

Wednesday, July 9, 2014

- 8:00 Continental Breakfast
- 8:30 Opening Remarks
Eser Tolunay, Ph.D., National Heart, Lung, and Blood Institute, Bethesda, MD

Session 1: GenTAC Research

Chairs: Scott LeMaire, M.D.; Alan Braverman, M.D.

- 8:40 Overview of research and new tools for clinical aortic research
Richard B. Devereux, M.D., Ph.D., Weill Cornell Medical Center, New York, NY
- 8:50 Vertebral Artery Tortuosity Index is a Novel Biomarker of Surgery and Aortic Dissection or Rupture in Children and Young Adults: Findings From the National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC)
Shaine A. Morris, M.D., Baylor College of Medicine, Houston, TX
- 9:00 Turner syndrome/CNV analysis
Siddharth Prakash, M.D., University of Texas Health Science Center, Houston, TX
- 9:10 Calcium channel blockers and aortic disease
Hal Dietz, M.D., Johns Hopkins University, Howard Hughes Medical Institute, Baltimore, MD
- 9:20 Incident Aortic Dissection Among Patients with Genetically Mediated Aortic Aneurysms – Initial Results from the GenTAC Registry
Jonathan Weinsaft, M.D. Ph.D., Weill Cornell Medical Center, New York, NY
- 9:30 Vascular surgery in patients with vascular Ehlers Danlos Syndrome
Sherene Shalhub, M.D., M.P.H., University of Washington, Seattle, WA
- 9:40 Question and Answer/Discussion
- 9:55 Break

Session 2: Genetic Predisposition to Thoracic Aortic Disease and Development of the Aorta

Chairs: Cheryl Maslen, Ph.D.; Peter Byers, M.D.

- 10:15 Arterial wall construction: mixing, sequential induction and controlled invasion
Daniel Greif, M.D., Yale University, New Haven, CT
- 10:30 Role of SCA1+ adventitial cells in vasculature
Mark Majesky, Ph.D., University of Washington, Seattle, WA
- 10:45 Myocardin-like Transcriptional Coactivators and the Pathogenesis of Heritable Forms of Arterial Aneurysm
Michael S. Parmacek, M.D., Hospital of the University of Pennsylvania, Philadelphia, PA
- 11:00 Novel genes and molecular pathways for familial thoracic aortic disease
Dianna Milewicz, M.D., Ph.D., University of Texas Health Science Center, Houston, TX
- 11:15 Novel genes and modifiers for Thoracic Aortic Disease
Catherine Boileau Pharm.D., Ph.D., Paris, France
- 11:45 Elucidation of a complex architecture for genetic modification of the TGF β vasculopathies
Juan F. Calderon, Ph.D., McKusick-Nathans Institute of Genetic Medicine, The Johns Hopkins University, Baltimore, MD
- 12:00 Bicuspid aortic valve: genome wide association study and rare variant analysis
Simon Body, MBChB, MPH, Brigham and Women's Hospital, Boston, MA
- 12:15 Modeling genetically triggered vascular diseases using human induced pluripotent stem cells
Sanjay Sinha, Ph.D., University of Cambridge, Cambridge, UK
- 12:30 Question and Answer/Discussion
- 1:00 Lunch





Session 3: Thoracic Aortic Disease in Mouse Models

Chairs: Hal Dietz, M.D., Alan Daugherty, Ph.D., Jennifer Habashi, M.D.

