CURRICULUM VITAE
Harry C. Dietz, III, M.D.

DEMOGRAPHIC INFORMATION

Current Appointments:
Investigator, Howard Hughes Medical Institute
Victor A. McKusick Professor of Medicine and Genetics
Johns Hopkins University School of Medicine,
Department of Genetic Medicine

Address:
Johns Hopkins University School of Medicine
Edward D. Miller Research Building
733 N. Broadway - MRB 539
Baltimore, MD 21205
Phone: (410) 614-0701
Fax (410) 614-2256
e-mail: hdietz@jhmi.edu

Education and Professional Training:

B.S.E.	8/76 - 6/80	Duke University, Biomedical Engineering Durham, North Carolina
M.D.	9/80 - 6/84	SUNY Upstate Medical University Syracuse, New York
	7/84 – 6/85	Internship in Pediatrics, Harriet Lane Service, Johns Hopkins Hospital, Baltimore, MD
	7/85 – 6/87	Residency in Pediatrics, Harriet Lane Service, Johns Hopkins Hospital, Baltimore, MD
	7/87 – 6/88	Residency in Anesthesia & Critical Care Medicine, Johns Hopkins Hospital, Baltimore, MD
	7/88 – 6/89	Chief Resident of Pediatrics, Harriet Lane Service, Johns Hopkins Hospital, Baltimore, MD
	7/89 – 6/92	Clinical Fellowship in Pediatric Cardiology, Johns Hopkins Hospital, Baltimore, MD
	7/90 – 6/92	Post-Doctoral Fellowship, Center for Medical Genetics, Johns Hopkins Hospital, Baltimore, MD

Professional Experience:

7/94 – pres. Director	Vascular Connective Tissue Disorder Clinic, Department of Genetic Medicine Johns Hopkins University School of Medicine, Baltimore, MD
4/97 – pres. Investigator	Howard Hughes Medical Institute Chevy Chase, MD
7/19 – pres. Professor	Victor A. McKusick Professor of Medicine and Genetics Department of Genetic Medicine, Departments of Pediatrics, Medicine and Molecular Biology & Genetics Johns Hopkins University School of Medicine, Baltimore, MD
7/04 – 6/19 Professor	Victor A. McKusick Professor of Medicine and Genetics Institute of Genetic Medicine, Departments of Pediatrics, Medicine and Molecular Biology & Genetics Johns Hopkins University School of Medicine, Baltimore, MD
6/99 - 6/19 Professor	Institute of Genetic Medicine, Departments of Pediatrics, Medicine, and Molecular Biology & Genetics Johns Hopkins University School of Medicine, Baltimore, MD
7/94 – 5/99 Assoc. Prof.	Center for Medical Genetics, Departments of Pediatrics, Medicine and Molecular Biology & Genetics Johns Hopkins University School of Medicine, Baltimore, MD
7/93 – 6/94 Asst. Prof.	Joint Appointment, Department of Medicine Johns Hopkins University School of Medicine, Baltimore, MD
7/92 – 6/94 Asst. Prof.	Division of Cardiology and Center for Medical Genetics, Department of Pediatrics Johns Hopkins University School of Medicine, Baltimore, MD

RECOGNITION

Honors and Distinctions:

SUNY Upstate School of Medicine:
1983 Alpha Omega Alpha
1984 Summa Cum Laude, Valedictorian, Class of 1984

The Johns Hopkins Hospital:
1988 Pediatric Chief Resident
1990 Harriet Lane Research Fellowship, Department of Pediatrics
1992 First Richard Starr Ross Research Scholar, Johns Hopkins Medical Institutions
2000 Director, William S. Smilow Center for Marfan Syndrome Research
2004 First Victor A. McKusick Professor of Medicine and Genetic
2009 Heritage Award

National and International:
1992 Richard D. Rowe Award, Society for Pediatric Research
1993 Young Investigator Award, Society for Pediatric Research
1993 Patrick John Niland Memorial Lecture, University of Michigan
1993 Professional Advisory Board, National Marfan Foundation
1993 Editorial Board, Genomics
1994 Antoine Marfan Award, National Marfan Foundation
1995 Visiting Professorship, Northwestern University School of Medicine
1996 Visiting Professorship, Cleveland Clinic
1997 Appointed to the Howard Hughes Medical Institute
1997 Elster Memorial Lecture, Mount Sinai University School of Medicine
1997 Visiting Professorship and Memorial Lecture, Hospital for Sick Children
1997 Visiting Professorship, Winner Memorial Lecture, Univ of Maryland School of Medicine
1997 Department of Energy BER 50th Anniversary Celebration, National Academy of Sciences
1997 Editorial Board, Human Molecular Genetics
1998 Chair, Professional Advisory Board, National Marfan Foundation
1999 Editorial Board, Circulation Research
2001 Pauline Wilson Horner Lecture, Case Western Reserve University
2001 Editorial Board, Pediatric Research
2002 Inductee, Society for Pediatric Research
2002 Chair, Agarini Foundation Scientific Committee
2002 Editorial Board, American Journal of Human Genetics
2003 Inductee, American Society for Clinical Investigation
2004 Board of Scientific Counselors, National Human Genome Research Institute
2005 Elected to the Board of the American Society for Clinical Investigation
2005 Inductee, American Association for the Advancement of Science
2005 Wilson Lecture, Case Western Reserve University
2005 Editorial Board, American Journal of Medical Genetics
2006 Recipient of the Curt Stern Award, American Society of Human Genetics
2006 Nahum J. Winer Memorial Lecture, New York Academy of Medicine
2006 Thomas P. Graham Jr. Lecture, Pioneer Lecture Series, Vanderbilt University
2006 Cazden Lecture, Oregon health Sciences University
2006 Meinhard Robinow Lecture, Dayton Children's Hospital
2007 Chair, Board of Scientific Counselors, National Human Genome Research Institute

2007 Elected to the Board of the American Society of Human Genetics
2007 Scientific Advisory Board, Center for Preterm Birth Research at Washington University in St. Louis
2007 Princess Lilian Foundation Professorship, Gent, Belgium
2007 Edmund R. McCluskey Memorial Lectureship, University of Pittsburgh
2007 Paulette Shirley Pritchett Endowed Lecture in Pathology, University of Alabama at Birmingham
2007 Isadore Rosenfeld Visiting Professorship, Weil Medical College of Cornell University
2007 Robert M. Jeresaty Lecture, St. Francis Medical Center, Hartford
2007 Congressional Biomedical Research Caucus Symposium, Joint Steering Committee on Public Policy
2007 Kathryn L. Ober Visiting Professor, Indiana University
2007 Editorial Board, Clinical and Translational Science
2008 Hero with a Heart Honoree, National Marfan Foundation
2008 Humanitarian Award, American Skin Association, New York
2008 Inductee, National Academy of Sciences Institute of Medicine
2009 Heritage Award, Johns Hopkins University
2009 George E. Brown Memorial Lecturer Award – American Heart Association
2009 Visiting Professorship, AHA/Council on Cardiovascular Disease in the Young
2009 Visiting Professorship, Imperial College (London)
2010 Niccolo Paganini Prize, SIMA Congreso Internacional de Síndrome de Marfan
2010 Visiting Professorship, (James Bass), Depart of Pediatrics, Madigan Army Medical Center
2010 Visiting Professorship, Anandi L. Sharma, Mount Sinai School of Medicine
2010 Art of Listening Award, Genetic Alliance
2011 Pruzansky Lecture (Annual Clinical Genetics Meeting) - ACMG
2011 Inductee, The National Academy of Sciences
2012 Colonel Harland Sanders Lifetime Achievement Award in Medical Genetics, March of Dimes and American College of Medical Genetics
2012 Winner - Taubman Prize for Excellence in Translation Medical Science
2013 Inductee – Association of American Physician
2013 Winner – Pasarow Award in Cardiovascular Research
2014 Winner – InBev - Baillet Latour Health Prize, Belgium
2014 Winner – Harrington Prize from the American Society for Clinical Investigation and the Harrington Institute
2014 Clinical Cardiology Laennec Clinician/Educator Lecture Award
2015 The Bernard and Joan Marshall Research Distinguished Investigator Award, University of Oxford
2015 Karsh Professorship Award, Children's Hospital of Eastern Ontario
2015 Recipient of the American Heart Association Research Achievement Award
2016 President, The American Society of Human Genetics
2019 Winner - ASHG Mentorship Award

EXTRAMURAL SPONSORSHIP

Current Research Support

Howard Hughes Medical Institute, Principal Investigator (8/97 - present)
Victor A. McKusick Professor of Medicine and Genetics Endowment (7/04 - present)
Ehlers-Danlos Syndrome Foundation (02/07 – present)
National Marfan Foundation (8/09 - present)
Loeys-Dietz syndrome Foundation (06/09 - present)
Coles Family Foundation (9/15 – present)
Alison Aldredge Fund for vascular Ehlers-Danlos syndrome - (11/15 - present)

The Feather Foundation Gift Fund (11/15 - present)
Daskal Fund for vascular Ehlers-Danlos syndrome (12/17 - present)
Kasper Fund for Loeys Dietz Syndrome (01/18 – present)
Erin G. and Douglas L. Becker Gift Account (12/19 – present)

Teaching Activities:

Preceptor, Predoctoral Research Training Programs
- Human Genetics
- Biochemistry, Cellular & Molecular Biology
- Cellular and Molecular Medicine

Fellowship Training
- Division of Pediatric Cardiology
- Division of Pulmonary and Critical Care Medicine
- Department of Surgery

Editorial Boards:
Editor, Science Translation Medicine
Editor, JCI

CLINICAL PRACTICE

Certification and Licensure:
American Board of Pediatrics
State of Maryland Medical License (D0038909)

Clinical Activities:
Director, Heritable Disorders of Connective Tissue Clinic, 7/94 - present
Weekly Clinic for the Evaluation of Families with Heritable Cardiovascular Disease

ORGANIZATIONAL ACTIVITIES

Professional Activities:

Heart Disease in the Young, American Heart Association Study Section (1996-1999)
Professional Advisory Board of the National Marfan Foundation (1997 – present)
Induced Mutant Repository Advisory Board, Jackson Laboratories
Member, Young Investigator's Day Award Review Committee
Member, David Israel Macht Memorial Research Prize Committee
Member, Michael A. Sarnoff Research Award Committee
Member, Human Genetics Selection Committee
Member, Medical Scientist Training Program Steering Committee (MD/PhD)
Associate Director, Human Genetics Predoctoral Training Program
American Society of Human Genetics Nomination Committee, 1999
The Genome Action Coalition (American Society of Human Genetics Representative)
Member, McKusick-Nathans Institute of Genetic Medicine Directorship Search Committee, 1999-2000
Member, Agarini Foundation Scientific Committee
Member, McKusick-Nathans Institute of Genetic Medicine Recruitment Committee
Member, Evaluation Committee for Specialized Centers of Research (SCOR) Program in Cardiovascular Disease, National Institutes of Health, Division of Heart and Vascular Diseases
Member, Mammalian Genetics Study Section, National Institutes of Health (2002-2004)
Member, Department of Molecular Biology and Genetics Chairman Recruitment Committee

Chair, Board of Scientific Counselors, National Human Genome Research Institute
Member, Advisory Board of the Gene Vector Core
Member, Board of American Society for Clinical Investigation
Director, William S. Smilow Center for Marfan Syndrome Research
Member, Professional Advisory Board of the Ehlers-Danlos Syndrome National Foundation
Member, Institute of Medicine (IOM) Committee on Accelerating Research and Development for Rare Diseases and Orphan Products
Chair, National of Arthritis and Musculoskeletal and Skin Diseases Advisory Council of the National Institutes of Health (NIAMS) 3/2010 – 9/2013
Member, Global Fibrosis Foundation Medical Advisory Council
Editor, Science Translation Medicine
ASHG Nominating Committee
Musculoskeletal P & F Advisory Board
Director, William S. Smilow Center for Marfan Syndrome Research
Member, Search and Screen Committee, Medical Physiology and Metabolism Section, National Academy of Sciences
Member, NiH Advisory Board for Clinical Research (ABCR)
Member, National Organization for Rare Disorders (NORD) Scientific Advisory Committee
President, The American Society of Human Genetics 2016
Member, Advisory Committee to the Deputy Director for Intramural Research (ACDDIR)
Member, NIH Board of Scientific Counselors for NHGRI
Member, NIH National Advisory Council for Human Genome Research (HGRAC)
Member, ASHG Nominating Committee
Member, Search Committee, Scientific Director Search (NHGRI)
Non-Executive Director (NED) – GlaxoSmithKline (2022)

Professional Societies:

The American Society of Human Genetics
Fellow of the American Heart Association
Charter Fellow of the Council on Basic Cardiovascular Sciences
Society for Pediatric Research
American Society for Clinical Investigation
Fellow of the American Association for the Advancement of Science
National Academy of Medicine
National Academy of Sciences
Association of American Physicians (AAP)

Ad Hoc Reviewer:
American Journal of Cardiology
American Journal of Human Genetics
American Journal of Medical Genetics
Archives of Pediatric and Adolescent Medicine
Cell
Circulation
Clinical Genetics
Developmental Dynamics
eLife
European Journal of Human Genetics
Human Molecular Genetics
Human Mutation
Journal of Clinical Investigation
Nature Genetics
Nature Medicine

New England Journal of Medicine
Nucleic Acids Research
Proceedings of the National Academy of Sciences (USA)
RNA
Science Translational Medicine

SCIENTIFIC MENTORING:

Predoctoral Graduate Trainees:

Zayd A. Eldadah, M.D./ Ph.D.
Medical Scientist Training Program, Institute of Genetic Medicine
The Johns Hopkins University School of Medicine, 1992 - 1996
Plenary Speaker, American Society of Human Genetics Meeting, 1994
Finalist, Predoctoral Basic Research Award,
American Society of Human Genetics, 1995

Nancy L. Jensen Biery, Ph.D.
Human Genetics Predoctoral Training Program,
The Johns Hopkins University School of Medicine, 1993 - 1999
Plenary Speaker, American Society of Human Genetics Meeting, 1999

Haley A. (Perlick) Laken, Ph.D.
Human Genetics Predoctoral Training Program,
The Johns Hopkins University School of Medicine, 1994 - 1999
Winner, Predoctoral Basic Research Award,
American Society of Human Genetics, 1996
Winner, Paul Ehrlich Award, Young Investigator's Day, 1998

Erick Noensie, Ph.D.
Human Genetics Predoctoral Training Program,
The Johns Hopkins University School of Medicine, 1995 – 2000

Pamela A. Frischmeyer, M.D./Ph.D.
Medical Scientist Training Program, Institute of Genetic Medicine
The Johns Hopkins University School of Medicine, 1996 - 2001
Associate, Howard Hughes Medical Institute
Winner, Predoctoral Basic Research Award,
American Society of Human Genetics, 2001
Winner, Michael A. Shanoff Award, Young Investigator's Day, 2002
(This is the highest award given to a scientist in training at Johns Hopkins University School of Medicine)

Dan E. Arking, Ph.D.
Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 1996 – 2002
Winner, Ruth L. Kirschstein National Research Service Award, 2003 – 2006
Finalist, American Society of Human Genetics,
Postdoctoral Presentation Award, 2005

Joshua T. Mendell, M.D./Ph.D.
Medical Scientist Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 1996 - 2004

Associate, Howard Hughes Medical Institute
National Science Foundation Young Science Scholar
Postdoctoral Basic Science Award, American Society of Human Genetics, 2003
Winner, Michael A. Shanoff Research Award, Young Investigators' Day,
Johns Hopkins University School of Medicine,, 2003
March of Dimes Basil O'Connor Scholar, 2004
Rita Allen Foundation Scholar, 2004
Allan C. Davis Medal, Outstanding Young Scientist in the
State of Maryland, 2007
Leukemia and Lymphoma Society Scholar, 2008
Howard Hughes Medical Institute Early Career Scientist, 2009
AACR Outstanding Achievement in Cancer Research Award, 2010
CPRIT Scholar in Cancer Research, 2011

Neda Sharifi, Ph.D.
Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2002 - 2007
Postdoctoral Fellow, Institute of Genetic Medicine,
Johns Hopkins University School of Medicine, 2006 - 2008

Mark Awad, M.D./Ph.D.
Medical Scientist Training Program,
Cellular and Molecular Medicine Graduate Program
The Johns Hopkins University School of Medicine, 2003 - 2006

Connie Ng (Hess), M.D.
Sarnoff Predoctoral Training Fellow, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2003 – 2004

K.C. Kent, Ph.D.
Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2004 - 2012

Timothy Cooper, DVM, Ph.D.
Cellular and Molecular Medicine Graduate Program,
The Johns Hopkins University School of Medicine, 2004 – 2007
Dean's Award for Excellence in Teaching, 2010
Junior Faculty Teaching Excellence Award, 2012
Basic Science, Society of Distinguished Educators, 2012
Current Position: Assistant Professor, Department of Comparative Medicine,
Hershey Medical Center College of Medicine

Matthew Feldman, M.A.
Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2004 - 2011

Tammy Holm, M.D./Ph.D.
Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2007 - 2009

Jefferson Doyle, M.D./Ph.D.
Cellular and Molecular Medicine Graduate Program
The Johns Hopkins University School of Medicine, 2007 – 2016
Winner, National Marfan Foundation Victor A. McKusick Fellowship, 2009 – 2011

Winner, Pre-Doctoral Translational Research Prize,
American Society of Human Genetics Meeting, 2009
Finalist, Translational Research Prize, American Society of Human Genetics, 2010
Scientific Seminar Series Award (2nd Place), Johns Hopkins Post-Doctoral Assoc., 2011
Winner, Michael A. Shanoff Award, Young Investigators' Day,
The Johns Hopkins School of Medicine, 2011
Winner, National Marfan Foundation (NMF),
Victor A McKusick Fellowship, 2011 - 2013
Funding: National Marfan Foundation Victor A. McKusick Fellowship Program

Elizabeth Gerber, M.D./Ph.D.
Medical Scientist Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2007 – 2013
Winner, Predoctoral Research Trainee Award,
American Society of Human Genetics, 2011
Winner, Paul Ehrlich Young Investigator Award, Young Investigators' Day,
The Johns Hopkins University School of Medicine, 2012
Funding: Scleroderma Research Foundation

Stefani Fontana, M.D./Ph,D.
Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2007 - 2013
Funding: The Smilow Center for Marfan Syndrome Research

David Kim, Ph.D.
Cellular and Molecular Medicine Graduate Program
The Johns Hopkins University School of Medicine, 2007 - 2015

Hamza Aziz, M.D.
Sarnoff Predoctoral Training Fellow,
The Johns Hopkins University School of Medicine, 2008 – 2009
Appleseed Resident Teaching Award, 2012

Juan Calderon, Ph.D.
Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2008 - 2014
Fulbright Scholar
Funding: Ehlers-Danlos Syndrome Gift Account

Shira Ziegler, M.D./Ph.D.
Medical Scientist Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2011 - 2017
Funding: Howard Hughes Medical Institute

Nicole Wilson, Ph.D.
Cellular and Molecular Medicine Graduate Program
The Johns Hopkins University School of Medicine, 2012 – 2017
Funding: Luducq Foundation

Benjamin Kang, Ph.D.
Cellular and Molecular Medicine Graduate Program
The Johns Hopkins University School of Medicine, 2012 – 2017
Funding: The National Marfan Foundation Gift Account

Suha Bachir, M.D.
Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2012 - 2014

Oscar Busnadiego Prieto, Ph.D.
Visiting Scientist, Institute of Genetic Medicine
The Johns Hopkins University School of Medicine, 2012
Funding: Fellowship provided by Spanish Ministry of Science and Innovation, EMBO (European Molecular Biology Organization)

James Beckett
Medical Scientist Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2014 – 2018
Funding: Scleroderma Research Foundation

Robert Wardlow
Cellular and Molecular Medicine Graduate Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2015 – 2018
Funding: Howard Hughes Medical Institute Scholarship (Gilliam Scholarship)

Adam Lee Johnson, M.D.
Medical Student Weill Cornell Medical College
HHMI Medical Research Fellow, 2016 - 2017
Funding: Howard Hughes Medical Institute

Joseph Shin
Medical Scientist Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2015 – 2019
Funding: Scleroderma Research Foundation

Manuel Senderos-Seman
Cellular and Molecular Medicine Graduate Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2015 – 2021
Funding: National Institutes of Health/NIH R01

Caitlin Bowen
Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2017 – 2020
Funding: National Marfan Foundation Grant

Emily Juzwiak
Cellular and Molecular Medicine Graduate Program, Department of Genetic Medicine
The Johns Hopkins University School of Medicine, 2019 –
Funding: National Institutes of Health/NiH R01

Cassie Parks
Medical Scientist Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2019 –
Funding: MSTP

Postdoctoral:

Robert A. Montgomery, M.D./Ph.D.
Postdoctoral Fellow, Department of Surgery and Center for Medical Genetics,
The Johns Hopkins University School of Medicine, 1995 - 1997
Winner, Postdoctoral Basic Research Award,
American Society of Human Genetics, 1996
Funding: NIH R01, individual foundation support
Current Position(s): Professor, Department of Surgery
Director, NYU Langone Transplant Institute

Pamela A. Frischmeyer, M.D./Ph.D.
Medical Scientist Training Program, Institute of Genetic Medicine
The Johns Hopkins University School of Medicine, 1996 - 2001
Associate, Howard Hughes Medical Institute
Winner, Predoctoral Basic Research Award,
American Society of Human Genetics, 2001
*Winner, Michael A. Shanoff Award, Young Investigator's Day, 2002
**(This is the highest award given to a scientist in training at Johns Hopkins University School of Medicine)*
Current Position: Chief, Food Allergy Research Unit, National Institute of Allergy and Infectious Diseases (NIAID)

Susan M. Medghalchi, Ph.D.
Associate, Howard Hughes Medical Institute, 1997 – 2000
Funding: Howard Hughes Medicine Institute
Current Position: Adjunct Professor of Biology, Stevenson University

Jun Zhang, Ph.D.
Associate, Howard Hughes Medical Institute
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 1998 - 2002
Funding: Howard Hughes Medicine Institute
Current Position: Associate Professor, Center of Emphasis in Cancer, Texas Tech University Health Sciences Center

Daniel P. Judge, M.D.
Postdoctoral Fellow, Division of Cardiology,
The Johns Hopkins University School of Medicine, 1998 - 2002
Funding: NIH R01, individual K08
Osler Science and Medicine Award, 2005
Current Position: Professor of Medicine in Cardiology, Medical University of South Carolina

Enid Neptune, M.D.
Postdoctoral Fellow, Division of Pulmonology,
The Johns Hopkins University School of Medicine, 1999 - 2002
Funding: NIH R01, individual K08
Current Position: Associate Professor of Medicine, Division of Pulmonology,
The Johns Hopkins University School of Medicine

Zayd A. Eldadah, M.D./Ph.D.
Postdoctoral Fellow, Center for Medical Genetics,
The Johns Hopkins University School of Medicine, 2000 - 2002
Winner, Postdoctoral Clinical Research Award,
American Society of Human Genetics, 2000
Funding: NIH R01, individual foundation support
Current Position: Director of Cardiac Electrophysiology at MedStar Heart & Vascular Institute

Denise Goh, M.D.
Postdoctoral Fellow, Medical Genetics Training Program,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2000 - 2002
Funding: Medical Genetics Training Grant, individual support from Singapore Government
Winner, Faculty Teaching Excellence Award, 2009 – 2010
Winner, Annual Teaching Award, 2010 - 2011
Current Position: Head, Division of Paediatric Genetics and Metabolism, Khoo Teck Puat-National University Children's Medical Institute, National University Health System

Joshua T. Mendell, M.D./Ph.D.
Medical Scientist Training Program,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 1996 - 2004
Associate, Howard Hughes Medical Institute
National Science Foundation Young Science Scholar
Postdoctoral Basic Science Award, American Society of Human Genetics, 2003
Winner, Michael A. Shanoff Research Award, Young Investigators' Day,
The Johns Hopkins University School of Medicine, 2003
March of Dimes Basil O'Connor Scholar, 2004
Rita Allen Foundation Scholar, 2004
Allan C. Davis Medal (Outstanding Young Scientist in the State of Maryland), 2007
Leukemia and Lymphoma Society Scholar, 2008
Howard Hughes Medical Institute Early Career Scientist, 2009
AACR Outstanding Achievement in Cancer Research Award, 2010
CPRIT Scholar in Cancer Research, 2011
Current Position: Professor and Vice-Chair, Department of Molecular Biology, UT Southwestern Medical Center

Bart Loeys, M.D./Ph.D.
Postdoctoral Fellow, Medical Genetics Training Program,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2002 - 2005
Funding: Medical Genetics Training Grant, NIH R01, individual support from Belgium Research Foundation

Laureate of the De Swerts Award, Royal Academy for Medicine, 2005
Laureate of the Jacqueline Bernheim Prize, Fund for Cardiac Surgery, 2006
Co-laureate of the Glaxo-Smith Kline for Medicine, Royal Academy for Medicine, 2007
Laureate of the Antoine Marfan Award, National Marfan Foundation, 2008
Laureate of the Inbev-Baillet-Latour Prize for Clinical Research, 2009
Current Position: Professor, University of Antwerp/Antwerp University Hospital

Melissa Loscalzo, M.D.

Postdoctoral Fellow, Medical Genetics Training Program,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2001 - 2004
Funding: Medical Genetics Training Grant
Current Position: Medical Director, Clinical Genetics Program,
Johns Hopkins All Children's Hospital
Assistant Professor, Johns Hopkin's Medicine

Ronald Cohn, M.D.

Postdoctoral Fellow, Combined Pediatric and Genetics Residency Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2004 – 2006
Co-recipient, Young Investigator Award, 7th International Symposium on the Marfan Syndrome, Gent, Belgium, 2005
Best Postdoctoral Research Presentation,
McKusick-Nathans Institute of Genetic Medicine, 2005
Johns Hopkins University School of Medicine Clinician Scientist Award, 2006
1st Annual Harvard-Partners Center for Genetics and Genomics Award In Med. Genetics, 2006
The Helen B. Taussig Award, Young Investigators' Day,
The Johns Hopkins University School of Medicine, 2006
Finalist, Postdoctoral Research Award,
American Society of Human Genetics, 2006
Mentored Clinical Investigator Career Development Award in Muscle Disease Research, 2006
NIH Director's Young Innovator Award, 2008
Funding: Medical Genetics Training Grant, NIH R01, individual K08
Current Position: Pediatrician-in-Chief, The Hospital for Sick Children

Junji Chen, Ph.D.

Associate, Howard Hughes Medical Institute,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2004 - 2008
Funding: Howard Hughes Medical Institute
Current Position: Research Scientist, Pharmaceutical International

Jennifer Habashi, M.D.

Postdoctoral Fellow, Division of Pediatric Cardiology,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2004 – 2007
Co-recipient, Young Investigator Award, 7th International Symposium on the Marfan Syndrome, Gent, Belgium, 2005
Winner, Postdoctoral Research Award,
American Society of Human Genetics, 2005
Winner, American Heart Association Outstanding Research Award, 2005

Winner, Outstanding Investigator Award, Ninth Annual Pediatric Cardiovascular Disease Symposium, Children's Hospital of Philadelphia, 2006
Francis F. Schwentker Award for Excellence in Pediatric Research, 2006
Victor A. McKusick Fellowship, National Marfan Foundation, 2006
Funding: NIH R01, McKusick Fund, RTI, Pediatric Cardiology Clinic
Current Position: Assistant Professor, Division of Pediatric Cardiology, The Johns Hopkins University School of Medicine.

Dongli Huang, Ph.D,
Associate, Howard Hughes Medical Institute,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2005 – 2008
Funding: Howard Hughes Medical Institute
Current Position: Associate Professor, Medical College of Wisconsin

Benjamin Brooke, M.D.
Postdoctoral Fellow, Graduate Training Program in Clinical Investigation, Bloomberg School of Public Health, Department of Surgery and
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2005 - 2011
Finalist, Postdoctoral Award, American Society of Human Genetics, 2006
Funding: GTPCI grant, University support, individual foundation support
Current Position: Chief, Division of Vascular Surgery, Department of Surgery, The University of Utah

Christel van Erp, Ph.D.
Postdoctoral Fellow, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2006 - 2011
Funding: NIH R01
Current Position: Physiotherapist Brisbane, Australia

David Loch, Ph.D.
Postdoctoral Fellow, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2006 - 2011
Funding: NIH R01
Current Position: Senior Associate/Patent Attorney, Spruson & Ferguson, Australia

Mark Lindsay, M.D./Ph.D.
Postdoctoral Fellow, Division of Pediatric Cardiology,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2007 - 2011
Funding: NIH K08, Pediatric Cardiology
Winner, Clinician Scientist Research Award, Johns Hopkins University School of Medicine, 2010
Fredman Fellow in Aortic Disease, Massachusetts General Hospital, 2012
Current Position: Assistant Professor, Massachusetts General Hospital

Ari Zaiman, M.D./Ph.D.
Assistant Professor, Division of Medical Pulmonology,
The Johns Hopkins University School of Medicine, 2009 - 2012
Funding: NIH K08, Adult Pulmonary
Current Position: Medical Director of Pulmonary Medicine, Critical Care Medicine, Greater Baltimore Medical Center/GMBC

Neda Sharifi, Ph.D.

Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2002 - 2007
Postdoctoral Fellow, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2006 - 2008
Current Position: Associate, Foley & Lardner LLP

Stefani Fontana, Ph.D.

Human Genetics Predoctoral Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2007 - 2013
Funding: The Smilow Center for Marfan Syndrome Research
Current Position: Resident, Plastic Surgery, Mount Sinai Health System

Elizabeth Gerber, M.D./Ph.D.

Medical Scientist Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2007 – 2013
Winner, Predoctoral Research Trainee Award,
American Society of Human Genetics, 2011
Winner, Paul Ehrlich Young Investigator Award, Young Investigators' Day,
Johns Hopkins University School of Medicine, 2012
Funding: Scleroderma Research Foundation
Current Position: Resident, (Psychiatry and Behavioral Sciences), The Johns Hopkins Hospital

Elena MacFarlane (Gallo), Ph.D.

Postdoctoral Fellow, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2008 –
Funding: Helen Hay Whitney Foundation Award
Current Position: Assistant Professor, Department of Genetic Medicine, Johns Hopkins University, School of Medicine

Rosanne Rouf, M.D.

Postdoctoral Fellow, Division of Cardiology, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2008 - 2010
Funding: Cardiology Clinical, Sarnoff Foundation, Division of Cardiology
Stanley J. Sarnoff Faculty Scholar Award, 2009
Stanley L. Blumenthal Cardiology Research Award,
First Prize in Basic Sciences, 2010
W. W. Smith Heart Research Award, 2011
International Congress of Human Genetics Young Investigator Award, 2011
Current Position: Assistant Professor, University of Michigan

Anthony Guerrerio, M.D/Ph.D.

Postdoctoral Fellow, Division of Pediatric Gastroenterology,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2008 - 2010
Funding: University and individual private foundation support
American Society of Human Genetics Postdoctoral Award, 2010
George Ferry Young Investigator Development Award, 2011
Current Position: Assistant Professor of Pediatric, Division of Gastroenterology,
The Johns Hopkins University School of Medicine

Juan Calderon, Ph.D.

Human Genetics Predoctoral Training Program, Institute of Genetic Medicine, The Johns Hopkins University School of Medicine, 2008 - 2014
Fulbright Scholar
Funding: Ehlers-Danlos Syndrome Gift Account
Current Position: Faculty of Medicine, University of Development, Centro de Genética y Genómica, (Chile)

Hans Bjornsson, M.D./Ph.D.

Postdoctoral Fellow, Combined Pediatric and Genetics Residency Training Program, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2010 – 2012
Chief Resident, Genetics Residency Training Program
Funding: Medical Genetics Training Grant
Francis F. Schwentker Award, Best Clinical Fellow Research, 2009
Winner, Young Investigator Research Grant Award,
American Academy of Pediatrics, 2011
Frank L. Coulson, Jr. Award for Clinical Excellence, 2012
William K. Bowes Award, 2014
Current Position: Assistant Professor/Pediatrics and Genetics, Institute of Genetic Medicine, The Johns Hopkins University School of Medicine, Clinical Director, Department of Genetics and Molecular Medicine, Landspitali University Hospital (Iceland), Adjunct Lecturer, Faculty Medicine, University of Iceland, Reykjavik

Alexander Doyle, M.D.,

Postdoctoral Fellow, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2010 - 2014
Funding: HHMI
Semifinalist, Trainee Research, International Congress of Human Genetics, 2011
Winner, Charles J. Epstein Postdoctoral Research Award,
American Society of Human Genetics, 2012
Current Position: Assistant Professor of Anesthesiology Resident, The Royal London Hospital

Fahrner, Jill, M.D./Ph.D.

Postdoctoral Fellow, Medical Genetics Training Program,
Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2011 - 2014
Chief Resident, Genetics Residency Training Program
Funding: Medical Genetics Training Grant, IGM Clinical Residents/Fellows Budget,
Current Position: Assistant Professor, Pediatrics/Genetics, The Johns Hopkins University School of Medicine, Institute of Genetic Medicine.

Xiaoli Zhong, Ph.D.

Postdoctoral Fellow, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2011 - 2015
Current Position: Assistant Professor, Nanjing Medical University, China

Hamza Aziz, M.D.
Visiting Scholar,
The Johns Hopkins University School of Medicine, 2012 – 2014
Appleseed Resident Teaching Award, 2012
Funding: Daland Fellowship, Leducq Foundation Grant
Current Position: Fellow, Surgery-Cardiac Surgery, The Johns Hopkins University School of Medicine

Russell Gould, Ph.D.
Postdoctoral Fellow, Institute of Genetic Medicine
The Johns Hopkins University School of Medicine, 2014 – 2017
Funding: Howard Hughes Medical Institute
Current Position, Scientist, Technology Development, Johnson & Johnson

David Kim, Ph.D.
Postdoctoral Fellow, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2015 - 2016
Current Position: Scientist III, Blade Therapeutics, Inc.

Guy Bar-Klein, Ph.D.
Postdoctoral Fellow, Institute of Genetic Medicine,
The Johns Hopkins University School of Medicine, 2015 – 2020
Funding: Howard Hughes Medical Institute
Current Position:

Varun Nagpal, Ph.D.
Postdoctoral Fellow Institute of Genetic Medicine
The Johns Hopkins University School of Medicine, 2015 – 2017
Funding: Scleroderma Research Foundation
Current Position: Scientist, Cascadian Therapeutics, Inc.

