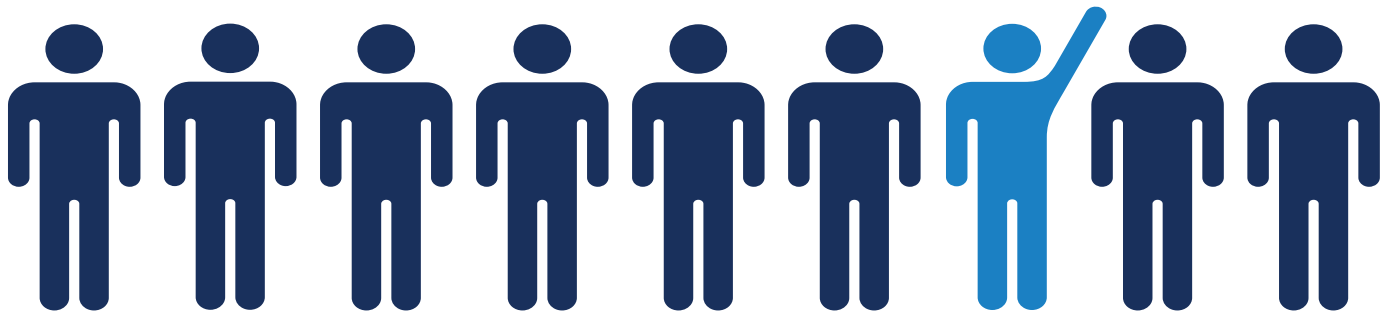




RARE DISEASE DAY

EDUCATIONAL CONFERENCE



FEBRUARY 26-28, 2021 | A VIRTUAL EVENT



[MICHMED.ORG/INTMEDCME](https://michmed.org/intmedcme)

PROGRAM SCHEDULE

FRIDAY, FEBRUARY 26, 2021

The Big Picture in Rare Diseases

10:30 am	Welcome and Announcements Sanjana Sathrasala & Donna Martin, MD, PhD
10:40	How Patients can Lead Drug Development - the Case of Black Bone Disease Nicholas Sireau, MSc, PhD
11:10	Keynote Lecture FV/FVIII and Cargo Receptors in the ER: From Bedside to Bench David Ginsburg, MD
12:00 pm	A Linguist's Reflections on the Word 'Rare' Anne Curzan
12:10	Patient & Caregiver Panel Moderator: Sanjana Sathrasala Patient Panel: Jolie Bailey; Jordan Cobe; Willow Stevens; Becky Strong & Liam Wolf Caregiver Panel: Terry Monkaba; Angela Rousse; Jennifer Shumsky & Rebecca Whitney
1:10	When Every Step is a Fight for Equity, the Value of Patient Advocacy Tanya Collin-Histed
1:35	Panel Discussion
1:45	Industry Exhibit Session

SATURDAY, FEBRUARY 27, 2021

Rare Lessons in Metabolism

10:30 am	Welcome and Announcements Dorene Markel, MS, MHSA
10:40	Diabetes Mellitus: Beyond Type 1 and Type 2 Toni Pollin, PhD
11:00	Monogenetic Obesity and New Treatments Martin Wabitsch, MD, PhD
11:20	Role of ApoC-III in Treatment of Familial Chylomicron Syndrome Joseph Witztum, MD
11:40	The Androgen Profile of 21-Hydroxylase Deficiency as an Example of Looking at Biomarkers in a Rare Disease Richard Auchus MD, PhD
12:00 pm	An Example of How Animal Models Help in Rare Diseases: Adipocyte-Specific Deletion of Lamin A/C as a Model for Familial Partial Lipodystrophy Type 2 Ormond MacDougald, PhD
12:15	Patient Perspectives Moderator: Elif A. Oral, MD Jason Coombs; Jessica Priest; Kristin Skiados and Kayce Trotti
1:00	Panel Discussion
1:45	Industry Exhibit Session

