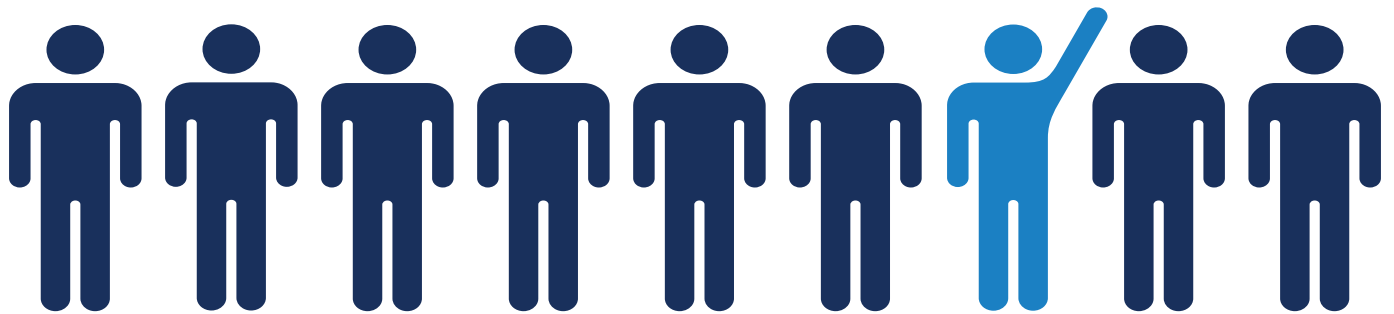




MICHIGAN MEDICINE
UNIVERSITY OF MICHIGAN

RARE DISEASE DAY

EDUCATIONAL CONFERENCE



FEBRUARY 25-26, 2022 | A VIRTUAL EVENT

michmed.org/intmedcme

PROGRAM AGENDA

FRIDAY, FEBRUARY 25, 2022

10:15 am	Welcome and Announcements Elif A. Oral, MD and Goutham Narla, MD, PhD
10:30	Insights from the Undiagnosed Diseases Network William Gahl, MD, PhD
11:10	FDA's Orphan Drug Designation Program Lewis Fermaglich, MD, MHA
11:50	Break
12:00 pm	Rare Disease Modeling: An Engineer's Perspective Jianping Fu, MS, PhD
12:40	Rare Advocacy Council At The State Level: Lessons From Michigan For National Stage Laura Teicher Bonnell
1:20	Panel Discussion William Gahl, MD, PhD; Lewis Fermaglich, MD, MHA; Jianping Fu, MS, PhD and Laura Teicher Bonnell
1:45	CME Session Adjourns
2:00 - 4:30	Bonus and Exhibitor Session

SATURDAY, FEBRUARY 26, 2022

10:20 am	Welcome and Announcements Elif A. Oral, MD and Peter Farrehi, MD
10:30	Full Spectrum Translation in Rare Disease Research Hans Bjornsson, MD, PhD
11:25	Break
11:30	Panel Discussion Hans Bjornsson, MD, PhD and Shigeki Iwase, PhD
12:00 pm	Family Dynamics and Difficult Moments: A Different Facet of Rare Diseases Eric Scott, PhD
12:30	Holistic Care Design for Chronic Intellectual Disabilities Sharon Lewis
1:00	Panel Discussion David Frame, MD; Eric Scott, PhD and Sharon Lewis
1:30	CME Session Adjourns
2:00 - 4:30	Bonus and Exhibitor Session

COURSE OBJECTIVES

At the conclusion of this seminar, participants will be able to:

- understand the national efforts for solving the mystery in the presentation of undiagnosed rare diseases and learn how to refer individual patients.
- appreciate how disciplines such as engineering is helping to solve puzzles and improve therapeutics in rare diseases.
- explain how disease modeling provides insights for breakthroughs for rare diseases.
- review the efforts of the Food and Drug Administration in the development of novel drugs for the rare diseases.
- discuss barriers/challenges in access to state-of-the-art care (in both diagnostics and therapeutics).
- brainstorm on the reinvention of care for patients with rare diseases and disabilities across lifetime.

