The NORD Rare Disease Centers of Excellence, in conjunction with the UT Southwestern Rare Disease Summer Fellowship, presents:

THE 2023 NORD RARE DISEASE SUMMER EDUCATION SERIES

In 2020, Dr. Angela Scheuerle, from the University of Texas Southwestern Medical Center, began a summer rare disease fellowship for 1st year medical students at UT Southwestern. During this fellowship,

her students are exposed to 3 weeks of laboratory genetics topics, followed by 3 weeks of clinical genetics topics, and finish off by attending the Bar Harbor McKusick Short Course. Throughout this fellowship, students rotate through different lab settings and clinics; their hands-on learning is complimented by genetics and rare disease topic lectures given by faculty at UT Southwestern and beyond.

In 2021, UT Southwestern Medical Center, along with 30 other academic institutions across the country, were designated NORD Rare Disease Centers of Excellence (RD CoE). Volunteer clinicians, genetic counselors, researchers, and other allied health professionals from the inaugural 31 sites (as of May 2023, the program now includes 40 sites) came together to begin tackling challenges facing the rare disease community, such as educating the next generation of genetic and rare disease specialists. The Medical Professional Education Working Group proposed expanding and more widely distributing the lecture series put together by Dr. Scheuerle. **This summer we bring you the NORD Rare Disease Summer Education Series.**

Thank you to all the NORD RD CoE volunteers for contributing to this year's summer education series! We could not have put on this program without you and appreciate your time and passion for education.

The National Organization for Rare Disorders (NORD) advances practical, meaningful, and enduring change so people with rare diseases can live their fullest and best lives. Every day, we elevate care, advance research, and drive policy in a purposeful and holistic manner to lift up the rare disease community. **Please send any questions, concerns, or general feedback about this Summer Series to nordcoe@rarediseases.org.**

The goal of this series is to increase your exposure to genetic and rare disease topics. Content for this series comes from any of our 40 Rare Disease Centers of Excellence and covers a wide range of information.

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Speaker(s)	Lecture Description	Streaming Links
Sarah Viall, MSN, Program Director, Clinical Metabolic Genetics Nicholas Ah Mew, MD, Director, Inherited Metabolic Disorders Program Debra Regier, MD, PhD, Medical Director Maura Carroll, MD, Primary Care Pediatrician *Rare Disease Institute at Children's National*	**Rare Disease Diagnosis and Testing Strategies** Apply the types of rare disease testing to specific case scenarios, Describe the risks and benefits of genetic testing, Understand the role of screening and testing in the diagnostic odyssey, Describe how using available practice guidelines can aid in primary care testing approaches	https://centers.rarediseases.org/event/nord-2023-summer-series-rare-disease-diagnosis-and-testing-strategies/
UT Southwestern Medical Center	**Cytogenetics I** A laboratory lecture introducing students to concepts in cytogenetics	https://centers.rarediseases.org/event/nord-2023-summer-series-cytogenetics-ii/
Speakers: Kimberly Chapman, MD, PhD, FAAP, FABIM, FABMGG, Attending Physician, Genetics and Metabolism Debra Regier, MD, PhD, Medical Director Eduardo Fox, MD, Pediatrician Danielle Starin, MS, RD, LD, Research Nutritionist & Metabolic Dietitian Leah Fleming, MD, Clinical Genetics and Medical Biochemical Genetics *Rare Disease Institute at Children's National*	**Culturally Competent Care in your Community** Define Culture, Describe how foods and food priorities vary in Cultures, Identify how you might implement a mechanism to learn about a rare disease family's cultural view point, Describe ways of understanding a patient's culture will improve your ability to provide rare disease care	https://centers.rarediseases.org/event/nord-2023-summer-series-culturally-competent-care-in-your-community/

