

We will start momentarily at 2pm ET



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National Chemistry Week

October 17-23, 2010



Theme: Behind the Scenes With Chemistry

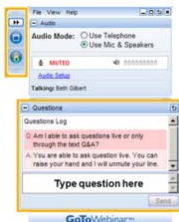
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ACS WEBINARS™ September 9, 2010



How Scientific Skills are Used in Advising the Movie Industry – Facts and Fiction



Speaker: Barry Byrne, Ph.D.
University of Florida



Moderator: Mark Griep, Ph.D.
University of Nebraska-Lincoln

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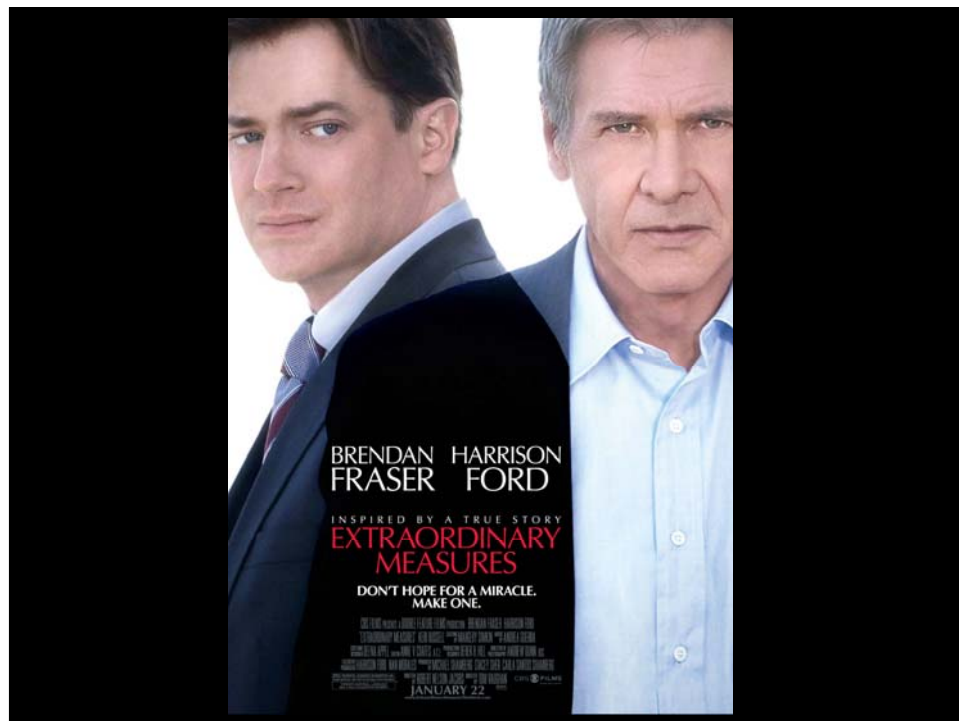
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*Treatment Strategies for Pompe Disease:
A Paradigm for Molecular Medicine*

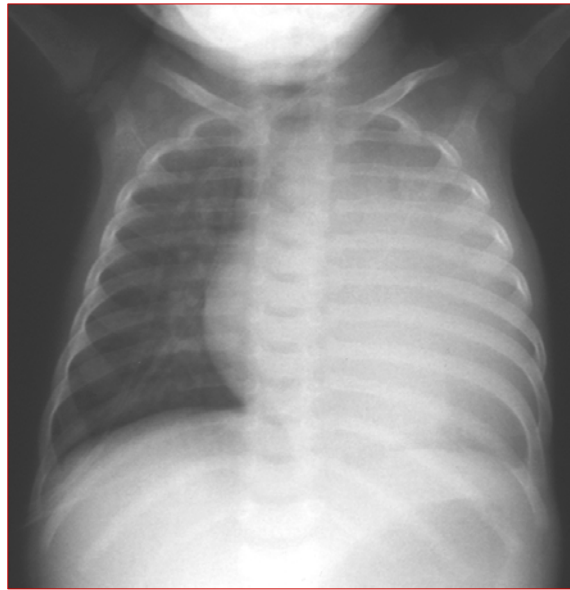
Barry J. Byrne, MD, PhD
Director, Powell Gene Therapy Center

