Medical Genetics (ACMG) Phoenix, AZ: 12-16 March, 2008 Abstract.Valayannopoulos V, Farr M, Tuberville S, et al. A follow-up of enzyme replacement therapy in two MPS VI patients’ with poorly engrafted bone marrow transplantation. 58th Annual meeting of the American Society of Human Genetics (ASHG) Annual Meeting. Philadelphia, PA: 11-15 November 2008. Abstract.**2007:**Magalhaes A, Teles E, Breda J, et al. Ophthalmologic evaluation of MPS VI patients following treatment with galsulfase enzyme replacement therapy. 2007 Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM). AbstractMiebach E, Thuemler A, Arash L, et. al. Adult patients with mucopolysaccharidosis VI. 2007 Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM). Abstract.Munoz M.V.R, Horovitz D, Vieira T, Costa R, Vedolin L, Fagondes S, Jardim L, Lierena J, Giugliani. Intrathecal Enzyme Replacement Therapy in a child with mucopolysaccharidosis VI and symptomatic spinal cord compression. Medical Genetics Service. Hospital de Clinicas de Porto Alegre, Porto Alegre, RS, Brazil 2007. Abstract. \*Sandberg S, Charnas L, Braulin E, et al. Treatment of multiple sulfatase deficiency with recombinant human arylsulfatase B. Mol Genet and Metab 2007; 92:S11-S34. Abstract 99.**2002:**Harmatz P, Whitley CB, Waber L, et.al. A Phase 1/2 study of enzyme replacement therapy (ERT) for mucopolysaccharidosis VI(MPS VI; Marteaux-Lamy syndrome): 48 week progress report. 52nd Annual Meeting of the American Society of Human Genetics (ASHG) Annual Meeting. Baltimore, MD: 15-19 October 2002. Abstract 2418 |  | **2005:****NCT00214773:** Mucopolysaccharidosis (MPS) VI Clinical Surveillance Program (CSP) |