UT Southwestern Medical Center	**Cytogenetics II** (*Cont.*) A laboratory lecture introducing students to concepts in cytogenetics	https://centers.rarediseases.org/event/nord-2023-summer-series-cytogenetics-i/
Speakers: Deborah Bilder, MD, Professor, Department of Psychiatry, Division of Child & Adolescent Psychiatry Debra Regier, MD, PhD, Medical Director Jamie Fraser, MD, PhD, FACMG, Director, Myelin Disorders Clinic Jenifer Jones-Dees, MD, Independent Pediatrician Christine Maccia, MS, CGC, Genetic Counselor *Rare Disease Institute at Children's National*	**Creation of a Patient-centered, Medical Rare Disease Home** Define a Medical Home, Describe how different groups of patients need different services in their medical home, Identify how to identify resources to equip a busy primary care practice to be a rare disease patient medical home, Describe how using cultural humility can aid in ensuring patients and families have access to meaningful medical homes	https://centers.rarediseases.org/event/nord-2023-summer-series-creation-of-a-patient-centered-medical-rare-disease-home/
Carla Cheatham, MD, MA, PhD, Assistant Professor; Debra Regier, MD, PhD, Medical Director *Rare Disease Insititute at Children's National*	**Support Mechanisms for the Providers of Rare Disease Care** Accept that every care provider can feel various emotions when managing rare disease patients, Identify feelings of insufficiency, sadness, or frustration when managing rare disease patients, Define boundaries in your own practice, Identify one way to increase your own boundaries in your practice.	https://centers.rarediseases.org/event/nord-2023-summer-series-support-mechanisms-for-the-providers-of-rare-disease-care/
Julie B. Eisengart, Ph.D., L.P., Associate Professor, Director - Neurodevelopmental Program in Rare Disease *University of Minnesota*	**Mucopolysaccharidosis disorders: Functional effects** This presentation will review measurement of neurocognition in MPS disorders, describe neurobehavioral manifestations of some MPS types, and outline important considerations in the multi-faceted impacts of these symptoms.	https://centers.rarediseases.org/event/nord-2023-summer-series-mucopolysaccharidosis-disorders-functional-effects/
Dawn Laney, MS, CGC, Director of Genetic Clinical Trial Center *Emory University*	**Fabry disease: From Clinical Trials to FDA Approved Therapies**	https://centers.rarediseases.org/event/nord-2023-summer-series-fabry-

	After a brief description of the root cause and natural history of Fabry disease, this lecture will focus on FDA approved therapies and those in clinical trials that are modifying the condition's natural history.	disease-from-clinical-trials-to-fda-approved-therapies/
Michele Disco, MS, CGC *Columbia University Medical Center*	**Williams Syndrome, Updates** Diagnosis and clinical experience with Williams syndrome, including healthcare maintenance	https://centers.rarediseases.org/event/nord-2023-summer-series-williams-syndrome-updates/
Thomas Cassini, MD, Medical Biochemical Genetics Fellow *National Institute of Health (NIH)*	**Metabolism Overview:** General Categorization of inborn errors of metabolism (IEMs), discussion of the pathways for acute IEMs, and more details on a few classic IEMs	https://centers.rarediseases.org/event/nord-2023-summer-series-metabolism-overview/
Speakers: Clayton Yates, Ph.D., Professor, Department of Biology and Center for Cancer, Tuskegee University Shawneequa Callier, MS, JD, Associate Professor, Department of Clinical Research and Leadership, George Washington University School of Medicine and Health Sciences Moderator: Charmaine Royal, Ph.D., Professor of African & African American Studies, Biology, Global Health, and Family Medicine & Community Health, Duke University. *Department of Molecular and Human Genetics at Baylor College of Medicine and Texas Children's Hospital: Evening with Genetics*	**Race and Genetics: Perspectives of Precision Medicine I** This webinar offers information on the historical, ethical, and legal perspectives resulting in the lack of diversity in genetic/genomic studies and includes an effective approach used in a current research study.	https://centers.rarediseases.org/event/nord-2023-summer-series-race-and-genetics-perspectives-of-precision-medicine-i/ *You can access this webinar at any time*
Speakers: Trudy Nyakambangwe, Founder and Director of Child and Youth Care Zimbabwe Taylor Kane, Founder and Executive Director, Remember the Girls	**Forgotten and Unseen: A Global Perspective on Life as an X-linked Carrier** Join IndoUSrare, Remember the Girls, and Child and Youth Care Zimbabwe for a discussion with X-linked Carriers from three different Continents.	https://centers.rarediseases.org/event/nord-2023-summer-series-forgotten-and-unseen-a-global-perspective-on-life-as-an-x-linked-carrier/