Hung Do, PhD





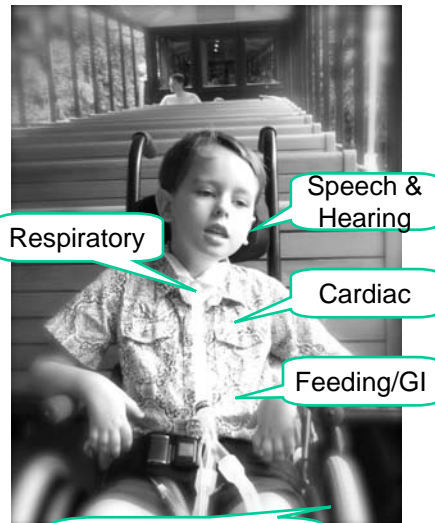
***Initial Presentation of Pompe Disease:
Fatal Cardiomyopathy in Infancy***





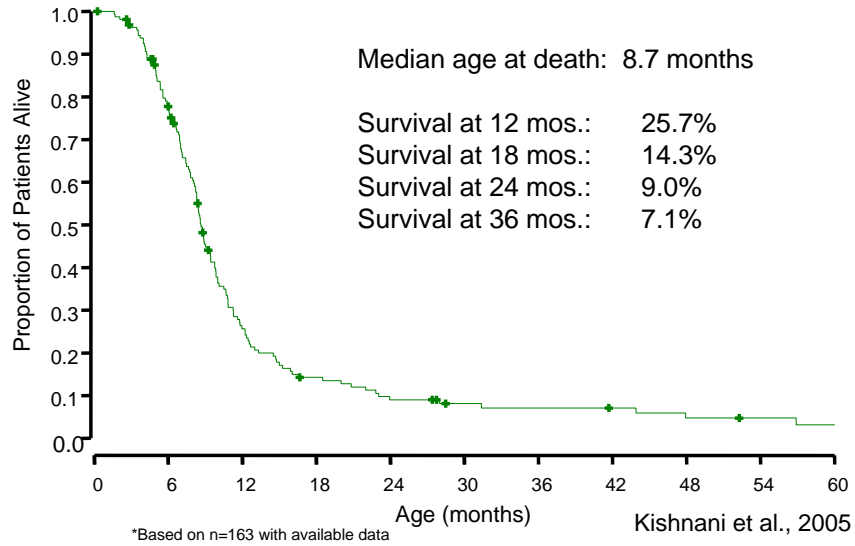
Background: Spectrum of Disease

- Mutations in GAA gene with lysosomal accumulation of glycogen
 - Loss of myofibrils and generalized weakness
 - 40% of wt activity is protective
- Heart disease: glycogen accumulation leading to increased cardiac mass and heart failure
- Musculoskeletal: severe weakness with early loss of motor milestones
- Respiratory disease: progressive loss of independent ventilation
 - 22/38 subjects in ERT studies are now using assisted ventilation or have died
 - All patients have a functional deficits in respiratory function