Ali Keramati, M.D.
Cardiology Medicine Fellow
The Johns Hopkins University School of Medicine, 2016 - 2017
Funding: Cardiology Department
Current Position: Clinical Fellow, Cardiology, The Johns Hopkins Hospital

Benjamin Kang, Ph.D.
Cellular and Molecular Medicine Graduate Program
The Johns Hopkins University School of Medicine, 2012 – 2017
Funding: The National Marfan Foundation Gift Account
Current Position: Clinical Fellow, Emory University

Katelynn Toomer, Ph.D.
Postdoctoral Fellow, Institute of Genetic Medicine
The Johns Hopkins University School of Medicine, 2018 – Present
Funding: Howard Hughes Medical Institute

Nicole Wilson, Ph.D.
Postdoctoral Fellow, Institute of Genetic Medicine
The Johns Hopkins University School of Medicine, 2019 – Present
Funding: Howard Hughes Medical Institute

RESEARCH ACTIVITIES

Publications:

Articles:

1. Dietz HC, Pyeritz RE, Hall BD, Cadle RG, Hamosh A, Schwartz J, Meyers DA, Francomano CA. The Marfan Syndrome Locus: Confirmation of Assignment to Chromosome 15 and Identification of Tightly Linked Markers at 15q15-21.3. **Genomics.** 9:355-361,1991. PMID: 2004786

2. Dietz HC, Cutting GR, Pyeritz RE, Maslen CL, Sakai LY, Corson GM, Puffenberger EG, Hamosh A, Nanthakumar EJ, Curristin SM, Stetten G, Meyers DA, Francomano CA. Marfan Syndrome Caused by a Recurrent de novo Missense Mutation in the Fibrillin Gene. **Nature.** 352:6337-337-9, 1991. PMID: 1852208

3. Kainulainen K, Steinmann B, Collins FS, Dietz HC, Francomano CA, Child A, Kilpatrick MW, Brock DJH, Keston M, Pyeritz RE, Peltonen L. Marfan Syndrome: No Evidence for Heterogeneity in Different Populations, and More Precise Mapping of the Gene. **Am J Hum Genet.** 49:662-667, 1991. PMID: 1882844 PMCID: PMC1683142

4. Hayward C, Keston M, Brock DJH, Dietz HC. Fibrillin (FBN1) Mutations in Marfan Syndrome. **Hum Mut.** 1:79, 1992. PMID: 1301195

5. Dietz HC, Saraiva JM, Pyeritz RE, Cutting GR, Francomano CA. Clustering of Fibrillin (FBN1) Missense Mutations in Marfan Syndrome Patients at Cysteine Residues in EGF-like Domains. **Hum Mut.** 1:366-374, 1992. PMID: 1301946

6. Dietz HC. Molecular Biology of Marfan Syndrome. **J Vasc Surg.** 15:927-928, 1992. PMID: 1578571

7. Dietz HC, Pyeritz RE, Francomano CA, Sakai LY, Corson GM, Kendzior RJ, Puffenberger EG, Cutting GR. Marfan Phenotype Variability in a Family Segregating a Missense Mutation in the Epidermal Growth Factor-like Motif of the Fibrillin Gene. **J Clin Invest.** 89:1674-1680, 1992. PMID: 1569206 PMCID: PMC443046. Free Full Text in Pub Med.

8. Dietz HC, McIntosh I, Sakai LY, Corson GM, Chalberg SC, Pyeritz RE, Francomano CA. Four Novel FBN1 Mutations: Significance for Mutant Transcript Level and EGF-like Domain Calcium Binding in the Pathogenesis of Marfan Syndrome. **Genomics.** 17:468-475, 1993. PMID: 8406497

9. Dietz HC, Valle D, Francomano CA, Kendzior RJ, Pyeritz RE, Cutting GR. The Skipping of Constitutive Exons In Vivo Induced by Nonsense Mutations. **Science.** 259:680-683, 1993. PMID: 8430317

10. Christiano AM, Greenspan DS, Hoffman GG, Zhang X, Tamai Y, Lin AN, Dietz HC, Hovnanian A, Uitto J. A Missense Mutation in Type VII Collagen in Two Affected Siblings with Recessive Epidermolysis Bullosa. **Nat Genet.** 4:62-66, 1993. PMID: 8513326

11. Corson GM, Chalberg SC, Dietz HC, Charbonneau NL, Sakai LY. Fibrillin Binds Calcium and is Encoded by cDNAs that Reveal a Multidomain Structure and Alternatively Spliced Exons at the 5' End. **Genomics.** 17:476-484, 1993. PMID: 7691719

12. Dietz HC, McIntosh I, Sakai LY, Corson GM, Chalberg SC, Pyeritz RE, Francomano CA. **Genomics.** 17(2):468-75, 1993. PMID: 8406497.

13. Jin Y, Dietz HC, Nurden A, Bray PF. Single-strand conformation polymorphism analysis is a Rapid and Effective Method for the Identification of Mutations and Polymorphisms in the Gene for GPIIIa. **Blood.** 82:2281-2288, 1993. PMID: 8400280

14. Aoyama T, Tynan K, Dietz HC, Francke U, Furthmayr H. Missense Mutations Impair Intracellular Processing of Fibrillin and Microfibril Assembly in Marfan Syndrome. **Hum Mol Genet.** 2:2135-2140, 1993. PMID: 8111384

15. Dietz HC, Ramirez F, Sakai LY. Marfan Syndrome and Other Microfibrillar Diseases. **Adv Hum Genet.** 22:153-186, 1994. PMID: 7762452

16. Dietz HC, Pyeritz RE. Molecular Genetic Approaches to the Study of Human Cardiovascular Disease. **Ann Rev Physiol.** 56:763-96, 1994. PMID: 8010760

17. Dietz HC, Pyeritz RE. Molecular biology -- to the Heart of the Matter (invited editorial). **N Engl J Med.** 330:930-932, 1994. PMID: 8114867

18. Nogee LM, Garnier G, Dietz HC, Singer L, Murphy AM, deMello DE, Colten HR. A Mutation in the Surfactant Protein B Gene Responsible for Fatal Neonatal Respiratory Disease in Multiple Kindreds. **J Clin Invest.** 93:1860-1863, 1994. PMID: 8163685 PMCID: PMC294267. Free full text in Pub Med.

19. Piersall LD, Dietz HC, Hall BD, Cadle RG, Pyeritz RE, Francomano CA, McIntosh I. Substitution of a cysteine residue in a non-calcium binding, EGF-like domain of fibrillin segregates with the Marfan syndrome in a large kindred. **Hum Mol Genet**. 3:1013-1014, 1994. PMID: 7951214

20. Aoyama T, Francke U, Dietz HC, Furthmayr H. Quantitative Differences in Biosynthesis and Extracellular Deposition of Fibrillin in Cultured Fibroblasts Distinguish Five Groups of Marfan Syndrome Patients and Suggest Distinct Pathogenic Mechanisms. **J Clin Invest.** 94:130-137, 1994. PMID: 8040255 PMCID: PMC296290. Free full Text in Pub Med.

21. Pereira L, Levran O, Ramirez F, Lynch JR, Sykes B, Pyeritz RE, Dietz HC. A Molecular Approach to the Stratification of Cardiovascular Risk in Families with Marfan's Syndrome. **N Engl J Med.** 331:148-153, 1994. PMID: 8008028. Free full Text in Pub Med.

22. Dietz H, Kendzior RJ, Jr. Maintenance of an Open Reading Frame as an Additional Level of Scrutiny During Splice Site Selection. **Nat Genet.** 8:183-188, 1994. PMID: 7842017

23. Dietz HC, Francke U, Furthmayr H, Francomano C, DePaepe A, Devereux R, Ramirez F, Pyeritz R. The question of heterogeneity in Marfan syndrome. **Nat Genet.** 9:228-31, 1995. PMID: 7773282

24. Dietz HC. New insights into the genetic basis of aortic aneurysms. **Monogr Pathol**. 37:144-155, 1995. PMID: 7603481

25. Dietz HC, Pyeritz RE. Mutations in the Human Gee for Fibrillin-1 (FBN1) in the Marfan Syndrome and Releated Disorders. **Hum Mol Genet.** 4:1799-1809, 1995. PMID: 8541880

26. Eldadah ZA, Brenn T, Furthmayr H, Dietz HC. Expression of a Mutant Human Fibrillin Allele Upon a Normal Human or Murine Genetic Background Recapitulates a Marfan Cellular Phenotype. **J Clin Invest.** 95:874-880, 1995. bi PMID: 7860770 PMCID: PMC295574. Free full Text in Pub Med.

27. Nijbroek G, Sood S, McIntosh I, Francomano CA, Bull E, Pereira L, Ramirez F, Pyeritz RE, Dietz HC. Fifteen Novel FBN1 Mutations Causing Marfan Syndrome Detected By Heteroduplex Analysis of Genomic Amplicons. **Am J Hum Genet.** 57:8-21, 1995. PMID: 7611299 PMCID: PMC1801235. Free full Text in Pub Med.

28. Eldadah ZA, Grifo J, Dietz HC. Marfan Syndrome as a Paradigm For Transcript-Targeted Preimplantation Diagnosis of Heterozygous Mutations. **Nature Med.** 1:798-803, 1995.
PMID: 7585183

29. Dietz HC. Guest Editor, Cardiovascular Abnormalities of the Marfan Syndrome in Infants and Children. **Prog Pediatr Cardiol.** 5:3, 1996.

30. Dietz HC. Molecular Etiology, Pathogenesis and Diagnosis of Marfan Syndrome. **Prog Pediatr Cardiol.** 5:159-166, 1996.

31. Polymeropoulos MH, Hurko O, Hsu F, Rubenstein J, Basnet S, Lane K, Dietz H, Spetzler RF, Rigamonti D. Linkage of the locus for cerebral cavernous hemangiomas to human chromosome 7q in four families of Mexican-American descent. **Neurology.** 48:752-7, 1997. PMID: 9065560 **J Clin Invest.** 98:1745-1754, 1996. PMID: 8878424 PMCID: PMC507612. Free full text in Pub Med.

32. Gott VL, Laschinger JC, Cameron DE, Dietz HC, Greene PS, Gillinov AM, Pyeritz RE, Alejo DE, Fleischer KJ, Anhalt GJ, Stone CD, McKusick VA. The Marfan Syndrome and the Cardiovascular Surgeon. **Eur J Cardiothorac Surg.** 10:149-158, 1996. PMID: 8664013. Free full text in Pub Med.

33. Sood S, Eldadah ZA, Krause WL, McIntosh I, Dietz HC. Mutations in Fibrillin-1 and the Marfanoid-Craniosynostosis (Shprintzen-Goldberg) Syndrome. **Nat Genet.** 12:209-211, 1996. PMID: 8563763

34. O'Brien K, Dietz HC, Romagnoli M, Eiden J. Evaluation of the inhA Gene and Catalase-Peroxidase Gene among Isoniazid-Sensitive and Resistant Mycobacterium Tuberculosis Isolates. **Mol Cell Probes.** 10:1-6, 1996. PMID: 8684371

35. DePaepe A, Devereux RB, Dietz HC, Hennekam RCM, Pyeritz RE (alphabetical order). Revised Diagnostic Criteria for the Marfan Syndrome. **Am J Med Genet.** 62:417-426, 1996. PMID: 8723076

36. Dietz HC, Hamosh A. Nonstop Treatment of Cystic Fibrosis (letter). **Nature Med.** 2:608-9, 1996. PMID: 8640540

37. Morales MM, Carrol TP, Morita T, Schweibert EM, Devuyst O, Wilson PD, Lopes AG, Stanton BA, Dietz HC, Cutting GR, Guggino WB. Both Wild-type and a Functional Isoform of

CFTR Are Expressed In Kidney. **Am J Physiol.** -- Renal Fluid and Electrolyte Physiology 39:F1038-F1048, 1996. PMID: 8764323

38. Perlick HA, Medghalchi SM, Spencer FA, Kendzior RJ Jr., Dietz HC. Mammalian Orthologues of a Yeast Regulator of Nonsense Transcript Stability. **Proc Natl Acad Sci.** 93:10928-10932, 1996. PMID: 8855285 PMCID: PMC38260. Free full text in Pub Med.

39. Jin Y, Dietz HC, Montgomery RA, Bell WR, McIntosh I, Coller B, Bray PF. Glanzmann Thrombasthenia: Cooperation Between Sequence Variants in Cis During Splice Site Selection. **J Clin Invest.** 15;98(8):1745-54, 1996. PMID 8878424.

40. Gillinov AM, Zehr KJ, Redmond JM, Gott VL, Dietz HC, Reitz BA, Laschinger JC, Cameron DE. Cardiac Operations in Children with Marfan's Syndrome: Indications and Results. **Ann Thorac Surg**. 64:1140-4 – 1144-5, 1997. PMID: 9354541

41. Dietz HC. Nonsense Mutations and Altered Splice-site Selection. **Am J Hum Genet.** 60:729-730, 1997. PMID: 9042933 PMCID: PMC1712508

42. Montgomery RA, Dietz HC. Inhibition of Fibrillin-1 Expression Using U1 snRNA as a Vehicle for the Presentation of Antisense Targeting Sequence. **Hum Mol Genet.** 6:519-25, 1997. PMID: 9097954. Free full text in Pub Med.

43. Pereira L, Andrikopoulos K, Tian J, Lee, SY, Keene DR, Ono R, Reinhardt DP, Sakai LY, Jensen Biery N, Bunton T, Dietz HC, Ramirez F. Targeting of the Gene Encoding Fibrillin-1 Recapitulates the Vascular Aspect of Marfan syndrome. **Nat Genet.** 17:218-222, 1997.
PMID: 9326947

44. Dietz H. Polishing The Cutting Edge of Gems (Invited Editorial). **Nat Genet.** 20:321-322, 1998. PMID: 9843197

45. Blaszczyk A, Tang YX, Dietz HC, Adler A., Berkeley AS, Krey LC, Grifo JA. Preimplantation genetic diagnosis of human embryos for Marfan's syndrome. **J Assisted Reproduction and Genet.**15:281-284, 1998. PMID: 9604760

46. Czaplinski K, Ruiz-Echevarria MJ, Paushkin SV, Han X, Weng Y, Perlick HA, Dietz HC, Ter-Avanesyan MD, Peltz SW. The Surveillance Complex Interacts with the Translation Release Factors to Enhance Termination and Degrade Aberrant mRNAs. **Genes Devel.** 12: 1665-77, 1998. PMID: 9620853 PMCID: PMC316864. Free full text in Pub Med.

47. Sun X, Perlick HA, Dietz HC, Maquat, LE. A Mutated Human Homologue to Yeast Upf1 Protein has a Dominant-Negative Effect on the Decay of Nonsense-Containing mRNAs in Mammalian Cells. **Proc Natl Acad Sci.** 95:10009-10014, 1998. PMID: 9707591 PMCID: PMC21452. Free full text in Pub Med.

48. Maron BJ, Moller JH, Seidman CE, Vincent GM, Dietz HC, Moss AJ, Sondheimer HM, Pyeritz RE, McGee G, Epstein AE. Impact of Laboratory Molecular Diagnosis on Contemporary Diagnostic Criteria for Genetically Transmitted Cardiovascular Diseases: Hypertrophic Cardiomyopathy, Long-QT Syndrome, and Marfan Syndrome. A Statement for Healthcare Professionals From the Councils on Clinical Cardiology, Cardiovascular Disease in the Young, and Basic Science, American Heart Association**. Circulation.** 1998 98(14): 1460-71. PMID: 9841131

49. Maron BJ, Moller JH, Seidman CE, Vincent GM, Dietz HC, Moss AJ, Sondheimer HM, Pyeritz RE, McGee G, Epstein AE. Impact of Laboratory Molecular Diagnosis on Contemporary Diagnostic Criteria for Genetically Transmitted Cardiovascular Diseases: Hypertrophic Cardiomyopathy, Long-QT Syndrome, and Marfan Syndrome. A Statement for Healthcare Professionals From the Councils on Clinical Cardiology, Cardiovascular Disease in the Young, and Basic Science, American Heart Association. **Circulation.** 1998:1460-1471, 1998. PMID: 9760303. Free full text in Pub Med.

50. Montgomery RA, Geraghty MT, Bull E, Gelb BD, Johnson M, McIntosh I, Francomano CA, Dietz HC. Multiple molelcular mechanisms underlying subdiagnostic variants of Marfan syndrome. **Am J Hum Genet.** 63(6):1703-11, 1998. PMID: 9837823

51. Cserhalmi-Friedman PB, McGrath JA, Mellerio JE, Romero R, Salas-Alanis JC, Paller AS, Dietz HC, Christiano AM. Restoration of Open Reading Frame Resulting from Skipping of an Exon with an Internal Deletion in the COL7A1 Gene. **Lab Invest.** 78:1483-1492, 1998. PMID: 9881948

52. Abounader R, Ranganathan S, Lal B, Fielding K, Book A, Dietz H, Burger P, Laterra J. Reversion of Human Glioblastoma Malignancy by U1 Small Nuclear RNA/Ribozyme Targeting of Scatter Factor/Hepatocyte Growth Factor and c-met Expression. **J Natl Cancer Inst.** 91:1548-1556, 1999. PMID: 10491431. Free full text in Pub Med.

53. Biery NLJ, Eldadah ZA, Spencer FS, Dietz HC. Revised Genomic Organization of the FBN1 and Significance for Regulated Gene Expression. **Genomics.** 56:70-77, 1999. PMID: 10036187

54. Pereira L, Lee SY, Gayraud B, Andrikopoulos K, Shapiro SD, Bunton T, Biery NJ, Dietz HC, Sakai LY, Ramirez F. Pathogenetic Sequence for Aneurysm Revealed in Mice underexpressing Fibrillin-1. **Proc Natl Acad Sci.** 96: 3819-3823, 1999. PMID: 10097121 PMCID: PMC22378. Free full text in Pub Med.

55. Frischmeyer PA and Dietz HC. Nonsense-Mediated mRNA Decay in Health and Disease. **Hum Mol Genet.** 8:1893-1900, 1999. PMID: 10469842

56. Marban E, Bolli R, Breitwieser G, Busse R, Dietz H, Endoh M, Finkel T, Kass D, Lowenstein C, Rabinovitch M, Tomaselli G. Under new management: A six-month progress report on Circulation Research. **Circ Res.** 86:111-3, 2000. PMID: 10666401. Free full text in Pub Med.

57. Marban E, Bolli R, Breitwieser G, Busse R, Dietz H, Endoh M, Finkel T, Kass D, Lowenstein C, Rabinovitch M, Tomaselli G. Circulation Research Editors' yearly report: 1999-2000. **Circ Res.**18;87(4):261-3, 2000. PMID: 10948056. Free full text in Pub Med.

58. Zhang J, Clatterbuck RE, Rigamonti D, Dietz HC. Mutations in KRIT1 in familial cerebral cavernous malformations. **Neurosurgery.** 46:1272-1279, 2000. PMID: 10807272

59. Youil R,Toner TJ, Bull E, Bailey AL, Earl CD, Dietz HC, Montgomery RA. Enzymatic mutation detection (EMD) of novel mutations (R565X and R1523X) in the FBN1 gene of patients with Marfan syndrome using T4 endonuclease VII. **Hum Mutat.** 16:92-3, 2000. PMID: 10874320

60. Dietz HC and Mecham RP. Mouse models of genetic diseases resulting from mutations in elastic fiber proteins. **Matrix Biol.** 19:481-488, 2000. PMID: 11068202

61. Mendell JT, Medghalchi SM, Lake RG, Noensie EN, Dietz HC. Novel Upf2p orthologues suggest a functional link between translation initiation and nonsense surveillance complexes. **Mol Cell Biol.** 20: 8944-8957, 2000. PMID: 11073994 PMCID: PMC86549. Free full text in Pub Med.

62. Gong W, Gottlieb S, Collins J, Blescia A, Dietz H, Goldmuntz E, McDonald-McGinn DM, Zackai EH, Emanuel BS, Driscoll DA, Budarf ML. Mutation analysis of TBX1 in non-deleted patients with features of DGS/VCFS or isolated cardiovascular defects. **J Med Genet.** 38:E45, 2001. PMID: 11748311 PMCID: PMC1734783. Free full text in Pub Med.

63. Bunton TE, Biery NJ, Myers L, Gayraud B, Ramirez F, Dietz HC. Phenotypic alteration of vascular smooth muscle cells precedes elastolysis in a mouse model of Marfan syndrome. **Circ Res.** 88:37-43, 2001. PMID: 11139471. Free full text in Pub Med.

64. Medghalchi SM, Frischmeyer PA, Mendell JT, Kelly AG, Lawler AM, Dietz HC. Rent1, a trans-effector of nonsense-mediated RNA decay, is essential for mammalian embryonic viability. **Hum Mol Genet.** 10:99-105, 2001. PMID: 11152657. Free full text in Pub Med.

65. Eldadah ZA, Hamosh A, Biery NJ, Montgomery RA, Duke M, Elkins R, Dietz HC. Familial tetralogy of Fallot caused by mutation in the Jagged1 gene. **Hum Mol Genet.** 10:163-169, 2001. PMID: 11152664. Free full text in Pub Med.

66. Zhang J, Clatterbuck RE, Rigamonti D, Dietz HC. Cloning of the murine KRIT1 cDNA reveals novel mammalian 5' coding exons. **Genomics.** 70:392-395, 2000. PMID: 11161791

67. Judge DP, Biery NJ, Dietz HC. Characterization of microsatellite markers flanking FBN1: Utility in the diagnostic evaluation for Marfan syndrome. **Am J Med Genet.** 99:39-47, 2001. PMID: 11170092

68. Noensie EN and Dietz HC. A strategy for disease gene identification through nonsense-mediated mRNA decay inhibition. **Nat Biotech.** 19(5):434-9, 2001. PMID: 11329012

69. Guo D, Hasham S, Kuang SQ, Vaughan CJ, Boerwinkle E, Chen H, Abeulo D, Dietz HC, Basson CT, Shete SS, Milewicz DM. Familial thoracic aortic aneurysms and dissections: Genetic heterogeneity with a major locus mapping to 5q13-14. **Circulation.** 103:2461-8, 2001. PMID: 11369686. Free full text in Pub Med.

70. Mendell JT and Dietz HC. When the Message Goes Awry: Disease-Producing Mutations that Influence mRNA Content and Performance. **Cell.** 107:411-414, 2001. PMID: 11719181

71. Zhang J, Clatterbuck RE, Rigamonti D, Chang DD, Dietz HC. Interaction between krit1 and icap1a infers perturbation of integrin b1-mediated angiogenesis in the pathogenesis of cerebral cavernous malformation. **Hum Mol Genet.** 10: 2953-60, 2001. PMID: 11741838. Free full text in Pub Med.

72. Arking DE, Krebsova A, Macek M Sr, Macek M Jr, Arking A, Mian IS, Fried L, Hamosh A, Dey S, McIntosh I, Dietz HC. Association of human aging with functional variant of klotho. **Proc Natl Acad Sci.** 99:856-861, 2002. PMID: 11792841 PMCID: PMC117395. Free full text in Pub Med.

73. Gott VL, Cameron DE, Alejo DE, Greene PS, Shake JG, Caparrelli DJ, Dietz HC. Aortic Root Replacement in 271 Marfan Patients: A 24-Year Experience. **Ann Thorac Surg.** 73:438-43, 2002. PMID: 11845856

74. Frischmeyer PA, van Hoof A, O'Donnell K, Guerrerio AL, Parker R, Dietz HC. An mRNA surveillance mechanism that eliminates transcripts lacking termination codons. **Science.** 295:2258-61, 2002. PMID: 11910109. Free full text in Pub Med.

75. van Hoof A, Frischmeyer PA, Dietz HC, Parker R. Exosome-mediated recognition and degradation of mRNAs lacking a termination codon. **Science.** 295:2262-4, 2002. PMID: 11910110. Free full text in Pub Med.

76. Erkula G, Jones KB, Sponseller PD, Dietz HC, Pyeritz RE. Growth and maturation in Marfan syndrome. **Am J Med Genet.** 109:100-15, 2002. PMID: 11977157

77. Slavotinek AM, Dubovsky E, Dietz HC, Lacbawan F. Report of a child with aortic aneurysm, orofacial clefting, hemangioma, upper sternal defect, and Marfanoid features: Possible PHACE syndrome. **Am J Med Genet.** 110:283-8, 2002. PMID: 12116239

78. Mendell JT, ap Rhys CMJ, Dietz HC. Separable roles for rent1/hUpf1 in altered splicing and decay of nonsense transcripts. **Science.** 298:419-422, 2002. PMID: 12228722. Free full text in Pub Med.

79. Neptune ER, Frischmeyer PA, Arking DE, Myers L, Bunton TE, Gayraud B, Ramirez F, Sakai LY, Dietz HC. Dysregulation of TGF-β activation contributes to pathogenesis of Marfan syndrome. **Nat Genet.** 33:407-11, 2003. PMID: 12598898

80. Arking DE, Becker DM, Yanek LR, Fallin D, Judge DP, Moy TF, Becker LC, Dietz HC. KLOTHO Allele Status and the Risk for Early-Onset Occult Coronary Artery Disease. **Am J Hum Genet.** 72:1154-61, 2003. PMID: 12669274 PMCID: PMC1180268. Free full text in Pub Med.

81. Hutchinson S, Furger A, Halliday D, Judge DP, Jefferson A, Dietz HC, Firth H, Handford PA. Allelic variation in normal human FBN1 expression in a family with Marfan syndrome: a potential modifier of phenotype? **Hum Mol Genet.** 12:2269-76, 2003. PMID: 12915484. Free full text in Pub Med.

82. Cserhalmi-Friedman PB, Djabali K, Dietz HC, Christiano AM. Strategy to assess the efficiency of U1 RNA-hammerhead ribozymes constructs using GFP-tagged targets. **Exp Dermatol.** 12:712-5, 2003. PMID: 14705813

83. Cattaneo SM, Bethea BT, Alejo DE, Spevak PJ, Clauss SB, Dietz HC, Gott VL, Cameron DE. Surgery for Aortic Root Aneurysm in Children: A 21-Year Experience in 50 Patients. **Ann Thorac Surg**. 77:168-76, 2004. PMID: 14726057

84. Abounader R, Montgomery R, Dietz H, Laterra J. Design and expression of chimeric U1/ribozyme transgenes. **Methods Mol Biol.** 252:209-19, 2004. PMID: 15017051

85. Judge DP, Biery NJ, Keene DR, Geubtner J, Myers L, Huso DL, Sakai LY, Dietz HC. Evidence for a critical contribution of haploinsufficiency in the complex pathogenesis of Marfan syndrome. **J Clin Invest**. 114:172-81, 2004. PMID: 15254584 PMCID: PMC449744. Free full text in Pub Med.

86. Bethea BT, Fitton TP, Alejo DE, Barrerio CJ, Cattaneo SM, Dietz HC, Spevak PJ, Lima JA, Gott VL, Cameron DE. Results of aortic valve-sparing operations: experience with remodeling and reimplantation procedures in 65 Patients. **Ann Thorac Surg.** 78:767-72, 2004. PMID: 15336989

87. Mendell JT, Sharifi NA, Meyer JL, Martinez-Murillo F, Dietz HC. Nonsense surveillance regulates expression of diverse classes of mammalian transcripts and mutes genomic noise. **Nat Genet.** 36:1073-8, 2004. PMID: 15448691. Free full text in Pub Med.

88. Ramirez F, Sakai LY, Dietz HC, Rifkin DB. Fibrillin microfibrils: multipurpose extracellular networks in organismal physiology. **Physiol Genomics.** 19:151-4, 2004. PMID: 15466717. Free full text in Pub Med.

89. Bektas A, Schurman SH, Sharov AA, Carter MG, Dietz HC, Francomano CA. Klotho gene variation and expression in 20 inbred mouse strains. **Mamm Genome.** 15:759-67, 2004. PMID: 15520879

90. Sonnenday CJ, Warren DS, Cooke SK, Dietz HC, Montgomery RA. A novel chimeric ribozyme vector produces potent inhibition of ICAM-1 expression on ischemic vascular endothelium. **J Gene Med.** 6:1394-402, 2004. PMID: 15538724

91. Ng CM, Cheng A, Myers LA, Martinez-Murillo F, Jie C, Bedja D, Gabrielson KL, Hausladen JMW, Mecham RP, Judge DP, Dietz HC. TGF-β-dependent pathogenesis of mitral valve prolapse in a mouse model of Marfan syndrome. **J Clin Invest.** 114:1586-1592, 2004. PMID: 15546004 PMCID: PMC529498. Free full text in Pub Med.

92. Marban E, Bolli R, Breitwieser G, Busse R, Dietz H, Endoh M, Finkel T, Griendling K, Kass D, Lowenstein C, Tomaselli G, Keehan KH. Circulation Research Editors' Annual Report for 2004. **Circ Res.** 96:269-271, 2005.

93. Arking DE, Atzmon G, Arking A, Barzilai N, Dietz HC. Association between a functional variant of KLOTHO gene and high-density lipoprotein cholesterol, blood pressure, stroke, and longevity. **Circ Res.** 96:412-8, 2005. PMID: 15677572. Free full text in Pub Med.

94. Jones KB, Myers L, Judge DP, Kirby PA, Dietz HC, Sponseller PD. Toward an understanding of dural ectasia: a light microscopy study in a murine model of Marfan syndrome. **Spine.** 30:291-3, 2005. PMID: 15682009

95. Foran JRH, Pyeritz RE, Dietz HC, Sponseller PD. Characterization of the Symptoms Associated With Dural Ectasia in the Marfan Patient. **Am J Med Genet.** 134:58-65, 2005. PMID: 15690402

96. Loeys BL, Chen J, Neptune ER, Judge DP, Podowski M, Holm T, Meyers J, Leitch CC, Katsanis N, Sharifi N, Xu L, Myers LA, De Backer J, Hellermans J, Chen Y, Davis EC, Webb CL, Kress W, Spevak PJ, Cameron DE, Coucke P, Rifkin DB, De Paepe, Dietz HC. A syndrome of altered cardiovascular, craniofacial, neurocognitive and skeletal development caused by mutations in TGFβR1 or TGFβR2. **Nat Genet.** 37:275-81, 2005. PMID: 15731757

97. Loscalzo ML, Van PL, Ho VB, Bakalov VK, Rosing DR, Malone CA, Dietz HC, Bondy CA. Association between fetal lymphedema and congenital cardiovascular defects in Turner syndrome. **Pediatrics.** 115:732-5, 2005. PMID: 15741379. Free full text in Pub Med.

98. Milewicz DM, Dietz HC, Miller DC. Treatment of aortic disease in patients with Marfan syndrome. **Circulation.** 111:150-7, 2005. PMID: 15781745. Free full text in Pub Med.

99. Miller NH, Justice CM, Marosy B, Doheny KF, Pugh E, Zhang J, Dietz HC, Wilson AF. Identification of candidate regions for familial idiopathic scoliosis. **Spine.** 30:1181-7, 2005. PMID: 15897833

100. Clotman F, Jacquemin P, Plumb-Rudewiez N, Pierreux CE, Van der Smissen P, Dietz HC, Courtoy PJ, Rousseau GG, Lemaigre FP. Control of liver cell fate decision by a gradient of TGFβ signaling modulated by One cut transcription factors. **Genes Devel.**1849-54, 2005. PMID: 16103213 PMCID: PMC1186184

101. Vricella LA, Williams JA, Ravekes WJ, Holmes KW, Dietz HC, Gott VL, Cameron DE. Early experience with valve-sparing aortic root replacement in children. **Ann Thorac Surg.** 80:1622-6; discussion 1626-7, 2005. PMID: 16242427

102. Ramirez F, Dietz HC. Therapy Insight: aortic aneurysm and dissection in Marfan's syndrome. **Nat Clin Prac Cardiovasc Med.** 1:31-36, 2004. PMID: 16265257

103. Dietz HC, Loeys B, Carta L, and Ramirez, F. Recent progress towards a molecular understanding of Marfan syndrome. **Am J Med Genet.** 139:4-9, 2005. PMID: 16273535

104. Judge DP, Dietz HC. Marfan's syndrome. **Lancet.** 366:1965-76, 2005. PMID: 16325700 PMCID: PMC1513064. Free article in Pub Med.

105. . JL, Leitch CC, Ansley SJ, May-Simera H, Lawson S, Lewis RA, Beales PL, Dietz HC, Fisher S, Katsanis N. Dissection of epistasis in oligogenic Bardet-Biedl syndrome. **Nature.** 439:326-30, 2006. PMID: 16327777

106. Sponseller PD, Jones KB, Ahn NU, Erkula G, Foran JRH, Dietz HC. Protrusio Acetabuli in the Marfan Syndrome: Age-related Prevalence and Associated Hip Function. **J Bone Joint Surg Am.** 88:486-495, 2006. PMID: 16510812

107. Coucke PJ, Willaert A, Wessels MW, Callewaert B, Zoppi N, De Backer J, Fox JE, Mancini GM, Kambouris M, Gardella R, Facchetti F, Willems PJ, Forsyth R, Dietz HC, Barlati S, Colombi M, Loeys B, De Paepe A. Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome. **Nat Genet.** 38:452-457, 2006. PMID: 16550171

108. Robinson PN, Arteaga-Solis E, Baldock C, Collod-Beroud G, Booms P, De Paepe A, Dietz HC, Guo G, Handford PA, Judge DP, Kielty CM, Loeys B, Milewicz DM, Ney A, Ramirez F, Reinhardt DP, Tiedemann K, Whiteman P, Godfrey M. The molecular genetics of Marfan syndrome and related disorders. **J Med Genet.** 43:769-87, 2006. PMID: 16571647 PMCID: PMC2563177. Free full text in Pub Med.

109. Habashi JP, Judge DP, Holm TM, Cohn RD, Loeys BL, Cooper TK, Myers L, Klein EC, Liu G, Calvi C, Podowski M, Neptune ER, Halushka MK, Rifkin DB, Carta L, Ramirez F, Huso DL, Dietz HC. Losartan, an AT1 antagonist, prevents aortic aneurysm in a mouse model in Marfan syndrome. **Science.** 312:117-121, 2006. PMID: 16601194 PMCID: PMC1482474. Free full text in Pub Med.

