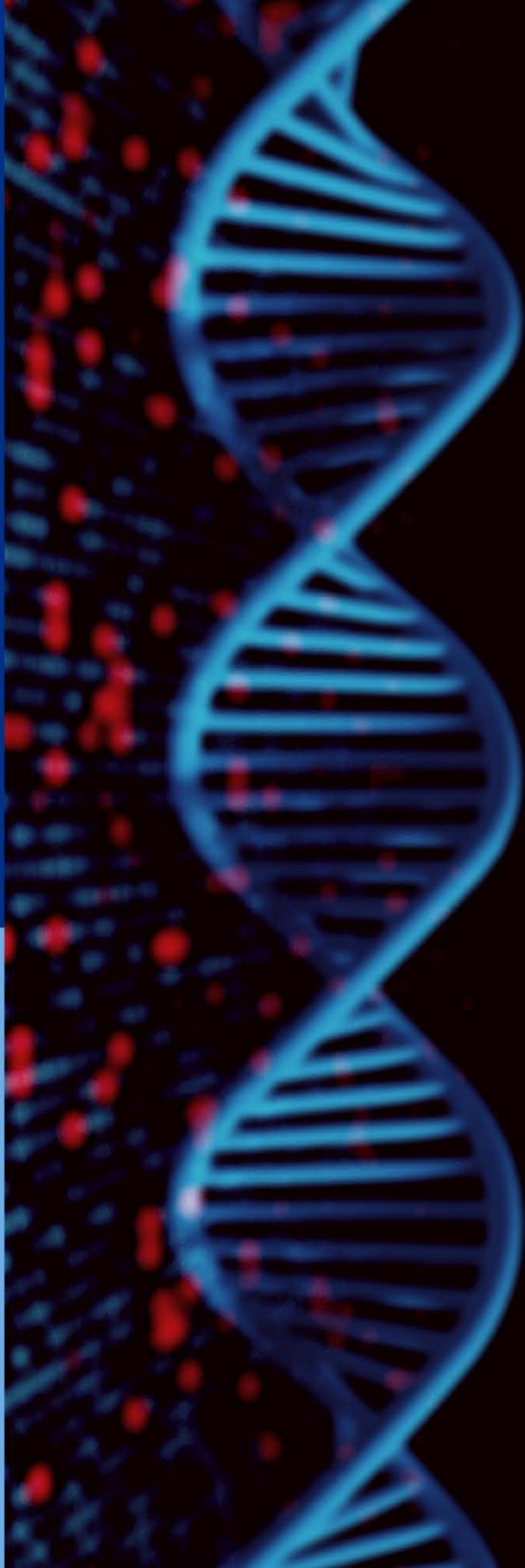
TEXAS GENETICS CONFERENCE

JULY 30, 2022

WARP SPEED GENETICS

FROM BENCH TO BEDSIDE

VIRTUAL • FREE



TEXAS Health and Human Services | Texas Department of State Health Services

Continuing education for multiple disciplines will be provided for this event.

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TEXAS Health and Human Services | Texas Department of State Health Services

Conference Schedule at a Glance

8:00-8:30 a.m.
Attendees Join

8:30-8:45 a.m.
Welcome and Introductions

8:45-9:45 a.m.
Non-Invasive Genetic Testing in Pregnancy: *What to Tell Parents*

9:55-10:55 a.m.
Newborn Screening and Genomics: *Ethical Issues, Resources and Tools*

11:05-12:05 p.m.
A New Era in Therapies for Genetic Disorders

12:15-1:15 p.m.
Working Lunch Panel
Parents Discuss Their Personal Journey with Genetic Testing and the Impact on the Family

1:25-2:25 p.m.
The Undiagnosed Network: *Past, Present, Future*

2:35-3:35 p.m.
Risk-Based Prevention: *Scaling Genomic Applications for Large Populations*

3:45-4:45 p.m.
Compounding Disparities in Genetic Testing for Cancer Predisposition in South Texas

4:45-5:00 p.m.
Conference Wrap Up and Closing Remarks

Warp Speed Genetics:
From Bench to Bedside

Fasten your seatbelts for a warp-speed tour of the evolving genetic landscape that is informing clinical practice in 2022. This conference will lay the foundation for genetic care across the lifespan, from non-invasive prenatal screening to ethical issues in newborn screening, and from cutting-edge gene therapies to cancer genetic predisposition testing "in the trenches." We will also be exploring the interface between research and clinical applications for advanced genetic testing, while recognizing the imperative to engage and include diverse populations in both clinical genetic care and research endeavors.