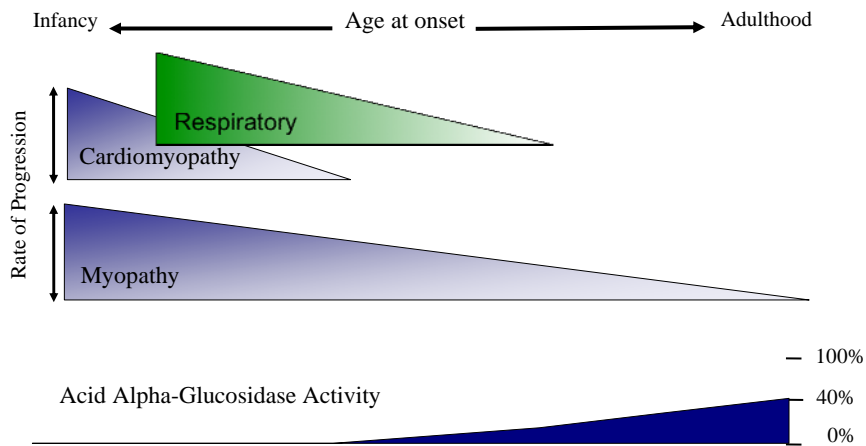


Musculoskeletal

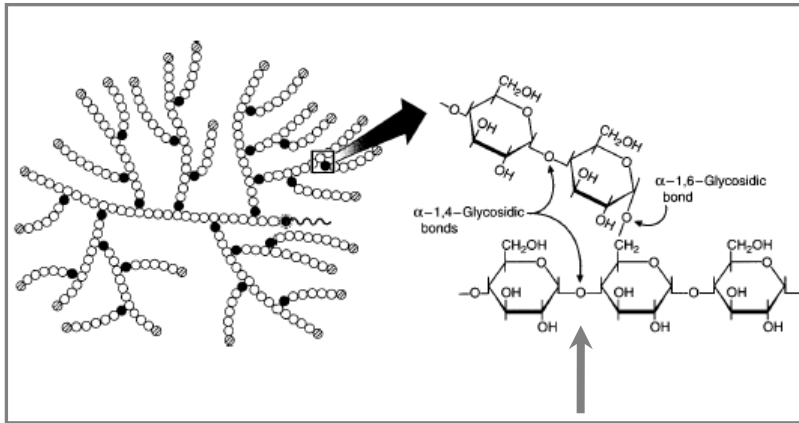
**Infantile-Onset Pompe Natural History Study:
Kaplan-Meier Plot of Time to Death***



Pompe Disease Spectrum



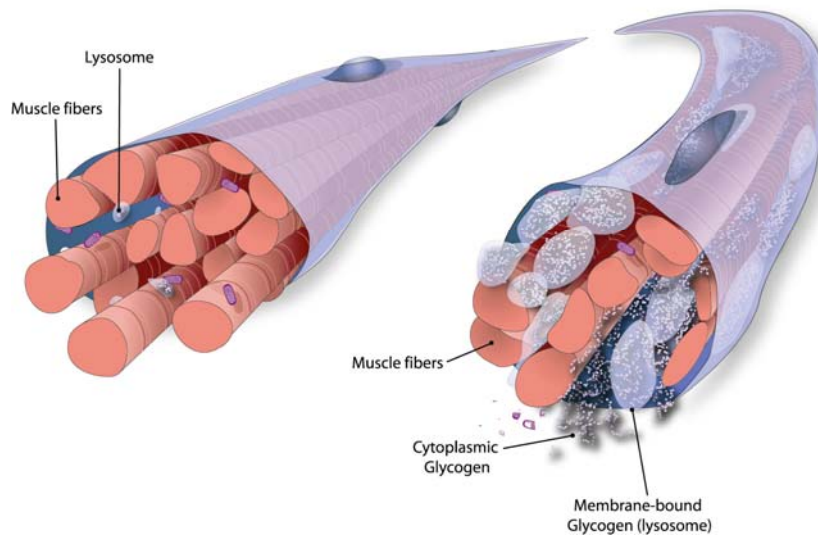
Glycogen



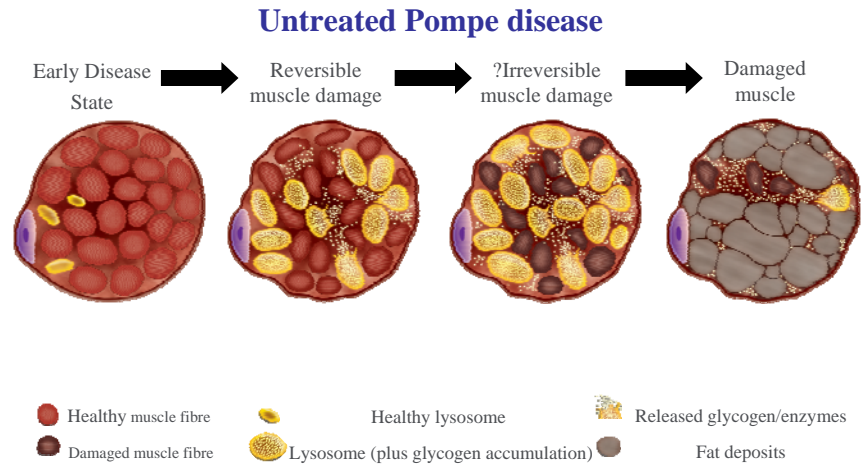
Defect in glycogen degradation due to GAA deficiency