110. Awad MM, Dalal D, Cho E, Amat-Alarcon N, James C, Tichnell C, Tucker A, Russell SD, Bluemke DA, Dietz HC, Calkins H, Judge DP. DSG2 mutations contribute to arrhythmogenic right ventricular dysplasia/cardiomyopathy. **Am J Hum Genet**. 79:136-42, 2006. PMID: 16773573 PMCID: PMC1474134. Free full text in Pub Med.

111. Ciaudo C, Bourdet A, Cohen-Tannoudji M, Dietz HC, Rougeulle C, Avner P. Nuclear mRNA degradation pathway(s) are implicated in Xist regulation and X chromosome inactivation. **PLoS Genet.** 2:e94, 2006. PMID: 16789828 PMCID: PMC1479048. Free full text in Pub Med.

112. De Backer J, Loeys B, Devos D, Dietz H, De Sutter J, De Paepe A. A critical analysis of minor cardiovascular criteria in the diagnostic evaluation of patients with Marfan syndrome. **Genet Med.** 8:401-8, 2006. PMID: 16845272

113. Patel ND, Williams JA, Barreiro CJ, Bethea BT, Fitton TP, Dietz HC, Lima JA, Spevak PJ, Gott VL, Vricella LA, Cameron DE. Valve-sparing aortic root replacement: early experience with the De Paulis Valsalva graft in 51 patients. **Ann Thorac Surg.** 82:548-53, 2006. PMID: 16863761

114. Loeys BL, Schwarze U, Holm T, Callewaert BL, Thomas GH, Pannu P, De Backer JF, Oswald GL, Symoens S, Manouvrier S, Roberts AE, Faravelli F, Greco MA, Pyeritz RE, Milewicz DM, Coucke PJ, Cameron DE, Braverman AC, Byers PH, De Paepe AM, Dietz HC. Aneurysm Syndromes Caused by Mutations in the TGF-β Receptor. **New Eng J Med.** 355:788-798, 2006. PMID: 16928994. Free full text in Pub Med.

115. Viassolo V, Lituania M, Marasini M, Dietz H, Benelli F, Forzano F, Faravelli F. Fetal aortic root dilation: a prenatal feature of the Loeys-Dietz syndrome. **Prenat Diagn.** 26:1081-3, 2006. PMID: 16981219

116. Guo G, Booms P, Halushka M, Dietz HC, Ney A, Stricker S, Hecht J, Mundlos S, Robinson PN. Induction of macrophage chemotaxis by aortic extracts of the mgR Marfan mouse model and a GxxPG-containing fibrillin-1 fragment. **Circulation.** 114:1855-62, 2006. PMID: 17030689 Free full text in Pub Med.

117. Jones KB, Sponseller PD, Erkula G, Sakai L, Ramirez F, Dietz HC 3rd, Kost-Byerly S, Bridwell KH, Sandell L. Symposium on the musculoskeletal aspects of Marfan syndrome: Meeting report and state of the science. **J Orthop Res.** 25:413-22, 2007. PMID: 17143900

118. Bee KJ, Wilkes D, Devereux RB, Lerman BB, Dietz HC, Basson CT. Structural and functional genetic disorders of the great vessels and outflow tracts. **Ann NY Acad Sci.** 1085:256-69, 2006. PMID: 17182942

119. Cohn RD, van Erp C, Habashi JP, Soleimani AA, Klein EC, Lisi MT, Gamradt M, Ap Rhys CM, Holm TM, Loeys BL, Ramirez F, Judge DP, Ward CW, Dietz HC. Angiotensin II type 1 receptor blockade attenuates TGFβ-induced failure of muscle regeneration in multiple myopathic states. **Nature Med.** 13:204-210, 2007. PMID: 17237794

120. Williams JA, Loeys BL, Nwakanma LU, Dietz HC, Spevak PJ, Patel ND, Francois K, De Backer J, Gott VL, Vricella LA, Cameron DE. Early surgical experience with Loeys-Dietz: a new syndrome of aggressive thoracic aortic aneurysm disease. **Ann Thor Surg.** 83:S757-63; discussion S785-90, 2007. PMID: 17257922

121. Zhang J, Rigamonti D, Dietz HC, Clatterbuck RE. Interaction between krit1 and malcavernin: implications for the pathogenesis of cerebral cavernous malformations. **Neurosurg.** 60:353-9, 2007. PMID: 17290187

122. Hanada K, Vermeij M, Garinis GA, de Waard MC, Kunen MG, Myers L, Maas A, Duncker DJ, Meijers C, Dietz HC, Kanaar R, Essers J. Perturbations of vascular homeostasis and aortic valve abnormalities in fibulin-4 deficient mice. **Circ Res.** 100:738-46, 2007. PMID: 17293478. Free full text in Pub Med.

123. Chung AW, Au Yeung K, Cortes SF, Sandor GG, Judge DP, Dietz HC, van Breemen C. Endothelial dysfunction and compromised eNOS/Akt signaling in the thoracic aorta during the progression of Marfan syndrome. **Br J Pharmacol.** 150:1075-83, 2007. PMID: 17339838 PMCID: PMC2013910. Free full text in Pub Med.

124. Holmes KW, Lehmann CU, Dalal D, Nasir K, Dietz HC, Ravekes WJ, Thompson WR, Spevak PJ. Progressive dilation of the ascending aorta in children with isolated bicuspid aortic valve**. Am J Cardiol.** 99:978-83, 2007. PMID: 17398196

125. Ramirez F, Dietz HC. Marfan syndrome: from molecular pathogenesis to clinical treatment. **Curr Opin Genet Dev.** 17:252-8, 2007. PMID: 17467262

126. Xie J, Bessling SL, Cooper TK, Dietz HC, McCallion AS, Fisher S. Manipulating mitotic recombination in the zebrafish embryo trough RecQ helicases. **Genetics.** 176:1339-42, 2007. PMID: 17483412 PMCID: PMC1894594. Free full text in Pub Med.

127. Johnson PT, Chen JK, Loeys BL, Dietz HC, Fishman EK. Loeys-Dietz Syndrome: MDCT Angiography Findings. **Am J Roentgenol.** 189:W29-35, 2007. PMID: 17579132.
Free full text in Pub Med.

128. Ramirez F, Sakai LY, Rifkin DB, Dietz HC. Extracellular microfibrils in development and disease. **Cell Mol Life Sci**. 64:2437-46, 2007. PMID: 17585369

129. Chung AW, Au Yeung K, Sandor GG, Judge DP, Dietz HC, van Breemen C. Loss of elastic fiber integrity and reduction of vascular smooth muscle contraction resulting from the upregulated activities of matrix metalloproteinase-2 and -9 in the thoracic aortic aneurysm in the Marfan syndrome. **Circ Research.** 101:512-22, 2007. PMID: 17641224. Free full text in Pub Med.

130. Loscalzo ML, Goh DL, Loeys B, Kent KC, Spevak PJ, Dietz HC. Familial Thoracic Aortic Dilation and Bicommissural Aortic Valve: A Prospective Analysis of Natural History and Inheritance. **Am J Med Genet.** 143:1960-7, 2007. PMID: 17676603

131. Faivre L, Collod-Beroud G, Loeys BL, Child A, Binquet C, Gautier E, Callewaert B, Arbustini E, Mayer K, Arlan-Kirchner M, Kiotsekoglou A, Comeglio P, Marziliano N, Dietz HC, Halliday D, Beroud C, Bonithon-Kopp C, Claustres M, Muti C, Plauchu H, Robinson PN, Ades LC, Biggin A, Benetts B, Brett M, Holman KJ, De Backer J, Coucke P, Francke U, De Paepe A, Jondeau G, Boileau C. Effect of Mutation Type and Location on Clinical Outcome in 1,013 Probands with Marfan Syndrome or Related Phenotypes and FBN1 Mutations: An International Study. **Am J Hum Genet.** 81:454-66, 2007. PMID: 17701892 PMCID: PMC1950837. Free full text in Pub Med.

132. Ramirez F, Dietz HC. Fibrillin-rich microfibrils: Structural determinants of morphogenetic and homeostatic events. **J Cell Physiol.** 213:326-30, 2007. PMID: 17708531 PMCID: PMC3042860

133. De Backer J, Loeys B, Leroy B, Coucke P, Dietz H, De Paepe A. Utility of molecular analyses in the exploration of extreme intrafamilial variability in the Marfan syndrome. **Clin Genet.** 72:188-98, 2007. PMID: 17718856

134. Judge DP, Dietz HC. Therapy of Marfan Syndrome. **Annu Rev Med.** 59:43-59, 2008. PMID: 17845137

135. Lacro RV, Dietz HC, Wruck LM, Bradley TJ, Colan SD, Devereux RB, Klein GL, Li JS, Minich LL, Paridon SM, Pearson GD, Printz BF, Pyeritz RE, Radojewski E, Roman MJ, Saul JP, Stylianou MP, Mahony L. Rationale and design of a randomized clinical trial of β-blocker therapy (atenolol) versus angiotensin II receptor blocker therapy (losartan) in individuals with Marfan syndrome. **Am Heart J.** 154:624-31, 2007. PMID: 17892982

136. Dietz HC. 2006 Curt Stern Award Address. Marfan syndrome: from molecules to medicines. **Am J Hum Genet.** 2007 81(4):662-7, 2007. PMID: 20529617

137. Xiong W, Knispel RA, Dietz HC, Ramirez F, Baxter BT. Doxycycline delays aneurysm rupture in a mouse model of Marfan syndrome. **J Vasc Surg.** 47:166-72, 2008. PMID: 18178469

138. Matt P, Habashi J, Carrel T, Van Eyk JE, Dietz HC. Recent Advances in Understanding the Marfan Syndrome – Should We Now Treat Surgical Patients with Losartan? **J Thorac Cardiovasc Surg.**135:389-94, 2008. PMID: 18242274

139. Neptune ER, Podowski M, Calvi C, Cho JH, Garcia JG, Tuder R, Linnoila RI, Tsai MJ, Dietz HC. Targeted disruption of NeuroD, a proneural basic helix-loop-helix factor, impairs distal lung formation and neuroendocrine morphology in the neonatal lung. **J Biol Chem.** 25;283:21160-9, 2008. PMID: 18339630 PMCID: PMC2475704. Free full text in Pub Med.

140. Phillips JA, Poling JS, Phillips CA, Stanton KC, Austin ED, Cogan JD, Wheeler L, Yu C, Newman JH, Dietz HC, Loyd JE. Synergistic heterozygosity for TGFβ1 SNPs and BMPR2 mutations modulates the age at diagnosis and penetrance of familial pulmonary arterial hypertension. **Genet Med.** 10:359-65, 2008. PMID: 18496036

141. Patel ND, Weiss ES, Alejo DE, Nwakanma LU, Williams JA, Dietz HC, Spevak PJ, Gott VL, Vricella LA, Cameron DE. Aortic root operations for Marfan syndrome: a comparison of the Bentall and valve-sparing procedures. **Ann Thorac Surg.** 852003-10; discussion 2010-1, 2008. PMID: 18498810

142. Brooke BS, Habashi JP, Judge DP, Patel N, Loeys B, Dietz HC 3rd. Angiotensin II blockade and aortic-root dilation in Marfan's syndrome. **New Eng J Med.** 358:2787-95, 2008. PMID: 18579813 PMCID: PMC2692965. Free full text in Pub Med.

143. Garringer HJ, Malekpour M, Esteghamat F, Mortazavi SM, Davis SI, Farrow EG, Yu X, Arking DE, Dietz HC, White KE. Molecular genetic and biochemical analyses of FGF23 mutations in familial turmoral calcinosis. **Am J Physiol Endocrinol Metab**. 295:E929-37, 2008. PMID: 18682534 PMCID: PMC2575904. Free full text in Pub Med.

144. Pearson G, Devereux R, Loeys B, Maslen C, Milewicz D, Pyeritz R, Ramirez F, Rifkin D, Sakai L, Svensson L, Wessels A, Van Eyk J, Dietz HC. Report of the National Heart, Lung and Blood Institute and National Marfan Foundation Working Group on Research in Marfan Syndrome and Related Disorders. **Circulation.** 12;118785-91, 2008. No abstract available. PMID: 18695204 PMCID: PMC2909440. Free full text in Pub Med.

145. Zhang J, Basu S, Rigamonti D, Dietz HC, Clatterbuck RE. Krit1 modulates beta1-integrin-mediated endothelial cell proliferation. **Neurosurgery**. 63:571-8; discussion 578, 2008. PMID: 18812969

146. Maleszewski J, Miller DV, Lu J, Dietz HC, Halushka MK. Histopathologic Findings in Ascending Aortas from Individuals with Loeys-Dietz syndrome (LDS). **Am J Surg Pathol.** 33:194-201, 2009. PMID: 18852674

147. Dabovic B, Chen Y, Choi J, Vassallo M, Dietz HC, Ramirez F, von Melchener H, Davis EC, Rifkin DB. Dual functions for LTBP in lung development: LTBP-4 independently modulates elastogenesis and TGF-beta activity. **J Cell Physiol.** 219:14-22, 2009. PMID: 19016471 PMCID: PMC2719250. Free full text in Pub Med.

148. Eagle KA, LeMaire SA, Volguina I, Basson CT, Devereux R, Weinsaft JW, McDermott D, Holmes KW, Dietz H, Ravekes W, Lurman K, Maslen CL, Song HK, Menashe V, Kushner JD, Pyeritz RE, Bavaria JE, Morales M, Milewicz DM, Baysinger-Morin K, Noll C, Tolunay HE, Desvigne-Nickens P, Mitchell M, Stylianou MP, Lapham C, Kroner BL, Brambilla D, Hendershot T, Ringer D, Cunningham M, Wills T, Kindem M. Rationale and design of the National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC). **Am Heart J.** 157:319-26, 2009. PMID: 19185640 PMCID: PMC2840718. Free full text in Pub Med.

149. Carta L, Smaldone, Zilberberg L, Loch D, Dietz HC, Rifkin DB, Ramirez F. p38 MAPK is an early determinant of promiscuous Smad2/3 signaling in the aortas of fibrillin-1 (Fbn1)-null mice. **J Biol Chem.** 284:5630-6, 2009. PMID: 19109253 PMCID: PMC2645821. Free full text in Pub Med.

150. Ramirez F, Dietz HC. Extracellular microfibrils in vertebrate development and disease processes. **J Biol Chem.** 29;284:14677-81,2009. PMID: 19188363
PMCID: PMC2685648 Free full text in Pub Med.

151. Volguina IV, Miller DC, Lemaire SA, Palmero LC, Wang XL, Connolly HM, Sundt TM 3rd, Bavaria JE, Dietz HC, Milewicz DM, Coselli JS. Valve-sparing and valve-replacing techniques for aortic root replacement in patients with Marfan syndrome: analysis of early outcome. **J Thorac Cardiovasc Surg.** 137(3):641-9, 2009. PMID: 19258081

152. Cameron DE, Alejo DE, Patel ND, Nwakanma LU, Weiss ES, Vricella LA, Dietz HC, Spevak PJ, Williams JA, Bethea BT, Fitton TP, Gott VL. Aortic root replacement in 372 Marfan patients: evolution of operative repair over 30 years. **Ann Thorac Surg.** 87:1344-9; discussion 1349-50, 2009. PMID: 19379862

153. Rodrigues VJ, Elsayed S, Loeys BL, Dietz HC, Yousem DM. Neuroradiologic manifestations of Loeys-Dietz syndrome type 1. **AJNR Am J Neuroradiol.** 30:1614-9, 2009. PMID: 19556353

154. Dang Y, Low WK, Xu J, Gehring NH, Dietz HC, Romo D, Liu JO. Inhibition of nonsense-mediated mRNA decay by the natural product pateamine A through eukaryotic initiation factor

4AIII. **J Biol Chem.** 28;284:23613-21, 2009. PMID: 19570977 PMCID: PMC2749136. Free full text in Pub Med.

155. Matt P, Schoenhoff F, Habashi J, Holm T, Van Erp C, Loch D, Carlson OD, Griswold BF, Fu Q, De Backer J, Loeys B, Huso DL, McDonnell NB, Van Eyk JE, Dietz HC; Circulating transforming growth factor-(beta) in Marfan syndrome. **Circulation**. 120:526-32, 2009. PMID: 19635970 PMCID: PMC2779568. Free full text in Pub Med.

156. Sponseller PD, Thompson GH, Akbarnia BA, Glait SA, Asher MA, Emans JB, Dietz HC 3rd. Growing rods for infantile scoliosis in Marfan syndrome. **Spine.** (Phila Pa 1976). 34:1711-5, 2009. PMID: 19770613

157. Patnaik S, Dietz HC, Zheng W, Austin C, Marugan JJ. Multi-Gram Scale Synthesis of FR180204. **J Org Chem**. 74:8870-3, 2009. PMID: 19852504 PMCID: PMC2862695. Free full text in Pub Med.

158. Matt P, Huso DL, Habashi J, Holm T, Doyle J, Schoenhoff F, Liu G, Black J, Van Eyk JE, Dietz HC. Murine model of surgically induced acute aortic dissection type A. **J Thorac Cardiovasc Surg**. 139:1041-7, 2010. PMID: 19910001

159. Jain D, Dietz HC, Oswald GL, Maleszewski JJ, Halushka MK. Causes and histopathology of ascending aortic disease in children and young adults. **Cardiovasc Pathol**. 20:15-25, 2011. PMID: 19926309

160. Dietz HC. TGF-Beta in the pathogenesis and prevention of disease: a matter of aneurysmic proportions. **J Clin Invest.** 120:403-7. doi: 10.1172/JC142014, 2010. PMID: 20101091 PMCID: PMC2810090. Free full text in Pub Med.

161. Loeys BL., Dietz HC. In Pagon RA, Bird TD, Dolar CR, Stephens K, Adam MP, editors. **GeneReviews™** (Internet). Seattle (WA): University of Washington, Seattle; 1993-. 2008 Feb 28 [updated 2008 Apr 29].

162. Dietz, HC. Marfan Syndrome. In: Pagon RA, Bird TD, Dolar CR, Stephens K, Adam MP, editors. **GeneReviews™** (Internet). Seattle (WA): University of Washington, Seattle; 1993-. 2001 Apr 18 [updated 2011 Dec 01].

163. Loeys BL, Gerber EE, Riegert-Johnson D, Iqbal S, Whiteman P, McConnell V, Chillakuri CR, Macaya D, Coucke PJ, DePaepe A, Judge DP, Wigley F, Davis EC, Mardon HJ, Handford P, Keene DR, Sakai LY, Dietz HC, Mutations in fibrillin-1 cause congenital scleroderma: stiff skin syndrome. **Science Transl Med.** 17;2:23ra20, 2010. PMID: 20375004 PMCID: PMC2953713 Free full text in Pub Med.

164. Renard J, Holm T, Veith R, Callewaert BL, Ades LC, Baspinar O, Pickart A, Dasouki M, Hoyer J, Rauth A, Trapane P, Earing MG, Coucke PJ, Sakai LY, Dietz HC, De Paepe AM, Loeys BL. Altered TGFbeta signaling and cardiovascular manifestations in patients with autosomal recessive cutis laxa type I caused by fibulin-4 dificiency. **Eur J Hum Genet.** 18:895-901, 2010. PMID: 20389311 PMCID: PMC2987390

165. Charbonneau NL, Jordan CD, Keene dR, Lee-Arteaga S, Dietz HC, Rifkin DB, Ramirez F, Sakai LY. Microfibril structure masks fibrillin-2 in postnatal tissues. **J Biol Chem.** 25:285:20242-51, 2010. PMID: 20404337 PMCID: PMC2888437. Free full text in Pub Med.

166. Cooper TK, Zhong Q, Krawczyk M, Tae HJ, Muller GA, Shubert R, Myers LA, Dietz HC, Talan MI, Briest W. The Haploinsufficient Col3al Mouse as a Model for Vascular Ehlers-Danlos Syndrome. **Vet Pathol.** 47:1028-39. 2010. PMID: 200587693

167. *Loeys BL, *Dietz HC, Braverman AC, Callewaert BL, De Backer J, Devereux RB, Hilhorst-Hofstee Y, Jondeau G, Faivre L, Milewicz DM, Pyeritz RE, Sponseller PD, Wordsworth P, De Paepe AM. The revised Ghent nosology for the Marfan syndrome. **J Med Genet.** 47:476-85, 2010. PMID: 20591885. **Both authors contributed equally to this work.*

168. Sponseller PD, Erkula G, Skollasky RL, Venuti KD, Dietz HC 3rd. Improving clinical recognition of Marfan syndrome. **J Bone Joint Surg AM.** 4;92:1868-75, 2010. PMID: 20686061

169. Erkula G, Sponseller PD, Paulsen LC, Oswald GL, Loeys BL, Dietz HC. Musculoskeletal findings of Loeys-Dietz syndrome. **J Bone Joint Surg Am**. 92:1876-83, 2010. PMID: 20686062

170. Dietz HC. New Therapeutic Approaches for Mendelian Disorders. **New England J Med.** 363:852-63, 2010. PMID: 20818846. Free full text in Pub Med.

171. Ting BL, Mathur D, Loeys BL, Dietz HC 3rd, Sponseller PD. The diagnostic value of the facial features of Marfan syndrome. **J Child Orthop.** 545-51, 2010. PMID: 22132032

172. Nistala H, Lee-Arteaga S, Carta L, Cook JR, Smaldone S, Siciliano G, Rifkin AN, Dietz HC, Rifkin DB, Ramirez F. Differential effects of alendronate and losartan therapy on osteopenia and aortic aneurysm in mice with severe Marfan syndrome. **Hum Mol Genet.** 19:4790-8, 2010. PMID: 20871099

173. Patel ND, Arnaoutakis GJ, George TJ, Allen JG, Aleja DE, Dietz HC, Cameron DE, Vricella LA. Valve-sparing aortic root replacement in children: intermediate-term results. Interact Cardiovasc **Thorac Surg.** 12:415-9, discussion 419, 2011. PMID: 21118834

174. Holm TM, Habashi JP, Doyle JJ, Bedja D, Chen Y, van Erp C, Lindsay M, Kim D, Schoenhoff F, Cohn RD, Loeys BL, Thomas C, Samarjit P, Marugan J, Judge DP, Dietz HC. Noncanonical TGFβ Signaling Contributes to Aortic Aneurysm Progression in Marfan Syndrome Mice, **Science.** 332:358-61, 2011. PMID: 21493862

175. Habashi JP, Doyle JJ, Holm TM, Aziz H, Scoenhoff F, Bedja D, Chen Y, Modiri AN, Judge DP, Dietz HC. Angiotensin II Type 2 Receptor Signaling Attenuates Aortic Aneurysm in Mice through ERK Antagonism. **Science.** 332:361-5, 2011. PMID: 21493863

176. Lindsay M, Dietz, HC. Lessons on the pathogenesis of aneurysm from heritable conditions. **Nature.** 473:308-16, 2011. PMID: 21593863

177. Frischmeyer-Guerrerio PA, Montgomery RA, Cooke SK, Warren DS, Sonnenday CJ, Guerrerio AL, Lutz J, Dietz HC. Perturbation of thymocyte development in nonsense-mediated decay (NMD)-deficient mice. **Proc Natl. Acad. of Sci.**108:10638-43, 2011. PMID: 21670277 PMIC: PMC3127929

178. Kuang SQ, Guo DC, Prakash SK, McDonald ML, Johnson RJ, Wang M, Regalado ES, Russell L, Cao JM, Kwartler C, Fraivillig K, Coselli JS, Safi HJ, Estrera AL, Leal SM, Lemaire SA, Belmont JW, Milewicz DM, GenTAC Investigators. Recurrent chromosome 16p13.1

duplications are a risk factor for aortic dissections. **PLoS Genet.** e1002118, 2011. PMID: 21698135

179. Mendoza DD, Kochar M, Devereux RB, Basson CT, Min JK, Dietz HC, Milewicz DM, Lemaire SA, Pyeritz RE, Bavaria JE, Masien CL, Song H, Kroner BL, Eagle KA, Weinsaft JW, GenTAC (National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions) Study Investigators. Impact of image analysis methodology on diagnostic and surgical classification of patients with thoracic aortic aneurysms. **Ann Thorac Surg.** 92:904-12. Epub 2011 Jul 2, 2011. PMID: 21723533

180. Patel ND, Arnaoutakis GJ, George TJ, Allen JG, Alejo DE, Dietz HC, Cameron DE, Vricella LA. Valve-sparing aortic root replacement in Loeys-Dietz syndrome. **Ann Thorac Surg.** 556-60, discussion 560-1, 2011. PMID: 21801912

181. Judge DP, Rouf R, Habashi J, Dietz HC. Mitral Valve Disease in Marfan Syndrome and Related Disroders. **J Cardiovasc Transl Res**., 741-7. Doi: 10.1007/s12265-011-9314-y, 2011. PMID: 21866385

182. McLoughlin D, McGuinness J, Byrne J, Terzo E, Huuskonen V, McAllister H, Black A, Kearney S, Kay E, Hill AD, Dietz HC, Redmond JM. Pravastatin reduces marfan aortic dilation. **Circulation.** 124 (11 Suppl): S168-73, 2011. PMID: 21911808

183. Renard M, Callewaert B, Baetens M, Campens L, Macdermot K, Fryns JP, Bonduelle M, Dietz HC, Gaspar IM, Cavaco D, Stattin EL, Schrander-Stumpel C, Coucke P, Loeys B, De Paepe A, De Backer J. Novel MYH11 and ACTA2 mutations reveal a role for enhanced TGFβ signaling in FTAAD. **Int J Cardiol.** 2011. PMID: 21937134

184. Kroner BL, Tolunay HE, Basson CT, Pyeritz RE, Holmes KW, Maslen CL, Milewicz DM, Lemaire SA, Hendershot T, Desvigne-Nickens P, Devereux RB, Dietz HC, Song HK, Mitchell M, Weinsaft JW, Ravekes W, Menashe V, Eagle KA. The National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC): Results from phase I and scientific opportunities in phase II. **Am Heart J.** 627-632.e.1, 2011. PMID: 21982653

185. Haskett D, Doyle JJ, Gard C, Chen H, Ball C, Estabrook MA, Encinans AC, Dietz HC, Utzinger U, Vande Geest JP, Azhar M. Altered tissue behavior of a non-aneurysmal descending thoracic aorta in the mouse model of Marfan Syndrome. **Cell Tissue Res.** 347(1):267-77, 2012. PMID: 22105919

186. Song HK, Kindem M, Bavaria JE, Dietz HC, Milewicz DM, Devereux RB, Eagle KA, Maslen CL, Kroner BL, Pyeritz RE, Holmes KW, Weinsaft JW, Menashe V, Ravekes W, Lemaire SA: Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions Consortium. Long-term implications of emergency versus elective proximal aortic surgery in patients with Marfan syndrome in the Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions Consortium Registry. **J Thorac Cardiovasc Surg.** 143(2):282-6, 2012. PMID: 22104675

187. van de Laar IM, van der Linde D, Oei EH, bos PK, Bessems JH, Bierma-Zeinstra SM, van Meer BL, Pais G, Oldenburg RA, Bekkers JA, Moeklelr A, de Graaf BM, Matyas G, Frohn-Mulder IM, Timmermans J, Hilhorst-Hofstee Y, Cobben JM, Bruggenwirth HT, van Laer L, Loeys B, DeBacker, J, Coucke PJ, Dietz HC, Willems PJ, Oostra BA, DePaepe A, Roos-Hesselink JW, Bertoli-Avella AM, Wessels MW. Phenotypic spectrum of the SMD3-related aneurysms-osteoarthritis syndrome. **J Med Genet.** 47-57, 2012. PMID: 22167769

188. Podowski M, Calvi C, Metzger S, Misono K, Poonyagariyagorn H, Lopez-Mercado A, Ku T, Lauer T, McGrath-Morrow S, Berger A, Cheadle C, Tuder R, Dietz HC, Mitzner W, Wise R, Neptune E. Angiotensin receptor blocker attenuates cigarette smoke-induced lung injury and rescues lung architecture in mice. **J Clin Invest.** 229-40, 10.1172/JCI146215, 2012. PMID: 22182843

189. Chu LC, Johnson PT, Dietz HC, Brooke BS, Arnaoutakis GJ, Black JH 3rd, Fishman EK. Vascular Complications of Ehlers-Danlos Syndrome: CT Findings. **AJR Am J Roentgenol**. 482-7, 2012. PMID: 22268198

190. Wei H, Bedja D, Koitabashi N, Xing D, Chen J, Fox-Talbot K, Rouf R, Chen S, Steenbergen C, Harmon JW, Dietz HC, Gabrielson KL, Kass DA, Semenza GL. Endothelial expression of hypoxia-inducible factor 1 protects the murine heart and aorta from pressure overload by suppression of TGF-β signaling. **Proc Natl Acad Sci.** U S A. 3;109(14):E841-50, 2012. PMID: 22403061

191. Doyle JJ, Gerber EE, Dietz HC. Matrix-dependent perturbation of TGFB signaling and disease. **FEBS Lett**. 4;586(14), 2012. PMID: 22641039

192. *Lindsay ME, *Schepers D, Bolar NA, Doyle JJ, Gallo E, Fert-Bober J, Kempers, MJE, Fishman E, Chen Y, Myers L, Bjeda D, Oswald G, Anderlid BM, Yang M, Bongers E, Timmers J, Braverman A, Canham N, Mortier G, Brunner H, Byers P, Van Eyk J, Van Laer L, *Dietz H, *Loeys BL. Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. **Nat. Genet.** 8:44(8):922-7, 2012. PMID: 22772368.
**These authors contributed equally to this work.*

193. Li Y, Foss CA, Summerfield DD, Doyle JJ, Torok CM, Dietz HC, Pomper, MG, Yu SM. Targeting collagen strands by photo-triggered triple-helix hybridization. **Proc Natl Acad Sci.** USA, 2012. PMID: 22927373

194. Gould RA, Sinha R, Aziz H, Rouf R, Dietz HC 3rd, Judge DP, Butcher J. Multi-scale biomechanical remodeling in aging and genetic mutant murine mitral valve leaflets: insights into Marfan syndrome. **PLoS One**. 2012 7(9):e44639, 2012. PMID: 22984535

195. van der Linde D, Verhagen JH, Moelker A, van de Laar IM, Van Herzeele I, De Backer J, Dietz HC, Roos-Hesselink JW. Aneurysm-osteoarthritis syndrome with visceral and iliac artery aneurysms. **J Vasc Surg.** 2012 pii: S0741-5214(12)01593-5, 2013. PMID: 22975338

196. Doyle, AJ, Doyle JJ, Bessling SI, Maragh S, Lindsay ME, Schepers D, Gillis E, Mortier G, Homfray T, Sauls Km, Norris R, Huso N, Leahy D, Mohr DW, Caulfied MJ, Scott AF, Destree A, Hennekam RC, Arn PH, Curry CJ, Van Laer L, McCallion AS, Loeys BL, Dietz HC. Mutations in the TGF-β repressor SKI cause Shprintzen-Goldberg syndrome with aortic aneurysm**. Nat Genet**. 2012 44(11):1249-54. PMID: 23023332

197. Nagy CD, Alejo De, Corretti MC, Ravekes WJ, Crosson JE, Spevak PJ, Ringel R, Carson KA, Khalil S, Dietz HC, Cameron DE, Vricella LA, Traill TA, Holmes KW. Tetralogy of Fallot and Aortic Root Dilation: A Long-Term Outlook. **Pediatr Cardiol.** 2013 34(4):809-16. PMID: 23080541

198. Kent KC, Crenshaw ML, Goh DL, Dietz HC. Genotype-phenotype correlation in patients with bicuspid aortic valve and aneurysm. **J Thorac Cardiovasc Surg.** Pii: S0022-52223(12)01214-7, 2012. PMID: 23102684

199. Holmes, KW, Maslen CL, Kindem M, Kroner BL, Song HK, Ravekes W, Dietz HC, Weinsaft JW, Roman MJ, Devereux RB, Pyeritz RE, Bavaria J, Milewski K, Milewicz D, Lemaire SA, Hendershot T, Eagle KA, Tolunay HE, Desvigne-Nickens P, Silberbach M; for GenTac Registry Consortium. GenTac registry report: Gender differences among individuals with genetically triggered thoracic aortic aneurysm and dissection. **Am J Med Genet A.** 2013 161A(4):779-86 PMID: 23444191

200. Wengrod J, Martin L, Wang D, Frischmeyer-Guerrerio P, Dietz HC, Gardner LB. The inhibition of nonsense mediated RNA decay activates autophagy. **Mol Cell Biol.** 2013 33(11):2128-35. PMID: 23508110

201. Liu X, Jin Z, O'Brien R, Bathon J, Dietz HC, Grote E, Van Eyk JE. Constrained selected reaction monitoring: Quantification of selected post-translational modifications and protein isoforms. **Methods.** 2013 15;61(3):304-12. PMID: 23523700

202. Lacro RV, Guey LT, Dietz HC, Pearson GD, Yetman AT, Gelb BD, Loeys BL, Benson DW, Bradley TJ, De Backer J, Forbus GA, Klein GL, Lai WW, Levine JC, Lewin MB, Markham LW, Paridon SM, Pierpont ME, Radojewski E, Selamet Tierney ES, Sharkey AM, Wechsler SB, Mahony L. Characteristics of children and young adults with Marfan syndrome and aortic root dilation in a randomized trial comparing atenolol and losartan therapy. **Am Heart J.** 2013, 165(5):828-835 e3. PMID: 23622922

203. Kane MS, Lindsay ME, Judge DP, Barrowman J, Ap Rhys C, Simonson L, Dietz HC, Michaelis S. LMNA-Associated Cardiocutaneous Progeria: An Inherited Autosomal Dominant Premature Aging Syndrome With Late Onset. **Am J Med Genet A.** 2013, 161A(7):1599-611. PMID: 23666920

204. Tan EW, Offoha RU, Oswald GL, Skolasky RL, Dewan AK, Zhen G, Shapiro JR, Dietz HC, Cao X, Sponseller PD. Increased fracture risk and low bone mineral density in patients with loeys-dietz syndrome. **Am J Med Genet A**. 2013. PMID: 23825031

205. Loeys BL, Mortier G, Dietz HC. Bone lessons from Marfan syndrome and related disorders: fibrillin, TGF-B and BMP at the balance of too long and too short. **Pediatr Endocrinol. Rev.** 2013, Suppl 2:417-23. PMID: 23858625

206. Frischmeyer-Guerrerio PA, Guerrerio AL, Oswald G, Chichester K, Myers L, Halushka MK, Oliva-Hemker M, Wood RA, Dietz HC. TGFβ Receptor Mutations Impose a Strong Predisposition for Human Allergic Disease. **Sci Transl Med.** 2013, 24:5(195):195ra94. PMID: 23884466

207. Liu D, Judge DP, Halushka MK, Ni J, Habashi JP, Moslehi J, Bedja D, Gabrielson KL, Xu H, Qian F, Huso D, Dietz HC, Germino GG, Watnick T. A Pkd1-Fbn1 Genetic Interaction Implicates TGF- β Signaling in the Pathogenesis of Vascular Complications in Autosomal Dominant Polycystic Kidney Disease. **J Am Soc Nephrol**. 2013, 25(1):81-91. PMID: 24071006

208. Gerber EE, Fontana SC, Gallo EM, Davis EC, Wigley F, Huso DL, Dietz HC. Integrin Modulating Therapies Prevent Fibrosis and Autoimmunity in Genetic Mouse Models of Scleroderma. **Nature.** 2013, 7;503(7474):126-30. PMID: 24107997

209. Kunkala MR, Schaff HV, Li Z, Volguina I, Dietz HC, Lemaire SA, Coselli JS, Connolly H. Mitral valve disease in patients with marfan syndrome undergoing aortic root replacement. **Circulation**. 2013, 10:128(11 Suppl 1):S243-7. PMID. 24030414.