Who Should Attend: physicians, nurses, nurse practitioners, physician assistants, certified nurse midwives, genetic counselors and others who may encounter patients with genetic disorders. Fields include, but are not limited to: general and family practice, pediatrics, obstetrics and gynecology, and genetics.

Cost: FREE! No cost to register for the online conference

For information email:
registrar@txgeneticsconference.com
or call 512.565.9500

For more information, or to register, check out the website: txgeneticsconference.com

Need help day of the event – email registrar@txgeneticsconference.com

Continuing Education Information

National Society of Genetic Counselors (NSGC)

Genetic Counselor CEUs: The National Society of Genetic Counselors (NSGC) has authorized Texas Department of State Health Services to offer up to 0.7 CEUs or 7 Category 1 contact hours for the activity Texas Genetics Conference – Warp Speed Genetics: From Bench to Bedside. The American Board of Genetic Counseling (ABGC) will accept CEUs earned at this program for the purposes of genetic counselor certification and recertification.

Texas Department of State Health Services

Continuing education credit/contact hours for this event are provided by The Texas Department of State Health Services, Continuing Education Service and include the following:

Continuing Medical Education:
The Texas Department of State Health Services, Continuing Education Service is accredited by the Texas Medical Association to provide continuing medical education for physicians.

The Texas Department of State Health Services, Continuing Education Service designates this live activity for a maximum of 7 *AMA PRA Category 1 Credit(s)*™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

This course has been designated by The Texas Department of State Health Services, Continuing Education Service for 6 credit(s) of education in medical ethics and/or professional responsibility.

Continuing Nursing Education:
The Texas Department of State Health Services, Continuing Education Service is accredited as a provider of continuing nursing education by the American Nurses Credentialing Center's Commission on Accreditation.

The Texas Department of State Health Services, Continuing Education Service has awarded 7 contact hour(s) of Continuing Nursing Education.

Social Workers:
The Texas Department of State Health Services, Continuing Education Service under sponsor number CS3065 has been approved by the Texas State Board of Social Worker Examiners (https://dshs.texas.gov/socialwork) to offer continuing education contact hours to social workers. The approved status of The Texas Department of State Health Services, Continuing Education Service expires annually on December 31. The Texas Department of State Health Services, Continuing Education Service has awarded 7 contact hour(s) of Continuing Social Work Education.

Certified in Public Health
On 10/18/2017 the Texas Department of State Health Services, Continuing Education Service was awarded multi-event provider status (ID #1360) by the National Board of Public Health Examiners to award CPH continuing education credits. This activity has been awarded 7 credits.

Certified Health Education Specialists:
Sponsored by The Texas Department of State Health Services, Continuing Education Service, a designated provider of continuing education contact hours (CECH) in health education by the National Commission for Health Education Credentialing, Inc. This program is designated for Certified Health Education Specialists (CHES®) and/or Master Certified Health Education Specialists (MCHES®) to receive up to 7 total Category I contact education contact hours. Provider ID# 98017

Licensed Marriage and Family Therapists:
The Texas Department of State Health Services, Continuing Education Service is considered a sponsor of continuing education for Licensed Professional Counselors according to the Texas Administrative Code, Title 22, Part 35, Chapter 801, Subchapter K, Rule §801.264 effective May 26, 2019, concerning continuing education sponsors. The Texas Department of State Health Services, Continuing Education Service has awarded 7 contact hour(s) for Licensed Marriage and Family Therapists.