Normal Muscle Cell

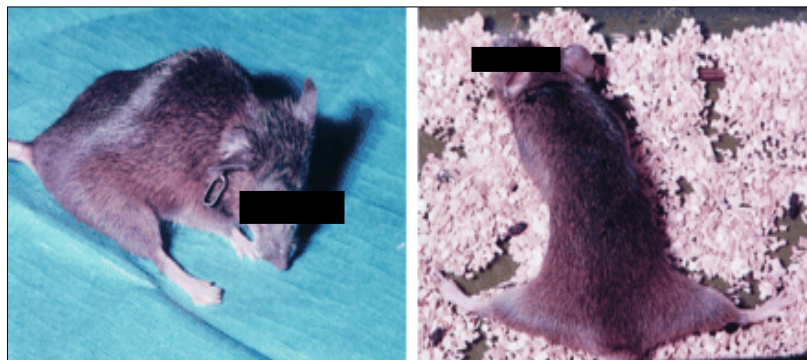
Affected Muscle Cell



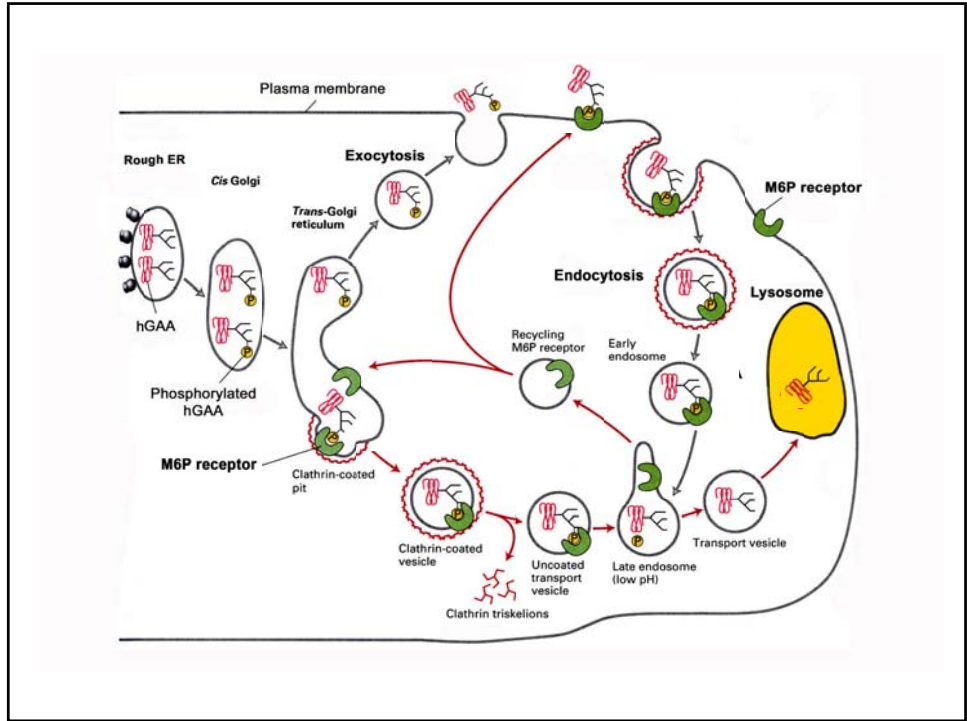
The natural course of Pompe disease is a progression from healthy muscle to muscle damage