Nirmala Parab, Active Member of Parent Project Muscular Dystrophy Moderator: Reena Kartha, MS, PhD, Associate Professor and Associate Director (Translational Pharmacology) *University of Minnesota/ IndoUSrare*		
Giavanna Verdi, MD, Pediatric-Medical Genetics Resident *University of Alabama at Birmingham*	**The Avenue to a Career in Medical Genetics** Guidance for first and second-year medical students in how to pursue a career in genetics. Bringing medical genetic awareness to the medical student audience, despite what specialty they decide to pursue.	https://centers.rarediseases.org/event/nord-2023-summer-series-the-avenue-to-a-career-in-medical-genetics/
UT Southwestern Medical Center	**Molecular Case Studies I**	https://centers.rarediseases.org/event/molecular-case-studies-i/
Omar Rahman, MD, Division Chief of Medical Genetics *NYP/Weill Cornell*	**Fetal Alcohol Spectrum Disorders: History, Evolution of Diagnosis, and Technology** An overview of the history of the identification of the effects of prenatal alcohol exposure will be presented. The evolution of the diagnostic criteria will be discussed. Finally, the role of technology such as AI in the diagnostic process will be reviewed.	https://centers.rarediseases.org/event/nord-2023-summer-series-fetal-alcohol-spectrum-disorders-history-evolution-of-diagnosis-and-technology/
Anna C. E. Hurst, MD, MS, FACMG *University of Alabama at Birmingham*	**Dysmorphology...With Dolls!** We will provide an overview of how to recognize and describe dysmorphic facial features using photographs of dolls as examples	https://centers.rarediseases.org/event/dysmorphology-with-dolls/

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Rebecca Ganetzky, MD *Children's Hospital of Philadelphia (CHOP)*	**Mitochondrial Disorders**	https://centers.rarediseases.org/event/mitochondrial-disorders/
Laura Klesse, MD, PhD *UT Southwestern Medical Center*	**Neurofibromatosis**	https://centers.rarediseases.org/event/neurofibromatosis/
Garrett Gotway, MD, PhD *UT Southwestern Medical Center*	**Skeletal Dysplasia**	https://centers.rarediseases.org/event/skeletal-dysplasia/
Susan Berry, MD *University of Minnesota*	**Newborn Screening**	https://nordcentral.org/event/newborn-screening/
Ralph DeBerardinis, MD, PhD *UT Southwestern Medical Center*	**Metabolomics**	https://centers.rarediseases.org/event/metabolomics/
Alicia Turner, MSN, APRN, FNP-C V. Reid Sutton, MD *Baylor College of Medicine*	**Evening with Genetics: Navigating New Therapies – 21st Century Treatments for Genetic Disorders**	https://centers.rarediseases.org/event/evening-with-genetics-navigating-new-therapies-21st-centuries-treatments-for-genetic-disorders/ *You can access this webinar at any time*
Jennifer Kalish, MD, PhD *Children's Hosptial of Philadelphia (CHOP)*	**Beckwith-Wiedemann Syndrome**	https://nordcentral.org/event/beckwith-wiedemann-syndrome/
Vinaya Murthy, MS, CGC *Indiana University* *University of California, San Francisco*	**Shaping the Genomics Workforce: The Power of Inclusion** This lecture will review the current landscape of the genomics workforce, describe patient-provider discordance in clinical genetics care, and discuss ways in which we can increase the diversity of the workforce through training and research partnerships	https://centers.rarediseases.org/event/shaping-the-genomics-workforce-the-power-of-inclusion/
Jennifer Ivanovich MS CGC, Cancer Genetic Counselor	**Pediatric Cancer Genetics**	https://centers.rarediseases.org/event/pediatric-cancer-genetics/