- 2:00 Mouse models: *TGFBR1*, *TGFBR2* and *FBN1* updates
Hal Dietz, M.D., Johns Hopkins University, Howard Hughes Medical Institute, Baltimore, MD
- 2:15 *Tgfr2* disruption in postnatal smooth muscle impairs aortic wall homeostasis
George Tellides, M.D., Ph.D., Yale University, New Haven, CT
- 2:30 *FBLN4*^{-/-} mouse model of thoracic aortic disease
Hiromi Yanagisawa, M.D., Ph.D., UT Southwestern, Dallas, TX
- 2:45 Fibrillin-1 is a component of the muscle mechanosignaling complex
Francesco Ramirez, Ph.D., Mount Sinai Medical Center, New York, NY
- 3:00 *Acta2* mutant mouse model
Jiyuan Chen, Ph.D. University of Texas Health Science Center at Houston
- 3:15 Angiotensin and TGF- β signaling in aortic disease
Alan Daugherty, Ph.D., D.Sc., University of Kentucky, Lexington, KY
- 3:30 Panel - Question and Answer/Discussion
- 3:45 Break
- 4:00 The role of ADAMTS proteins in thoracic aortic disease
Scott LeMaire, M.D., Baylor College of Medicine, Houston, TX
- 4:15 Pregnancy and the Marfan mouse
Jennifer Habashi, M.D., Johns Hopkins University, Baltimore, MD
- 4:30 *Structural alterations in the aorta due in mouse models of aortic disease*
Elaine Davis, Ph.D., McGill University, Montreal, Canada,

Contributed Abstracts

- 4:45 Cardiovascular disease in a mouse model with a germline *FBN1* knockin mutation
Marjolijn Renard, Ph.D., Shriners Hospital for Children, Portland, OR
- 5:05 TGF- β signaling overdrive contributes to the pathology of Loeys-Dietz syndrome
Elena M. Gallo, Ph.D., McKusick-Nathans Institute of Genetic Medicine, The Johns Hopkins University, Baltimore, MD
- 5:15 KLF4 Regulates ascending aortic aneurysm formation via caspase 3 regulation.
Morgan Salmon, Ph.D., University of Virginia, Charlottesville, VA
- 5:25 Critical role of NLRP3 inflammasome in thoracic aortic aneurysm and dissection formation in a mouse model
Ying Shen, M.D., Ph.D., Baylor College of Medicine, Houston, TX
- 5:35 Panel - Question and Answer/Discussion