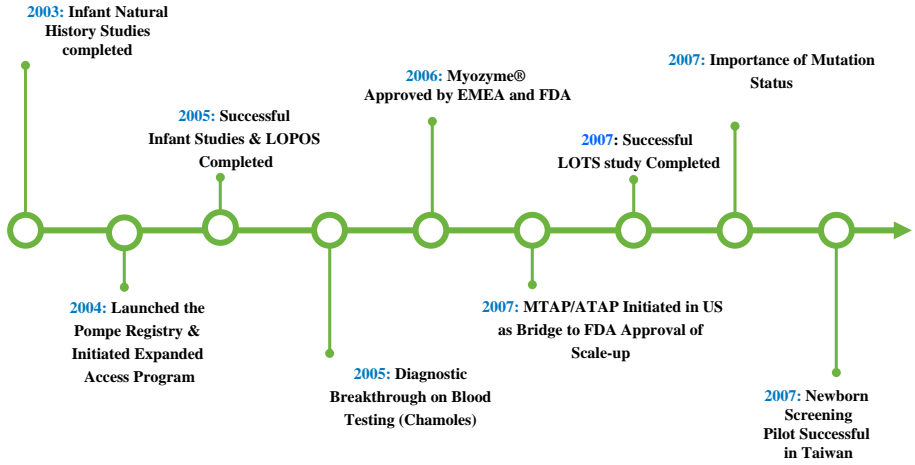
***GAA^{-/-} Mouse Model: Essential tool
in evaluation of therapeutic strategies***



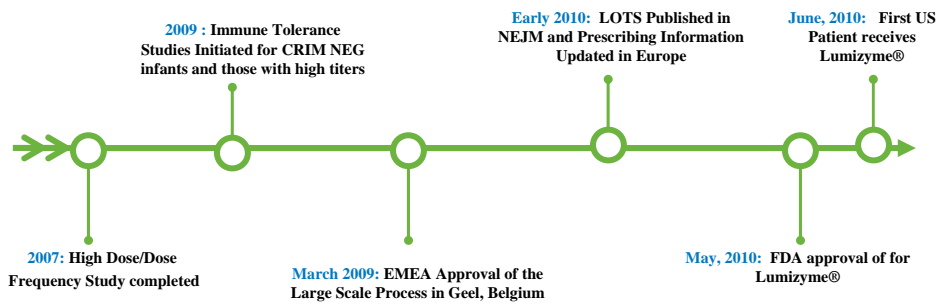
Raben et al., 1998



Product Development History



Product Development History

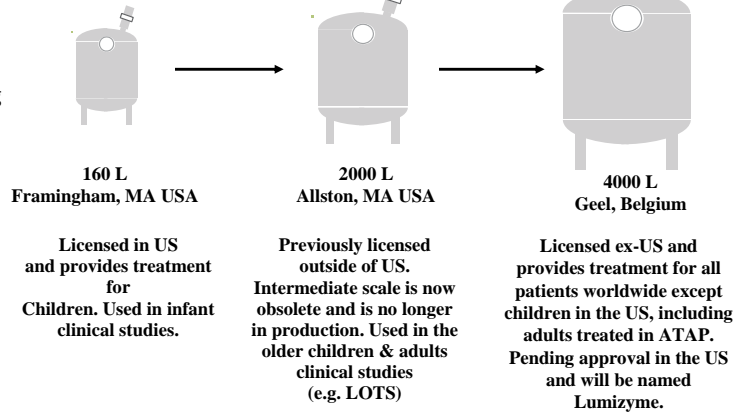


Myozyme (for children <8 yrs and exUS adults)
Lumizyme (>8 yrs in US)