TARGET AUDIENCE

House Officers, Medical Students, Nurses and Other Allied Healthcare Professionals, Physicians, Social Workers, and Patients and Caregivers.

ACCREDITATION

The University of Michigan Medical School is accredited by the Accreditation Council (ACCME) for Continuing Medical Education to provide continuing medical education for physicians. The University of Michigan Medical School designates this live activity for a maximum of 6.25 *AMA PRA Category 1 Credits*. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

ABIM MOC CREDIT

Successful completion of this CME activity, which includes participation in the evaluation component, enables the participant to earn up to 6.25 Medical Knowledge MOC points in the American Board of Internal Medicine's (ABIM) Maintenance of Certification (MOC) program. Participants will earn MOC points equivalent to the amount of CME credits claimed for the activity. It is the CME activity provider's responsibility to submit participant completion information to ACCME for the purpose of granting ABIM MOC credit.

SPEAKERS

Laura Teicher Bonnell
President
Bonnell Foundation

Hans Bjornsson, MD, PhD
Professor of Translational Medicine and Pediatrics
University of Iceland
Associate Professor of Genetics
Johns Hopkins Medicine

Peter Farrehi, MD
Clinical Associate Professor
Division of Cardiology
Department of Internal Medicine
Michigan Medicine

Lewis Fermaglich, MD, MHA
Orphan Products Medical Officer
Food and Drug Administration

David Frame, MD
Course Co-Director
Clinical Assistant Professor of Pharmacy
Clinical Pharmacist
Michigan Medicine

Jianping Fu, MS, PhD
Professor of Mechanical Engineering
Professor of Biomedical Engineering
Professor of Cell and Developmental Biology
Michigan Medicine

William Gahl, MD, PhD
Director of Undiagnosed Disease Program
Senior Investigator Medical Genetics Brand
Head Human Biochemical Genetics Section
National Human Genome Research Institute

Shigeki Iwase, PhD
Associate Professor
Department of Human Genetics
Department of Pediatrics
Michigan Neuroscience Institute Affiliate
Michigan Medicine

Sharon Lewis
Principal
Health Management Associates

Goutham Narla, MD, PhD
Professor of Internal Medicine
Professor of Human Genetics
Chief, Division of Genetic Medicine, Department of Internal Medicine
Associate Director, Medical Scientist Training Program (MSTP)
Michigan Medicine

Eric Scott, PhD
Clinical Associate Professor
C.S. Mott Children's Hospital
Michigan Medicine

PLANNING COMMITTEE

Elif A. Oral, MD
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Janice Farrehi, MD
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Department of Pediatrics
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Christina Vallianatos, PhD
Research Assistant Iwase Lab
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Emily Vandervoort
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BONUS SESSION

Join us after the CME portion of the course for specialty talks by KDM5C intellectual disability researchers and families, community leaders and Students for Rare (NORD).

FRIDAY, FEBRUARY 25, 2022

- 2:00 pm **Welcome and Announcements**
Elif A. Oral, MD
- 2:10 **Community Leaders and Patient Champions: Navigating Resources for Rare**
Janice Farehi, MD; Evelyn Argirokastritis; Katheryn Elibri Frame, DO; Phoebe Martin, MD and Daniel Dry Dock Shockly

