



2012 Research Fund Update on the GECHO project

Contribution of Genetic Variation in Oxidative Stress Response Pathways to Functional Outcomes in Pediatrics with Single Ventricle Defects: The GECHO project

Progress Report:

There has been significant progress made in the GECHO project over the past year and the Faith's Angels funding is a valuable piece of this success.

The Long-term goal of GECHO is to assess the contribution of genetic polymorphisms in oxidative stress response pathways to specific short and intermediate term outcomes in patients with single ventricle disease such as hypoplastic left heart syndrome. Identification of genetic traits predisposing patients to adverse outcomes may enable development of individualized treatment protocols targeting those pathways most "at-risk" in each patient.

Progress To-date:

- Recruited 2 additional Retrospective sites in addition to CHOP & Columbia, in the original proposal:
 - Emory University (EM), Atlanta GA
 - Medical College of Wisconsin (MCW), Madison, WI
- Instituted monthly all-sites conference calls to aid problem-solving and build consensus.
- Received 104 retrospective patient samples previously enrolled in the Pediatric Heart Network (PHN) Infants with Single Ventricle (ISV) trial for genetic analysis.
- Received 346 additional retrospective patient samples previously enrolled in the PHN Single Ventricle Reconstruction (SVR) trial for genetic analysis.
- Performed genetic analysis on 107 retrospective patient samples previously enrolled in the Pediatric Heart Network (PHN) Infants with Single Ventricle (ISV) trial and nearly 300 additional retrospective patient samples previously enrolled in the PHN Single Ventricle Reconstruction (SVR) trial. The effect of APOE polymorphism was reproduced for the first time in this analysis and thus forwarding our understanding of this finding. A manuscript related to this work is in progress.
- Received 200 DNA Retrospective samples and clinical data from Emory.
- MCW Retro: MCW has 144 total retrospective cases and have provided their genetic data but are still working on compilation and transfer of their clinical data to us, with an anticipated delivery by 10/31/13.
- Dr. Christen Willer has replaced Dr. Sebastian Zoellner (away on sabbatical) to coordinate merging the genetic and clinical data. A medical summer student (Will Kurtz) was matched to Drs. Wilder and Russell. Will's project included assisting in developing the Xcel spreadsheet format and data elements to transfer from MCW for both the genetic and clinical data spreadsheets.

Faith's Angels Inc. is a 501c3 nonprofit organization.

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