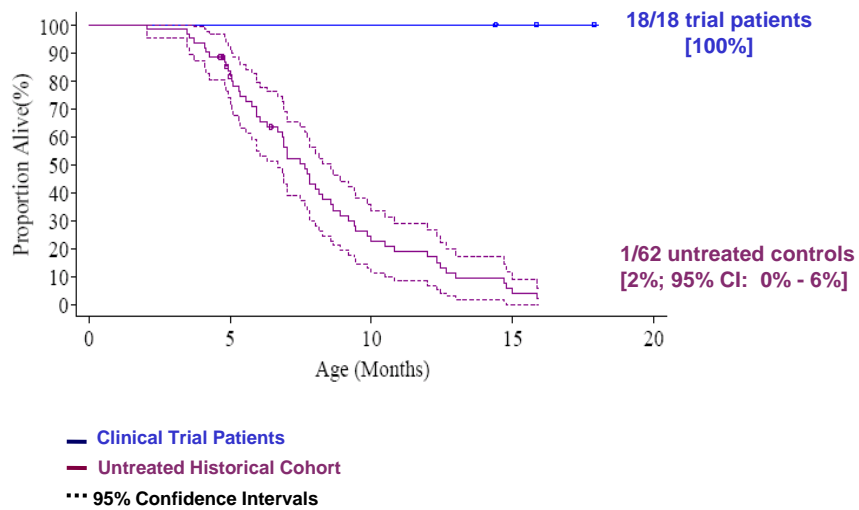
Increasing Capacity to Meet Needs Worldwide

**Bioreactor
Manufacturing
Scale**

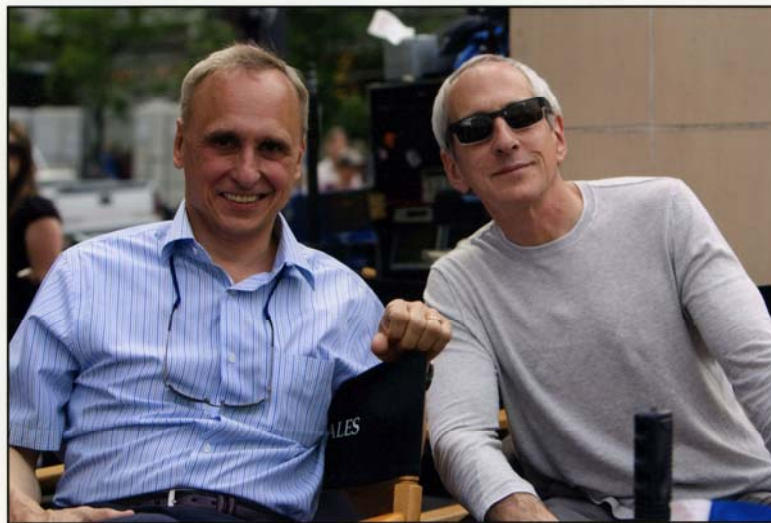
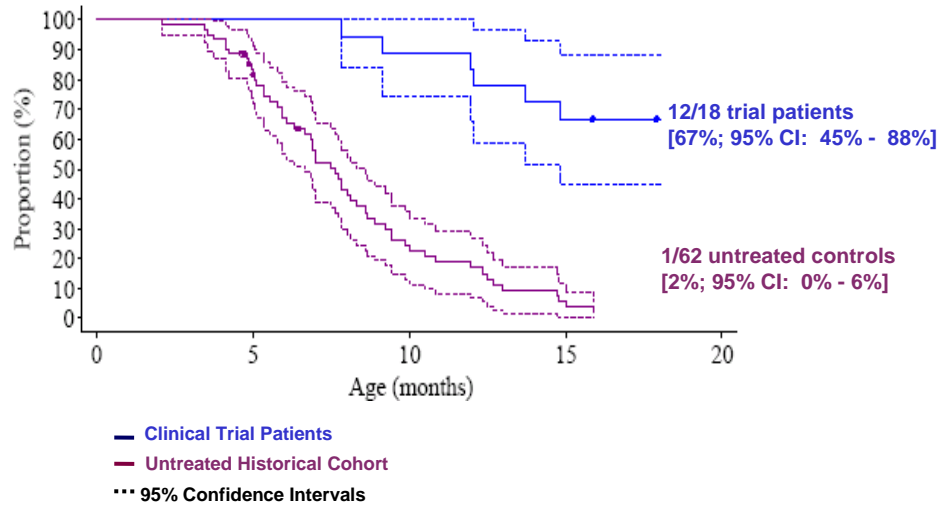