Licensed Professional Counselors:
The Texas Department of State Health Services, Continuing Education Service is considered a sponsor of continuing education for Licensed Professional Counselors according to the Texas Administrative Code, Title 22, Part 30, Chapter 681, Subchapter J, Rule § §681.142. The Texas Department of State Health Services, Continuing Education Service has awarded 7 contact hours of Continuing Education.

Licensed Psychologists:
The Texas Department of State Health Services is authorized by the Texas State Board of Examiners of Psychologists as a (c)(2)(A) provider of professional development hours for licensed psychologists. Per the Texas State Board of Examiners of Psychologists Act and Rules, at least half (10) of the required 20 hours of professional development must be obtained by a provider listed in section (c)(2)(A). The Texas Department of State Health Services, Continuing Education Service has awarded 7 contact hour(s) for Licensed Psychologists.

Certificate of Attendance:
The Texas Department of State Health Services, Continuing Education Service has designated 7 hour(s) for attendance.

Conference Program

8:00 a.m. - 8:30 a.m.

Attendees Join Platform

8:30 a.m. - 8:45 a.m.

Welcoming Remarks and Introductions

Debra Freedenberg, MD, PhD
Medical Director
Newborn Screening & Genetics
Texas Department of State Health Services
Debra Freedenberg, MD, PhD is the Medical Director of the Newborn Screening Program and Genetics at the Texas Department of State Health Services. She has experience in academic, private practice, and public health settings covering all areas of genetics practice.

Dr. Freedenberg is board certified in pediatrics, clinical genetics, clinical molecular genetics, and medical biochemical genetics. She has particular expertise in short-term and long-term follow-up of children identified by newborn screening and serves on many national and regional committees related to newborn screening and genetic services.

8:45 a.m. - 9:45 a.m.

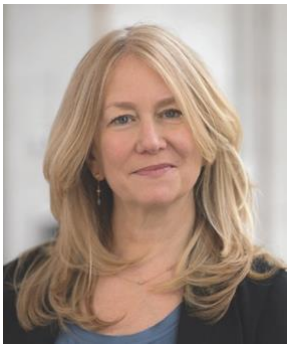
Non-Invasive Genetic Testing in Pregnancy: *What to Tell Parents*

Susan Klugman, MD, FACOG, FACMG
Montefiore Medical Center - Albert Einstein College of Medicine

In the past, non-invasive prenatal tests were only recommended for women at high risk for carrying a baby with a chromosomal abnormality. High-risk pregnancies include moms-to-be who are 35 or older; previously had a child with a genetic disorder; have a family history of a genetic condition; or concern about the results of another prenatal test. Today, genetic screening is offered to all pregnant women and usually discussed during the first prenatal visit. The decision to test is a personal one, and it is important for the family to understand the meaning of a test that is low-risk, not-resulted or high-risk. The counseling phase is especially crucial. Attend this session to review available tests, current guidelines, and learn how to discuss the testing process and results with parents-to-be.

Session Objectives:

- Describe available genetic tests, including the biologic specimen and basis for each test and the steps in the analysis and disclosure of results.
- Describe the role of the physician, nurse, genetic counselor or clinic in working with families whose baby or minor child has received clinically actionable findings unrelated to the initial reason for testing.
- List three issues of concern that may arise for families when genetic testing is done.
- List three ethical issues that may arise related to genetic testing.



Susan Klugman, MD, FACOG, FACMG, serves as Director, Reproductive and Medical Genetics, and Program Director, Medical Genetics and Genomics Residency at Montefiore. She is also Professor, Obstetrics and Gynecology and Women's Health at our Albert Einstein College of Medicine. Dr. Klugman's clinical focus is on the evaluation of patients and families at risk for genetic disorders during pregnancy or preconception, including prenatal diagnosis and consultation for patients undergoing assisted reproductive technologies. She also provides evaluations for patients at risk for hereditary cancer syndromes. Dr. Klugman's research focuses on novel methods for prenatal screening, testing and diagnosis as well as the assessment of new modalities for diagnoses and treatment of hereditary cancer syndromes. Her work has been published in peer-reviewed journals, book chapters, and other print publications. She has completed six-year-terms on both the Committee on Genetics of the American Board of Obstetrics and Gynecology and the Residency Review Committee for Medical Genetics for the Accreditation Council of Graduate Medical Education. In 2021 she served as President Elect and, in 2023, will serve as President of the organization.