210. Gallo EM, Loch DC, Habashi JP, Calderon JF, Chen Y, Bedja D, van Erp C, Gerber EE, Parker SJ, Sauls K, Judge DP, Cooke SK, Lindsay ME, Rouf R, Myers L, Ap Rhys CM, Kent KC, Norris RA, Huso DL, Dietz HC. Angiotensin II-dependent TGF-β signaling contributes to Loeys-Dietz syndrome vascular pathogenesis. **J Clin Invest.** 2014, 2;124(1):448-60. PMID: 24355923

211. Li W, LiQ, Jiao Y, Qin L, Ali R, Zhou J, Ferruzzi J, Kim RW, Geirsson A, Dietz HC, Offermanns S, Humphrey JD, Tellides G. Tgfbr2 disruption in postnatal smooth muscle impairs aortic wall homeostasis. **J Clin Invest**. 2014, 124(2):755-67. PMID: 24401272

212. Maccarrick G, Black JH 3rd, Bowdin S, El-Hamamsy I, Frischmeyer-Guerrerio PA, Guerrerio AL, Sponseller PD, Loeys B, Dietz HC 3rd. Loeys-Dietz syndrome: a primer for diagnosis and management. **Genet Med.** 2014 16(8):576-87. PMID: 24577266

213. Bai S, Lozada A, Jones MC, Dietz HC, Dempsey M, Das S. Mandibuloacral Dysplasia Caused by LMNA Mutations and Uniparental Disomy. **Case Rep Genet**. 2014. PMID: 24639906. PMCID: PMC3930135.

214. Coselli JS, Volguina IV, Lemaire SA, Sundt TM, Connolly HM, Stephens EH, Schaff HV, Milewicz DM, Vricella LA, Dietz HC, Minard CG, Miller DC, on behalf of the Aortic Valve Operative Outcomes in Marfan Patients Study Group. **J Thorac Cardiovasc Surg.** 2014 Jun:147(6):1758-66.e1-4. PMID: 24655904

215. Ganesh SK, Morissette R, Xu Z, Schoenhoff F, Griswold BF, Yang J, Tong L. Yang ML, Hunker K, Sloper L, Kuo S, Raza R, Milewicz DM, Francomano CA, Dietz HC, Van Eyk J, McDonnell NB. Clinical and biochemical profiles suggest fibromucular dysplasia is a systemic disease with altered TGF-β expression and connective tissue features. **FASEB J.** 2014 28(8):3313-24. PMID 24732132

216. Schepers D, Doyle AJ, Oswald G, Sparks E, Myers L, Willems PJ, Mansour S, Simpson MA, Frysira H, Maat-Kievit A, Van Minkelen R, Hoogeboom JM, Mortier GR, Titheradge H, Brueton L, Starr L, Stark Z, Ockeloen C, Lourenco CM, Blair E, Hobson E, Hurst J, Maystadt L, Destree A, Girisha KM, Miller M, Dietz HC, Loeys B, Van Laer L. The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzen-Goldberg syndrome. **Eur J Hum Genet.** 2015 23(2):224-8. PMID: 24736733.

217. Chu LC, Johnson PT, Dietz HC, Fishman EK. CT angiographic evaluation of genetic vascular disease: role in detection, staging, and management of complex vascular pathologic conditions. **AJR Am J Roentgenol**. 2014, 202(5):1120-9. PMID: 24758669

218. MacCarrick G, Loeys B, Dietz HC 3rd. Response to pyeritz et Al. **Genet Med.** 2014 16(8):642-4. PMID: 25093569

219. Lindsay ME, Dietz HC. The Genetics Basis of Aortic Aneurysm. **Cold Spring Harb Perspect Med.** 2014 4(9):a015909. PMID: 25183854

220. Bjornnson HT, Benjamin JS, Zhang L, Weissman J, Gerber EE, Chen YC, Vaurio RG, Potter MC, Hansen KD, Dietz HC. Histone deacetylase inhibition rescues structural and functional brain deficits in a mouse mouse of Kabuki syndrome. **Sci Transl Med.** 2014 1:6(256):256ra135. PMID: 25273096

221. Chetaille P, Preuss C, Burkhard S, Côté JM, Houde C, Castilloux J, Piché J, Gosset N, Leclerc S, Wünnemann F, Thibeault M, Gagnon C, Galli A, Tuck E, Hickson GR, El Amine N, Boufaied I, Lemyre E, de Santa Barbara P, Faure S, Jonzon A, Cameron M, Dietz HC, Gallo-

McFarlane E, Benson DW, Moreau C, Labuda D; FORGE Canada Consortium, Zhan SH, Shen Y, Jomphe M, Jones SJ, Bakkers J, Andelfinger G. Mutations in SGOL1 cause a novel cohesinopathy affecting heart and gut rhythm. **Nat Genet.** 2014, 46(11):1245-9. PMID: 25282101

222. Lacro RV, Dietz HC, Sleeper LA, Yetman AT, Bradley TJ, Colan SD, Pearson GD, Selamet Tierney ES, Levine JC, Atz AM, Benson DW, Braverman AC, Chen S, De Backer J, Gelb BD, Grossfeld PD, Klein GL, Lai WW, Liou A, Loeys BL, Markham LW, Olson AK, Paridon SM, Pemberton VL, Pierpont ME, Pyeritz RE, Radojewski E, Roman MJ, Sharkey AM, Stylianou MP, Wechsler SB, Young LT, Mahony L; Pediatric Heart Network Investigators. Atenolol versus losartan in children and young adults with Marfan's syndrome. **N Engl J Med**. 2014 27;37(22):2061-71. PMID: 25405392.

223. Lacro RV, Dietz HC, Mahony L. Atenolol versus Losartan in Marfan's Syndrome. **N Engl J Med.** 2015 5;372(10):980-1. PMID: 25738680.

224. Fuhrhop SK, McElroy MJ, Dietz HC 3rd, MacCarrick GL, Sponseller PD. High prevalence of cervical deformity and instability requires surveillance in loeys-dietz syndrome. **J Bone Joint Surg Am.** 2015 4;97(5):411-9. PMID: 25740032.

225. Crosas-Molist E, Meirelles T, Lopez-Luque J, Serra-Peinado C, Selva J, Caja L, Gorbenko Del Blanco D, Uriarte JJ, Bertran E, Mendizabal Y, Hernandez V, Garcia-Calero C, Busnadiego O, Condom E, Toral D, Castella M, Forteza A, Navajas D, Sarri E, Rodriguez-Pascual F, Dietz HC, Fabregat I, Egea G. Vascular smooth muscle cell phenotypic changes in patients with marfan syndrome. **Arterioscler Thromb Vasc Biol.** 2015 35(4):960-72. PMID: 25593132

226. Bertoli-Avella AM, Gillis E, Morisaki H, Verhagen JM, de Graff BM, van de Beek G, Gallo E, Kruithof BP, Venselaar H, Myers LA, Laga S, Doyle AJ, Oswald G, van Cappenllen GW, Yamanka I, van der Helm RM, Beverloo B, de Klein A, Pardo L, Lammens M, Evers C, Devriendt K, Dumoulein M, Timmermans J, Bruggenwirth HT, Verheijen F, Rodrigus I, Baynam G, Kempers M, Saenen J, Van Craenenbroeck EM, Minatoya K, Matsukawa R, Tsukube T, Kubo N, Hofstra R, Goumans MJ, Bekkers JA, Roos-Hesselink JW, van de Laar IM, Dietz HC, Van Laer L, Moresaki T, Wessels MW, Loeys BL. Mutations in a TGF-β Lingand, Cause Syndromic Aortic Aneurysms and Dissections. **J Am Coll Cardiol.** 2015 7;65(13):1324-36. PMID: 25835445.
PMCID: PMC4380321

227. Dietz HC. Potential Phenotype-Genotype Correlation in Marfan Syndrome: When Less is More? **Circ Cardiovasc Genet.** 2015 8(2):256-60. PMID: 25901038.

228. Proost D, Vandeweyer G, Meester JA, Salemink S, Kempers M, Ingram C, Peeters N, Saenen J, Vrints C, Lacro RV, Roden D, Wuyts W, Dietz HC, Mortier G, Loeys BL, Van Laer L. Performant Mutation Identification using Targetd Next Generation Sequencing of Fourteen Thoracic Aortic Aneurysm Genes. **Hum Mutat**. 2015 PMID: 25907466

229. Teixido-Tura G, Almeida AL, Choi EY, Gjesdal O, Jacobs DR Jr, Dietz HC, Liu K, Sidney S, Lewis CE, Garcia-Dorado D, Evangelista A, Gidding S, Lima JA. Determinants of Aortic Root Dilatation and Reference Values Among Young Adults Over a 20-Year Period: Coronary Artery Risk Development in Young Adults Study. **Hyertension**. 2015 115.05156. PMID: 25941347

230. Pitcher A, Emberson J, Lacro RV, Sleeper LA, Mahony L, Pearson GD, Groenink M, Mulder BJ, Zwinderman AH, DeBacker J, De Paepe AM, Arbustini E, Erdem G, Jin XY,

Flather MD, Mullen MJ, Child AH, Forteza A, Evangelista A, Chui HH, wu MH, Sandor G, Bhatt AB, Creager MA, Devereux RB, Loeys B, Forfar JC, Neubauer S, Watkins H, Boileau C, Jondeau G, Dietz HC, Baigent C. Design and rationale of prospective, collaborative meta-analysis of all randomized controlled trials of angiotensin receptor antagonists in Marfan syndrome, based on individual patient data: A report from the Marfan Treatment Trialists' Collaboration. **Am Heart J.** 2015 169(5):605-12. 2015 Feb 12. PMID: 25965707.

231. Busnadiego O, Gorbenko Del Blanco D, Gonzalez-Santamaria J, Habashi JP, Calderon JF, Sandoval P, Bedja D, Guinea-Viniegra J, Lopez-Cabrera M, Rosell-Garcia T, Snabel JM, Hanemaaijer R, Forteza A, Dietz HC, Egea G, Rodriguez-Pascual F. Evelated expression levels of lysyl oxidases protect against aortic aneurysm progression in Marfan syndrome. **J Mol Cell Cardiol.** 2015 85:48-57 PMID: 25988230.

232. Dietz HC. One intergrin to rule them all? **Sci Transl Med**. 2015 20;7(288):288fs21 PMID: 25995220

233. Dewan AK, Tomlison RE, Mitchell S, Boh BC, Yung RM, Kumar S, Tan EW, Faugere MC, Dietz HC 3rd, Clemens TL, Sponseller PD. Dysregulated TGFB signaling alters bone microstructure in a mouse model of Loeys-Dietz syndrome. **J Orthop Res**. 2015 1447-54. 234. PMID: 26173585

235. Levine RA, Hagege AA, Judge DPm Padala M, Dal-Bianco JP, Aikawa E, Beaudoin J, Bischoff J, Bouatia-Naji N, Bruneval P, Butcher JT, Carpentier A, Chaput M, Chester AH, Clusel C, Delling FN, Dietz HC, Dina C, Durst R, Fernandez-Friera L, Handschumacher MD, Jenson MO, Jeunemaitre XP, Marec HL, Tourneau TL, Markwald RR, Mérot J, Messas E, Milan DP, Neri T, Norris RA, Peal D, Perrocheau M, Probst V, Pucéat M, Rosenthal N, Solis J, Schott JJ, Schwammenthal E, Salugenhaupt SA, Song JK, Yacoub MH, Leducq Mitral Transatlantic Network. Mitral valve disease-morphology and mechanisms. **Nat Rev Cardiol.** 2015 12:689-710. PMID 26483167

236. Doyle JJ, Doyle AJ, Wilson NK, Habashi JP, Bedja D, Whitworth RE, Lindsay ME, Schoenhoff F, Myers L, Huso N, Bachir S, Squires O, Rusholme B, Ehsan H, Huso D, Thomas CJ, Caulfield MJ, Van Eyk JE, Judge DP, Dietz HC, GenTAC Registry Consortium; MIBAVA Leducq Consortium. A deleterious gene-by-environment interaction imposed by calcium channel blockers in Marfan syndrome. **Elife**. 2015 10.7554/eLife.08648. PMID: 26506064 PMCID: PMC4621743

237. Bennett CL, Aziz H, Sparks E, Shah T, Yoder M, MacCarrick G, Dietz HC. Massive hemoptysis in Loeys-Dietz syndrome. **Am J Med Genet A.** 2015 10.1002/ajmg.a.37487 PMID: 26614122

238. Price J, Magruder JT, Young A, Grimm JC, Patel ND, Alejo D, Dietz HC, Vricella LA, Cameron DE. Long-term outcomes of aortic root operations for Marfan syndrome: A comparison of Bentall versus aortic valve-sparing procedures. **J Thorac Cardiovasc** Surg. 2015. pii: S0022-5223(15)02131-5. PMID: 26704057

239. Rao SS, Venuti KD, Dietz HC 3rd, Sponseller PD. Quantifying Health Status and Function in Marfan Syndrome. **J Surg Orthop Adv**. 2016 Spring;25(1):34-40. PMID: 27082886

240. Weinsaft JW, Devereux RB, Preiss LR, Feher A, Roman MJ, Basson CT, Geevarghese A, Ravekes W, Dietz HC, Holmes K, Habashi J, Pyeritz RE, Bavaria J, Milewski K, LeMaire SA, Morris S, Milewicz DM, Prakash S, Maslen C, Song HK, Silberbach GM, Shohet RV, McDonnell N, Hendershot T, Eagle KA, Asch FM, GENTAC Registry Investigators. Aortic

Dissection in Patients With Genetically Mediated Aneurysms: Incidence and Predictors in the Gen TAC Registry. **J Am Coll Cardiol**. 2016 14;67(23):2744-54. PMID 27282895

241. Bressner JA, MacCarrick GL, Dietz HC, Sponseller, PD. Management of Scoliosis in Patients With Loeys-Dietz Syndrome. **J Pediatr Orthop.** 2017. PMID: 27379784

242. Guerrerio AL, Frischmeyer-Guerrerio PA, Huang C, Wu Y, Haritunians T, McGovern DP, MacCarrick GL, Brant SR, Dietz HC. Increased Prevalence of Inflammatory Bowel Disease in Patients with Mutations in Genes Encoding the Receptor Subunits for TGFβ. **Inflamm Bowel Dis**. 2016 22(9):2058-62. PMID: 27508510.

242. Roman MJ, Pugh NL, Hendershot TP, Devereux RB, Dietz H, Holmes K, Eagle KA, LeMaire SA, Milewicz DM, Morris SA, Pyeritz RE, Ravekes WJ, Shohet RV, Silberbach M GenTAC Investigators. Aortic Complications Associated With Pregnancy in Marfan Syndrome: the NHLBI National Registry of Genetically Triggered Aortic Aneurysms and Cardiovascular Conditions (GenTAC). **J Am Heart Assoc.** 2016 1:5(8). PMID 27515814

243. Meester JA, Vandeweyer G, Pintelon I, Lammens M, Van Hoorick L, De Belder S, Waitzman K, Young L, Markham LW, Vogt J, Richer J, Beauchesne LM, Unger S, Superti-Furga A, Prsa M, Dhilon R, Reyniers E, Dietz HC, Wuyts W, Mortier G, Verstraeten A, Van laer L, Loeys BL. Loss-of-function mutations in the X-linked biglycan gene cause a severe syndromic form of thoracic aortic aneurysms and dissections. **Genet Med.** 2016 10.1038/gim. PMID: 27632686.

244. Maleki S, Kjellgvist S, Paloschi V, Mange J, Branca RM, Du L, Hultenby K, Petrini J, Fuxe J, MIBAVA Leducq Consortium, (Collaborators: Dietz HC, Loeys B, Van Laer L, McCallion AS, Mertens L, Mital S, Mohamed SA, Andelfinger G.), Lehtio J, Franco-Cereceda A, Eriksson P, Bjorck HM. Mesenchymal state of intimal cells may explain higher propensity to ascending aortic aneurysm in bicuspid aortic valves. **Sci Rep**. 2016 6:35712. doi: 10.1038/srep35712.
PMID: 27779199

245. Patel ND, Crawford T, Magruder JT, Alejo DE, Hibino N, Black J, Dietz HC, Vricella LA, Cameron DE. Cardiovascular operations for Loeys-Dietz syndrome: Intermediate-term results. J **Thorac Cardiovasc Surg**. 2016 153:406-412 PMID: 27955909.

246. Vricella LA, Ravekes WA, Arbustini E, Jaquiss RD, Mavroudis C, Dietz HC, Jacobs ML, Hibino N, Cameron DE. Simplified mitral valve repair in pediatric patients with connective tissue disorders. **J Thorac Cardiovasc Surg.** 2017. 153(2):399-403. PMID: 28104197.

247. Dietz HC. Marfan Syndrome. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors (GeneReviews), Updated 2017. PMID: 20301510.

248. MacFarlane EG, Haupt J, Dietz HC, Shore EM. TGF-β Family Signaling in Connective Tissue and Skeletal Diseases. **Cold Spring Harb Perspect Biol.** 2017 pii: a022269. Doil 10.1101/cshperspect.a022269. PMID: 28246187.

249. Dietz HC. 2016 Presidential Address: Let's Make Human Genetics Great (Again): The Importance of Beauty in Science. **Am J Hum Genet.** 2017 100(3):379-384. doi: 10.1016/j.ajhg.2017.01.009. PMID: 28257683.

250. Patel ND, Alejo D, Crawford T, Hibino N, Dietz HC, Cameron DE, Vricella LA. Aortic Root Replacement for Children With Loeys-Dietz Syndrome. **Ann Thorac Surg.** 2017. PMID: 28366467

251. Roohi J, Kang B, Bernard D, Dietz HC, Brody LC. Moderately Elevated Homocysteine Does Not Contribute to Thoracic Aortic Aneurysm in Mice. **J Nutr**. 2017. 147(7):1290-1295. Doi: 10.3945/jn. 117.251173. PMID: 28539414.

251. Ziegler S, Ferreira CR, Gallo MacFarlane E, Riddle RC, Tomlinson R, Martin L, Ma C, Sergienko E, Pinkerton AB, Millan JL, Gahl WA, Dietz HC. Ectopic calcification in pseudoxanthoma elasticum responds to inhibition of tissue non-specific alkaline phosphatase. **Sci Transl Med**. 2017 9(393). pii: eaal1669. PMID: 28592560.

252. Gillis E, Kumar AA, Luyckx L, Preuss C, Cannaerts E, van de Beek G, Wieschendorf B, Alaerts M, Bolar N, Vandeweyer G, Meester J, Wunnemann F, Gould RA, Zhurayev R, Zerbino D, Mohamed SA, Mital S, Mertens L, Bjorck HM, Franco-Cereceda A, McCallion AS, Van Laer L, Verhagen JMA, van de Laar IMBH, Wessels MW, Goudot G, Nemcikova M, Krebsova A, Kempers M, Salemink S, Duijnhouwer T, Jeunemaitre X, Albuisson J, Eriksson P, Andelfinger G, Dietz HC, Verstraeten A, Loeys BL, Mibava Leducq Consortium. **Front Physiol.** 2017 8:400 doi: 10.3389. PMID: 28659821. PMCID: PMC5469151.

253. Rouf R, MacFarlane EG, Takimoto E, Chaudhary R, Nagpal V, Rainer PP, Bindman JG, Gerber EE, Bedja D, Schiefer C, Miller KL, Zhu G, Myers L, Amat-Alarcon N, Lee DI, Koitabash N, Judge DP, Kass DA, Dietz HC. Nonmyocyte ERK1/2 signaling contributes to load-induced cardiomyopathy in Marfan mice. **JCI Insight.** 2017 3;2 PMID: 28768908. PMCID: PMC5543913.

254. Krepp JM, Roman MJ, Devereux RB, Bruce A, Prakash SK, Morris SA, Milewicz DM, Holmes KW, Ravekes W, Shohet RV, Pyeritz RE, Masien CL, Kroner BL, Eagle KA, Preiss L, Asch FM, GenTAC Investigator. Ravekes W, Dietz HC, Holmes KW, Habashi J, Milewicz DM, Prakash SK, LeMaire SA, Morris SA, Maslen CL, Song HK, Silberbach GM, Pyeritz RE, Bavaria JE, Milewski K, Devereux RB, Weinsaft JW, Roman MJ, Shohet R, McDonnell N, Asch FM, Eagle KA, Tolunay HE, Desvigne-Nickens P, Tseng H, Kroner BL. Bicuspid and unicuspid aortic valves: Different phenotypes of the same disease? Insight from the GenTAC Registry **Congenit Heart Dis**. 2017 doi: 10.1111/chd. 12520. PMID: 28805011.

255. Gillis E, Kumar AA, Luyckx l, Preuss C, Cannaerts E, van de Beek G, Wieschendorf B, Alaerts M, Bolar N, Vandeweyer G, Meester J, Wunnemann F, Gould RA, Zhurayev R, Zerbino D, Mohamed SA, Mital S, Mertens L, Bjorck HM, Franco-Cereceda A, McCallion AS, Van Laer L, Verhagen JMA, van de Laar IMBH, Wessels MW, Messas E, Goudot G, Nemcikova M, Krebsova A, Kempers M, Salemink S, Duijnhouwer T, Jeunemaitre X, Albuisson J, Eriksson P, Andelfinger G, Dietz HC, Verstraeten A, Loeys BL, Mibava Leducq Consortium. Corrigendum: Candidate Gene Resequencing in a Large Bicuspid Aortic Valve-Associated Thoracic Aortic Aneurysm Cohort: SMAD6 as an Important Contributor. **Front Physiol.** 2017 8:730 doi: 10.3389. PMID: 28659821.

256. Björck HM1, Du L2, Pulignani S3, Paloschi V2, Lundströmer K2, Kostina AS4,5,6, Österholm C7,8, Malashicheva A4,5,6, Kostareva A4,5, Evangelista A9, Teixidó-Tura G9, Maleki S2, Franco-Cereceda A8, Eriksson P2; Mechanistic Interrogation of Bicuspid Aortic Valve associated Aortopathy (MIBAVA) Leducq Consortium. Collaborators (8) Dietz HC, Loeys B, Van Laer L, McCallion AS, Mertens L, Mital S, Mohamed SA, Andelfinger G. Altered DNA methylation indicates an oscillatory flow mediated epithelial-to-mesenchymal

transition signature in ascending aorta of patients with bicuspid aortic valve. Sci Rep. 2018 9;8(1):2777. PMID: 29426841.

257. Kirby DJ, Dietz HC, Sponseller PD. Spondylolisthesis is Common, Early, and Severe in Loeys-Dietz Sydrome. **J Pediatr Orthop** 2018 doi: 10.1097 2018. PMID: 298889773.

258. Chu LC, Haroun RR, Beaulieu RJ, Black JH 3rd, Dietz HC, Fishman EK. Carotid Artery Tortuosity Index Is Associated With the Need for Early Aortic Root Replacement in Patients With Loeys-Dietz Syndrome. **J Comput Assist Tomogr**. 2018 doi: 10.1097 PMID: 29901510.

259. Hoskoppal A, Menon S, Trachtenberg F, Burns KM, DeBacker J, Gelb BD, Gleason M, James J, Lai WW, Liou A, Mahony L, Olson AK, Pyeritz RE, Sharkey AM, Stylianou M, Wechsler SB, Young L, Levine JC, Tierney ESS, Lacro RV, Bradley TJ, *Pediatric Heart Network Investigators. Predictors of Rapid Aortic Root Dilation and Referral for Aortic Surgery in Marfan Syndrome. **Pediatr Cardiol.** 2018. doi: 10.1007/s00246-018-1016-6. PMID: 29948025

***Collaborators:**
Pearson G, Stylianou M, Pemberton V, Mahony L, Sleeper L, Tennstedt S, Colan S, Klein G, Guey L, Wruck L, Travison T, Chen S, Gerstenberger E, Olesker T, Teitel DF, Newburger J, Lacro RV, King M, Dunbar-Masterson C, Handisides J, Posa A, Nang Q, Hass C, Hsu D, Lai W, Hellenbrand W, Printz B, Roman MJ, Devereux R, Korsin R, Sherwood G, Vetter V, Paridon S, Gleason M, Pyeritz R, Mirarchi N, DiLullo S, Ejembi A, Morgan R, Morrison T, Benson DW, Border W, Cnota J, Heydarian H, James J, Hamstra M, Hogan K, Bogenschutz L, Anderson PAW, Li JS, Wechsler SB, Cook A, Sang C, Covitz W, Xu M, Sutton LJ, Crawford K, Roberts S, Palmer D, Saul JP, Atz A, Forbus G, Atz T, Infinger P, Choudhury A, Minich L, Williams R, Yetman A, Shearrow M, Robinson M, Porter J, McCrindle B, Bradley TJ, Russell J, Colman J, Radojewski E, Khaikin S, Slater N, Dietz HC, Ravekes WJ, Rykiel M, Sparks E, Oswald G, Leadroot J, Canter C, Sharkey A, Braverman A, Rainey C, Jefferies JL, Slesnick T, Liou A, Martinez H, Menesses A, Tenende T, Liang D, Merkel E, Loeys B, De Backer J, Cobben JM, Sluysmans T, De Paepe A, De Nobele S, Gelb B, Srivastava S, Mendiz-Ramdeen T, Weismann C, Lawrence E, Chin S, Ko H, Yau JL, Webber S, Drant S, Luce J, Stiegler K, Markham L, Kinnard C, Stewart C, Sommers S, Madison C, Young L, Domenico M, Waitzman K, Lozano C, Pierpont ME, Baker C, Zielinski E, Velden HV, Overman A, Lewin M, Olson A, Payne A, Rimoin D, Pariani M, Siegel R, Rafique A, Grossfeld P, Smith A, McLees-Palinkas T, Colan SD, Selamet Tierney ES, Levine J, Trevey S, Rivera M, Artman M, Austin E, Baldwin HS, Bernstein D, Feltes T, Johnson J, Klitzner T, Krischer J, Matherne GP, Zahka KG, Kugler J, Driscoll DJ, Galantowicz M, Hunsberger SA, Knight TJ, Taylor H.

260. Parker SJ, Stotland A, MacFarlane E, Wilson N, Orosco A, Venkatraman V, Madrid K, Gottlieb RA, Dietz HC, Van Eyk JE. Proteomics Reveals Rictor as a Non-Canonical TGFB Signaling Target During Aneurysm Progression in Marfan Mice. **Am J Physiol Heart Circ Physiol** 2018 1;315:H1112-H1126 PMID: 30004239

261. Renard M, Francis C, Ghosh R, Scott AF, Witmer PD, Adès LC, Andelfinger GU, Arnaud P, Boileau C, Callewaert BL, Guo D, Hanna N, Lindsay ME, Morisaki H, Morisaki T, Pachter N, Robert L, Van Laer L, Dietz HC, Loeys BL, Milewicz DM, De Backer J. Clinical Validity of Genes for Heritable Thoracic Aortic Aneurysm and Dissection. **J Am Coll Cardiol.** 2018. 7;72(6):605-615 doi: 10.1016. PMID 30071989

262. Baban A, Magliozzi M, Loeys B, Adorisio R, Alesi V, Secinaro A, Corica B, Vricella L, Dietz HC, Drago F, Novelli A, Amodeo A.First evidence of maternally inherited mosaicism in

TGFBR1 and subtle primary myocardial changes in Loeys-Dietz syndrome: a case report. **BMC Med Genet**. 2018 15;19(1):170. Doi: 10.1186/s12881-018-0661-2. PMID: 30219046 PMCID: PMC6139163

263. Handisides JC, Hollenbeck-Pringle D, Uzark K, Trachtenberg FL, Pemberton VL, Atz TW, Bradley TJ, Cappella E, De Nobele S, Groh GK, Hamstra MS1, Korsin R1, Levine JC, Lindauer B1, Liou A1, Neal MKM, Markham LW, Morrison T, Mussatto KA, Olson AK, Pierpont MEM, Pyeritz RE, Radojewski EA, Roman MJ, Xu M, Lacro RV; *Pediatric Heart Network Investigators. Health-Related Quality of Life in Children and Young Adults with Marfan Syndrome. **J Pediatr**. 2018 204:250-255 PMID: 30270167

***Pediatric Heart Network Investigators:**

Pearson G, Stylianou M, Mahony L, Sleeper L, Tennstedt S, Colan S, Klein G, Guey L, Wruck L, Travison T, Chen S, Gerstenberger E, Olesker T, Teitel DF, Newburger J, King M, Dunbar-Masterson C, Posa A, Nang Q, Hass C, Hsu D, Lai W, Hellenbrand W, Printz B, Devereux R, Sherwood G, Vetter V, Paridon S, Gleason M, Mirarchi N, DiLullo S, Ejembi A, Morgan R, Benson DW, Border W, Cnota J, Heydarian H, James J, Hogan K, Bogenschutz L, Benham MP, Barnard T, Anderson PAW, Li JS, Wechsler SB, Cook A, Sang C, Covitz W, Sutton LJ, Crawford K, Roberts S, Palmer D, Saul JP, Atz A, Forbus G, Infinger P, Choudhury A, Minich L, Williams R, Yetman A, Shearrow M, Robinson M, Porter J, McCrindle B, Russell J, Colman J, Khaikin S, Slater N, Dietz HC, Ravekes WJ, Rykiel M, Sparks E, MacCarrick G, Leadroot J, Canter C, Sharkey A, Braverman A, Rainey C, Jefferies JL, Slesnick T, Martinez H, Menesses A, Tenende T, Liang D, Merkel E, Loeys B, De Backer J, Cobben JM, Sluysmans T, De Paepe A, Gelb B, Srivastava S, Mendiz-Ramdeen T, Weismann C, Lawrence E, Chin S, Ko H, Le Yau J, Webber S, Drant S, Luce J, Stiegler K, Kinnard C, Stewart C, Sommers S, Madison C, Young L, Domenico M, Waitzman K, Lozano C, Baker C, Zielinski E, Velden HV, Overman A, Lewin M, Payne A, Rimoin D, Pariani M, Siegel R, Rafique A, Grossfeld P, Smith A, McLees-Palinkas T, Colan SD, Tierney ESS, Trevey S, Rivera M, Artman M, Austin E, Baldwin HS, Bernstein D, Feltes T, Johnson J, Klitzner T, Krischer J, Matherne GP, Zahka KG, Kugler J, Driscoll DJ, Galantowicz M42, Hunsberger SA, Knight TJ, Taylor H.

264. Gould RA, Aziz H, Woods CE, Seman-Senderos MA, Sparks E, Preuss C, Wunnemann F, Bedja D, Moats CR, McClymont SA, Rose R, Sobeira N, Ling H. MacCarrick G, Kumar AA, Luyckx I, Cannaerts E, Verstraeten A, Njork HM, Lehsau AC, Jaskula-Ranga V, Lauridsen H, Shah AA, Bennett CL, Ellinor PT, Lin H, Isselbacher EM, Lino-Cardenas CL, Butcher JT, Hughes GC, Lindsay ME, Baylor-Hopkins Center for Mendelian Genomics, MIBAVA Leducq Consortium, Mertens L, Franco-Cereceda A, Verhagen-MA J, Wessels M, Mohamed SA, Eriksson P, Mital S, Van Laer L, Loeys BL, Andelfinger G, McCallion AS, Dietz HC. *ROBO4* Variants Predispose Individuals to Bicuspid Aortic Valve and Thoracic Aortic Aneurysm. **Nature Gen.** 2018 doi: 10.1038/s41588-018-0265. PMID: 30455415

265. Williams D, Lindley KJ, Russo M, Habashi J, Dietz HC, Braverman AC. Pregnancy after Aortic Root Replacement in Marfan's Syndrome: A Case and Review of the Literature. **AJP Rep.** 2018 8(4):3234-e240 doi: 10.1055/s-038-1675347. PMID: 30473905

266. Yan J, Lehsau AC, Sauer B, Pieper B, Mohamed SA. Collaborators: Loeys BL, Dietz HC, Van Laer L, McCallion AS, Eriksson P, Franco-Cereceda A, Mertens L, Mital S, Mohamed SA, Andelfinger G., Comparison of biomechanical properties in ascending aortic aneurysms of patients with congenital bicuspid aortic valve and Marfan syndrome. **Int J Cardiol.** 2018. pii: S0167-5273(17)35074-X. doi: 10.1016/j.ijcard.2018.11.102. PMID: 30527531.

266. MacFarlane EG, Parker SJ, Shin JY, Ziegler SG, Creamer TJ, Bagirzadeh R, Bedja D, Chen Y, Calderon JF, Lindsay ME, Habashi J, Dietz HC. Lineage-Specific events in aortic root aneurysm pathogenesis in Loeys-Dietz syndrome. **J Clin Invest** 2019 pii: 123547 doi: 10.1172/JCI123547. PMID: 30614814.