Power Presentations from Labs and Research Groups
- 2:55 **The Differences of Sex Development Translational Research Network (DSD-TRN): Addressing Barriers to Discovery and Improved Pathways of Care**
Bryan Sack, MD
Contributors: B. Sack; M. Gardner; L. Mohnach; and D.E. Sandber
- 3:00 **Atypical Diabetes Genetics Clinical Program: Genetic Counseling for Hereditary Lipodystrophy Syndromes**
Marie-Louise Henry, MS, CGC
Contributors: Colby Chase, Wendy Uhlmann, Veronica Greve; and Kristen Lee
- 3:05 **Characterizing Malnutrition-Related Diabetes Mellitus as a Distinct Form of Diabetes**
Meredith Hawkins, MD, MS
Contributors: Eric-Lontchi-Yimagou; Riddhi Dasgupta; Shajith Aroop; Sylvia Kehlenbrink; Sudha Koppaka; Anankasha Goyal; Michelle Carey; Matthews Kurian; Daniel Stein; Anneka Wickramanayake; Nihal Thomas; and Meredith Hawkins
- 3:10 **Gene Expression Analysis in Familial Partial Lipodystrophy Type 2 (FPLD2)**
Rebecca Schill, PhD
Contributors: Rebecca L. Schill; Jessica N. Maung; Maria C. Foss Freitas; Akira Nishii; John Bonoris; Callie A. S. Corsa; Carolyn Walsh; Elif A. Oral; and Ormond A. MacDougald
- 3:15 **Nuclear Envelope Proteins in Rare and Common Metabolic Liver Disease**
Graham Brady, MD, PhD
Contributors: Kapil Upadhyay, Bishr Omary, Roland Foisner, Elizabeth Speliotes, Xiaomeng Du
- 3:20 **A Truncating Mutation of EBF2 May Disrupt Adipocyte Differentiation in Lipodystrophy**
Maria Cristina Foss de Freitas, MD, PhD
Contributors: Maria Foss-Freitas, Noel Wys, Elif A. Oral, Tae-Hwa Chun
- 3:25 **Joint Discussion**
Bryan Sack, MD; Mary Louise Henry, MS, CGC; Meredith Hawkins, MD, MS; Rebecca Schill, PhD; Graham Brady, MD, PhD and Maria Cristina Foss de Freitas, MD, PhD
- 3:30 **Exhibitor and Interest Group Breakout Rooms**

SATURDAY, FEBRUARY 26, 2022

- 2:00 pm **Welcome and Announcements**
Elif A. Oral, MD
- 2:10 **Basic Research: The Power of Animal Models and their Promise in Rare Disease**
Shigeki Iwase, PhD and Julie Secombe, PhD
- 2:50 **Q&A**
Moderators: Shigeki Iwase, PhD and Julie Secombe, PhD
- 3:00 **KDM5C Intellectual Disability Family Panel**
Moderator: Christina Vallianatos, PhD
- 4:00 **Exhibitor and Interest Group Breakout Rooms**

SPEAKERS

- Evelyn Argirokastritis**
President and Founder
Race for Immunology
- Graham Brady, MD, PhD**
Clinical Lecturer
Division of Gastroenterology
Department of Internal Medicine
Michigan Medicine
- Maria Cristina Foss de Freitas, MD, PhD**
Division of Metabolism, Endocrinology & Diabetes
Michigan Medicine
- Janice Farrehi**
Staff Physician, Infection Prevention Officer
Department of Internal Medicine
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- Katheryn Elibri Frame, DO**
Emergency Medicine Specialist
St. Joseph Mercy Hospital
- Meredith Hawkins, MD, MS**
Professor
Department of Medicine (Endocrinology)
Harold and Muriel Block Chair in Medicine
Director, Global Diabetes Institute
- Shigeki Iwase, PhD**
Associate Professor
Department of Human Genetics
Department of Pediatrics
Michigan Neuroscience Institute Affiliate
Michigan Medicine
- Marie-Louise Henry, MS, CGC**
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Clinical Instructor
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Michigan Medicine
- Phoebe Martin, MD**
Emergency Medicine and Hospitalist
Arkansas Children's Hospital
- Elif A. Oral, MD**
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Albert Einstein College of Medicine
- Dan Dry Dock Shockley**
Genetic Hereditary Testing (MiGHT) Advisory Board
Michigan Medicine
- Christina Vallianatos, PhD**
Research Assistant Iwase Lab
Michigan Medicine

REGISTRATION

CME/MOC RATES

Full Course:

Physician	\$60
Physician-in-Training, Retired Physician, RN, and Allied Health	\$40
Michigan Medicine Faculty	\$20

Register and Pay Online

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PATIENTS, FAMILY AND STUDENT REGISTRATION

[Link to Register](#)

Free

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