SUNDAY, FEBRUARY 28, 2021

Rare Lessons in Immunology

10:30 am	Welcome and Announcements Kelly Walkovich, MD
10:40	Finding Nemo: My Son's Fight for a Diagnosis Evelyn Argirokastritis
11:10	Role of Genetic Testing in Rare Immunological Disorders Mark Hannibal MD, PhD
11:40	Immunology Keynote: ALPS spectrum V. Koneti Rao, MD
12:40 pm	Panel Discussion and Clinical Care, Patient and Caregiver Perspective Moderator: Kelly Walkovich, MD Panel: Evelyn Argirokastritis; David Frame, MD; Mark Hannibal, MD, PhD; Joseph Jacher, MS,CGC & V. Koneti Rao, MD, FRCPA
1:40	Meeting Wrap Up: Future of Rare Moderator: James Shayman, MD
2:00	Course Adjourns

COURSE OBJECTIVES

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

- appreciate the state of genomic and big data era in driving progress in diagnostics, and therapeutics in rare and orphan diseases.
- review the importance of rare diseases for scientific progress and social justice.
- review the challenges of the patient and care-giver journey.
- discuss barriers/challenges in access to state-of-the-art care (in both diagnostics and therapeutics) across the globe.
- review barriers in therapeutic development for rare diseases.
- provide mentorship opportunities for trainees of all levels (high-school to postdoctoral fellows) in research and advocacy for rare diseases.

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 9.25 *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 9.25 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

Evelyn Argirokastritis

Mother, Caregiver & Founder of Race For Immunology

Richard Auchus MD, PhD

Professor
Division of Metabolism, Endocrinology & Diabetes
Department of Internal Medicine
Michigan Medicine

Tanya Collin-Histed

CEO
International Gaucher Alliance

Anne Curzan

Dean
Professor of English Language and Literature, Linguistics, and Education
College of Literature, Science, and the Arts
University of Michigan

David Frame, MD

Course Co-Director
Clinical Assistant Professor of Pharmacy and Clinical Pharmacist, Michigan Medicine

David Ginsburg, MD

James V Neel Distinguished University Professor Internal Medicine & Human Genetics
Warner-Lambert/Parke-Davis Professor of Medicine Research Professor,
Life Sciences Institute Investigator, Howard Hughes Medical Institute
Michigan Medicine

Mark Hannibal MD, PhD

Associate Professor
Clinical Genetics, Pediatrics
C.S. Mott Children's Hospital
Michigan Medicine

Joseph Jacher, MS, CGC

North American Marketing Manager
Blueprint Genetics

Ormond MacDougald, PhD

Professor
Division of Molecular & Integrative Physiology
Department of Internal Medicine
Michigan Medicine

Dorene Markel, MS, MHSA

Managing Director, Elizabeth Weiser Caswell Diabetes Institute
Director, The Brehm Center
Assistant Research Scientist, Department of Learning Health Sciences
Michigan Medicine

Donna Martin, MD, PhD

Chair, Department of Pediatrics
Ravitz Foundation Endowed Professor of Pediatrics and Communicable Diseases
Professor of Human Genetics

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)
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Course Director
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Toni Pollin, PhD

Associate Professor
Epidemiology & Public Health,
Track Leader, Human Genetics; Program in Epidemiology and Human Genetics, Graduate Program in Life Sciences
University of Maryland School of Medicine

V. Koneti Rao, MD, FRCPA

Staff Clinician, Primary Immune Deficiency Clinic (ALPS Clinic)
National Institute of Allergy and Infectious Diseases

James Shayman, MD

Professor of Internal Medicine and Pharmacology
Agnes C. And Frank D. McKay Professor, Medical School
University of Michigan

Nicholas Sireau, MSc, PhD

CEO and Chairman of the trustees
Alkaptonuria Society

Martin Wabitsch, MD, PhD

Head of the Division of Pediatric Endocrinology and Diabetes
Department of Pediatrics and Adolescent Medicine
Division of Pediatric Endocrinology and Diabetes
Ulm University Medical Center

Kelly Walkovich, MD

Associate Professor
Pediatric Hematology/Oncology
C.S. Mott Children's Hospital
Michigan Medicine

Joesph Witztum, MD

Distinguished Professor of Medicine
University of California San Diego

RARE DISEASE ADVOCATES

Jolie Bailey
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Jason Coombs
Terry Monkaba
Jessica Priest
Angelo Rousse
Jennifer Shumsky

Kristin Skiados
Becky Strong
Willow Stevens
Kayce Trotti
Rebecca Whitney
Liam Wolf

PLANNING COMMITTEE

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RATES

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and Allied Health..... \$40

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Faculty..... \$20

Patients, Families & Students \$0

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