267. Luyck I, Kumar AA, Reyniers, Dekeyser E, Vanderstraeten K, Vandeweyer G, Wunnemannn F, Preuss C, Mazzella JM, Goudot G, Messas E, Albuisson J, Jeunemaitre X, Eriksson P, Mohamed SA, Kempers M, Salemink S, Duijnhouwer A, Angelfinger G, Dietz HC, Verstraeten A, Van Laer L, Loeys. *MIBAVA Leducq Consortium. Copy number variation analysis in bicuspid aortic valve-related aortopathy identifies TBX20 as a contributing gene. **Eur J Hum Genet.** 2019 10.1038/s41431-019-0364-7. PMID: 30820038.

***Collaborators:** Zhurayev R, Zerbino D, Mital S, Mertens L, Franco-Cereceda A, Verhagen JMA, van de Laar IMBH, Wessels MW, Nemcikova M, Krebsova A.

268. Park JS, Oh Y, Park YJ, Park O, Yang H, Slania S, Hummerss LK, Shah AA, An HT, Jang J, Horton MR, Shin J, Dietz HC, Song E, Na DH, Park EJ, Kim K, Lee KC, Roschke VV, Hanes J, Pomper MG, Lee S. Targeting of dermal myofibroblasts through death receptor 5 arrests fibrosis in mouse models of scleroderma. **Nat Commun.** 2019 8;10(1):1128 1038/41467-019-09101-4. PMID: 30850660 PMC:6408468.

269. Fraser CD 3rd, Liu RH, Zhou X, Patel ND, Pierre AS, Jacobs ML, Dietz HC, Habashi J, Hibino N, Cameron DE, Vricella LA. Valve-sparing aortic root replacement in children Outcomes from 100 consecutive cases. **J Thorac Cardiovasc Surg.** 2019 157(3): 1100-1109. PMID: 30982542.

270. Habashi JP, Gallo-MacFarlane E, Bagizadeh R, Bowen C, Huso N, Chen Y, Bedja J, Creamer TJ, Rykiel G, Manning M, Huso D, Dietz HC. Oxytocin Antagonism Prevents Pregnancy-Associated Aortic Disease in a Mouse Model of Marfan Syndrome. **Sci Transl Med.** 2019 1;11(490). PMID: 31043570

271. Elliott MB, Ginn B, Fukunishi T, Bedja D, Suresh A, Chen T, Inoue T, Dietz HC, Santhanam L, Mao HQ, Hibino N, Gerecht S. Regenerative and durable small-diameter graft as an arterial conduit. **Proc Natl Acad Sci** U S A. 2019 116(26):12710-12719. PMID: 31182572.

272. Shin JY, Beckett JD, Bagirzadeh R, Creamer TJ, Shah AA, McMahan Z, Paik JJ, Sampedro MM, MacFarlane EG, Beer MA, Warren D, Wigley FM, Dietz HC. Epigenetic activation and memory at a *TGFB2* enchancer in systemic sclerosis. **Sci Transl Med**. 2019 19;11(4997) PMID: 31217334

273. Kim DH, Beckett JD, Nagpal V, Seman-Senderos MA, Gould RA, Creamer TJ, MacFarlane EG, Chen Y, Bedja D, Butcher JT, Mitzner W, Rouf R, Hata S, Warren DS, Dietz HC. Calpain 9 as a Novel Therapeutic Target TGFβ -Induced Mesenchymal Transition and Fibrosis. **Sci Transl Med.** 2019 11(501). PMID: 31316008.

274. Nissen AP, Thanh Truong VT, Alhafez BA, Puthumana JJ, Estrera AL, Body SC, Prakash SK, BAVCon Investigators; *GenTAC Registry Investigators. Surgical repair of bicuspid aortopathy at small diameters: Clinical and institutional factors. **J Thorac Cardiovasc. Surg** 2019 pii: S0022-523(19)31583-1. PMID: 31543305

***GenTAC Registry Investigators:**

Bossone E, Citro R, Muehlschlegel JD, Shahram JT, Nguyen TB, Stefano V, Gilon ND, Durst R, de Vincentiis C, Pluchinotta FR, Sundt TM, Michelena HI, Limongelli G, McCarthy PM, Malaisrie SC, Bavishi A, Bissell MM, Huggins GS, Dagenais F, Corte AD, Girdsaukas E, Yang B, Eagle K, Prakash SK, Milewicz DM, Nguyen TC, Sandhu HK, Safi HJ, Denny JC, Evangelista A, Galian-Gay L, Eagle KA, Ravekes W, Dietz HC, Holmes KW, Habashi J, Milewicz DM, Prakash SK, LeMaire SA, Coselli JS, Morris SA, Maslen CL, Song HK, Silberbach GM, Pyeritz RE, Bavaria JE, Milewski K, Devereux RB, Weinsaft JW, Roman MJ, Shohe RV, McDonnell N, Asch FM, Tolunay HE, Tseng H, Kroner BL.

275. Schoenhoff FS, Alejo DE, Black JH, Crawford TC, Dietz HC, Grimm JC, Magruder JT, Patel ND, Vricella LA, Young A, Carrel TP, Cameron DE. Management of the aortic patients with Loeys-Dietz syndrome. **J Thorac Cardiovasc Surg.** 2019 pii: S0022-5223 31737-4 PMID: 31627951

276. Li J, Ritelli M, Ma CS, Rao G, Habib T, Corvilain E, Bougarn S, Cypowyj S, Grodicka L, Levy R, Beziat V, Shang L, Payne K, Avery DT, Migaud M, Boucherit S, Boughhorbel S, Guennoun A, Chrabieh M, Rapaport F, Bigio B, Itan Y, Boisson B, Cormier-Daire V, Syx D, Malfait F, Zoppi N, Abel L, Freiberger T, Dietz HC, Marr N, Tangye SG, Columbi M, Casanova JL, Puel A. Chronic mucocutaneous candidiasis and connective tissue disorder in humans with impaired JNK1-dependent responses to IL-17A/F and TGF-β. Sci Immunol. 2019 29;4 PMID: 31784499

277. Bowen CJ, Calderon Gladrosic JF, Burger Z…et al, Dietz HC. Targetable cellular signaling events mediate vascular pathology in vascular Ehlers-Danlos syndrome. **J Clin Invest.** 2020 3;130(2):686-698 PMID: 31639107

278. Wunnemann F, Ta-Shma A, Preuss C, Leclerc S, van Vliet PP, Oneglia a, Thibeault M, Nordquist E, Lincoln J, Scharfenberg F, Becker-Pauly C, Hofmann P, Hoff K, Audain E, Kramer HH, Makalowski W, Nir A, Gerety SS, Hurles M, Comes J, Fournier A, Osinska H, Robins J, Puceat M. *MIBAVA Leducq Consortium principal investigators, Elpeleg O, Hitz MP, Andelfinger G. **Nat. Genet.** 2020 Jan. 52:40-47. PMID: 31844321

***Collaborators:**
Dietz HC, McCallion AS, Andelfinger G, Loeys BL, Van Laer L, Eriksson P, Mohamed SA, Mertens L, Franco-Cereceda A, Mital S.

279. Frischmeyer-Guerrerio PA, MacCarrick G, Dietz HC, Stewart FD, Guerrerio AL. Safety and Outcome of Gastrostomy Tube Placement in Patients With Loeys-Dietz Syndrome. **BMC Gastroenterol**. 2020 Mar 12;20(1):71. PMID: 32164578

280. Asgari S, Luo Y, Akbari A, Belbin GM, Xinyi L, Harris DN, Selig M, Bartell E, Calderon R, Slowikowski K, Contreras C, Yataco R, Galea JT, Jimenez J, Coit JM, Farronay C, Nazarian RM, O'Connor TD, Dietz HC, Hirschhorn JN, Heinner G, Lecca L, Kenny EE, Freeman EE, Murray MB, Raychaudhuri S. A Positively Selected FBN1 Missense Variant Reduces Height in Peruvian Individuals. **Nature** 2020 Jun;582(7811):234-239. PMID: 32499652

281. Van Driest SL, Sleeper LA, Gelb BD, Morris SA, Dietz HC, Forbus GA, Goldmuntz E, Hoskoppal A, James J, Lee TM, Levine JC, Li JS, Loeys BL, Markham LW, Meester JAN, Mital S, Mosley JD, Olson AK, Renard M, Shaffer CM, Sharkey A, Young L, Lacro RV, Roden DM. Variants in ADRB1 and CYP2C9: Association With Response to Atenolol and Losartan in Marfan Syndrome. **J Pediatr.** 2020 Jul;222:213-220.e5. PMID: 32586526

282. Anderson NK, Juzwiak EE, Dietz HC. A seX(X/Y) Article on Marfan Syndrome. **J Am Heart Assoc.** 2020 Oct 20;9(20). PMID: 33059494

283. Ziegler SG, MacCarrick G., Dietz HC., Toward Precision Medicine in Vascular Connective Tissue. **AM J Med Genet A.** 2021 10.1002/ajmg.a.62462. PMID 34428348

284. Hamosh A, Amberger JS, Bocchini CA, Bodurtha J, Bult CJ, Chute CG, Cutting GR, Dietz HC, Firth HV, Gibbs RA, Grody WW, Haendel MA, Lupski JR, Posey JE, Robinson PN, Schrimi LM, Scottt AF, Sobreira NL, Valle D, Wu N, Rasmussen SA. Response to Biesecker et. Al. **Am J Hum Genet.** 2021 108(9):1807-1808. PMID 3448655.

285. Coselli JS, Volguina IV, LeMaire SA, Connolly HM, Sundt TM, Milewicz DM, Dietz HC, Amarasekara HS, Green SY, Zhang Q, Schaff HV, Miller DC. Aortic Valve Operative Outcomes in Marfan Patients Study Group. Midterm outcomes of aortic root surgery in patients with Marfan syndrome: A prospective, multicenter, comparative study**. J Thorac Cardiovasc Surg.** 2021 (Online ahead of print). PMID: 34629178

286. Collaco JM, Raraigh KS, Betz J, Aksit MA, Blau N, Brown J, Dietz HC, MacCarrick G, Nogee LMM, Sheridan MB, Vernon HJ, Beaty TH, Louis TA, Cutting GR. Accurate assignment of disease liability to genetic variants using only population data. **Genet Med.** 2021 17;S1098-3600(21)01126-6 (Online ahead of print). PMID: 34906463

287. Guerrerio AL, Mateja A, Rasooly M, Levin S, Magnani A, Dempsey C, MacCarrick G, Dietz HC, Brittain E, Boyce AM, Frischmeyer-Guerrerio PA. Predictors of low bone density and fracture risk in Loeys-Dietz syndrome. Genet Med. 2021 16;S1098-3600(21)05352-1. (Online ahead of print). PMID: 34906513

288. Meester JAN, Peeters S, Van Den Heuvel L, Vandeweyer G, Fransen E, Cappella E, Dietz HC, Forbus G, Gelb BD, Goldmuntz E, Hoskoppal A, Landstrom AP, Lee T, Mital S, Morris S, Olson AK, Renard M, Roden DM, Singh MN, Tierney ESS, Tretter JT, Van Driest SL, Willing M, Verstraeten A, Van Laer L, Lacro RV, Loeys BL. Molecular characterization and investigation of the role of genetic variation in phenotype variability and response to treatment in a large pediatric Marfan syndrome cohort. Gent Med. 2022 17;S1098-3600(21)05474-5. PMID 35058154

289. Cottrill EJ, Bowen CJ, Pennington ZA, Murray JA, Rajkovic CJ, Dietz HC 3rd, Sponseller PD. Tendon Healing in a Mouse Model of Loeys-Dietz Syndrome: Controlled Study Using a Patellar Tendon Transection Model. J Pediatr Orthop. 2022 (Online ahead of print). PMID: 35442932

Invited Book Chapters:

1. Dietz HC. "Fluids and Electrolytes" in The Harriet Lane Handbook, Eleventh Edition, Rowe PC, (editor), Chicago, Yearbook Medical Publishers, 1987.

2. Dietz HC. "Pediatric Sedation" in The Harriet Lane Handbook, Eleventh Edition, Rowe PC, (editor), Chicago, Yearbook Medical Publishers, 1987.

3. Dietz HC and Oski FA. "Differential Diagnosis" in Principles and Practice of Pediatrics, Oski FA, (editor-in-chief), DeAngelis C, Feigin R, and Warshaw J. (editors), Philadelphia, J. B. Lippincott Co., 1989.

4. Dietz HC, Pyeritz RE. "The Marfan Syndrome" in Connective Tissue and Its Heritable

Disorders, Wiley-Liss, pp. 437-468, 1993.

5. Dietz HC. "Coma" in Primary Pediatric Care, Second Edition, Hoekelman, R.A. (editor-in-chief), Blatman S, Friedman S, Nelson N, and Seidel, H. (editors), St. Louis, C.V. Mosby Co., 1994.

6. Dietz HC. "Dehydration" in Primary Pediatric Care, Second Edition, Hoekelman, R.A. (editor-in-chief), Blatman S, Friedman S, Nelson N, and Seidel H. (editors), St. Louis, C.V. Mosby Co., 1994.

7. Dietz HC, Hamosh A. "Heritable Metabolic Heart Disease" in Critical Heart Disease in Infants and Children, Nichols DG, Cameron DE, Greely WJ, Lappe DG, Underleider RN, Wetzel RC (editors), St. Louis, Mosby-Yearbook, 1995.

8. Dietz HC. "New Insights into the Genetic Basis of Aortic Aneurysms" in Cardiovascular Pathology: Clinicopathologic Correlations and Pathogenic Mechanisms, Schoen FJ, Gimbrone MA (editors), Baltimore, Williams and Wilkins, 1995.

9. Dietz HC. "Marfan Syndrome" in Molecular Genetics and Gene Therapy of Cardiovascular Diseases, Mockrin SC (editor), New York, Marcel Dekker, Inc. pp 219-237, 1996.

10. Dietz HC, McKusick VA. "Inherited Vascular Disease" in Molecular Basis of Cardiovascular Disease" A Companion to Braunwald's Heart Disease, Chien, K.R. (editor), W.B. Saunders Company, Philadelphia, pp. 167-187, 1999.

11. Wright MJ and Dietz HC. "Heritable Disorders of Connective Tissue" in Oski's Pediatrics: Principles and Practice, Third Edition, McMillan JA, DeAngelis CD, Feigin RD and Warshaw JB (Editors), Philadelphia, Lippincott Williams and Wilkins, pp. 1892-1899, 1999.

12. Pyeritz RE, Dietz HC. "The Marfan Syndrome" in The Metabolic and Molecular Bases of Inherited Disease, Eighth Edition, vol. 4, Scriver CR, Beaudet AL, Sly WS, and Valle D (Editors), New York, McGraw-Hill, pp. 5287-5311, 2001.

13. Pyeritz RE and Dietz HC. "Marfan Syndrome and Other Microfibrillar Disorders" in Connective Tissue and Its Heritable Disorders, Second Edition, Royce PM and Steinmann B (Editors), John Wiley & Sons, New York, pp. 585-626, 2002.

14. Dietz HC. "Marfan Syndrome" in NORD Guide to Rare Diseases, Lippincott, Williams & Wilkins. Philadelphia, pp. 218-9, 2003

15. Dietz HC. "Marfan Syndrome" in GeneClinics: Medical Genetics Knowledge Base (www.geneclinics.org).

16. Mendell JT and Dietz HC. "Nonsense-mediated mRNA Decay" in Encyclopedia of the Human Genome, Nature Publishing Group.

17. Loeys BL and Dietz HC. "*TGFBR 1/2* and Loeys-Dietz Syndrome" in Inborn Errors of Development, Second Edition, Epstein CJ, Erickson RP, and Wynshaw-Boris A (Editors), Oxford University Press, New York, pp. 417-424, 2008.

18. De Backer J, De Paepe D, Loeys BL, Dietz HC. "Echocardiography in Pediatric and Congenital Heart Disease" from Fetus to Adult, Lai WL, Mertens LL, Cohen MS, Geva T (Editors). Wiley-Blackwell, pp. 537-546, 2009.

19. Doyle JJ, and Dietz HC. "Marfan Syndrome" in Nelson Textbook of Pediatrics, 19th Edition, Kliegman RM, Stanton B, St. Geme J, Schor NF, and Behrman RE, (Editors), Elsevier Saunders, Pennsylvania, pp. 2440 - 2446, 2011

20. Doyle JJ, and Dietz HC. "Marfan Syndrome and Related Disorders" in Metabolic and Molecular Bases of Inherited Disease, (8th ed.), Scriver C, Sly WS, Childs B, Beaudet AL, Valle D, and Knizler KW, (Editors), McGraw-Hill, New York, 2001, Chapter 206, (Updated November 2012)

21. Gerber EE and Dietz HC. "Fibrosis: Insights from the Stiff Skin Syndrome" in Scleroderma in From Pathogenesis to Comprehensive Management, Varga J, Denton CP, and Wigley FM, (Editors), Springer, New York, Chapter 22, 2012

22. Cohn RD, Dietz HC., "Molecular pathogenesis of skeletal muscle abnormalities in marfan syndrome" in Muscle, Vol. 2. p. 995-1007 7 p. 2012

23. Lindsay ME and Dietz HC. "The Genetic Basis of Aortic Aneurysm" in Biology of the Heart K.R. Chien, K. Mummery (Editors) Cold Spring Harbor Laboratory Press, 2013

24. Van Laer L, Dietz HC, Loeys B. "Loeys-Dietz syndrome" in Advances in Experimental Medicine and Biology, Vol. 802 p. 95-105 11p., 2014.

25. Lindsay ME and Dietz Harry C. "Congential Thoracic Aortic Aneurysm" in Monograms in Human Genetics. Karger Press, 2015

Abstracts:

1. Dietz HC, Francomano CA, Cutting GR, Maslen CL, Sakai LY, Pyeritz RE. The Fibrillin Locus and Heritable Disorders of Connective Tissue: Genotype-Phenotype Associations. **Proceedings of the Greenwood Genetics Center** 1991. (Platform presentation at the 12th Annual David W. Smith Workshop on Malformations and Morphology, Lake Arrowhead UCLA Conference Center, September, 1991)

2. Kainulainen K, Steinmann B, Collins F, Dietz HC, Francomano CA, Child A, Kilpatrick MW, Brock DJ, Keston M, Kaitila I, Pyeritz RE, Peltonen L. Marfan Syndrome: No Evidence for Heterogeneity in Different Populations and More Precise Mapping of the Gene. **Am J Hum Genet** 49:16S, 1991. (Platform Presentation at the International Society of Human Genetics Meeting, Washington, D.C., October, 1991)

3. Dietz HC, Pyeritz RE, Sakai LY, Francomano CA, Cutting GR. Variability of Cellular and Clinical Phenotype in the Marfan Syndrome: Three Novel Mutations in the Fibrillin Gene. **Pediatr Res** 31:132A, 1992. (Platform presentation at the Society for Pediatric Research Meetings, Baltimore, MD, May 1992)

4. Dietz HC, Sakai LY, Cutting GR, Pyeritz RE, Francomano CA. A Frameshift Mutation in the Fibrillin Gene: Reduced Transcript, Stable Truncated Protein, and Mild Marfan Phenotype. **Clin**

Res 40:294A, 1992. (Platform presentation at the American Federation of Clinical Research Meetings, Baltimore, MD, April 1992)

5. Pyeritz RE, Gott VL, Dietz HC, Cameron D, Cutting GR, Reitz BA, Francomano CA. Management of the Marfan Syndrome: Bench to Bedside. **The Journal of Japanese College of Angiology** 32:814, 1992. (Platform presentation at the Japanese Vascular Society Meeting, Tokyo, Japan, August 1992)

6. Dietz HC, Pyeritz RE, Cutting GR, Francomano CA. Mutations in the Fibrillin Gene and Variability of the Marfan Syndrome. **J Mol Cell Cardiol** 24 (supplement V):S.76, 1992. (Platform presentation at the International Society of Heart Research: European Section, Heidelberg, Germany, October 1992)

7. Dietz HC, Pyeritz RE, Sakai LY, Cutting GR, Francomano CA. Fibrillin 15 gene defects causing the Marfan Syndrome. (Platform presentation at the Fourth International Conference on the Molecular Biology and Pathology of the Extacellular Matrix, Philadelphia PA, June 1992)

8. Dietz HC, Francomano CA, Sakai LY, Cutting GR, Pyeritz RE, Maumenee IH. Mutations in the Fibrillin Gene are Associated with Diverse Ophthalmologic and Other Clinical Manifestations of Marfan Syndrome. (Platform presentation at the IX Meeting of the International Society of Genetic Eye Disease and the VI International Symposium on Retinoblastoma. Siena, Italy, 1992.)

9. Dietz HC, Francomano CA, Kendzior RJ, Pyeritz RE, Cutting GR. Skipping of a Constitutive Exon in Vivo Rescues the Open Reading Frame of a Nonsense Allele in a Patient with Marfan Syndrome. **Am J Hum Genet** 51:A200, 1992. (Platform presentation at the American Society of Human Genetics, San Francisco, CA, October 1992).

10. Francomano CA, Saraiva JM, Cutting GR, Pyeritz RE, Dietz HC. Clustering of Fibrillin 15 Missense Mutations in Marfan Syndrome Patients at Cysteine Residues in EGF-like Domains. **Am J Hum Genet** 51:A17, 1992. (Platform presentation at the American Society of Human Genetics Meeting, San Francisco, CA, October 1992)

11. Dietz HC, Pyeritz, RE, Sakai LY, Bull EC, Francomano CA. Discordant Marfan Syndrome Severity in Two Patients with Frameshift Mutations in the Fibrillin Gene (FBN1) and Severe Reduction of Mutant Allele Transcript. **Am J Med Genet** (suppl.) 1993. (Platform presentation at the International Marfan Syndrome Symposium, San Francisco, CA, October 1993)

12. Dietz HC, Saraiva JM, Pyeritz RE, Cutting GC, Francomano CA. Clustering of Fibrillin-1 Missense Mutations in Marfan Syndrome Patients at Cysteine Residues in EGF-like Domains. **Am J Med Genet** (suppl.) 1993. (Platform presentation at the International Marfan Syndrome Symposium, San Francisco, CA, October 1993)

13. Dietz HC, Kendzior RJ, Francomano CA, Pyeritz RE, Cutting GR. A Nonsense Mutation Induces Skipping of a Constitutive Fibrillin (FBN1) Exon In Vivo in a Patient with Marfan Syndrome. **Am J Med Genet** (suppl.) 1993. (Platform presentation at the International Marfan Syndrome Symposium, San Francisco, CA, October 1993)

14. Dietz HC, Cutting GR, Francomano CA, Pyeritz RE. Variability of Cellular and Clinical Phenotype in the Marfan Syndrome: Five Novel Mutations in the Fibrillin Gene. **Proceedings of the Greenwood Genetics Center** 12:73-74, 1993. (Platform presentation at the David W. Smith Workshop on Malformations and Morphology, Lake Arrowhead UCLA Conference Center, September, 1992)

15. Eldadah ZA, McIntosh I, Pyeritz RE, Francomano CA, Dietz HC. Four Novel FBN1 Mutations Implicate Mutant Transcript Level and EGF-like Domain Calcium Binding in the Pathogenesis of Marfan Syndrome. **Am J Hum Genet** 53:A1154, 1993. (Platform presentation at the American Society of Human Genetics, New Orleans, LA, October 1993)

16. Nogee LM, Murphy AM, Dietz HC. Identification of a Common Mutant Allele In the Pulmonary Surfactant B (SP-B) Gene in Patients with Neonatal Alveolar Proteinosis. **Am J Hum Genet** 53:A93, 1993. (Platform presentation at the American Society of Human Genetics, New Orleans, LA, October 1993)

17. Dietz HC, Kendzior RJ Jr, Eldadah ZA. Nonsense Mutations Act In Cis to Alter Splice-Site Selection: Verification in a Heterologous Expression System. **Am J Hum Genet** 53:A675, 1993. (Platform presentation at the American Society of Human Genetics, New Orleans, LA, October 1993)

18. Eldadah ZA, Dietz HC. Production of a Marfan Cellular Phenotype by Expression of a Mutant Human Fibrillin Allele Upon a Normal Human or Mouse Genetic Background. **Pediatr Res** 35(supp):151A, 1994. (Platform presentation at the Society for Pediatric Research Meeting, Anaheim, CA, May 1994)

19. Kendzior RJ, Valle D, Dietz HC. Maintenance of Open Reading Frame: An Additional Level of Scrutiny for Exon Definition During RNA Splicing. **Pediatric Res** 35(supp):152A, 1994. (Platform presentation at the Society for Pediatric Research Meeting, Anaheim, CA, May 1994)

20. Nijbroek G, Bull E, Dietz HC. Identification of a Hypermutable CpG Dinucleotide in FBN1: De Novo Transition as the Cause of Marfan Syndrome in Three Sporadic Cases. (Platform presentation at the 3rd International Symposium on the Marfan Syndrome, Berlin, Germany, September 1994)

21. Nijbroek G, Pereira L, Ramirez F, Dietz HC. MDE Heteroduplex Analysis of PCR Products Spanning Each FBN1 Exon Greatly Increases Mutation Detection Efficiency in MFS. (Platform presentation at the 3rd International Symposium on the Marfan Syndrome, Berlin, Germany, September, 1994.)

22. Pereira L, Levran O, Ramirez F, Lynch JR, Sykes B, Pyeritz RE, Dietz HC. A Molecular Approach to the Stratification of Cardiovascular Risk Within Families With the Marfan Syndrome. (Platform presentation at the 3rd International Symposium on the Marfan Syndrome, Berlin, Germany, September, 1994)

23. Eldadah ZA, Brenn T, Furthmayer H, Dietz HC. Expression of a Mutant Fibrillin Allele on a Normal Human or Murine Genetic Background Reproduces the Marfan Cellular Phenotype. (Platform presentation at the 3rd International Symposium on the Marfan Syndrome, Berlin, Germany, September, 1994)

24. Eldadah ZA, Brenn T, Furthmayer H, Dietz HC. Expression of a Mutant Fibrillin Allele on a Normal Human or Murine Genetic Background Reproduces the Marfan Cellular Phenotype. **Am J Hum Genet** 55:A4, 1994. (Plenary presentation at the American Society of Human Genetics Meeting, Montreal, Canada, October, 1994)

25. Nijbroek G, Pereira L, Ramirez F, Dietz HC. MDE Heteroduplex Analysis of PCR Products Spanning Each Exon of the Fibrillin (FBN1) Gene Greatly Increases the Efficiency of Mutation Detection in the Marfan Syndrome. **Am J Hum Genet** 55:A233, 1994. (Poster presentation at the American Society of Human Genetics Meeting, Montreal, Canada, October, 1994)

26. Eldadah ZA, Grifo J, Dietz H. Preimplantation Diagnosis of Marfan Syndrome: Obstacles and Corrective Strategies. **Pediatr Res** 37(part 2):148A, 1995. (Platform presentation at the Society for Pediatric Research, San Diego, CA, 1995).

27. Dietz HC, Kendzior RJ, Aberdam D, Baudoin C, Ciatti S, Guggino W, Meneguzzi G, Uitto J, Christiano A. Intermolecular recombination and use of reverse complement splice sites in the regulated expression of a chimeric transcript *in vivo*. **Pediatr Res** 37(part 2):147A, 1995. (Platform presentation at the Society for Pediatric Research, San Diego, CA, 1995)

28. Nogee LM, Jacobstein D, Dietz HC. Allelic hetergeneity in surfactant protein B (SP-B) deficiency. **Pediatr Res** 37(part 2): 344A, 1995. (Platform presentation at the Society for Pediatric Research, San Diego, CA, 1995

29. Eldadah ZA, Grifo JA, Dietz HC. Marfan syndrome as a paradigm for transcript-targeted pre-implantation diagnosis of heterozygous mutations. **Am J Hum Genet** 57(suppl):A32, 1995. (Platform presentation at the American Society of Human Genetics Meeting, Minneapolis, MN, October 1995) **Finalist, Predoctoral Clinical Award.**

30. Dietz HC, Sood S, McIntosh I. The phenotypic continuum associated with FBN1 mutations includes the Shprintzen-Goldberg syndrome. **Am J Hum Genet** 57(suppl): A211, (Poster presentation at the American Society of Human Genetics Meeting, Minneapolis, MN, October 1995)

31. Perlick HA, Medghalchi SM, Spencer FA, Dietz HC. Cloning and characterization of a human regulator of nonsense transcript stability. **Am J Hum Genet** 59:A32, 1996. (Platform presentation at the American Society of Human Genetics Meeting, San Francisco, CA, October 1996) **Winner, Predoctoral Basic Award.**

32. Biery NLJ, Pereira L, Bunton T, Sakai LY, Ramirez F, Dietz HC. Phenotypic Characterization of a mouse model of Marfan Syndrome. **Am J Hum Genet** 59:A46, 1996. (Platform presentation at the American Society of Human Genetics Meeting, San Francisco, CA, October 1996)

33. Montgomery RA, Dietz HC. U1 snRNA as a vehicle for the delivery of antisense targeting sequences. **Am J Hum Genet** 59:A10, 1996. (Platform presentation at the American Society of Human Genetics Meeting, San Francisco, CA, October 1996) **Winner, Postdoctoral Basic Award.**

34. Arking DE, Perlick HA, Dietz HC. Direct evidence for nuclear nonsense surveillance. **Am J Hum Genet** 61:A25, 1997. (Platform presentation at the American Society of Human Genetics Meeting, Baltimore, MD, October 1997) **Nominated, Predoctoral Basic Award.**

35. Perlick HA, Dietz HC. Dissociation of translation and nonsense surveillance. Presentation at the Third Annual Meeting of the RNA Society, Madison, WI, May 1998.

36. Sun X, Perlick HA, Dietz HC, Maquat LE. The human homologue of yeast Upf1 protein functions in mammalian cells to mediate the decay of nonsense-containing RNAs. Presentation at the Third Annual Meeting of the RNA Society, Madison, WI, May 1998.

37. Biery N, Eldadah Z, Spencer F, Dietz HC. Revised genomic structure of FBN1 and significance for regulated expression. **Am J Hum Genet** 63:A176, 1998. (Poster presentation at the American Society of Human Genetics Meeting, Denver, CO, October 1998).

38. Perlick HA, Dietz HC. Ongoing nonsense mediated RNA decay despite severe impairment of translation. **Am J Hum Genet** 63:A189, 1998. (Poster presentation at the American Society of Human Genetics Meeting, Denver, CO, October 1998).

39. Mendell JT, Rashid A, Bull E, Jabs EW, Dietz HC. Examination of mutant *Gus* and TCOF1 transcripts reveals cooperation between tissue- and allele-specific factors indetermining the efficiency of nonsense-mediated RNA decay. **Am J Hum Genet** 63:A56, 1998. (Platform presentation at the American Society of Human Genetics Meeting, Denver, CO, October 1998).

40. Montgomery R, Geraghty MT, Bull E, Dietz HC. Multiple molecular mechanisms underlying variants of the Marfan syndrome. **Am J Hum Genet** 63:A375, 1998. (Poster presentation at the American Society of Human Genetics Meeting, Denver, CO, October 1998).

41. Medghalchi S and Dietz HC. Rent1, a Mammalian trans-effector of Nonsense-Mediated RNA Decay, is Essential for Early Embryonic Viability. **Proceedings of the Fourth Annual RNA Society Meeting**, p. 411, 1999. (Poster presentation at the RNA '99 Meeting, Edinburgh, Scotland, June 1999).

42. Medghalchi S, Lake R, and Dietz HC. Trans- and Cis-effectors of Nonsense-Mediated Decay in Schizosaccharomyces Pombe. **Proceedings of the Fourth Annual RNA Society Meeting**, p. 412, 1999. (Poster presentation at the RNA '99 Meeting, Edinburgh, Scotland, June 1999).

43. Medghalchi S and Dietz HC. Rent1, a Mammalian Trans-Effector of Nonsense-Mediated RNA Decay is Essential for Cellular Viability. **Am J Hum Genet** 65:A80, 1999. (Poster presentation at the American Society of Human Genetics Meeting, San Francisco, CA, October 1999.)

44. Arking DE, Macek M Jr, Krebsova A, Macek M Sr, Arking A, Mcintosh I, and Dietz HC. Genetic Association with Human Survival and Aging at the Klotho Locus. **Am J Hum Genet** 65:A195, 1999. (Poster presentation at the American Society of Human Genetics Meeting, San Francisco, CA, October 1999.)

45. Cserhalmi-Friedman PB, Dietz HC, and Christiano AM. Application of Mutation Specific Hammerhead Ribozymes as Gene Therapy Tools for Skin Diseases. **Am J Hum Genet** 65:A308, 1999. (Poster presentation at the American Society of Human Genetics Meeting, San Francisco, CA, October 1999.)

46. Biery N, Bunton T, Keene D, Sakai L, Gayraud B, Ramirez F, and Dietz HC. Dedifferentiation of Vascular Smooth Muscle Cells Initiates Vascular Disease in an Animal Model of Marfan Syndrome. **Am J Hum Genet** 65:A18, 1999. (Plenary Slide Session at the American Society of Human Genetics Meeting, San Francisco, CA, October 1999.)

47. Mendell JT, Medghalchi S, Lake RG, and Dietz HC. Structural and Functional Analysis of A Novel Mammalian Trans-Effector of Nonsense-Mediated RNA Decay. **Am J Hum Genet** 65:A376, 1999. (Poster presentation at the American Society of Human Genetics Meeting, San Francisco, CA, October 1999.)

48. Neptune ER, Bunton TE, Biery NJ, Frischmeyer PA, Gayraud B, Ramirez F, and Dietz HC. TGF-Beta mediates failure of lung branching morphogenesis in fibrillin-1 deficient mice. (American Lung Association/American Thoracic Society International Conference, Toronto, May 2000.)

49. Mendell JT, Noensie EN, Lake RG, Medghalchi SM, and Dietz HC. Rent2, a novel mammalian trans-effector of nonsense-mediated RNA decay. **Proceedings of the Fifth Annual RNA Society Meeting**, p. 187, 2000. (Poster presentation at the RNA 2000 Meeting, Madison, WI, May 2000)

50. Montgomery RA, Cooke SK, Ratner LE, and Dietz HC. A novel approach to knocking-out ICAM-1 expression in the vascular endothelium. (Presentation at the International Transplantation Congress, Rome, August 2000.)

51. Eldadah Z, Hamosh A, Vermeiren F, and Dietz HC. Tetralogy of Fallot with characteristic facies: A novel syndrome caused by mutation in *Jagged1*. **Am J Hum Genet** 67:A22, 2000. (Slide Session at American Society of Human Genetics Meeting, Philadelphia, PA, October 2000.)

