Pompe disease is caused by pathogenic variations in the acid alpha-glucosidase (GAA) gene. Close to 500 different GAA gene variations have been identified in families with this disorder.

Glycogen Storage Diseases are devastating, rare orphan diseases for which there is no cure and treatment regimes are suboptimal. The two most common ones are Von Gierke’s and Pompe’s disease. Myozyme and Lumizyme are two approved treatments for Pompe’s (Sanofi-Genzyme) that are somewhat effective, but not curative.

Here at CBI we have had the privilege to work on a clinical product for a Glycogen Storage Disease in which we developed unique methods and conducted histology, developed a very high quality PAS EPON method to stain thin section muscle biopsies and conducted transmission electron microscopy on patient samples.

Below are some representative histology, thin sections and TEM of GSD specimens that we prepared and evaluated in our laboratory. Digital image analysis and histo-morphometry were conducted on the TEM samples below.

**Pompe Disease**

Histopathology on Human Muscle Biopsies from patients affected with a Glycogen Storage Disease.

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**TEM micrograph showing ultrastructural changes in the lysosomes typical of Pompe’s Disease.**

**PAS on ultrathin plastic sections of a Pompe’s patient.**

**PAS on ultrathin plastic sections of a severely affected Pompe’s patient.**

**Glycogen Storage Disease**

Histopathology on Human Muscle Biopsies from patients affected with a Glycogen Storage Disease.

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Be sure to check out the movie: **EXTRAORDINARY MEASURES**

The film is about parents who form a biotechnology company to develop a drug to save the lives of their children, who have a life-threatening disease. The film is based on the true story of John and Aileen Crowley, whose children have Pompe’s disease.

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