


Research Team Update

Dan Lambert
Lead, Research Team




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Outline


- Thank You
- Accomplishments
- Progress
- We Need You!
- Future Plans



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Who we are...



Christy Johnson for Caden
Brian Mannix for Chris
Jo Beth Southard for Sarah
Alison Clark for Denis
Dan Lambert for Eric
Denise Wilburn for Grant
Kristine Saylor for Jake




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Program Goal

- Fund projects to improve lives of galactosemics
- Four kinds of project designs are eligible for grants:
 - research studies
 - clinical projects
 - educational projects
 - collaborative meetings



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Thanks to: Donors

- Over \$214,419 has been donated to date
- \$94,500 donated in 2010/11
- \$94,500 donated in 2011/12
- 12 total donors/fundraisers



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Thanks to: Experts

We couldn't provide grants without:

- Proposals from committed professionals
- Peer Review of proposals by professionals in Galactosemia and related fields
- Researchers, clinicians, other experts in any aspect of Galactosemia:

Please consider becoming a GF Peer Reviewer if you haven't already!




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Research Funded to Date

- Initial investments
 - Select targeted opportunities
- Research Grants Program – 3 Rounds
 - 1st Round (2007): 1 grant
 - 2nd Round (2009): 2 grants
 - 3rd Round (2010): 3 grants




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Initial Research Fund Investments

- \$70,000: Galactosemia Center of Excellence
 - Provided support in establishing new Center at Boston Children’s Hospital
 - *Supported by Sean & Kristine Lydon*
- \$25,000: Support to Dr. Stanton Segal
 - Support research on Galactosemia at Children’s Hospital of Philadelphia (2006)




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Round 1: Funded Project

- Title: Toward Intervention for Galactosemia
- PI: Dr. Judith Fridovich-Keil, Emory
- Total Award: \$50,000
- Summary of Work:
 - Goal: take important steps toward ultimate goal to identify a drug or pill to speed up the metabolism of gal-1P in CG patients
 - Follow-up: Seeking NIH or Emory University funding to continue work




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Round 2: Funded Projects

- Title: Health and Psychosocial Outcomes in Adults with Classic Galactosemia
- PI: Dr Gerard Berry, Boston Children’s
- Total Award: \$30,000
- Summary of Work:
 - Goal: Characterize health & psychosocial status of ~30 adults with Galactosemia; Assess male and female fertility
 - Results/Follow-up: Future NIH proposal




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Round 2: Funded Projects

- Title: Review of Outcomes and Development of Guidelines for Management of Educational and Behavioral Challenges
- PI: Dr. Susan Waisbren, Boston Children’s
- Total Award: \$29,919
- Summary of Work:
 - Goal: Carry out retrospective chart review of all CG patients at Boston Children’s Hospital; Develop educational materials for families, teachers and other school personnel




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Round 3: Funded Projects

- Title: Evaluation of Nutrition Recommendations for Classic Galactosemia
- PI: Dr. Sandy van Calcar, U of Wisconsin
- Total Award: \$18,000
- Summary of Work:
 - Goals:
 - Improve consensus on dietary treatment of Galactosemia
 - Measure galactose in sodium caseinate and aged cheeses
 - Develop chart review process to compare diet records of estimated dietary galactose intake with (1) gal-1-p and urine galactitol concentrations and (2) long-term outcome
 - Follow-up: Writing and submitting a peer-reviewed publication outlining findings and recommendations



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Round 3: Funded Projects

- Title: Computational Biology Strategy for the Development of Ligands of GALK Enzyme as Potential Drugs for People with Classical Galactosemia
- PI: Dr. Anna Marabotti, *Inst. Of Biomedical Technology*
- Total Award: \$41,500
- Summary of Work:
 - **Goals:** Search for GALK inhibitors, which avoid or reduce GALT accumulation, by using computational biology to simulate the interactions of millions of compounds with GALK in relatively short time
 - **Follow-up:** Results should save future researchers money and time by highlighting potentially relevant molecules for in-depth study.

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Round 3: Funded Projects

- Title: The Brain and Neuro-functioning in Adults with Galactosemia
- PI: Dr. Susan Waisbren
- Total Award: \$34,997
- Summary of Work:
 - **Goals:** Explore whether MRI and EEG evaluations help understand CG neuropsychological problems; Examine differences between hemispheres of brains
 - **Follow-up:** Results will provide a framework for future studies in galactosemia

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We Need You!

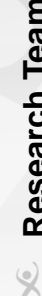
Galactosemia Professionals

- We want to fund your projects
 - Fill out a proposal and send it to us
 - www.galactosemia.org/grant_detail.asp
- We need peer reviewers:
 - Physicians, Dieticians, Researchers, etc.
 - Contact us at research@galactosemia.org

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Future Plans

- Round 4 Research Grant Program
 - **Formal Announcement Today -- \$75,000**
- Improving our communications
 - Web summaries
 - Grants underway
 - Completed grants
 - **Help needed here**
- Collaborating with any large-scale NIH project that includes galactosemia

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