52. Noensie EN and Dietz HC. A strategy for disease gene identification through NMD inhibition (GINI). **Am J Hum Genet** 67:A74, 2000. (Slide Session at American Society of Human Genetics Meeting, Philadelphia, PA, October 2000.)

53. Frischmeyer PA, O'Donnell K, and Dietz HC. Nonstop decay: A novel mRNA decay pathway. **Am J Hum Genet** 67:A205, 2000. (Slide Session at American Society of Human Genetics Meeting, Philadelphia, PA, October 2000.)

54. Neptune E, Frischmeyer PA, Arking DE, Bunton TE, Myers L, Ramirez F, and Dietz HC. TGFβ mediates impaired alveologenesis in fibrillin-1 deficient mice. **Am J Hum Genet** 67:A288, 2000. (Slide Session at American Society of Human Genetics Meeting, Philadelphia, PA, October 2000.)

55. Levy HP, Johnston JJ, Davis J, Balog J, Rose PS, Schäffer A, Dietz HC, and Francomano CA. A new connective tissue disorder with features overlapping those of Ehlers-Danlos, Marfan and Stickler syndromes. **Am J Hum Genet** 67:A545, 2000. (Poster Presentation at American Society of Human Genetics Meeting, Philadelphia, PA, October 2000.)

56. Arking DE, Krebsova A, Macek M Jr., Macek M, Arking A, Mian IS, Fried LP, Hamosh A, Dey S, McIntosh I, and Dietz HC. Mutations in Klotho are associated with survival in diverse human populations. **Am J Hum Genet** 67:A1231, 2000. (Poster Presentation at American Society of Human Genetics Meeting, Philadelphia, PA, October 2000.)

57. Judge DP, Biery NJ, and Dietz HC. Characterization of microsatellite markers flanking FBN1: Utility in the diagnostic evaluation for Marfan syndrome. **Am J Hum Genet** 67:A1331, 2000. (Poster Presentation at American Society of Human Genetics Meeting, Philadelphia, PA, October 2000.)

58. Judge DP, Martin S, Williams NR, Montgomery RA, Dietz HC, Chatterjee S. Neutral sphingomyelinase-deficient cells are resistant to apoptosis caused by pro-inflammatory cytokines. (Poster presentation at the American Heart Association Scientific Sessions, New Orleans, LA, November 2000.)

59. Zhang J, Clatterbuck RE, Rigamonti D, Chang DD, and Dietz HC. Novel insights regarding the pathogenesis of cerebral cavernous malformation (CCM). (Platform presentation at American Society of Human Genetics Meeting, San Diego, CA, October 2001.)

60. Judge DP, Biery NJ, Myers L, Sakai LY, and Dietz HC. Novel murine models of Marfan syndrome provide insight into disease pathogenesis. (Platform presentation at American Society of Human Genetics Meeting, San Diego, CA, October 2001.)

61. Neptune ER, Judge DP, Ramirez F, Sakai LY, and Dietz HC. Novel therapeutic strategies in Marfan syndrome (MFS). (Platform presentation at American Society of Human Genetics Meeting, San Diego, CA, October 2001.)

62. Mendell JT, Laken HA, and Dietz HC. rent1, a mammalian *trans*-effector of nonsense-mediated mRNA decay, shuttles between the nucleus and cytoplasm. (Platform presentation at American Society of Human Genetics Meeting, San Diego, CA, October 2001.) **Nominated, Predoctoral Basic Science Award.**

63. Frischmeyer PA, Montgomery RA, Cooke SK, Warren DS, Sonnenday CJ, and Dietz HC. Perturbation of thymic development in nonsense-mediated decay-deficient mice. (Platform presentation at American Society of Human Genetics Meeting, San Diego, CA, October 2001.) **Nominated, Predoctoral Basic Science Award.**

64. Arking DE and Dietz HC. Functional dissection of a human klotho allele that influences longevity. (Platform presentation at American Society of Human Genetics Meeting, San Diego, CA, October 2001.)

65. Gong W, Gottlieb S, Collins J, Blescia A, Dietz HC, McDonald-McGinn DM, Zackai EH, Emanuel BS, Driscoll DA, and Budarf ML. Mutation screening and further analysis of *TBX1* in non-deleted patients with Di-George/Velocardiofacial syndrome (DGS/VCFS). (American Society of Human Genetics Meeting, San Diego, CA, October 2001.)

66. Frischmeyer PA, Montgomery RA, Cooke SK, Warren DS, Sonnenday CJ, Guerrerio AL, Morrell C, and Dietz HC. RNA-Initiated Allelic Exclusion in Nonsense-Mediated Decay (NMD)-Deficient Mice. (American Society of Human Genetics Meeting, Baltimore, MD, October 2002.)

67. Goh D, Han L, Judge DP, Geubtner JA, McIntosh I, Patel A, Thomas GH, Basson CT, Milewicz DM, Dietz HC. Linkage of Familial Bicupsid Aortic Valve with Aortic Aneurysm to Chromosome 15q. (American Society of Human Genetics, Baltimore, MD, October 2002.)

68. Arking DE, Becker DM, Yanek LR, Judge D, Moy TF, Becker LC, Dietz HC. *KLOTHO* allele status as a potent and environmentally modified risk factor for atherosclerosis. (American Society of Human Genetics, Baltimore, MD, October 2002.)

69. Mendell JT, ap Rhys CMJ, Dietz HC. Direct involvement of the nonsense surveillance machinery in nonsense-mediated altered splicing. (American Society of Human Genetics, Baltimore, MD, October 2002.)

70. Frischmeyer-Guerrerio PA, Montgomery RA, Cooke SK, Warren DS, Sonnenday CJ, Guerrerio AL, Dietz HC. A translation-independent role for TCRb mRNA in fetal thymic development. (American Society of Human Genetics, Los Angeles, CA, November 2003.)

71. Judge DP, ap Rhys CMJ, Guerrerio P, Geubtner J, Zhang J, Cheng A, Dietz HC. Perturbation of alternative splicing in a novel cardiocutaneous progeria syndrome caused by mutation in lamin A/C. (American Society of Human Genetics, Los Angeles, CA, November 2003.)

72. Mendell JT, Sharifi NA, Dietz HC. Nonsense surveillance regulates diverse classes of physiologic transcripts and mutes a noisy genome. (American Society of Human Genetics, Los Angeles, CA, November 2003.)

73. Neptune ER, Russell AC, Cho JH, Tsai MJ, Dietz HC. Identification of neuroD as a downstream effector of abnormal lung morphogenesis in a mouse model of Marfan syndrome. (American Society of Human Genetics, Los Angeles, CA, November 2003.)

74. Zhang J, Clatterbuck RE, Basu S, Rigamonti D, Dietz HC. Perturbation of multiple beta-1 integrin-mediated signaling cascades in the pathogenesis of cerebral vascular malformations. (American Society of Human Genetics, Los Angeles, CA, November 2003.)

75. Zhang J, Sharmila B, Rigamonti D, Dietz HC, Clatterbuck RE. Krit1 plays a major role in β1 integrin-modulated cell proliferation and survival: implications for cerebral cavernous malformation (CCM). (Neurosurgical Society of America Annual Meeting, Santa Fe, New Mexico, 2004.)

76. Zhang J, Sharmila B, Rigamonti D, Clatterbuck RE, Dietz HC. Pathogenesis of cerebral cavernous malformation: Depletion of Krit1 leads to perturbation of β1 integrin-mediated endothelial cell mobility and survival. *Am J Hum Genet*, S61 (Slide session) (suppl), 2004.

77. Ng CM, Cheng A, Judge DP, Dietz HC. TGFβ-dependent pathogenesis of mitral valve prolapse in a mouse model of Marfan syndrome. (American Society of Human Genetics 54th Annual Meeting, Toronto, Ontario, Canada, October 2004, oral)

78. Zhang J, Sharmila B, Rigamonti D, Dietz HC, Clatterbuck RE. Depletion of Krit1 leads to perturbation of β1 integrin-mediated endothelial cell angiogenesis in the pathogenesis of cerebral cavernous malformation (CCM). *Stroke* 36(2) S36 (Slide session), 2005.

79. Couke PJ, Willaert A, Calleweart B, Wessels MW, Mancini GM, De Backer J, Fox JE, Kambouris M, Gardella R, Barlati S, Colombi M, Dietz HC, Loeys B, De Paepe A. Mutations in GLUT10/SLC2A10, a facilitative glucose transporter, cause arterial tortuosity syndrome. (American Society of Human Genetics, Salt Lake City, UT, October 2005.)

80. Cohn RD, Pardo J, Loeys BL, Holm TM, Judge DP, Dietz HC. TGFβ-induced failure of satellite cell performance and muscle regeneration in mouse models of Marfan syndrome and other myopathic states. (American Society of Human Genetics, Salt Lake City, UT, October 2005.)

81. Habashi JP, Holm T, Loeys BL, Bedja D, Neptune ER, Judge DP, Dietz HC. AT1 blockade rescues multisystem manifestations of Marfan syndrome independent of hemodynamic effects. (American Society of Human Genetics, Salt Lake City, UT, October 2005.)

82. Badano JL, Leitch CC, Ansley SJ, May-Simera H, Lewis RA, Beales PL, Dietz HC, Fisher S, Katsanis N. Dissection of epistasis in an oligogenic disorder. (American Society of Human Genetics, Salt Lake City, UT, October 2005.)

83. Loeys B, Chen J, Scharze U, De Backer J, Braverman A, McDonnell N, Coucke P, Holm T, Thomas G, Francomano C, Byers P, De Paepe A, Dietz H. Phenotypic spectrum and pathogenesis of Loeys-Dietz aortic aneurysm syndrome. (American Society of Human Genetics, Salt Lake City, UT, October 2005.)

84. Moslehi J, Wang C, Judge D, Piontek K, Deng J, Germino G, Watnick T, Dietz HC. Interaction between Pkd1 and Fbn1 in the pathogenesis of aortic aneurysm. (American Society of Human Genetics, Salt Lake City, UT, October 2005.)

85. Holm TM, Loeys B, Habashi J, Meyers L, Bedja D, Neptune E, Judge D, Dietz H. Epistatic interaction between fibrillin-1 and effectors of TGFβ signaling in the pathogenesis of aortic aneurysm in Marfan syndrome. (American Society of Human Genetics, Salt Lake City, UT, October 2005.)

86. Brooke BS, Habashi JP, Dietz HC. Losartan decreases aortic root dilation in Marfan syndrome. (American Society of Human Genetics, New Orleans, LA, October 2006.)

87. Cohn RD, Habashi JP, Loeys BL, Klein EC, Gamradt M, Holm TM, Judge DP, Dietz HC. Angiotensin II Type 1 Receptor Blockade Improves TGFβ-induced Failure of Muscle Regeneration in the mdx Mouse Model of Duchenne Muscular Dystrophy. (American Society of Human Genetics, New Orleans, LA, October 2006.)

88. Sharifi NA, Dietz HC, Coupling of UPR induction with Aminoglycosides: A Potential Therapeutic Strategy for Genetic Disorders Caused by Nonsense Mutations. (American Society of Human Genetics, New Orleans, LA, October 2006.)

89. Habashi JP, Gamradt M, Awad M, Klein E, Bedja D, Dietz HC. Losartan modifies the predisposition for dissection in a mouse model of Marfan syndrome. (American Society of Human Genetics, New Orleans, LA, October 2006.)

90. Loeys B, Schwarze U, Holm T, Callewaert B, Thomas G, De Backer J, Coucke P, Braverman, De Paepe A, Dietz H. A critical evaluation of phenotypes associated with mutations in the TGFβ receptor genes. (American Society of Human Genetics, New Orleans, LA, October 2006.)

91. Schwarze U, Riegert-Johnson DL, Dietz HC, Byers PH. Symptomatic monoallelic dimorphic mosaicism resulting from early embryonic mitotic mutations. (American Society of Human Genetics, New Orleans, LA, October 2006.)

92. Huang D, Spencer F, Dietz HC. Nonsense-mediated mRNA decay modulates cellular fate in response to DNA damage. (Platform presentation at the American Society of Human Genetics, San Diego, CA, October 2007.)

93. Loeys B, Riegert D, Whiteman P, McConnell V, Coucke PJ, De Paepe A, Judge D, Handford P, Sakai L, Dietz HC. Domain-specific mutations in FBN1 cause a congenital form of scleroderma: Stiff Skin Syndrome. (Platform presentation at the American Society of Human Genetics, San Diego, CA, October 2007).

94. Kent KC, Loscalzo ML, Goh DLM, Cutting AL, Dietz HC. Genotype-phenotype correlation in patients with bicuspid aortic valve and aneurysm. (Poster presentation at the American Society of Human Genetics, San Diego, CA, October 2007).

95. Matt P, Habashi J, Holm T, Klein E, Gamradt M, Huso D, Van Eyk J, Dietz H. Circulating TGF-beta as a prognostic and monitoring biomarker of the aortic root dilatation and losartan therapy in Marfan syndrome. (Poster presentation at the American Society of Human Genetics, San Diego, CA, October 2007).

96. Klein EC, Cohn RD, van Erp C, Holm TM, Habashi JP, Myers L, Huso DL, Dietz HC. Crosstalk between the angiotensin II, TGFb and Wnt signaling cascades inhibits preadipocyte differentiation in Marfan syndrome. (Poster presentation at the American Society of Human Genetics, San Diego, CA, October 2007).

97. Cameron DE, Alejo DE, Patel ND, Nwakanma LU, Weiss ES, Vricella LA, Dietz HC, Spevak PJ, Williams JA, Bethea BT, Fitton TP, Gott VL. Aortic Root Replacement in 372 Marfan Patients: Evolution of Operative Repair Over 30 Years. (The J. Maxwell Chamberlain Memorial Paper for Adult Cardiac Surgery at the 44th Annual Meeting of the Society of Thoracic Surgeons, Ft. Lauderdale, FL, January 2008.

98. Habashi, JP, Doyle JJ, Bedja D, Dietz HC. AGTR2 is a genetic and therapeutic modifier of Marfan syndrome. (Poster presentation at The American Society of Human Genetics Meeting 2008, Philadelphia, PA, November 2008).

99. Loeys, B, Callewaert B, De Backer, J. Faivre L, Jondeau G, Devereux R, Pyertiz R, Sponseller P, Wordsworth P, Milewicz D, Dietz HC, De Paepe A. Towards a revised Ghent nosology for the Marfan syndrome. (Poster presentation at The American Society of Human Genetics Meeting 2008, Philadelphia, PA, November 2008)

100. Lindsay ME, Loch DC, Chen Y, Dietz HC. A Developmentally-Imposed Fixed Alteration in Cellular Identity Contributes to Pathogenesis in Marfan Syndrome (MFS). (Poster presentation at The American Society of Human Genetics Meeting 2008, Philadelphia, PA, November 2008)

101. Cooper TK, Zhong Q, Schwarze U, Pepin M, Byers P, Dietz HC. Altered Cytokine Signaling in Vascular Ehlers Danlos Syndrome (vEDs). (Poster presentation at The American Society of Human Genetics Meeting 2008, Philadelphia, PA, November 2008)

102. Loch D, Chen J, Dietz HC. Pathogenesis of Loeys-Dietz Syndrome and Therapeutic Implications. (Poster presentation at The American Society of Human Genetics Meeting 2008, Philadelphia, PA, November 2008).

103. Patel ND, Kim DH, Lindsay ME, Holm TM, John A, Garbarini J, Goldmuntz E, Dietz HC. Filamin A mutations cause tetralogy of Fallot with ascending aortic aneurysm. (Poster presentation at The American Society of Human Genetics Meeting 2008, Philadelphia, PA, November 2008)

104. van Erp C, Marx-Rattner R, Dietz H, Cohn R. Evaluation of the JNK Signaling Cascade in Muscular Dystrophies. (Poster presentation at The American Society of Human Genetics Meeting 2009, Honolulu, Hawaii)

105. Loch D, Habashi J, Dietz H. Interrogation of the Pathogenesis of Loeys-Dietz Syndrome Using an Allelic Series of Mutant Mice. (Poster presentation at The American Society of Human Genetics Meeting 2009, Honolulu, Hawaii)

106. Doyle J, Pardo-Habashi J, Holm T, Bedja D, Dietz H. Dissection of a Modifier Network in Marfan Syndrome Reveals New Therapeutic Targets. (Plenary Platform presentation at The American Society of Human Genetics Meeting 2009, Honolulu, Hawaii). ***Winner, Pre-Doctoral Translational Research Award.***

107. Milewicz D, Pannu H, Pyeritz R, Basson C, Dietz H, Maslen C, Eagle K, Kroner B, and the GenTac consortium. Sequencing Known Genes for Thoracic Aortic Disease in the GenTac cohort (Genetically Triggered Thoracic Aortic Aneurysms). (Poster presentation at The American Society of Human Genetics Meeting 2009, Honolulu, Hawaii)

108. Lisi, E, Dietz H, Oswald G, Cohn R. Hypotonia As A Feature of Connective Tissue Disorders: Lessons from A Hypotonia Specialty Clinic. (Poster presentation at The American Society of Human Genetics Meeting 2009, Honolulu, Hawaii)

109. Lindsay ME, Domian IJ, Chien KR, Dietz H. Developmental Underpinnings of Apparently Acquired Aortic Aneurysm in Marfan Syndrome Revealed by Analysis of Mutant Embryonic Stem Cells. (Platform presentation at The American Society of Human Genetics Meeting 2009, Honolulu, Hawaii)

110. Kim D, Patel N, Lindsay M, Goldmuntz E, John A, Garbarini J, Dietz H. Calpain as a Therapeutic Target in Inherited Aortic Aneurysm: Lessons from Rare Mendelian Disorders. (Platform presentation at The American Society of Human Genetics Meeting 2009, Honolulu, Hawaii)

111. Loch D, Gallo E, Dietz H. Elucidation of Mechanism and Therapy for Loeys-Dietz Syndrome Using an Allelic Series of Mutant Mice (Abstract presentation at The American Society of Human Genetics Meeting 2010, Washington, DC)

112. Gerber E, Huso D, Loeys B, Davis E, Wigley F, Dietz H. Elucidation of Sensors and Effectors in Matrix Equilibrium Derives Novel Therapeutic Strategies for Scleroderma. (Abstract presentation at The American Society of Human Genetics Meeting 2010, Washington, DC)

113. Dietz H, Doyle J, van Erp C, Aziz H. Inhibition of TGFβ Signaling Prevents Myopia in a Mouse Model of Marfan Syndrome. (Abstract presentation at The American Society of Human Genetics Meeting, 2010, Washington, DC)

114. Doyle J, Habashi J, Lindsay M, Bedja D, Dietz H. Calcium Channel Blockers Exacerbate Aortic Disease and Cause Premature Lethality in Marfan Syndrome. (Abstract presentation at The American Society of Human Genetics Meeting 2010, Washington, DC)

115. Guerrerio AL, Guerrerio P, Myers L, Halushka MK, Dhilon G, Anders R, Oliva-Hemker M, Wood RA, Dietz HC. A Genetically Defined Mendelian Presentation of Asthma, Food Allergy, Eosinophilic Esophagitis and Inflammatory Bowel Disease. (Abstract presentation at ASHG 2010, Washington, DC)

116. Gerber EE, Huso D, Loeys B, Davis E, Wigley F, Dietz HC. Treating stiff skin syndrome; Study of a rare Mendelian disorder reveals novel therapeutic strategies for complex acquired scleroderma. (Abstract presentation at The American Society of Human Genetics Meeting 2011, Montreal, Canada). ***Winner, ASHG Predoctoral Trainee Award*.**

117. Doyle A, Kent KC, Dietz HC. A major modifier locus for vascular disease in Marfan syndrome. (Abstract presentation at American Society of Human Genetics Meeting 2011, Montreal, Canada).

118. Habashi, J, Dietz HC. A mechanism and treatment strategy for pregnancy-associated aortic dissection in Marfan syndrome. (Platform presentation at The American Society of Human Genetics Meeting 2012, San Francisco, CA).

119. Doyle A, Dietz, HC. Heterozygous Germline Mutations In A Prototypical TGFβ Repressor Cause Shprintzen-Goldberg Syndrome With Aortic Aneurysm. (Abstract presentation at The American Society of Human Genetics Meeting 2012, San Francisco, CA), ***Winner, Charles J. Epstein Trainee Award for Excellence in Human Genetics Research.***

220. Calderon J, Dietz HC, Identification of a major genetic modifier in mouse models of TGFβ vasculopathies. (Poster presentation at The American Society of Human Genetics Meeting 2012, San Francisco, CA).

221. Gerber EE, Gallo EM, Fontana SC, Davis EC, Zhong X, Wigley FM, Huso DL, Dietz HC. Integrin Modulating Therapies Prevent Fibrosis and Autoimmunity in Genetic Mouse Models of Scleroderma. (Platform presentation at The American Society of Human Genetics Meeting 2013, Boston, MA).

222. Bjornsson HT, Benjamin JS, Zhang L, Gerber EE, Chen Y, Potter MC, Dietz HC. A mouse model of Kabuki syndrome demonstrates defective hippocampal neurogenesis rescued with treatment with AR-42, a histone deacetylase inhibitor. (Platform presentation at The American Society of Human Genetics Meeting 2013, Boston, MA).

223. Ziegler S, Dietz HC. Delineation of renal-independent disease mechanism underlying Hyperphosphatemic Familial Tumoral Calcinosis caused by GALNT3 mutations. (Poster presentation at the American Society of Human Genetics Meeting 2013, Boston, MA).

224. Calderon J, Dietz HC. Robust epistasis between the genes encoding in TGFβ effector and its regulatory microRNA governs modification of cardiovascular phenotypes in TGFβ vasculopathies. (Platform presentation at The American Society of Human Genetics Meeting 2013, Boston, MA). ***Winner, Charles J. Epstein Trainee Award for Excellence in Human Genetics Research.***

225. Doyle JJ, Doyle AJ, Wilson N, Bedja D, Pardo-Habashi J, Myers L, Braunstein K, Huso N, Bachir S, Squires O, Rusholme B, George A, Lindsay M, Huso D, Thomas C, Judge D, Dietz HC. ERK Activation Unifies Deleterious Gene-by-Gene and Gene-by-Environment Interactions in Marfan Syndrome. (Platform presentation at The American Society of Human Genetics Meeting 2013, Boston, MA).

226. Habashi JP, Oswald GL, Holmes KW, Reynolds EM, LeMaire S, Ravekes W, McDonnell NB, Maslen C, Shohet RV, Pyeritz RE, Devereux R, Milewicz DM, Dietz HC, Prevalence and Predictors of Pneumothorax in Patients with Connective Tissue Disorders Enrolled in the GenTAC (National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions), (Platform presentation at The American Society of Human Genetics Meeting 2013, Boston, MA).

227. Gallo MacFarlane E, Habashi JP, Chen Y, Bedja D, Dietz HC. Paracrine TGFβ-signaling overdrive between cells of different embryonic origin promotes aneurysm pathology in the aortic root of Loeys-Dietz syndrome mouse models. (Platform presentation at the 18th International Vascular Biology Meeting (NAVBO), 2015, Cape Cod, MA).

228. Wilson N, Doyle J, Antonescu C. Wheelan S, Bedja D, Dietz HC, MIBAVA Leducq Consortium. A Gene-by-Environent Interaction informs Aortic Segment-Specific Vulnerability for Aneurysm Formation in Mendelian Aoropathies. (Poster presentation at The American Society of Human Genetics Meeting 2015, Baltimore).

229. Gould RA, Hamza A, Kumar A, Preuss C, Woods C, Ling H, Sobreira N, Mohamed SA, Franco-Cereceda A, Angelfinger G, McCallion AS, Eriksson P, Van Laer L, Loeys BL, MIBAVA Leducq Consortium, Dietz HC. ROBO-SLIT Mutations Predispose Individuals to Bicuspid Aortic Valve with Ascending Aortic Aneurysm. (Platform presentation at The American Society of Human Genetics Meeting 2015, Baltimore, MD).

230. Ziegler SG, Ferreira CR, Creamer TJ, Warren DS, Goff L, inkerton AB, Millan JL, Gahl WA, Dietz HC. Toward identification of the pathogenic cell type driving calcification in pesudoxanthoma elasticum. (Plantform presentation at the American Society of Human Genetics Meeting 2015, Baltimore, MD).

231. Gallo MacFarlane, E, Habashi JP, Chen Y, Bedja D, Dietz HC. Genetic, developmental and paracrine interactions in the complex pathogenesis of heritable aneurysm conditions. (Platform presentation at the American Society of Human Genetics Meeting 2015, Baltimore, MD).

232. Doyle A, Doyle J, Wardlow R, Wilson N, Bedja D, Lindsay M, Habashi J, Myers L, Braunstein K, Bachir S, Huso N, Squires O, Rusholme B, George A, Caulfield M, Judge D, Dietz H. Identification of major genetic modifiers of vascular disease in Marfan Syndome mice. (Platform presentation at the American Society of Human Genetics Meeting 2015, Baltimore, MD).

233. Ziegler SG, Ferreira CR, Creamer TJ, Warren DS, Goff L, inkerton AB, Millan JL, Gahl WA, Dietz HC. Toward identification of the pathogenic cell type driving calcification in pesudoxanthoma elasticum. (Platform presentation at the National American Vascular Biology Organization Annual Meeting. 2015, Cape Cod).

234. Beckett JD, Gerber L, Huso D, Dietz H. Implication of Innate Immunity in the Pathogenesis of Scleroderma. (Platform and Poster presented at Keystone Conference: Immunity in Skin Development, Homeostasis and Disease, 2016 Tahoe City, CA)

234. Ziegler SG, Ferreira, CR, Gallo-MacFarlane E, Riddle RC, Tomlinson R, Martin L, Ma C, Sergienko E, Pinkerton AB, Millan JR, Gahl WA, Dietz HC. Pseudoxanthoma elasticum: Dysregulation of local ATP metabolism and treatment with a tissue non-specific alkaline phosphatase inhibitor. (Platform presentation at the Johns Hopkins Cardiovascular Research annual retreat, 2016 Baltimore, MD).

235. Kang B.E., Bedja D., Dietz H.C., Application of a Conditional Allelic Series of the Sloan-Kettering Institute Proto-oncogene (SKI) to Mechanistically Dissect the TGFB Vasculopathies. (Platform presentation at the American Society of Human Genetics Meeting 2016, Vancouver, Canada).

236. Gould RA., Au D., Migliorini M., Sobreira N., MacCarrick G., Lopez-Gutierrez J., Muratoglu S., Strickland D., Dietz HC. A rare pediatric mendelian presentation of abdominal aortic aneurysm informs the predisposition for a common but complex cardiovascular disease. (Platform presentation Presented at the American Society of Human Genetics Meeting 2016, Vancouver, Canada.

237. Ziegler SG, Ferreira CR, MacFarlane-Gallo, E, Riddle RC., Tomlinson R, Ma C, Sergienko E, Pinkerton AB, Millan J., Gahl WA, Dietz HC. Pseudoxanthoma elasticum (PXE): Dysregulation of local ATP metabolism and treatment with a tissue non-specific alkaline phosphatase (TNAP) inhibitor. (Poster presentation at the American Society of Human Genetics Meeting 2016, Vancouver, Canada).

238. Wardlow RD, Doyle JJ, Doyle AJ, Wilson NK, Bedja D, Dietz HC. Functional characterization for modifier loci for Marfan syndrome reveals novel therapeutic strategies. (Platform presentation at the American Society of Human Genetics Meeting 2017, Orlando, FL).

239. Wilson N, Doyle JJ, MacFarlane -Gallo E, Bagirzadeh R, Yazdanifar G, Bedja J, Cooke S, Dietz HC, MIBAVA Leducq Consortium. Mechanistic Interrogation of a Gene-by-Environment Interaction Informs the Pathogenesis and Treatment of Mendelian Aneurysm Disorders. (Platform presentation at the American Society of Human Genetics Meeting 2017, Orlando, FL).

240. Kang BE, Bedja J, Bagirzadeh R, Dietz HC. Epigenetic Modulation of the Pathogenesis and Treatment of Inherited Aortic Aneurysm Conditions. (Poster presentation at the American Society of Human Genetics Meeting 2017, Orlando, FL).

241. Bowen CJ, Rykiel G, Giadrosic JC, Habashi J, Helmers M, Dietz HC,. Inhibition of oxytocin signaling prevents pregnancy-associated aortic dissection in a novel mouse model of vascular Ehlers-Danlos Syndrome. (Plenary presentation at the American Society of Human Genetics Meeting 2018, San Diego, CA).

242. Seman-Senderos MA, Kim DH, Beckett JD, Nagpal V, Gould RA, Creamer TJ, Chen Y, Bedja D, Butcher JT, Mitzner W, Rouff R, Hata S, Warren DS, Dietz HC. A new mechanism and vulnerability for fibrosis. (Poster presentation at the American Society of Human Genetics Meeting 2018, San Diego, CA).

243. Kang BE, Bagirzadeh R, Bedja D, Dietz HC. Rational therapeutic epigenetic modulation in the treatment of syndromic thoracic aortic aneurysm. (Platform presentation at the American Society of Human Genetics Meeting 2018, San Diego, CA).

244. Rykiel G, Bowen CJ, Giadrosic JC, Helmers M, Dietz HC,. Mechanistic and therapeutic

interrogation of a novel mouse model of vascular Ehlers-Danlos syndrome. (Platform presentation at the American Society of Human Genetics Meeting 2018, San Diego, CA).

245. Shin J, Beckett JD, Shah A, McMahan Z, Paik J, Sampedro M, Warren DS, Dietz HC, Wigley F. "Potential Genetic and Epigenetic Therapies of Disease". (Platform presentation at the American Society of Human Genetics Meeting 2018, San Diego, CA).

236. Bowen CJ, Burger Z, Rykiel G, Calderon-Giadrosic JF, Wilson NK, Dietz HC. "A mechanism and treatment strategy for the sexual dimorphism seen in vascular Ehlers-Danlos syndrome". (Poster presentation at the American Society of Human Genetics Meeting 2019, Houston, TX).

237. Wilson NK, Doyle JJ, Gallo-MacFarlane E, Bagirzadeh R, Bedja D, Bowen CJ, Toomer KA, Creamer TJ, Cooke SK, MIBAVA, Dietz HC. Elucidation of mechanism for accentuation of aortic aneurysm predisposition in males reveals a novel therapeutic strategy for heritable aortopathies. (Poster presentation at the American Society of Human Genetics Meeting 2019, Houston, TX).

238. Bowen CJ, Burger Z, Rykiel G, Calderon JF, Doyle JJ, Zhang X, Dietz HC. A gene, variant and mechanism for a potent protective modifier of vascular Ehlers-Danlos syndrome. (Poster presentation at the American Society of Human Genetics Meeting 2020, Virtual).

239. Sorber R, Bowen CJ, Burger Z, Rykiel G, Calderon Giadrosic JF, Dietz HC. Treatment with high dose vitamin C demonstrates a sexually dimorphic survival benefit in a mouse model of severe vascular Ehlers-Danlos syndrome. (Poster presentation at the American Society of Human Genetics Meeting 2020, Virtual)

240. Bowen CJ., Sober R, Burger Z, Rykiel G, Calderon Giadrosic JF, Doyle JJ, Zhang X, Dietz HC. Map2K6 is a Potent Protective Genetic Modifier of Arterial Rupture and Death in Vascular Ehlers-Danlos Syndrome Mice. (Poster presentation at HHMI Science Meeting 2022, Virtual).

Invited Lectures:

1. "A Linkage Map of the Marfan Locus." Banbury Conference: Marfan Syndrome, Cold Spring Harbor, New York, April 1991.

2. "Fibrillin Gene Defects in the Marfan Syndrome." Gordon Conference: Elastin, Meriden, New Hampshire, July 1991.

3. "Molecular Etiology of Marfan Syndrome." Grand Rounds, Shriners Hospital for Crippled Children, Portland, Oregon, November 1991.

4. "Molecular Genetics of the Marfan Syndrome." National Institutes of Health Frontiers in Science Conference: Molecular Genetics of Vascular Disease, Bethesda, Maryland, February 1992.

5. "Marfan Syndrome." Grand Rounds, Department of Pediatrics, The Johns Hopkins University School of Medicine, Baltimore, April 1992.

6. “Marfan Syndrome.” Grand Rounds, Department of Pediatrics, Washington University School of Medicine, St. Louis, Missouri, May 1992.

7. “Molecular Biology of Marfan Syndrome.” Grand Rounds, Department of Pediatrics. Georgetown University School of Medicine, Washington, D.C., September 1992.

8. “Molecular Biology of Connective Tissue Diseases Affecting the Heart.” American Academy of Pediatrics Annual Meeting, San Francisco, October 1992.

9. “Fibrillin Mutations in the Marfan Syndrome: Lessons for PXE?” Workshop on Pseudoxanthoma Elasticum, Philadelphia, June 1992.

10. “Molecular Basis of the Marfan Syndrome.” Sixth Annual Patrick John Niland Memorial Lecture, Ann Arbor, Michigan, March 1993.

11. “Molecular Pathology of the Marfan Syndrome.” Gordon Research Conference on Collagen, New London, New Hampshire, July 1993.

12. “Molecular Pathology of the Extracellular Microfibril.” Gordon Research Conference on Elastin (Session Coordinator, Speaker), Meriden, New Hampshire, August 1993.

13. “Molecular Basis of Marfan Syndrome.” American Heart Association Postgraduate Seminar, Atlanta, November 1993.

14. “Molecular Biology of Marfan Syndrome.” Plenary Lecture at the 50th Anniversary Meeting of the Asociacion De Medicos Del Hospital Infantil De Mexico. Mexico City, November 1993.

15. “Molecular Pathology of Marfan Syndrome.” Keystone Symposia: Molecular Biology of Human Genetic Disease, Copper Mountain, Colorado, January 1994.

16. “New Insights Into the Genetic Basis of Aortic Aneurysms.” United States and Canadian Academy of Pathology, Cardiovascular Pathology: Clinicopathologic Correlations and Pathogenetic Mechanisms, San Francisco, March 1994.

17. “The Molecular Basis of Cardiovascular Disease.” University of Nebraska Medical Center, Omaha, March 1994.

18. “Marfan Syndrome: Clinical Molecular Interface.” American College of Cardiology, Atlanta, March 1994.