9:45 a.m. – 9:55 a.m.

Break

9:55 – 10:55 a.m.

Newborn Screening and Genomics: *Ethical Issues, Resources and Tools*

Amy Brower, PhD
American College of Medical Genetics and Genomics

The increasing use of genomics in newborn screening has accelerated the need for families and patient advocates to engage with public health experts, clinicians and researchers. This session will highlight innovative approaches to creating resources and tools to support these efforts, and will include an important discussion on ethical considerations in light of increasing awareness surrounding health care disparities with population-based genomic screening. The presentation will provide a comprehensive overview and update for professionals who care for families with conditions identified through newborn screening.

Session Objectives:

- Describe the key components of newborn screening in the United States.
- List at least two emerging principles related to the use of genomics in the neonatal period.

In her role at the American College of Medical Genetics and Genomics (ACMG), Dr. Brower's work focuses on creating informatics tool and an expert network of clinicians, researchers and public health teams to advance research in newborn screening. Dr. Brower was a member of the Human Genome Project team and spent a decade in the industry developing molecular diagnostic tools.

She is a former member of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ADHDNC) and a member of the Follow-up and Treatment Subcommittee and co-chair of its Quality Measures and Health Outcome Workgroup. She serves several advisory boards focused on incorporating genomic medicine into clinical care to improve health outcomes. Dr. Brower lectures on genomics, neuroscience, embryology and child development for doctoral-level programs.

10:55 a.m. – 11:05 a.m.

Break

11:05 a.m. - 12:05 p.m.

A New Era in Therapies for Genetic Disorders

Jerry Vockley, MD, PhD, FACMG
Division Director, Genetic and Genomic Medicine, Cleveland Family Endowed Chair in Pediatric Research
Professor of Pediatrics and Human Genetics, and Director, Center for Rare Disease Therapy

For decades, gene therapy has been the ultimate goal for therapy of genetic disorders, replacing or augmenting a damaged copy of a gene with one that works, leading to a cure. While gene therapy has indeed transitioned from pipe dream to evolving reality, new insights into the pathophysiology of genetic disease have provided additional opportunities to develop new therapies in a more rapid fashion than the traditional approach of undirected screening of chemical libraries for molecules with appropriate biological function. This talk will review the scope of new treatments that promise to revolutionize the field of clinical genetics, transforming it from a primarily diagnostic one, to one with a focus on therapy.

Session Objectives:

- Trace the path of development of new treatments based on cellular pathophysiology
- Define the need for novel approaches to clinical trials for rare diseases

Jerry Vockley, MD, PhD, FACMG, is the Division Director, Genetic and Genomic Medicine, Cleveland Family Endowed Chair in Pediatric Research, Professor of Pediatrics and Human Genetics, and Director, Center for Rare Disease Therapy. Dr. Vockley received his undergraduate degree at Carnegie Mellon University in Pittsburgh, Pennsylvania, and received his degree in Medicine and Genetics from the University of Pennsylvania School of Medicine in Philadelphia, Pennsylvania.

He is internationally recognized as a leader in the field of inborn errors of metabolism. His lab has been responsible for identifying multiple new genetic disorders, many of them defects in mitochondrial energy metabolism, and he has published nearly 300 scientific articles in peer review journals. His current research focuses on the molecular architecture of mitochondrial energy metabolism, in which he is breaking new ground in describing the role of dysfunction of mitochondrial energy metabolism in such common conditions as diabetes, obesity, and Alzheimer disease, and branched chain amino acid metabolism.

He also is a leader in the development and testing of novel therapeutic agents for treating inborn errors of metabolism. He is the principal investigator on four NIH grants and a co-investigator on seven others.