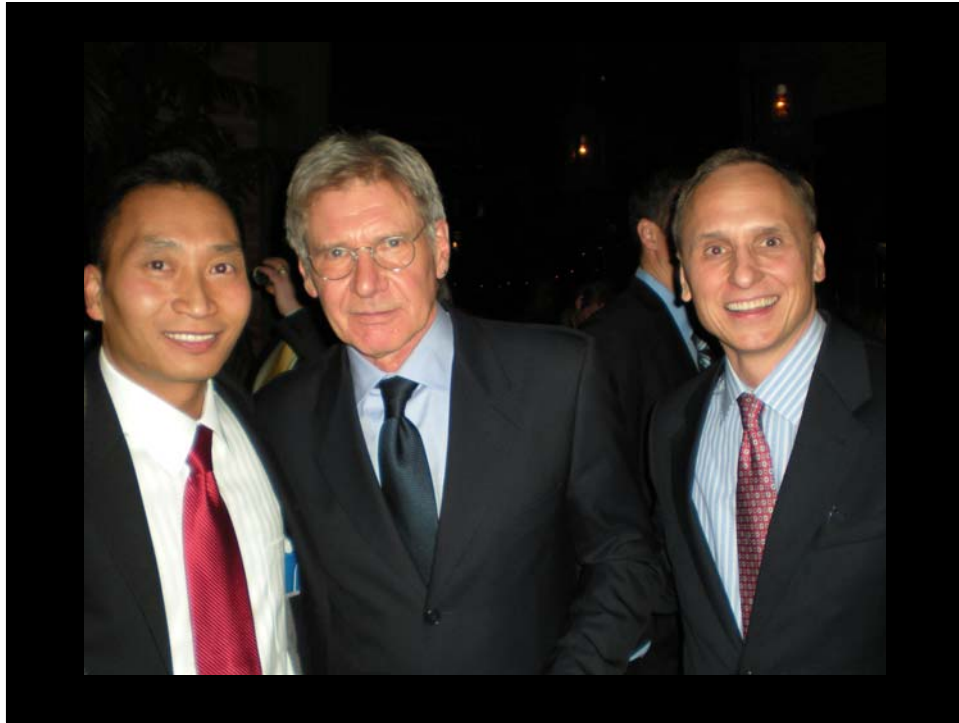
AGLU 1602:

Overall Survival at 18 Months of Age (1 yr ERT)



AGLU 1602:
Survival Free of ANY Ventilation at 18 Months





Pompe Disease

Summary

- Pompe disease is a **continuum** of clinical phenotypes, ranging from rapidly progressive infantile-onset to more slowly progressive, late-onset disease.
- All patients share a common pathophysiology: deficiency in GAA, leading to **glycogen accumulation** in the lysosome.
- **Early diagnosis** is the key to optimal patient management.
- Current management consists of **multidisciplinary** supportive measures.
- Pivotal study of ERT completed and next generation therapies are under study.



Acknowledgements

University of Florida

Cathryn Mah
David Fuller
Denise Cloutier
Darien Falk
Julie Berthy
Lee Ann Lawson
Stacy Porvasnik
Kerry O. Cresawn
Christina A. Pacak
Thomas J. Fraitcs, Jr.
Lara De Ruisseau
Brian Cleaver
Nathalie Clement

Genzyme Corporation

1602-1702 PIs:

Priya Kishnani, Durham, US
Marc Nicolino, Lyon, FR
Ed Wraith, Manchester, UK
Nancy Lesley, Cincinnati, US
Paul Hwu, Taipei, TW

NIH Collaborators

Nina Raben and Paul Plotz

Children's Hospital Boston

Carolyn Spencer
Jami Levine
Steve Colan
Charlie Berul

Supported by NIH/NHLBI, NIDDK, NCRR, American Heart Association, Muscular Dystrophy Association, Genzyme and the University of Florida.



Q&A SESSION



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Thursday, September 16, 2010, 2-3 p.m. EDT

Diversity: Old Topic – Fresh Conversations

Lolita Chandler, adjunct instructor at Cornell University, and Cathy Bristow, Founder of BRIDGES



Thursday, September 23, 2010, 2-3 p.m. EDT

Demystifying SBIR/STTR Grant Applications and Processes for Entrepreneurs and Small Businesses

Dr. Josephine Yuen, Program Director, National Science Foundation.



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