19. “Maintenance of an Open-Reading Frame as an Additional Level of Scrutiny During Splice-Site Selection.” Young Investigator Award Lecture, Society for Pediatric Research Meeting, Washington, DC, May 1994.

20. “The Basics of Molecular Biology.” Plenary Educational Symposium, Society for Pediatric Research, Seattle, May 1994.

21. “Molecular Basis of Marfan Syndrome.” Fifth International Congress on the Extracellular Matrix, Philadelphia, June 1994.

22. “Cellular and Transgenic Models of Marfan Syndrome.” Third Annual Bristol-Myers Squibb Symposium on Cardiovascular Biology. Brigham & Women's Hospital, Harvard Medical School, Boston, September 1994.

23. “Fibrillin Mutations in the Marfan Syndrome.” Third International Symposium on the Marfan Syndrome (speaker and session chair), Berlin, September 1994.

24. Grand Rounds, Departments of Pediatrics and Human Genetics. Rainbow Babies Hospital, Cleveland, Ohio, February, 1995.

25. Scientific Conference on the Molecular, Cellular, and Functional Aspects of Cardiovascular Development, American Heart Association (speaker and session chair), New Orleans, March 1995.

26. Visiting Professorship, Department of Vascular Surgery, Northwestern University, Chicago, April 1995.

27. “Molecular Biology of Marfan Syndrome.” Samsung Medical Center and Research Institute, Opening Ceremony Symposium: An Approaching Era of Molecular Medicine, Seoul, Korea, April 1995.

28. “Marfan Syndrome.” International Nomenclature Committee, Heritable Disorders of Connective Tissue, Ghent, Belgium, April 1995.

29. Scientific Conference on Vascular Biology, Society for Pediatric Research, San Diego, May 1995.

30. Scientific Conference on Heritable Disorders of the Extracellular Matrix, National Institutes of Health, Bethesda, Maryland, June 1995.

31. Plenary Lecture: Western Thoracic Society, Couer d’Alene, Idaho, June 1995.

32. “Molecular Pathogenesis of Marfan Syndrome and Related Disorders.” Gordon Conference: Elastin, Meriden, New Hampshire, July 1995.

33. “Molecular Pathogenesis of Marfan Syndrome and Related Disorders.” Gordon Conference: Molecular Biology, Salve Regina, Rhode Island, August 1995.

34. “Molecular Biology of Congenital Heart Disease.” American Academy of Pediatrics, San Francisco, October 1995.

35. “Marfan Syndrome.” Plenary Lecture at the American Heart Association Meeting, Dallas, January 1996.

36. Grand Rounds: Department of Biochemistry, University of Maryland, Baltimore, April 1996.

37. “Model Systems in the Study of the Pathogenesis of Marfan Syndrome and Related Disorders.” Frontiers of Science, Transgenic Models of Cardiovascular Disease, Washington D.C., May 1996.

38. “Molecular Biology of Marfan Syndrome.” Plenary Lecture at the American Society of Hypertension Meeting, New York, May 1996.

39. Visiting Professor, Department of Molecular Cardiology, Cleveland Clinic, Cleveland, Ohio, June 1996.

40. ”Scientific Update on Marfan Syndrome.” National Marfan Foundation National Meeting, Houston, July 1996.

41. “Traditional and Novel Molecular Mechanisms in the Pathogenesis of Marfan Syndrome and Related Disorders.” Instructor and featured speaker, 37th Annual Jackson Laboratory Short Course in Medical and Experimental Mammalian Genetics, Bar Harbor, Maine, July 1996.

42. “Patients and Patience in the Study of Marfan Syndrome and Related Disorders.” Third International Symposium on the Marfan Syndrome (session chair and speaker), Davos, Switzerland, August 1996.

43. “Marfan Syndrome.” Vascular Biology Meeting, Seattle, September 1996.

44. “Nonsense-mediated perturbation of RNA processing.” Department of Biochemistry and Genetics Seminar Series, University of Nebraska, Omaha, September 1996.

45. “Nonsense-mediated perturbation of RNA processing.” Department of Biochemistry, University of Medicine and Dentistry of New Jersey, Rutgers, New Jersey, November 1996.

46. “Nonsense RNA surveillance in health and disease.” Yeast Genetics and Human Disease: Conference of the American Society for Microbiology (Member, Scientific Organizing Committee and Speaker), Baltimore, November 1996.

47. “Traditional and novel molecular mechanisms in the Pathogenesis of Marfan Syndrome and Related Disorders.” Howard Hughes Symposium Series, University of Iowa School of Medicine, February 1997.

48. “Molecular Biology of Marfan Syndrome and Therapeutic Applications.” Elster Memorial Lecture, Departments of Cardiology and Molecular Biology, Mount Sinai University School of Medicine, New York, March 1997.

49. “Molecular Biology of Marfan Syndrome and Therapeutic Applications.” Mead Johnson Clinical Scholars Research Symposium (Featured Speaker).

50. “The Biology of Nonsense (Mutations).” Mead Johnson Clinical Scholars Research Symposium (Featured Speaker), Charleston, South Carolina, April 1997.

51. “Genomic Approaches For the Functional Analysis of Data Derived from the Human Genome Projects.” Department of Energy BER 50th Anniversary Celebration (Featured Speaker), National Academy of Science, Washington, DC, May 1997.

52. “Molecular Biology of Marfan Syndrome.” Memorial Lectureship, Department of Pediatric Cardiology, Sick Kids Hospital, Toronto, Ontario, June 1997.

53. “Molecular Advances in Marfan Syndrome.” National Marfan Foundation Meeting (Featured Speaker), Stanford University School of Medicine, Stanford, California, July 1997.

54. “Traditional and Novel Molecular Mechanisms in the Pathogenesis of Marfan Syndrome and Related Disorders.” Instructor and featured speaker, 38th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 1997.

55. “Molecular Biology of Marfan Syndrome.” Meeting on Obstructive and Obliterative Vascular Disease (featured speaker), National Heart, Lung and Blood Institute, National Institutes of Health, Bethesda, Maryland, September 1997.

56. “Molecular Biology of Marfan Syndrome.” Grand Rounds Speaker and Visiting Professor, Winner Memorial Lecture, University of Maryland School of Medicine, Baltimore, October 1997.

57. “Molecular Mechanisms of Inherited Cardiovascular Disorders.” Division of Cardiology CME Course (featured speaker), Johns Hopkins University School of Medicine, Baltimore, February 1998.

58. “Marfan Syndrome.” Banbury Conference on the Marfan Syndrome (Featured Speaker, Coordinator), Cold Spring Harbor, New York, April 1998.

59. “Cerebrovascular Manifestations of Connective Tissue Disease.” First International Congress Genetics in Neuroscience, Terni, Italy, June 1998.

60. “Pediatric Concerns in the Marfan Syndrome.” National Marfan Conference, New York, July 1998.

61. “Molecular Biology of the Marfan Syndrome.” Instructor and featured speaker, 39th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 1998.

62. “Post-Transcriptions Control of Gene Expression.” Instructor and featured speaker, 39th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 1998.

63. “Molecular Advances in the Marfan Syndrome.” International Marfan Syndrome Society Meeting (Keynote Speaker), Helsinki, August 1998.

64. “Mutational Screening.” Positional Cloning Course, Cold Spring Harbor Laboratory, Cold Spring Harbor, New York, October 1998.

65. “Clinical Correlates of the Molecular Basis of Marfan Syndrome.” Pediatric Grand Rounds, St. Agnes Hospital, Baltimore, Maryland, February 1999.

66. “New Insight into the Pathogenesis of Marfan Aortic Aneurysms.” Session Coordinator and Speaker, Vascular Biology Meeting, Washington DC, April 1999.

67. Testimony before the U.S. House of Representatives Committee on Appropriations on Behalf of the Coalition for Heritable Disorders of Connective Tissue, Washington DC, April 1999.

68. “Marfan Syndrome: Investigations Involving Animal Models and Gene Therapy.” Fifteenth Annual National Marfan Foundation Conference, Pittsburgh, Pennsylvania, July 1999.

69. “Reality and Prospects for Genetic Testing for Marfan Syndrome and Related Connective Tissue Disorders.” Fifteenth Annual National Marfan Foundation Conference, Pittsburgh, Pennsylvania, July 1999.

70. “Clinical and Molecular Genetics of Marfan Syndrome.” Instructor and featured speaker, 40th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 1999.

71. “Post-Transcriptional Control of Gene Expression.” Instructor and featured speaker, 40th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 1999.

72. “Nonsense RNA Metabolism in Health and Disease.” Gordon Conference: Molecular Biology, Session Coordinator and Invited Speaker, Salve Regina, Rhode Island, August 1999.

73. “New Insights Into the Pathogenesis of Marfan Syndrome.” Annual Scientific Meeting of Howard Hughes Medical Institute, Chevy Chase, Maryland, March 2000.

74. “Higher Order Pathogenesis of the Marfan Syndrome.” First Symposium of the International Society for Matrix Biology, Invited Speaker, Philadelphia, Pennsylvania June 2000.

75. “Future Directions in Clinical Care and Research.” Sixteenth Annual National Marfan Foundation Conference, Omaha, Nebraska, July 2000.

76. “Molecular and Clinical Genetics of Marfan Syndrome.” Instructor and featured speaker, 41st Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2000.

77. “Post-Transcriptional Control of Gene Expression.” 41st Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2000.

78. “Higher Order Pathogenesis of the Marfan Syndrome.” International Conference on Biology and Pathology of the Extracellular Matrix, Featured Speaker and Workshop Leader, St. Louis, Missouri, October 2000.

79. “Pathogenic Mechanisms in the Marfan Syndrome.” UCLA Human Genetics Fall 2000 Speaker Series, Los Angeles, California, October 2000.

80. “Molecular and Genetic Basis of Inherited Aortic Diseases.” Seminar on Inherited Diseases of the Ascending Aorta - Etiology, Diagnosis, and Management, American Heart Association 73rd Scientific Sessions, Invited Speaker, New Orleans, Louisiana, November 2000.

81. “What Can FBN1-Targeted Mice Tell Us About the Marfan Syndrome?” Third Conference on Heritable Disorders of Connective Tissue, Bethesda, Maryland, November 2000.

82. “Mechanism, Clinical Implications and Experimental Applications of Nonsense-Mediated mRNA Decay.” National Human Genome Research Institute Division of Intramural Research Seminar Series, Bethesda, Maryland, January 2001.

83. “Genetics, Geneticists and the Marfan Syndrome.” The Third World Congress of Pediatric Cardiology and Cardiac Surgery, Toronto, Ontario, May 2001.

84. “Post-Transcriptional Control of Gene Expression.” Instructor and featured speaker, 42nd Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2001.

85. “Clinical and Molecular Genetics of Marfan Syndrome.” Instructor and featured speaker, 42nd Annual Short Course in Medical Experimental and Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2001.

86. “Mouse Models Reveal Novel Pathogenetic Mechanisms in the Marfan Syndrome.” Gordon Conference: Elastin and Elastic Fibers, Session Coordinator and Invited Speaker, Meriden, New Hampshire, July 2001.

87. “New Insights into the Pathogenesis of Marfan Syndrome.” Seventeenth Annual National Marfan Foundation Conference, Seattle, Washington, August 2001.

88. “New Insights into the Pathogenesis of Marfan Syndrome.” Baylor College of Medicine, Houston, Texas, October 2001.

89. “Marfan Syndrome: Beyond the Gene.” The Sixth Annual Pauline Wilson Horner Genetics Symposium, Case Western Reserve University, Cleveland, Ohio, October 2001.

90. “Molecular Biology of the Marfan Syndrome.” Cornell University, New York, New York, October 2001.

91. “Biologic and Medical Significance of Mammalian Nonsense-mediated mRNA Decay.” Genetics Colloquium Presentation, University of Wisconsin, Madison, Wisconsin, October 2001.

92. “Nonsense mRNA Metabolism in Health and Disease.” PTC Therapeutics, Edison, New Jersey, December 2001.

93. “The Aorta as a Biomatrix: Lessons from Marfan Syndrome and Related Connective Tissue Disorders.” Developmental Biology Seminar, Cincinnati Children’s Hospital, Cincinnati, Ohio, February 2002.

94. “Mouse Models for Marfan Syndrome.” Session Organizer and Invited Speaker, Keystone Symposium, Santa Fe, New Mexico, February 2002.

95. "Bench to Bedside: Genetic Basis for Marfan." New York Academy of Medicine, New York, New York, May 2002.

96. 3rd International Congress in Genetics and Regeneration in Neuroscience and the 1st Satellite Symposium "New Frontiers in Surgical Neurology." Session Chairman and Invited Speaker for Closing Remarks, Terni, Italy, June 2002.

97. "Post-Transcriptional Control of Gene Expression." Instructor and featured speaker, 43rd Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2002.

98. "Molecular Biology of Marfan Syndrome." Instructor and featured speaker, 43rd Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2002.

99. "Current Research in the Marfan Syndrome." Eighteenth Annual National Marfan Foundation Conference, Host, Speaker, and Workshop Leader, Baltimore, Maryland, July 2002.

100. Planning Workshop on Relating Genetic Variation to Health and Disease. National Human Genome Research Institute, NIH, Bethesda Hyatt Hotel, Bethesda, Maryland, August 2002.

101. NIH/NIAMS ZAR1-TAS-B (02) Study Session, Silver Spring, Maryland, August 2002.

102. “Bedside to the Gene and Back—Structuring a Career in Investigative Medicine.” University of Washington Pathology Department Retreat, Leavenworth, Washington, October 2002.

103. “Molecular basis of NMD in human cells and strategies for rescue of mRNA.” 52nd Annual Meeting of The American Society of Human Genetics Session, Co-Moderator and Speaker. Baltimore, Maryland, October 2002.

104. “Pulling Out All the Stops in mRNA Surveillance.” M.D.-Ph.D. Symposium, Airlie Conference Center, Airlie, Virginia, October 2002.

105. “Making Sense of Nonsense: On the Mechanism and Role of Mammalian Nonsense RNA Surveillance.” The University of Texas Southwestern Medical Center at Dallas, Dallas, Texas, November 4, 2002.

106. “Fibrillin-1 in the Pathogenesis of Marfan Syndrome: Not Just Another Pretty Structural Protein.” The University of Texas Southwestern Medical Center at Dallas, Dallas, Texas, November 2002.

107. Session Chairman on “Degenerative and Chronic diseases Involving ECM.” American Society for Matrix Biology Annual Meeting, Houston, Texas, November 2002.

108. “Dysregulation of Cytokine Activation in the Pathogenesis of Marfan Syndrome and Related Disorders.” Cardiovascular Research Meeting, Washington University, St. Louis, Missouri, January 2003.

109. “Pulling Out All the Stops in mRNA Surveillance.” Cell Biology Department Seminar. Washington University, St. Louis, Missouri, January 2003.

110. “Pathogenesis of Cardiopulmonary Disease in Marfan Syndrome.” American Society for Clinical Investigation & Association of American Physicians Joint Meeting, Chicago, Illinois, April 2003.

111. “Genes That Alter Vascular Development and Gene Therapy.” Pediatric Academic Societies’ Annual Meeting, Seattle, Washington, May 2003.

112. “Pulling Out All the Stops in mRNA Surveillance.” Rutgers University, Department of Cell Biology and Neuroscience Seminar Series, Piscataway, New Jersey, May 2003.

113. “Research Update in Marfan Syndrome.” The Canadian Marfan Association, Halifax Medical Conference Day, Halifax, Nova Scotia, June 2003.

114. “Diagnosis of the Marfan Syndrome.” National Marfan Foundation Nineteenth Annual National Conference, Speaker and Workshop Leader, Chicago, Illinois, July 2003.

115. “Post-Transcriptional Control of Gene Expression.” Instructor and featured speaker, 44th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2003.

116. “Physiologic Substrates for mRNA Surveillance: Mechanistic and Therapeutic Implications.” Gordon Research Conference on Human Genetics and Genomics, Colby College, Waterville, Maine, August 2003.

117. “Pathophysiology of Microfibril/Elastic Fiber Architecture.” Speaker and Chair, The Cleveland Clinic, Marfan Syndrome and Connective Tissue Disorders, Cleveland, Ohio, September 2003.

118. “Molecular Genetics of Marfan’s and Future Development.” Speaker and Chair, The Cleveland Clinic, Marfan Syndrome and Connective Tissue Disorders, Cleveland, Ohio, September 2003.

119. “After the gene: Marfan syndrome as a paradigm for dissection of pathogenesis and elucidation of new treatment strategies.” 19th Annual Medical Student Research Day, Wake Forest University School of Medicine, Winston-Salem, North Carolina, October 2003.

120. NIH Study Section for Mammalian Genetics. Washington, DC, October 2003.

121. "Pathogenesis of Aortic Aneurysms in Polycystic Kidney Disease." American Society of Nephrology, 36th Annual Meeting and Scientific Exposition, San Diego, CA, November 2003.

122. NIH/NHLBI Program Project Grant Review Consultant. Columbia, Maryland, March 2004.

123. NIH Study Section for Mammalian Genetics. Bethesda, Maryland, February 2004.

124. "Interrogation of the mechanism and physiologic importance of nonsense surveillance." Columbia University, College of Physicians and Surgeons, New York, NY, March 2004.

125. "Novel insights into pathogenesis and therapy of Marfan syndrome." Speaker, Keystone Symposium, Molecular Biology of Cardiac Disease, Keystone, Colorado, March 2004.

126. "Cytokine dysregulation and altered tissue morphogenesis contribute to pathogenesis in Marfan syndrome." Genetic and Developmental Basis of Pediatric Disease Symposium, Washington University in St. Louis, St. Louis, Missouri, April 2004.

127. The Sarnoff Endowment's 24th Annual Scientific Meeting. Co-Moderator, Roundtable Luncheon Session I, "Laboratory-based Research Careers." Georgetown University Conference Center, Washington, DC, April 2004.

128. "Marfan Syndrome: From Pathogenetic Interrogation to Rational Therapeutic Strategies." The Sarnoff Endowment's 24th Annual Scientific Meeting. Georgetown University Conference Center, Washington, DC, April 2004.

129. "Physiologic Substrates for Nonsense RNA Surveillance: Mechanistic Insights and Pathogenic Implications." University of Utah School of Medicine Seminar Series, Salt Lake City, Utah, May 2004.

130. "Physiologic substrates for nonsense RNA surveillance: how and why?" Seminar, Western General Hospital, Edinburgh, Scotland, June 2004.

131. "The molecular genetics of Marfan syndrome and its future." Seminar, Mt. Sinai School of Medicine, New York, June 2004.

132. 2004 FASEB Summer Conference. Chair, Session 7, "Post-transcriptional Regulation of Gene Expression: Mechanisms of mRNA decay." Omni Tucson Resort & Spa, Tucson, Arizona, June 2004.

133. "New insights into the pathogenesis of Marfan syndrome." Pediatric Grand Rounds, Cedars-Sinai Medical Center, National Marfan Foundation 20th Annual Conference, Los Angeles, CA, July 2004.

134. "Overview of Marfan Syndrome: Clinical Features and Natural History." Beverly Hilton Hotel, National Marfan Foundation 20th Annual Conference, Los Angeles, CA, July 2004.

135. "Research Update and Therapeutic Advances." Beverly Hilton Hotel, National Marfan Foundation 20th Annual Conference, Los Angeles, CA, July 2004.

136. “Post-transcriptional Control of Gene Expression.“ Instructor and featured speaker, 45th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2004.

137. Site visit review of the Medical Genetics Branch of the National Human Genome Research Institute of the National Institutes of Health. Bethesda, MD, September 2004.

138. “New insights regarding the physiologic and medical significance of nonsense surveillance.” Howard Hughes Scientific Meeting, Chevy Chase, MD, October 2004.

139. “Marfans and Fibrillin.” American College of Rheumatology Annual Scientific Meeting, San Antonio, TX, October 2004.

140. American Society of Human Genetics 54th Annual Meeting, Toronto, Ontario, Canada, October 2004.

141. National Human Genome Research Institute Board of Scientific Counselors Meeting and Scientific Retreat, Gettysburg, PA November 2004.

142. “On the pathogenesis and treatment of genetically-induced thoracic aortic aneurysm.” Medicine Grand Rounds, Johns Hopkins University School of Medicine, January 2005.

143. “On the pathogenesis and treatment of genetically-imposed thoracic aortic aneurysm and dissection.” Genetic Counseling Seminar, National Human Genome Research Institute of the National Institutes of Health, Bethesda, MD, February 2005.

144. “New insights into the pathogenesis and treatment of ascending aortic aneurysm.” Howard Hughes Medical Institute-National Institutes of Health Formal Science Dinner Lecture, National Institutes of Health, Bethesda, MD, March 2005.

145. “On the pathogenesis and treatment of genetically-induced thoracic aortic aneurysm.” Pediatrics Grand Rounds, Johns Hopkins University School of Medicine, March 2005.

146. “New insights into the pathogenesis and treatment of Marfan syndrome.” 85th Annual Meeting of the American Association for Thoracic Surgery, San Francisco, CA, April 2005.

147. “When to kill the messenger: On the mechanism and purpose of mammalian nonsense surveillance.” The University Lecture Series, The University of Texas Southwestern Medical Center at Dallas, Dallas, TX, April 2005.

148. “New insights into the pathogenesis and treatment of ascending aortic aneurysm in Marfan syndrome and related disorders.” American Heart Association, 6th Annual Conference on Arteriosclerosis, Thrombosis and Vascular Biology, Washington, DC, April 2005.

149. “New insights into the pathogenesis and treatment of human aortic aneurysm syndromes.” Howard Hughes Medical Institute Scientific Meeting, Chevy Chase, MD, May 2005.

150. “New insights into the pathogenesis and treatment of Marfan syndrome.” European School of Genetic Medicine, 18th Course in Medical Genetics, Bertinoro di Romagna, Italy, May 2005.

151. “Combining a career in clinical and research genetics.” European School of Genetic Medicine, 18th Course in Medical Genetics, Bertinoro di Romagna, Italy, May 2005.

152. “The ABCs of NMD.” Johns Hopkins University School of Medicine, Institute of Genetic Medicine, Current Topics in Clinical Genetics Lecture Series, Baltimore, MD, May 2005.

153. “Fatal aortic dissection in 37-year-old female – lessons on inequality of ascending aortic aneurysm.” Johns Hopkins University School of Medicine, Biennial Lecture – Etiology and Management of Aortic Aneurysms, Baltimore, MD, June 2005.

154. “Post-transcriptional Control of Gene Expression.“ 46th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2005.

155. “Molecular genetics and genomics approaches to human disease: Marfan syndrome as a model.” Instructional and featured speaker, 46th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2005.

156. “Marfan Syndrome: What is New in Research.” 21st Annual National Marfan Conference, Speaker and Workshop Leader, St. Louis, MO, August 2005.

157. “The pathogenesis of MFS: pathways from nucleotide to the bedside and beyond.” 7th International Research Symposium on Marfan Syndrome, Co-chair and speaker, Ghent, Belgium, September 2005.

158. NIH/National Human Genome Research Institute Site Visit, Bethesda, MD, September 2005.

159. NIH/Chemical Genomics Center Site Visit, Rockville, MD, September 2005.

160. Moderator, Session No. 36, Cardiovascular Development and Disease. American Society of Human Genetics, Salt Lake City, UT, October 2005.

161. NIH/National Human Genome Research Institute Retreat, Gettysburg, PA, November 2005.

162. “Management of Marfan syndrome: Insights derived from mouse models.” American Heart Association, Dallas, TX, November 2005.

163. “TGF-β in Connective Tissue Disease: Marfan Syndrome.” American College of Rheumatology, San Diego, CA, November 2005.

164. “A New Syndrome Combining Cardiovascular Disease and Retinal Detachments.” 18th Current Concepts in Ophthalmology Baltimore Meeting, The Johns Hopkins Wilmer Eye Institute, Baltimore, MD, December 2005.

165. “TGFβ antagonism in the treatment of the multisystem pathogenesis of Marfan syndrome and related disorders.” Seminar, Genzyme Corporation, Framingham, MA, December 2005.

166. NIH - Marfan Subcommittee Meeting, Bethesda, MD, December 2005.

167. “New Therapies for Marfan Syndrome Derived from Interrogation of Disease Pathogenesis.” Wilson Symposium, Case Western Reserve University, Cleveland, OH, December 2005.

168. “Dysregulation of TGFβ signaling in the multisystem pathogenesis of Marfan syndrome and related disorders.” CVRC Seminar, Massachusetts General Hospital, Cardiovascular Research Center, Boston, MA, February 2006.

169. “New insights regarding the pathogenesis and treatment of Marfan syndrome and related disorders.” Grand Rounds, Massachusetts General Hospital, Boston, MA, March 2006.

170. “Use of animal models to dissect human aortic aneurysm syndromes.” Comparative Medicine Seminar Series, Johns Hopkins University, Department of Comparative Medicine, Baltimore, MD, March 2006.

171. “New insights into the multisystem pathogenesis of Marfan syndrome.” Musculoskeletal Disorders Seminar, University of Pennsylvania, Philadelphia, PA, April 2006.

172. “Important recent advances in the cause and medical management of Marfan and Loeys-Dietz aortic root aneurysms.” Cardiac Surgery Grand Rounds, Johns Hopkins Hospital, Baltimore, MD, May 2006.

173. “From pathogenetic insight to rational therapeutic strategies for Marfan syndrome and related disorders.” Cardiovascular Grand Rounds at the Brigham and Women’s Hospital, Boston, MA, May 2006.

174. “TGFβ in the Pathogenesis of Cystic Fibrosis: Lessons from Marfan syndrome and Related Disorders.” Cystic Fibrosis Foundation, Williamsburg Conference, Williamsburg, VA, June 2006.

175. “Elucidation of the pathogenesis of Marfan syndrome and related aneurysm phenotypes derives novel therapeutic strategies.” Pediatric Basic Research Conference, Mount Sinai School of Medicine, New York, NY, June 2006.

176. “TGFβ in the Pathogenesis of Marfan syndrome and related connective tissue disorders.” NIH/NIAID, Laboratory of Clinical Infectious Diseases, Bethesda, MD, June 2006.

177. “Pathogenesis of Marfan Syndrome.” Ophthalmologic Research & Clinical Management Workshop, 22nd Annual National Marfan Conference, Philadelphia, PA, July 2006.

178. “Losartan and Atenolol Clinical Trial.” Speaker and Workshop Leader, 22nd Annual National Marfan Conference, Philadelphia, PA, July 2006.

179. “Post-transcriptional Control of Gene Expression. “ Instructional and featured speaker, 47th Annual Short Course in Medical and “Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2006.

180. “Marfan syndrome.” Instructional and featured speaker, 47th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2006.

181. “New insights regarding the pathogenesis and treatment of Marfan syndrome and related disorders.” Pediatric Grand Rounds, A.I. DuPont Hospital for Children, Wilmington, DE, August 2006.

182. “New insights regarding the pathogenesis and treatment of Marfan syndrome and related disorders.” Nahum J. Winer Memorial Lecture, The New York Academy of Medicine, New York, NY, September 2006.

183. “Marfan syndrome: from molecules to medicine.” Cardiovascular Seminar Series, Department of Cardiology, Children’s Hospital Boston, Boston, MA, September 2006.

184. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Canadian Marfan Association Conference, Toronto, Canada, September 2006.

185. “Genetic Advances in Marfan Syndrome.” Canadian Marfan Association Conference, Toronto, Canada, September 2006.

186. American Society of Human Genetics. Invited Speaker, New Orleans, LA, October 2006.

187. “ATI antagonists in the treatment of the multisystem manifestations of Marfan syndrome and related disorders.” The Drug Repositioning Summit: Finding New Routes to Success, Cambridge Healthtech Institute, Philadelphia, PA, October 2006.

188. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” 3rd Annual Thomas P. Graham, Jr. Lecturer and Discovery Lecture Series Speaker, The Pediatric Heart Institute, Children’s Hospital at Vanderbilt, Nashville, TN, October 2006.

189. “Marfan Syndrome and Related Disorders.” 2006 Cazden Lecturer, Department of Pediatrics Grand Rounds, Oregon Health & Science University, Doernbecher Children’s Hospital, Portland, OR, October 2006.

190. “Marfan Syndrome: From Molecules to Medicines.” 2006 Cazden Lecturer, Oregon Health & Science University, Doernbecher Children’s Hospital, Portland, OR, October 2006.

191. “Pathogenesis and Treatment Strategies for Marfan Syndrome and Other TGFβ Vasculopathies.” The American Society for Matrix Biology Biennial National Meeting 2006, Nashville, TN, October 2006.

192. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Keynote Speaker, National Human Genome Research Institute Board of Scientific Counselors Meeting and Scientific Retreat, Gettysburg, PA, November 2006.

193. “Marfan Syndrome and Connective Tissue Disorders.” American Heart Association Scientific Sessions 2006 Program Participant, Chicago, IL, November 2006.

194. “Marfan Syndrome and Related Disorders: From Molecules to Medicine.” Keynote Speaker, Meinhard Robinow Grand Rounds Lectureship, The Children’s Medical Center, Dayton, OH, November 2006.

195. “TGF-β Vasculopathies.” Resident Core Conference, The Children’s Medical Center, Dayton, OH, November 2006.

196. “Marfan Syndrome and Related Disorders: From Molecules to Medicine.” Cardiology Grand Rounds, Johns Hopkins University SOM, Baltimore, MD, January 2007.

197. “Marfan syndrome and Related Disorders: From Molecules to Medicines.” Keynote Speaker, 39th Annual Edmund R. McCluskey Memorial Lecture, University of Pittsburgh Medical Center, Pittsburgh, PA, January 2007.

198. Local Chapter of the National Marfan Foundation. Co-host and Invited Speaker, Johns Hopkins University SOM, Baltimore, MD, January 2007.

199. “Angiotensin II Type 1 Receptor Blockade Attenuates TGFβ-Induced Failure of Muscle Regeneration in Multiple States.” Howard Hughes Medical Institute Scientific Meeting, Chevy Chase, MD, January 2007.

200. 3rd International Marfan Syndrome Nosology Consortium. Co-Chair, Gent, Belgium, February 2007.

201. American Society of Clinical Investigation. Mid-winter Council Meeting, Yountville, CA February 2007.

202. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Princess Lilian Foundation Visiting Professor, Gent, Belgium, March 2007.

203. “Marfan Syndrome: From Molecules to Medicine to Muscular Dystrophy.” Speaker/Moderator 2007 Annual Clinical Genetics Meeting, Nashville, TN, March 2007.

204. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Grand Rounds, Mercy Medical Center, Des Moines, IA, March 2007.

205. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Invited Lecturer, 16th Annual Paulette Shirley Pritchett Endowed Lecture in Pathology, University of Alabama at Birmingham, Birmingham, AL, April 2007.

206. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” The 2007 Isadore Rosenfeld Visiting Professor, Weill Medical College of Cornell University, New York, NY, April 2007.

207. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Cardiology Grand Rounds, Beth Israel Deaconess Medical Center, Boston, MA, April 2007.

208. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Institute of Genetic Medicine Seminar Series, Johns Hopkins University SOM, Baltimore, MD, April 2007.

209. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Seminar Children’s Hospital Research Foundation, Cincinnati, OH, April 2007.

210. “Marfan Syndrome and Its Cardiovascular Complications.” 43rd Annual Robert M. Jeresaty, M.D. Cardiovascular Symposium, St. Francis Hospital and Medical Center, Hartford, CT, April 2007.

211. “New Insights in the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders: A Journey from Bedside to Bench to Bedside.” Pediatric Grand Rounds, Rady Children’s Hospital-San Diego, San Diego, CA, May 2007.

212. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” GGPD Seminar Speaker, Children’s Hospital of Philadelphia, Philadelphia, PA, May 2007.

213. “The Individualized Treatment of Aortic Aneurysm.” 2007 Biennial McKusick-Nathans Institute of Genetic Medicine Lecture Series, Johns Hopkins University School of Medicine, Baltimore, MD, June 2007.

214. “Marfan Syndrome.” The Society for Vascular Medicine and Biology 18th Annual Scientific Session, Keynote Speaker, Baltimore, MD, June 2007.

215. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Guest Speaker, UCLA Intercampus Medical Genetics Graduation, Los Angeles, CA, June 2007.

216. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Guest Speaker, Centro Malattie Genetiche Cardiovascolari, IRCCS Policlinico San Matteo, Pavia, Italy, June 2007.

217. 23rd Annual National Marfan Conference, Speaker and Workshop Leader, Stanford, CA, June 2007.

218. “Post-transcriptional Control of Gene Expression.“ Instructional and featured speaker, 48th Annual Short Course in Medical and “Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2007.

219. “Marfan Syndrome: Disease mechanisms and new ideas about treatment.” Instructional and featured speaker, 48th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2007.

220. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Gordon Research Conference: Human Genetics and Genomics. Salve Regina University, Newport, RI, July 2007.

221. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Gordon Research Conference: Elastin and Elastic Fibers. University of New England, Biddeford, ME, July 2007.

222. “Marfan Syndrome.” Co-chair, Session III: Pleiotropic Diseases and Systemic Induction of Retinal Diseases. Arvo Summer Eye Research Conference, Monterey, CA, August 2007.

223. “A Therapy for Marfan’s Syndrome.” Invited to address the Congressional Biomedical Research Caucus and the Joint Steering Committee for Public Policy. Rayburn Building, Washington, DC, September 2007.

224. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Guest Lecturer, Midwestern Vascular Surgical Society, Chicago, IL, September 2007.

225. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Dermatology Grand Rounds, Johns Hopkins University SOM, Baltimore, MD, September 2007.

226. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Invited Speaker, The British Society for Human Genetics, University of York, Manchester, England, September 2007.

227. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Pediatric Grand Rounds and Guest Lecturer, Kathryn L. Ober Lectureship on Marfan Syndrome, Indiana University School of Medicine, Indianapolis, IN, September 2007.

228. “TGFβ-induced failure of tissue regeneration: Lessons from Mendelian disorders.” Invited Speaker, Whitehead Symposium XXV, Massachusetts Institute of Technology, Boston, MA, October 2007.

229. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Pediatric Grand Rounds, NY Presbyterian/Weill Cornell Medical College, New York, NY, October 2007.

230. “Marfan Syndrome: From Molecules to Medicines.” Invited Speaker and Co-Chair, Clinical Cardiovascular Genomics – Clinical Genomics to Function, Cold Spring Harbor Laboratory, Cold Spring Harbor, NY, October 2007.