Indiana University School of Medicine - Department of Medical and Molecular Genetics	A review of general information about pediatric cancer followed by examples of pediatric cancer predisposition syndromes to explain the care of children and their families with hereditary cancer.	
Jennifer Heeley, MD *Washington University in St. Louis*	**Differences of Sex Development** Review of biological and psychosocial sexual development, and discussion of the multidisciplinary team approach to management of DSDs	https://centers.rarediseases.org/event/differences-in-sex-development/
Nathaniel H Robin, MD, Professor and Clinical Division Director *University of Alabama at Birmingham*	**The Genetics Approach to Tall/Short Stature** A review of the genetics evaluation to small and large sizer, with a focus on clinical evaluation, especially growth parameters.	https://centers.rarediseases.org/event/the-genetics-approach-to-tall-short-stature/
Lakshmi Mehta, MD. Clinical Professor of Pediatrics *Columbia University Medical Center*	**Genetic Causes of Hearing Loss** Hearing loss affects almost 1:500 people. Genetic causes are particularly common in infants with congenital hearing loss. We will review the approach to identifying such causes and how knowledge of a genetic cause can help in patient care	https://centers.rarediseases.org/event/genetic-causes-of-hearing-loss/
Pongtawat Lertwilaiwittaya, MD, Combined Internal Medicine-Medical Genetics PGY-3 *University of Alabama at Birmingham*	**Adult Genetics Overview** The lecture will introduce learners to the Internist - Geneticist pathway. We will focus on common clinical encounters in the Adult clinic with touches in undiagnosed diseases aspect and precision medicine. The session will conclude with career path (Residency application) and Q&A from the audience	https://centers.rarediseases.org/event/adult-genetics-overview/
Amitha L. Ananth, MD, Assistant Professor *University of Alabama at Birmingham*	**Rett Syndrome and Related Disorder** Reviews the history of Rett syndrome, brief discussion of the genetics of the condition, discussion of disorders once thought to be variants of Rett syndrome, now known to be their own conditions (CDKL5, FOXG1)	https://centers.rarediseases.org/event/rett-syndrome-and-related-disorders/
Beth Conover, MS, APRN, CGC, Associate Professor UNMC *University of Nebraska Medical Center Monroe-Meyer Institute*	**Teratology** Discuss various hazardous exposures to pregnant people, consider strategies to effectively convey teratogen information, learn about resources for current and comprehensive teratogen information	https://centers.rarediseases.org/event/teratology/
Lois J. Starr, MD, PhD *University of Nebraska Medical Center Monroe-Meyer Institute*	**Genetic Aortopathies** Discuss the genetic basis and phenotypic spectrum of the more common aortopathies	https://centers.rarediseases.org/event/genetic-aortopathies/

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Joanna Spaulding, MS, CGC, Assistant Professor *University of Nebraska Medical Center, Munroe-Meyer Institute*	**Approaches to Risk Assessment: Cytogenetics and Beyond** Prognosis, susceptibility, recurrence...genetics is all about risk. Using examples from commonly-seen cytogenetic abnormalities, we'll learn how to identify and evaluate these risks. We'll also discuss how to communicate effectively with patients about risk and how these considerations can be used to guide testing and clinical care.	https://centers.rarediseases.org/event/risk-assessment-cytogenetics-and-beyond/
Leslie Dunnington, MS, CGC, Assistant Professor *UT Houston*	**Genetics of Huntington's Disease** We will review Huntington's Disease including signs and symptoms, genetics and special considerations in genetic testing.	https://centers.rarediseases.org/event/genetics-of-huntingtons-disease/
Sara Berger, MS, CGC, Lecturer in Genetic Counseling *Columbia University Medical Center*	**Screening for Genetic Disorders** This lecture focuses on the principles of genetic screening including screening vs. diagnostic testing, clinical validity and utility, sensitivity and specificity, and positive vs. negative predictive values. This also discusses newborn screening and carrier screening as well as ethical considerations.	https://centers.rarediseases.org/event/screening-for-genetic-disorders/
Kristen Fishler, CGC, Genetic Counselor *University of Nebraska Medical Center*	**Rapid Genomic Testing** We will discuss rapid genomic testing including exome and genome as well as studies which have investigated the yield of these tests in the inpatient unit.	https://centers.rarediseases.org/event/rapid-genome-testing/