12:05 – 12:15 p.m.

Break

12:15 – 1:15 p.m.

Lunch Session: Parents Discuss Their Personal Journey with Genetic Testing and the Impact on the Family

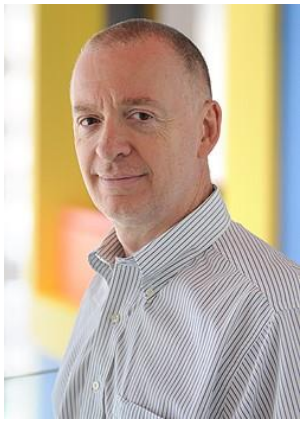
Carlos Bacino, MD, FACMG
Texas Children's Hospital, Baylor College of Medicine And Patient Families

While advances in genomic technology and an increase in the number of gene-disease associations have helped reduce the number of individuals living without a diagnosis, the journey can still be difficult. Genetic diagnoses or test results have significant and far-reaching implications for family members.

Some parents hope that their child's test will have a direct effect on their child's diagnosis; others believe that results will be definitive; while others recognize testing limitations. During this panel discussion, a physician and patients will discuss the impact of genetic testing results on families and reflect on their personal journey.

Session Objective

- Describe the role of the physician, nurse, geneticist or clinic in working with families whose baby or minor child has received clinically actionable findings unrelated to the initial reason for testing.
- Recognize the role of clinicians in discussing results of medically prescribed and commercially available molecular tests



Dr. Bacino is involved in the diagnosis and management of patients with birth defects and a variety of genetic disorders. He participates in subspecialty clinics, such as the Skeletal Dysplasia and Craniofacial Clinics. As the Medical Director of the Kleberg Cytogenetics Laboratory, he has a particular interest in structural chromosome abnormalities and genomic disorders, as well as the mechanism of origin of these chromosome anomalies. In the area of clinical genetics, he is actively involved in the diagnosis and management of pediatric patients with birth defects and rare genetic disorders. He is also interested in the clinical aspect of bone disorders and participates at the Skeletal Dysplasia Clinic at TCH. Currently, he supervises and trains medical students, residents, and fellows.

As the Medical Director of the BMGL Cytogenetics Laboratory, he has a particular interest in structural chromosome abnormalities and genomic disorders (contiguous gene deletion/duplication syndromes).

1:15 – 1:25 p.m.

Break

1:25 – 2:25 p.m.

The Undiagnosed Diseases Network: *Past, Present, Future*

Brendan Lee, MD, PhD
Department of Molecular and Human Genetics, Baylor College of Medicine

The National Institutes of Health is a multi-center collaboration focused on applying in depth clinical phenotyping and multi-omic analysis to make disease diagnosis in patients, while advancing scientific discovery and treatments. The Common Fund's Undiagnosed Diseases Network (UDN) is a research study to improve the level of diagnosis of rare and undiagnosed conditions.

In the United States, it has been estimated that approximately 25 million Americans suffer from a rare disorder. The UDN established a nationwide network of clinicians and researchers who use both basic and clinical research to uncover the underlying disease mechanisms associated with these conditions.

Session Objectives:

- Describe the approach and yield of different multi-omic technologies for diagnosing genetic disease
- Identify how model organisms can be used to determine pathogenicity of variants of uncertain significance identified during genome sequencing.

Brendan Lee, MD, PhD is a pediatrician and geneticist. The overall mission of his research program is to translate the study of structural birth defects and inborn errors of metabolism into a basic understanding of development, disease and novel therapeutic approaches. In the area of metabolism, he has applied genetic approaches to the study of biochemical genetic disorders (specifically urea cycle disorders) as models of complex disease (those involving nitric oxide dysregulation).

This has led his team to study the consequences of metabolic derangements broadly in the endocrine, cardiovascular, skeletal, renal and neurological systems. In the area of structural birth defects, they have studied paracrine and endocrine signaling pathways that regulate skeletal development including morphogens (TGFb, Wnt and Notch), and extracellular matrix proteins and their modifications (e.g., collagen prolyl-hydroxylation) that contribute to the human skeletal dysplasias, including brittle bone diseases. These developmental pathways have led them to ask how their dysregulation contributes to common diseases, such as osteoporosis, osteoarthritis, and bone cancer.