6:00 Cocktail Hour and Poster Session



Poster Presentations

- 1. Pregnancy and the Risk of Aortic Dissection: a Case-Crossover Analysis**
Hooman Kamel, Mary J. Roman, Richard B. Devereux
- 2. Twins with Progressive Thoracic Aortic Aneurysm, Recurrent Dissection and ACTA2 Mutation**
Robert B. Hinton, Amy Shikany, Benjamin J. Landis, Jeanne F. James, Stephanie M. Ware
- 3. Open Repair of Descending Thoracic or Thoracoabdominal Aortic Aneurysms in Patients With Chronic DeBakey Type I Dissection: Early and Long-term Outcomes.**
Michael S. Hughes, Joseph S. Coselli, Susan Y. Green, Samantha Zarda, Courtney Nalty, Matt D. Price, Ourania Preventza, Kim I. de la Cruz, Scott A. LeMaire
- 4. NLRP3 Inflammasome Degrades Contractile Proteins Leading to Thoracic Aortic Biomechanical Failure in a Murine Model**
Yanqiu Zheng, Darrell Wu, Michael Hughes, Lin Zhang, Joseph S. Coselli, Ying H. Shen, Scott A. LeMaire
- 5. Changes in Gender-Related Differences in Acute Aortic Dissection over Time**
Stephen Philip, Linda Pape, Toru Suzuki, Lori Conklin, James Froehlich, Rossella Fattori, Eva Kline-Rogers, Justin Jabara, Guillaume Jondeau, Hasan Siddiqi, Eduardo Bossone, Daniel Montgomery, Eric Isselbacher, Christoph A. Nienaber
- 6. Decreased TGF β activity is associated with formation of thoracic aortic aneurysm in patients with bicuspid aortic valve**
Per Eriksson, Valentina Paloschi, Jesper Robert Gådin, Shaukat Khan, Hanna Björck, Lei Du, Shohreh Maleki, Joy Roy, Jan H.M. Lindeman, Salah A. Mohamed, Takeshi Tsuda, Anders Franco-Cereceda
- 7. Role for Notch1 Signaling in Ascending Aortic Aneurysms**
Sara N. Koenig, Patrick Rowland, Nianyuan Huang, Vidu Garg
- 8. New Genes in Old Families with Heritable Thoracic Aortic Disease**
Julie De Backer, L. Campens, B. Callewaert, L. Muino Mosquera, S. De Nobele, A. De Paepe, P. Coucke
- 9. Mutation detection rate and -characteristics in TAA related disorders: results from NGS panel testing J**
Julie De Backer, L. Campens, L. Muiño Mosquera, B. Callewaert, M. Baetens, M. Renard A. De Paepe, P. Coucke
- 10. Comprehensive Genetic Screening for Thoracic Aortic Aneurysm via Whole Exome Sequencing**
Bulat A. Ziganshin, Allison E. Bailey, Celinez Coons, Daniel Dykas, Lokman Tanriverdi, Lucy Liu, Maryann Tranquilli, Petros Tspirouras, Allen E. Bale, John. A. Elefteriades
- 11. Advanced glycation end products and their soluble receptors: A novel risk factor in the pathogenesis of aortic aneurysm**
Bulat A. Ziganshin, Kailash Prasad, Abdullah Sarkar, Courtney Mayerick, Mohammad A. Zafar, Ahmed Shoker, Hamdi El Moselhi, Maryann Tranquilli, John A. Elefteriades
- 12. Comparison of 3-Dimensional Echocardiography and Cardiac Magnetic Resonance Imaging for the Assessment of Aortic Dilation**
Justin Uhl, Luciana Young, Jennifer Acevedo, Elizabeth Cappella, Karen Rychlik, Grace Choi
- 13. Captopril Treatment Worsens Aortic Remodeling Associated with Aortic Constriction and Aneurysm Formation in Acta2^{-/-} Mice**
Andrew Peters, Jiyuan Chen, Siddharth Prakash, Shao-Qing Kuang, Corey Reynolds, Dianna Milewicz



- 14. Integrin interaction with fibrillin-1 triggers an integrated response in microRNA expression**
Dieter Reinhardt, Karina Zeyer, Heena Kumra, Amani Hassan
- 15. SM22 knockout mouse model: the roles of cytoskeletal remodeling-induced oxidative stress and inflammation in aneurysm formation**
Li Li, Xiaohua Dai, Neeraja Priyanka Annam, Gannon Curtis, Pamela C Udeh-Ubaka
- 16. The Turner Resource Network (TRN): A Collaboration to Promote Excellence in Turner syndrome (TS) Science and Health Policy**
Michael Silberbach, Cheryl Maslen, Dianna M. Milewicz
- 17. Premature Aortic Smooth Muscle Cell Differentiation Contributes to Matrix Dysregulation in Marfan Syndrome**
Matthew Dale, Wanfen Xiong, Trevor Meisinger, Jeffrey Carson, B. Timothy Baxter
- 18. Echocardiography Underestimates Aortic Size in Children with Bicuspid Aortic Valve**
Glen Iannucci, Timothy Slesnick, James Parks, William Mahle, Denver Sallee,





GenTAC Thursday, July 10, 2014

8:00 Continental Breakfast

Session 4: Losartan Trials, Cohort Studies, and Biomarkers

Chairs: Reed Pyeritz, M.D., Guillaume Jondeau, M.D.,

- 8:30 Timeline for US trial outcome and ancillary studies
Ronald Lacro, M.D., Harvard University, Boston, MA
- 8:45 Update on meta-analysis of the losartan trials world wide
Alex Pitcher, M.A., BMBCh, MRCP, University of Oxford, Oxford, UK
- 9:00 Overview of mouse drug trials – what are they telling us?
Francesco Ramirez, Ph.D., Mount Sinai Medical Center, New York, NY
- 9:15 Aortic Disease Outcomes Associated with ACTA2 mutations
Ellen Regalado, M.S., C.G.C., University of Texas Health Science Center, Houston, TX
- 9:30 IRAD update
Kim Eagle, M.D., University of Michigan, Ann Arbor, MI