231. 2007 National Society of Genetic Counselors Annual Education Conference. Speaker and co-chair of EBS Session 2: Connective Tissue Disorders: Navigating the Clinical Diagnosis and Counseling Maze, Kansas City, MO, October 2007.

232. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." Invited Speaker, Multidisciplinary Cardiovascular Disease Conference, Carolinas HealthCare System. Charlotte, NC, October 2007.

233. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." Translational Research Speaker, Trans-NIH Intramural Research Initiatives Retreat, Bethesda, MD, October 2007.

234. American Society of Human Genetics 57th Annual Meeting, San Diego, CA, October 2007.

235. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." Pediatric Grand Rounds, Johns Hopkins University SOM, Baltimore, MD, October 2007.

236. "Marfan Syndrome: From Molecules to Medicines." Invited Speaker, Lab Links Symposium, *Cell* Press, Boston, MA, November 2007.

237. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." Invited Speaker, Genzyme Corporation, Framingham, MA, November 2007.

238. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." Welch Center Grand Rounds, Johns Hopkins University SOM, Baltimore, MD January 2008.

239. "Marfan Syndrome: From Molecules to Medicines." Speaker and Session Chair, Keystone Symposium 2008 B2: TGF-β Family in Homeostasis and Disease, Santa Fe, NM, February 2008.

240. "Interrogation of the Pathogenesis of Stiff Skin Syndrome: A Congenital Form of Scleroderma." Scleroderma Research Foundation Scientific Workshop, San Francisco, CA, February 2008.

241. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." IGM Distinguished Speakers Seminar Series, University of Southern California, Los Angeles, CA, April 2008.

242. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." Medical Grand Rounds, Johns Hopkins University School of Medicine, Baltimore, MD, April 2008.

243. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." University Program in Genetics and Genomics, Duke University, Durham, NC, May 2008.

244. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." New York University School of Medicine, Keynote Speaker, The Honors Program Lecture Series, New York, NY, May 2008.

245. "Marfan Syndrome and Related Disorders: From Molecules to Medicines." Medical Genetics Grand Rounds, Emory University School of Medicine, Atlanta, GA, May 2008.

246. "TGFβ and disease: lessons from Mendelian disorders." Seminar, Emory University School of Medicine, Atlanta, GA, May 2008.

247. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Molecular Medicine Lecture, University of Washington, Seattle, WA, May 2008.

248. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Keynote Speaker, Genetics and Clinical Aspects of Connective Tissue Disease, 14th Canadian Connective Tissue Conference, McGill University, Montreal, Canada, June 2008.

249. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” NHLBI Seminar Series, NIH, Bethesda, MD, June 2008.

250. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Guest Speaker, Cohn Lecture at SUNY Stony Brook, NY, June 2008.

251. “Genetics and Treatment of Diseases of the Arterial Wall.” Featured Speaker, First NIH MD/PhD Molecular Medicine Symposium, NIH/NIAMS, Bethesda, MD, June 2008.

252. 24th Annual National Marfan Conference, Speaker and Workshop Leader, Boston, MA, July 2008.

253. “Post-transcriptional Control of Gene Expression". Instructional and featured speaker, 49th Annual Short Course in Medical and “Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2008.

254. “Marfan Syndrome: Disease mechanisms and new ideas about treatment.” Instructional and featured speaker, 49th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2008.

255. “Investigations into the pathogenesis of stiff skin syndrome. Trinity College, Cambridge England, August 2008.

256. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Medical University of South Carolina (MUSC), Charleston, S.C., October 2008.

257. “Marfan Syndrome and Related Disorders: From Molecules to Medicines” (Otto Wolff Lecture), ICH/GOSH Institute of Child Health/Great Ormond Street Hospital, London, England, November 2008.

258. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” ASHG, 2008, Philadelphia, PA, November 2008.

259. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” CITP/Clinical Investigator Training Program, Boston, MA, November 2008.

260. “Cycle of discovery from genes to mechanisms to therapies” (Gene Findings and Mechanisms of Disease). Leducq Mitral Networking Autumn Meeting, Boston, MA, December 2008.

261. “Insights from Marfan Syndrome: Potential from integration into clinical studies.” Leduc Mitral Networking Autumn Meeting, Boston, MA, December 2008.

262. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” NiH Clinical Center: Astute Clinician Lectures, Bethesda, MD, January 2009.

263. “TGF-beta, Microfibrils and Connective Tissue Homeostasis.” Keystone Symposia, Fibrosis, Keystone, CO, January 2009.

264. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” University of Toronto Medical Imaging Strategic Retreat, Toronto, Canada, February 2009.

265. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Stanford Cardiovascular Institute, Stanford University, San Francisco, CA February 2009.

266. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Children’s National Medical Center, Bethesda, MD, March 2009.

267. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” University of Maryland School of Medicine, Baltimore, MD, April 2009.

268. “Interrogation of the Pathogenesis of Stiff Skin Syndrome: A Congenital Form of Scleroderma.” Scleroderma Research Foundation, San Francisco, CA, April 2009.

269. “Marfan disease as paradigm for the transition from medical genetics to genetic medicine.” European School of Genetic Medicine, Bertinoro, Italy, April 2009.

270. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” University of Michigan, Ann Arbor, MI, May 2009.

271. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” The Dr. Jennifer Ann Kierson Memorial Pediatric Grand Rounds Series at Sinai Hospital, Baltimore, MD, May 2009.

272. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Ganz Lecture, Mass. General Hospital (MGH), Boston, MA, June 2009.

273. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Medical and Population Genetics (MPG) Series, The Broad Institute, Cambridge, MA, June 2009.

274. "Post-transcriptional control of gene expression". Instructional and featured speaker, 50th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2009

275. "TGFbeta Signaling and Marfan Syndrome". Instructional and featured speaker, 50th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2009

276. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Clinical Research Seminar, Baylor College of Medicine, Houston, TX, September 2009.

277. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Genetics and Genomics of Vascular Disease Workshop, NAVBO, Cape Code, MA, September 2009.

278. “Therapeutic Insights for Aortic Aneurysm Derived from Interrogation of the Pathogenesis of Marfan Syndrome.” International Aortic Disease Summit, Baltimore, MD, September 2009.

279. “Novel Therapeutic Targets Marfan Syndrome.” International Aortic Disease Summit, Baltimore, MD, September 2009.

280. “The TGFbeta Vasculopathies: From Molecules to Medicines.” President’s Research Seminar, Memorial Sloan-Kettering Institute, New York, NY, September 2009.

281. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” 25th Ernst Klenk Symposium in Molecular Medicine, Extracellular Matrix in Health and Disease, Cologne, Germany, October 2009.

282. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Medical Grand Rounds at UC San Diego, San Diego, CA, October 2009

283. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Molecular Biology Institute Seminar Series, UCLA, Los Angeles, CA, October 2009.

284. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Eight Annual New Principal Investigators Meeting, Canadian Institutes of Health Research, Institute of Genetics and Institute of Musculoskeletal Health and Arthritis, Toronto, CA, November 2009.

285. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” Vascular Lectureship, University of Toronto, Toronto, CA, November 2009.

286. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” BCVS Brown Lecture, American Heart Association Scientific Sessions 2009, Orlando, FL, November 2009.

287. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” 2009 Abelson Advancing Science Salon Event (Translation Medicine and Human Health), Washington, DC, November 2009.

288. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” James Bass Visiting Professorship, Madigan Army Medical Center, Tacoma, WA, December 2009

289. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” St. Christopher’s Hospital for Children/Heart Center for Children, Philadelphia, PA, January 2010

290. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” Temple University, Philadelphia, PA, January 2010

291. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” The Anandi L. Sharma Visiting Professorship, “Controversies in Cardiology”, Mount Sinai Medical Center, New York, NY, February 2010

292. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” Lady Davis Institute for Medical Research, Jewish General Hospital for McGill University (Distinguished Seminar Series) Montreal, Canada, March 2010.

293. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.”Biomedical Discovery Seminar, University of Iowa, Iowa City, Iowa, March 2010.

294. “Parsing of Angiotensin Signaling cascades in the Pathogenesis and Preventions of Aneurysm”. Robert E. Olson Lecture, St. Louis University School of Medicine, St. Louis, MO, April 2010.

295. “The TGFβ Vasculopathies”, The ASCI/AAP Joint Meeting (“Cell Biology, Signaling and Human Disease”), The Fairmont Chicago, Chicago, IL, April 2010.

296. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” 1st Marfan International Congress. SIMA (Congreso Internacional Sindrome de Marfan), Alicante, Spain, May, 2010.

297. "Post-transcriptional Control of Gene Expression". Instructional and featured speaker, 51st Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2010.

298. "TGFbeta Signaling and Marfan Syndrome". Instructional and featured speaker, 51st Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2010.

299. “Marfan Syndrome; from pathogenesis to treatment”. Cutting Edge Rheumatology, The 2010 Frank Wollheim Symposium Lund, Sweden, August, 2010.

300. “Marfan Syndrome and Related Disorders: From Molecules to Medicines.” 4th Annual Case Cardiovascular Center Research Review. Case Western Reserve University, School of Medicine, Cleveland, Ohio, September, 2010.

301. “Matrix-Dependent Perturbation of TGFβ Signaling and Disease”. Extracellular Matrix as a Structural Mediator of Morphological and Homeostatic Information. Foundation des Treilles, Paris, October, 2010.

302. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” David G. Marsh Symposium. The Asthma and Allergy Center, Johns Hopkins Bayview Campus, October, 2010.

303. “What is the Role for Losartan?” and “Where Do We Stand with Basic Science Research for MFS and LDS?” Management of Connective Tissue Disorders and Latest Research: Bicuspid Valves, Marfan Syndrome, Loeys-Dietz Syndrome, Ehlers-Danlos and Related Diseases, Cleveland Clinic Heart & Vascular Institute, November, 2010.

304. “Genetics of Aortic Aneurysm Disease”. VEITH Symposium (37th Annual Symposium on Vascular and Endovascular Issues, Techniques, Horizons. The Hilton New York, New York. November, 2010.

305. “TGFβ in the Pathogenesis and Prevention of Aneurysm: Embracing Paradox.”, Moore Lecture, Washington University School of Medicine, St. Louis, MO, November 2010.

306. “TGFβ in the Pathogenesis and Prevention of Aneurysm: Embracing Paradox”, Fall 2010 Seminar at Columbia University, New York, NY. November 2010.

307. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” Medical Genetics and Genomic Medicine: From Diagnosis to Treatment, NiH, Bethesda, MD. December, 2010.

308. “State-of-the-art review of disease mechanisms revealed by fibrillin-1 mutation: canonical and non-canonical TGF-β pathways and therapeutic opportunities” and “Hematopoietic cell infiltration: A theme shared with Marfan syndrome”. Leducq MITRAL Network Meeting, Boston, MA, December 2010.

309. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” Grand Rounds, Washington Hospital/Georgetown University Hospital Cardiology Program, Washington, DC, January 2011.

310. “Dissection of a Modifier Network Informs the Pathogenesis and Treatment of Marfan Syndrome”. Keystone Symposia (Extracellular Matrix and Cardiovascular Remodeling), Tahoe City, CA, January 2011.

311. “TGFβ in the Pathogenesis and Prevention of Aneurysm: Embracing Paradox.” Gladstone Institute of Cardiovascular Disease, San Francisco CA, January 2011.

312. “Layered Mechanistic Complexity in TGFβ-Induced Disease States: Heritable Connective Tissue Disorders”. Systems Biology and Connective Tissue Disease Meeting, Washington, DC, February 2011.

313. “TGF Beta Vasculopathies”. 2011 ACMG Annual Clinical Genetics Meetings, Vancouver, Canada, March 2011.

314. “Marfan Syndrome and Related Disorders; From Molecules to Medicines.” 23rd Annual Eastern Society for Pediatric Research Meeting, Philadelphia, PA, March 2011.

314. “Marfan Syndrome and Related Disorders; From Molecules to Medicines”. Dean’s Distinguished Lecture Series, University of Kentucky, Lexington KY, April 2011.

316. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”. 2011 Visiting Clinical Geneticists, Harvard Medical School, Boston, MA, April 2011

317. “What Does This Have to do with Marfan Syndrome?: Insights into Common Diseases from the Comprehensive Study of a Rare Mendelian Disorder”. Medical and Population Genetics Program Meeting, Broad Institute, Cambridge, MA, April 2011.

318. “Marfan Syndrome and Related Disorders: From Molecules to Medicines”. Heart and Vascular Institute Grand Rounds, Cleveland Clinic, Cleveland, OH, April, 2011

319. “Dissection of a Modifier Network Informs the Pathogenesis and Treatment of Marfan Syndrome”. Weinstein Cardiovascular Development Conference, Cincinnati, OH, May 2011.

320. “New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”. Jack Peter Lecture, Mount Sinai School of Medicine, New York, NY, May 2011.

321. “Marfan Syndrome and Related Disorders: From Molecules to Medicines”, Medicine Grand Rounds. “TGFBeta in the Pathogenesis of Disease: Embracing Paradox”, Rehfuss Lecture, Jefferson Medical College, Philadelphia, PA, June 2011.

322. “Interrogation of the Pathogenesis of Stiff Skin Syndrome: A Cogential Form of Scleroderma”. Genzyme Corporation, Framingham, MA, June 2011.

323. “Found in translation: new insights regarding the pathogenesis of Marfan syndrome and related disorders”, Translation Opportunities for The Heritable Disorders of Connective Tissue, Shriners Hospitals for Children, Portland, OR, June 2011.

324. "Post-transcriptional Control of Gene Expression". Instructional and featured speaker, 52ndAnnual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2011.

325. "TGFbeta Signaling and Marfan Syndrome". Instructional and featured speaker, 52nd

Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2011.

326. “Matrix-Dependent Perturbation of TGFbeta Signaling and Disease”. FASEB 2011, The TGF-B Superfamily: Signaling in Development and Disease, Lucca, Italy, August 2011.

327. “Found in Translation: New Insights into the Mechanism and Treatment of Marfan Syndrome and Related Disorders”. HHMI Cloister Scholars, NiH, Bethesda, MD, September 2011.

328. “Found in Translation: New Insights into the Mechanism and Treatment of Marfan Syndrome and Related Disorders”. Mount Sinai School of Medicine Graduate School of Biological Sciences 11th Annual MD/PhD Retreat, Ellenville, NY, September 2011.

329. “Marfan Syndrome and Related Disorders: From Molecules to Medicines”. FEBS, Cell Biology and Pharmacology of Mendelian Disorders. Vico Equense, Italy, October 2011.

330. “From Bedside to Bench to Bedside for Aortic Disease”. American Heart Association Scientific Sessions 2012, Orlando, FL, November 2011.

331. “State-of-art review of disease mechanisms revealed by fibrillin-1 mutation: TGF-B pathways and therapeutic opportunities”. Leducq Mitral Network Meeting, Baltimore, MD, December 2011.

332. “Genetic Mechanisms of Marfan Syndrome”. Keystone Symposium, Cardiovascular Development and Regeneration. Taos, New Mexico, January 2012.

333. “Found in Translation: New Insights into the Mechanism and Treatment of Marfan Syndrome and Related Disorders”. Human Genome Meeting, Genetics and Genomics in Personalised Medicine, Sydney, Australia, March 2012.

334. “Found In Translation: Emerging Concepts in the Pathogenesis and Treatment of Inherited Forms of Aortic Aneurysm”, Cardiology in the Capital 2012, Mayo Clinic, Washington, DC, April 2012.

335. “Rational therapeutics for genetic conditions”, Genomic Lecture Series, Surburban Hospital, Bethesda, MD, May 2012.

336. “Found In Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”, Special Seminar Series for the Center for Genomic Medicine, Rockefeller University, New York, NY, May 2012.

337. “The Marfan Saga: State-of-the art review of disease mechanisms and therapeutic opportunities”, Heart Valve Summit, Faculty of Medicine of the Hebrew University, Israel, May 2012.

338. “Marfan Syndrome and Related Disorders: From Molecules to Medicines”, Heart Valve Summit, Faculty of the Hebrew University, Israel, May 2012.

339. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”, Paul Dudley White Lecture, Massachusetts General Hospital, Boston, MA, June 2012.

340. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan

Syndrome and Related Disorders", European Human Genetics Conference (ESHG) 2012, Nuremberg, Germany, June 2012.

341. "Post-transcriptional Control of Gene Expression". Instructional and featured speaker, 53rd Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2012.

342. "TGFbeta Signaling and Marfan Syndrome". Instructional and featured speaker, 53rd Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2012.

343. 33rd Annual David W. Smith Workshop on Malformations and Morphogenesis, Buford, GA, August 2012.

344. "Pathologic roles of TGF-β in Marfan's syndrome and DMD", Antibody Targeting of TGF-β in Musculoskeletal Diseases, Genzyme, Framington, MA, August 2012.

345. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders", Taubman Prize for Excellence in Translational Medical Science, University of Michigan Medical School, Ann Arbor, MI, October 2012.

346. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders", 7th Annual Institute for Translational Medicine and Therapeutics (ITMAT), Perelman School of Medicine, Philadelphia, PA, October 2012.

347. "Context-specified Mechanisms for TGFB-induced Perturbation of Morpogenic and Homeostatic Events", UT Southwestern, University Lecture Series, Dallas, TX, March 2013.

348. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders", UT Southwestern, University Lecture Series, Dallas, TX, March 2013.

349. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders", Texas Children's Hospital, Houston, TX, April 2013.

350. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders", Verge Symposium, Cornell University, Ithaca, NY, April 2013.

351. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders", Aab Cardiovascular Research Institute, University of Rochester, West Henrietta, NY, May 2013.

352. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders", Medicine Grand Rounds, Cleveland Clinic, Cleveland, OH, May 2013.

353. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders", 1st Rare Diseases Summer School, Rare Disease Zurich, Switzerland, July, 2013.

354. ""Post-transcriptional Control of Gene Expression". Instructional and featured speaker, 54th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2013.

355. "Found in Translation: new Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders". Instructional and featured speaker, 54th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2013.

356. “The Extracellular Matrix in Homeostatic, Autoimmune and Fibrotic Disease Processes”, CV Society and Medical Grand Rounds, Mayo Clinic, Rochester, MN, September 2013.

357. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”, Robert L. Frye Lectureship, Mayo Clinic, Rochester, MN, September 2013.

358. “TGF-β and Vascular Disease”, Regulation of Transforming Growth Factor Beta Activity and Cardiovascular Disease, Fondation des Treilles, Nice, France, September 2013.

359. “Exploration of the Downstream Effectors of Diverse TGF beta-Mediated Diseases”, Regulation of Transforming Growth Factor Beta Activity and Cardiovascular Disease Fondation des Treilles, Nice, France, October 2013.

360. “Why Do Some Aortas Dilate”, Canadian Cardiovascular Society, Montreal, Canada, October 2013.

361. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”, University of Florida, Surgery Grand Rounds, Gainesville, FL, November 2013.

362. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”, American Society of Nephrology/Kidney Week, Atlanta, GA, November 2013.

363. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”, Canadian College of Medical Genetics, Toronto, Canada, November 2013.

364. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”, 9th Pan Pacific Connective Tissue Societies Symposium, Hong Kong, November 2013.

365. “New Insights Regarding the Pathogenesis and Modification of Marfan Syndrome and Related Disorders”, 78th Annual Scientific Meeting of the Japanese Circulation Society, Tokyo, Japan, March 2014.

366. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders”, Charles R. Ross Memorial, Student Research Celebration, Upstate Medical University, Syracuse, NY, April 2014.

367. “The Extracellular Matrix in Homeostatic, Autoimmune and Fibrotic Disease Process, Christian J. 2014 Lambertsen Honorary Lecture, University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA, April 2014.

368. “The Extracellular Matrix in Homeostatic, Autoimmune and Fibrotic Disease Processes”, Robert Berliner Professorship, Yale University, New Haven, CT, April 2014.

369. “Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders. American Heart Association, BCVS Meeting (Basic Cardiovascular Sciences), Las Vegas, NV, July, 2014

370. "Post-transcriptional Control of Gene Expression". Instructional and featured speaker, 55th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2014.

371. "TGFbeta Signaling and Marfan Syndrome". Instructional and featured speaker, 55th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2014.

372. "Marfan & Loeys-Dietz", 5th Annual New England Symposium on Marfan Syndrome & Related Disorders. Ambrey Genetics, Marfan Foundation, MA Chapter and the NH/VT Network of the Marfan Foundation, Manchester, NH, September 2014.

373. "Conditional Provocations in a Knock-In-Mouse Model of Marfan Syndrome" (Basic Science Session), "New Therapeutic Opportunities in MFS and LDS as Revealed by Modifier Studies in Patients and Mouse Models" (Mechanisms and Options for Therapeutic Interventions Session), 9th International Research Symposium on Marfan Syndrome and Related Disorders, The Marfan Foundation, Paris (France), September 2014.

374. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders. Centro de Biología Molecular "Severo Ochoa" Consejo Superior de Investigaciones Científicas Madrid, Spain, October 2014.

375. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan syndrome and Related Disorders". Stevenson Lecture, University of Western Ontario, London, Ontario, November 2014.

376. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan syndrome and Related Disorders". Laennec Clinician/Educator Lecture, American Heart Association Scientific Sessions, Chicago, IL, November 2014.

377. "The Extracellular Matrix in Homeostatic, Autoimmune and Fibrotic Disease". Northwestern Lectures in Life Sciences, Northwestern University, Chicago, IL, January 2015.

378. "Personalized Surgery – Genomics and Immunotherapy for Surgical Diseases" (SUS Presidential Session), 10th Academic Surgical Congress, Las Vegas, NV. February 2015

379. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders". Yale Pediatric Grand Rounds, New Haven, CT, February 2015.

380. "TGFbeta in the Pathogenesis of Disease: A Matter of Aneurysmic Proportions". The Deuel Conference on Lipids, Monterrey, CA, March 2015.

381. "The Extracellular Matrix in Homeostatic, Autoimmune and Fibribotic Disease Processes". Annual Aaron I. Grollman Visiting Professorship Lecture (Graduated Program in Life Sciences), University of Maryland School of Medicine, Baltimore, MD, March 2015.

382. "New Insights into the Pathogenesis and Treatment of Scleroderma". Yale Tissue Fibrosis Symposium. Yale University, Orange, CT, April 2015.

383. "Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders. 30th Annual Edward Massie Visiting Professor at Washington University School of Medicine. St. Louis, MO, May 2015.

384. “Of Mice and Modifiers in Inherited Aneurysm Conditions”. Seminar, 30th Annual Edward Massie Visiting Professor, Washington University School of Medicine, St. Louis, MO, May 2015.

385. "Post-transcriptional Control of Gene Expression". Instructional and featured speaker, 56th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2015.

386. "TGFBeta Signaling and Marfan Syndrome". Instructional and featured speaker, 56th Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, Maine, July 2015.

387. “TGFβ in the Pathogenesis of Inherited Vasculopathies: A Matter of Aneurysmic Proportions”, LDS Day, Oxford University, London UK, September 2015.

388. “Found in Translation: New Insights into the Pathogenesis and Treatment of Aortic Aneurysm Syndromes”, Bernard and Joan Marshall Distinguished Investigator Lecture at British Society of Cardiovascular Research (BSCR) Meeting, University of Glasgow, Scotland, September 2015.

389. “Found in Translation: New Insights into the Pathogenesis and Treatment of Aortic Aneurysm Syndromes”, Karsh Visiting Professor, Children’s Hospital of Eastern Ontario (CHEO), Ottawa, Ontario, September 2015.

390. “Of Mice and Modifiers in Inherited Aneurysm Conditions”, University of Chicago, Department of Chicago, Chicago, IL, October 2015.

391. “TGFβ in the Pathogenesis and Treatment of Aortic Aneurysm: A Matter of Aneurysmic Proportions”, Cardiology Grand Rounds, Johns Hopkins University School of Medicine, Baltimore, MD, October 2015.

392. “Pathogenesis and Treatment of Fibrotic Conditions”, The Robert A. Welch Foundation 59th Conference On Chemical Research “Next Generation Medicine”, Houston, TX, October 2015.

393. “TGFβ in Inherited Vasculopathies: A Matter of Aneurysmic Proportions”, Inaugural Gale and Ira Drukier Lecture in Children’s Health, Weill Cornell Medical College, New York, NY, November 2015.

394. “Marfan Syndrome and Loeys-Dietz Syndrome”, 16th Annual International Symposium on Cogenital Heart Disease (All Children’s Hospital), St. Petersburg, FL, February 2016.

395. “The Extracellular Matrix in Homeostatic, Autoimmune and Fibrotic Disease Processes”, UCSF Seminars in Biomedical Sciences seminar series at University of California, San Francisco, CA, April 2016.

396. “TGFβ In Inherited Vasculopathies: A Matter of Aneurysmic Proportions”, Genomics of Rare Disease: Beyond the Exome 2016, Wellcome Genome Campus, Cambridge, UK, April 2016.

397. “Inherited Disease of The Aorta”, Aortic Symposium 2016, New York, NY, May 2016.

398. “Genetics of Aortopathies in Childhood”, AATS/STS Cogenital Heart Disease Symposium, Baltimore, MD, May 2016

399. "New insights into the pathogenesis and treatment of Marfan syndrome and other presentations of thoracic aortic aneurysm". The Allied Genetics Conference (TAGC), Orlando, FL, July 2016.

400. "New Genes and Mechanisms for Thoracic Aortic Aneurysm". GenTAC Thoracic Aortic Disease Summit, Washington, DC, September 2016.

401. "New Insights into the pathogenesis and treatment of fibrosis". Genomics of Common Diseases, Baltimore, MD, September 2016.

402. "Connective Tissue Disorders and Cardiac Surgery (A Success Story)". 2016 Congenital Heart Surgeons Society Meeting, Chicago, IL, October 2016.

403. "TGFβ in the Pathogenesis of Inherited Vasculopathies: A matter of Aneurysmic Proportions", UCLA Molecular Biology Institute, Los Angeles, CA, November 2016.

404. "New Insights into the Mechanisms and Prevention of Fibrotic Disease", 2016 American College of Rheumatology Annual Meeting, Washington, DC, November 2016.

405. "TGFβ in the Pathogenesis of Inherited Vasculopathies: A matter of Aneurysmic Proportions", Houston, TX, James T. Willerson Cardiovascular Seminar, Texas Heart Institute, February 2017.

406. "New Mechanisms and Vulnerabilities for Fibrosis", Consortium for Fibrosis Research & Translation, University of Colorado Anschutz Medical Campus, Denver, CO, April 2017.

407. "Genetics of Aortic Diseases", 2017 Padua Course on Paediatric Cardiology, University of Padua, Padova, Italy, April, 2017.

408. "TGFβ in the Pathogenesis of Inherited Vasculopathies: A matter of Aneurysmic Proportions", Cardiology Grand Rounds, Vanderbilt University Medical Center, Nashville, TN, April 2017.

409. "Of Mice and Modifiers in Inherited Aneurysm Conditions", Medicine Grand Rounds, Vanderbilt University Medical Center, Nashville, TN, April 2017.

410. "TGFβ in the Pathogenesis of Inherited Vasculopathies: A matter of Aneurysmic Proportions", Cardiology Grand Rounds, Duke University Medical Center, Durham, NC, May 2017.

411. "TGFbeta in Inherited Vascuopathies: A Matter of Aneurysmic Proportions. Keystone Symposia (Angiogenesis and Vascular Diseases), Santa Fe, NM, May 2017.

412. "Developmental syndromes and genetic alterations in the TGF-B pathway", Keynote Speaker, FASEB The TGF-B Superfamily: Signaling in Development and Disease", Lisbon, Portugal, July 2017.

413. "The Importance of Paradox and Beauty in Science", GSK Sharing Science, Collegeville, PA, September 2017

414. "TGF-Beta in the Pathogenesis of Inherited Vasculopathies: A matter of Aneurysmic Proportions", John Keith Lecture, Canadian Cardiovascular Congress (CCC),, Vancouver, Canada, October 2017

415. "Genetic Considerations in Aortopathy", Focus on: Aortopathy in the Young, (Breakout Session), Cardiology 2018 - 21st Annual Update on Pediatric and Congenital Cardiovascular Disease, Children's Hospital of Philadelphia, Scottsdale, AZ. February 2018

416. "Leveraging Nature's Success: Lessons from Modifiers of Cardiovascular Disease", Cardiology 2018 - 21st Annual Update on Pediatric and Congenital Cardiovascular Disease, Children's Hospital of Philadelphia, Scottsdale, AZ. February 2018.

417. "Research Update on Marfan Syndrome and Related Disorders", 3rd Annual Colorado Marfan Syndrome and Related Connective Tissue Disorder Symposium, Children's Hospital, Aurora, CO. April 2018.

418. "Leveraging Nature's Success: Lessons from Modifiers of Cardiovascular Disease", Dean's Distinguished Seminar Series, University of Colorado, School of Medicine (Anschutz Medical Campus), Aurora, CO. April 2018.

419. "New Mechanisms and Vulnerabilities for Fibrotic Diseases"**,** Rare Musculoskeletal Disease Symposium, University of Pennsylvania, Philadelphia, PA, June 2018.

420. "Molecular determinants of regional predisposition for aortic aneurysm", Cardiovascular Research Seminar Series (CVRC), Massachusetts General Hospital, Boston, MA, September, 2018.

421. "Leveraging Nature's Success: Lessons from Modifiers of Aortic Aneurysm", Grand Rounds, Massachusetts General Hospital, Boston, MA , September 2018.

422. "Vascular Connective Tissue Disorders: New Insights from Basic Science", 8th Annual New England CTD Meeting, Manchester, NH, September 2018.

423. "New Mechanisms & Vulnerabilities in Fibrosis", American College of Rheumatology Annual Meeting (ACR/ARHP) Meeting, Chicago, IL, October 2018.

424. "Leveraging Nature's Success: Lessons from Modifiers of Cardiovascular Disease", Thomas D. Gelehrter, M.D. Lecture in Medical Genetics, University of Michigan Medical School, Ann Arbor, MI, October 2018.

425. "A Mechanistic Approach to Heritable Thoracic Aortic Disorders: From Gene to Mouse to Patient", 2018 Heritable Aortic Disorders Symposium, Toronto Western Hospital, Toronto, CA, November, 2018.

426. "Leveraging Nature's Success: Lessons from Modifiers of Cardiovascular Disease", Robert Boxer Pediatric Grand Rounds at Cohen Children's Medical Center, New York, February, 2019.

427. "Leveraging Nature's Success: Insights from Modifiers of Marfan Syndrome and Related Disorders", 49th Annual Meeting of the Japanese Society of Cardiovascular Surgery. Okayama, Japan, February, 2019.

428. "Leveraging Nature's Success: Lessons from Modifiers of Cardiovascular Disease", 40th Minhas Lecturer, Pediatric Cardiology Grand Rounds, University of Louisville, Louisville, KY, April, 2019.

429. "TGFβ and Angiotensin Receptor Blockers in Marfan Syndrome: Not for the Faint of Heart", 28th Annual Joel G. Hardman Student-Invited Pharmacology Forum, Vanderbilt in Nashville, TN, April 2019.

430. "Molecular Regulation of Vascular Wall Growth and Remodeling", The 2019 Robert Grover Conference Series, (ATS Assembly on Pulmonary Circulation), Sedalia, CO, September 2019.

431. “Leveraging Nature’s Success: Lessons from Modifiers of Cardiovascular Disease”, Mass General Medical Grand Rounds Clinical Research Day, Massachusetts General Hospital, Boston, MA, October 2019.

432. “Leveraging Nature’s Success: Lessons from Modifiers of Aortic Disease”, Grand Rounds at the John Hopkins Bayview Medical Center, Baltimore, MD, October 2019.

433. “Leveraging Nature’s Success: Lessons from Modifiers of Cardiovascular Disease”, Vascular Biology Meeting (NAVBO), Pacific Grove, CA, October 2019.

434. Keynote Speaker, First International Symposium on Renovascular Hypertension, Taubman Institute Medicine, Ann Arbor, MI, October 2019.

435. “Leveraging Nature’s Success: Insights from Modifiers of Marfan Syndrome and Related Disorders”, Translational Medicine Seminar Series, George Washington University School of Medicine, Washington, DC, December 2019.

436. “New Mechanism and Vulnerabilities of Fibrosis”, Keystone Symposia (Stromal Cells in Immunity and Disease/Fibrosis and Tissue Repair: From Molecules and Mechanics to Therapeutic Approaches), Victoria, BC Canada, January 2020.

437. "New Insights into the Evaluation and Management of Pediatric Vascular Connective Tissue Disorders" Irene Uchida Lectureship (Pediatrics Grand Rounds) @ Univ. of Manitoba, Canada. Virtual/March 2021.

438. “Leveraging Nature’s Success: Lessons from Genetic Modification of Vascular Connective Tissue Disorders", Research Rounds - CHRIM (Children's Hospital Research Institute of Manitoba), Virtual/March 2021.

439. “A Broad Mechanism and Potential Treatment Strategy for Fibrosis”, Hopkins Conte Digestive Diseases Basic & Translational Core Center, Virtual/March 2021.

440. “Leveraging Nature’s Success: Insights from Modifiers of Marfan Syndrome and Related Disorders”, Distinguished Lecture Seminar Series, Department of Human Genetics, Emory University, Virtual/March 2021.

441. “Leveraging Nature’s Success: Lessons from Modifiers of Marfan Syndrome and Related Aneurysm Conditions”, Boston University School of Medicine Cardiovascular Institute Seminar Series, Virtual/April 2021.

442. “Aneurysms and Associated Syndromes” Cardiogenetic: Getting to the Heart of Diagnosis and Management. The Baltimore-Washington Genetics Group Educational Series, Virtual/September 2021.

443. “Leveraging Nature’s Success: Lessons from Modifiers of Marfan Syndrome and Related Vascular Disorders”, Johns Hopkins All Children’s 10th Annual Research Symposium, Virtual/October 2021.

443. “Leveraging Nature’s Success: Lessons from Modifiers of Marfan Syndrome and Related Aneurysm Conditions”. Stanford Medicine Pediatric Grand Rounds, Virtual/January 2022.

444. “Leveraging Nature’s Success: Lessons from Modifiers of Vascular Connective Tissue Disorders”, Osteogenesis Imperfecta Foundation Scientific Meeting, April 2022, Chicago, IL