2:25 – 2:35 p.m.

Break

2:35 – 3:35 p.m.

Risk-Based Prevention: *Scaling Genomic Applications for Large Populations*

Alice Zhou, PhD
Color

Genomic medicine promises to improve health outcomes through risk-based prevention and tailored treatment. However, genomic research has long focused on European ancestry populations, threatening the ability of genomic medicine to benefit diverse populations.

One effort to include diverse populations in genomic medicine research is the *All of Us* Research Program - a national initiative aiming to enroll over one million individuals across the United States and return information to improve health. In order to successfully implement this program at scale, participant input was imperative. Results from comprehension testing of health reports to be returned to *All of Us* participants will be discussed in this session.

Session Objectives:

- Describe approaches and considerations for building, scaling, and maintaining population genomics programs.
- Specify results from comprehension testing of health reports for the All of Us Research Program.

Alicia Zhou, PhD, is the Chief Science Officer at Color. She is responsible for leading research collaborations with academic institutions, biopharmaceutical companies, and large population initiatives. She and her team initiate and execute academic collaborations towards driving improved population health outcomes. In addition, Dr. Zhou is an active part of Color's market development team for population genomics and population research. Dr. Zhou received her PhD at Harvard and performed her postdoctoral work at UCSF with an emphasis on cancer biology.

3:35 – 3:45 p.m.

Break

3:45 – 4:45 p.m.

Compounding Disparities in Genetic Testing for Cancer Predisposition in South Texas

Gail Tomlinson, MD, PhD
Division Chief of Pediatric Hematology-Oncology, Mays Cancer Center

The historical lack of access to both genetic services and lack of research enrollment is intertwined with the lack of information which can be provided back to patients. This session will present experience from a recent outreach project targeted at our underserved, primarily Latinx, population of South Texas. We will discuss means of successfully reaching individuals across a large geographic area of 22 counties, including border and rural areas. We provided patient education in a variety of community small-group settings, as well as physician offices, regional medical centers, and mammography centers. Our education is focused on knowledge of explaining hallmarks of common hereditary cancers, and extended opportunities for individuals to self-refer, or for medical professionals to refer patients for genetic risk assessment. We will discuss the impact or our outreach program and the challenges of cancer genetic counseling in an underserved population.

Session Objectives:

- Describe the genetic architecture observed in the Hispanic population of South Texas
- Explain how Variants of Uncertain Certainty compound existing disparities in cancer screening and prevention.
- Recognize instances in which an individual with an uncertain variant will need to undergo cancer screening as if they are high-risk

Gail Tomlinson, MD, PhD serves as the Division Chief of Pediatric Hematology-Oncology and is also currently the co-Director of the Population Science and Prevention Program at the Mays Cancer Center and part of the high-risk Genetic High-Risk Cancer Screening team. Since her arrival in 2007, she has also served as Interim Director of the GCCRI and Interim Chair of the Department of Pediatrics. She is currently the holder of the Greehey Distinguished Chair in Cancer and Genetics. Dr. Tomlinson is board certified in pediatrics and pediatric hematology-oncology.

Her clinical interests are in guiding individuals and families who may be at risk for cancer because of genetic predisposition and leads a multi-site program working with individuals and families throughout South Texas. Her research background is in the genetics and biology of liver tumors in children. She has recently initiated an investigation of predisposing factors underlying the increased incidence of leukemia in children of Hispanic background in South Texas and maintains a growing registry and biobank for children and adolescents with cancer. She has served on multiple national and state committees focusing on advancing research, education, and advocacy in childhood cancer and has published over 180 scientific papers. She currently is a member of the National Cancer Institute's Board of Scientific Counselors.

4:45 – 5:00 p.m.

Conference Wrap-Up / Closing Remarks