- 9:45 Panel - Question and Answer/Discussion

- 10:15 Biomarkers for aortic disease
Lynn Sakai, Ph.D., Shriners Research Center, Portland, OR
- 10:30 Preliminary Biomarkers for identification of human ascending thoracic aortic aneurysm
James D. McCully, Ph.D., Harvard University, Cambridge, MA

Contributed Abstracts

- 10:45 Aortic Wall Delayed Gadolinium Enhancement in Thoracic Aortic Aneurysm Patients
Hadas Shiran, M.D., Stanford University, Palo Alto, CA
- 10:55 Aortic size indices: A more comprehensive evaluation of aortic risk
Arturo Evangelista, M.D., Hospital General Universitari Vall d'Hebron, Barcelona, Spain
- 11:05 Pediatric nonsyndromic thoracic aortic aneurysm: opportunities for early clinical stratification and therapy?
Benjamin J. Landis, M.D., Cincinnati Children's Hospital Medical Center, Cincinnati, OH
- 11:15 Prevalence of aortopathy in patients with hereditary hemorrhagic telangiectasia
Christina Rigelsky, M.S., Cleveland Clinic, Cleveland, OH
- 11:25 Panel - Question and Answer/Discussion
- 11:45 Lunch

Session 5 Imaging and Surgical Outcomes

Chairs: Craig Miller, M.D. and Federico Asch, M.D.

- 1:00 Evolving Guidelines: Multimodality imaging of the thoracic aorta
Steven Goldstein, M.D., Washington Hospital Center, Washington, DC
- 1:15 Novel MRI for aortic physiology
Rachel Clough, M.D., Ph.D., King's College, London, UK
- 1:30 Aortic Size & Dissection – What Do We Really Know?
Richard B. Devereux, M.D., Ph.D., Weill Cornell Medical Center, New York, NY
- 1:45 Imaging of Aortic Disease with bicuspid aortic valve
Arturo Evangelista, M.D., Hospital General Universitari Vall d'Hebron, Barcelona, Spain
- 2:00 Update from the Aortic Valve Operative Outcomes in Marfan Patients Study
Scott LeMaire, M.D., Baylor College of Medicine, Houston, TX
- 2:15 Indications and Contraindications of TEVAR for Genetically Triggered Aortic Aneurysmal Disease
Thomas G. Gleason, M.D., University of Pittsburgh, Pittsburgh, PA



- 2:30 Tirone David V-SARR for Patients with Bicuspid Aortic Valves. Corollary: Should all aortic root surgery be performed only in established centers of excellence?
D. Craig Miller, M.D., Stanford University, Palo Alto, CA
- 2:45 Aortic replacement in Bicuspid Aortic Valve: How big? How much?
Thor Sundt, III, M.D., Harvard University, Boston, MA
- 3:00 Emergency stent-grafting (TEVAR) for complicated acute type B dissections in genetically triggered aortic disease
Karianna Milewski, M.D., Ph.D., University of Pennsylvania Medical School, Philadelphia, PA
- 3:15 TEVAR vs. Open surgery for chronic Type B
Eric Roselli, M.D., Cleveland Clinic, Cleveland, OH
- 3:30 Panel - Question and Answer/Discussion

Closing Remarks:

- 3:45 Overview and Future Directions for GenTAC
Kim Eagle, M.D., University of Michigan, Ann Arbor, MI,
Patrice Desvigne-Nickens, M.D., National Heart, Lung, and Blood Institute, Bethesda, MD
- 4:00 Adjourn

