

**The McKusick-Nathans Department of Genetic Medicine
Presents
Inaugural Advances in Genetic and Genomic Medicine 2025 Conference
December 10th 2025**

8:00 AM 8:30 AM Registration and Continental Breakfast

Session 1: Opening Session

Start	End	Title	Speaker
8:30 AM	8:35 AM	Welcome and Conference Goals	Nara Sobreira, MD PhD
8:35 AM	8:45 AM	Annual Departmental Update	Dan Arking, PhD
8:45 AM	8:50 AM	Introduction to 2025 Morning Keynote Speaker	Kim Doheny, PhD
8:50 AM	9:20 AM	2025 Morning Keynote Address Development of Elamipretide as a treatment for Barth syndrome	Hilary Vernon, MD PhD
9:20 AM	9:30 AM	Discussion / Question & Answer	Andy McCallion, PhD

9:30 AM 9:45 AM Break

Session 2: Rare Disease

Moderator: Jefferson Doyle

Start	End	Title	Speaker
9:45 AM	9:55 AM	Myelin lipid composition in adrenoleukodystrophy Mentor: Ali Fatemi, MD (KKI)	Nadav Weinstock, MD PhD
9:55 AM	10:00 AM	Discussion / Question & Answer	Krista Schatz, MS CGC
10:00 AM	10:10 AM	Cell death pathways in lower motor neurons in spinal muscular atrophy Mentor: Charlotte Sumner, MD PhD	Madeline Aldridge, BS
10:10 AM	10:15 AM	Discussion / Question & Answer	Elena Gallo, PhD
10:15 AM	10:25 AM	Mitophagy as a developmental pathology in the heart in Barth syndrome Mentor: Hilary Vernon, MD PhD	Madison Santizo, BA
10:25 AM	10:30 AM	Discussion / Question & Answer	Cesar Fortes-Lima, PhD
10:30 AM	10:40 AM	Uncovering disease mechanisms with long-read and methylation patterns Mentor: Nara Sobreira, MD PhD	Gabriel Gama, PhD
10:40 AM	10:45 AM	Discussion / Question & Answer	Hungoo Lee, PhD
10:45 AM	10:55 AM	A human iPSC-derived skeletal muscle model of Barth Syndrome Mentor: Hilary Vernon, MD PhD	Valeria A. Silva
10:55 AM	11:00 AM	Discussion / Question & Answer	Tao Wang, MD PhD

11:00 AM 11:15 AM Break / Refreshments

Session 3: Complex Disease

Moderator: Nara Sobreira

Start	End	Title	Speaker
11:15 AM	11:25 AM	Uncovering genetic predictors of kidney dysfunction in sickle cell disease Mentor: Ambroise Wonkam, MD PhD	Fujr Osman
11:25 AM	11:30 AM	Discussion / Question & Answer	Christy Smith, ScM CGC
11:30 AM	11:40 AM	Genetic studies of hearing loss in African populations Mentor: Ambroise Wonkam, MD PhD	Abdoulaye Yalcouye, MD PhD
11:40 AM	11:45 AM	Discussion / Question & Answer	Amy Patterson, MS CGC
11:45 AM	11:55 AM	Uncovering the nuclear genetic basis of mitochondrial DNA heteroplasmy Mentor: Dan Arking, PhD	Rama Alhariri
11:55 AM	12:00 PM	Discussion / Question & Answer	Lisa Kratz, PhD
12:00 PM	12:10 PM	Clonal hematopoiesis is adaptive and convergent Mentor: Mary Armanios, MD PhD	Eric Liu, MS
12:10 PM	12:15 PM	Discussion / Question & Answer	Neeraj Sharma, DVM PhD
12:15 PM	12:25 PM	HIF1-regulated PPME1 expression promotes breast cancer via AKT/ β Catenin Mentor: Greg Semeza, MD PhD	Yajing Lyu
12:25 PM	12:30 PM	Discussion / Question & Answer	Jefferson Doyle, MD PhD

12:30 PM 1:30 PM Lunchbreak and Poster Session in Turner Concourse

12:30 PM	1:30 PM	Lunchbreak and Poster Session in Turner Concourse	
Session 4: Genetic Diagnostics			Moderator: Jefferson Doyle
Start	End	Title	Speaker
1:30 PM	1:40 PM	Deep learning prediction of transposable element exonization in human genomes Mentor: Liliana Florea, PhD	Zitong He
1:40 PM	1:45 PM	Discussion / Question & Answer	Mike Beer, PhD
1:45 PM	1:55 PM	CRISPRi/a in zebrafish for systemic biological exploration of genes implicated in disease Mentor: Andy McCallion, PhD	Nelson Barrientos
1:55 PM	2:00 PM	Discussion / Question & Answer	Shira Ziegler, MD PhD
2:00 PM	2:10 PM	SpliSync: Genomic language model-driven splice correction of long RNA sequencing reads Mentor: Liliana Florea, PhD	Wui Wang Lui
2:10 PM	2:15 PM	Discussion / Question & Answer	Winston Timp, PhD
2:15 PM	2:25 PM	Novel EV-based biomarkers from iPSC neurons for Alzheimer's disease Mentor: Vasiliki Machairaki, MSc PhD	Waqar Ahmed, PhD
2:25 PM	2:30 PM	Discussion / Question & Answer	Weiyi Mu, ScM
2:30 PM	2:40 PM	Investigating the role of CFTR in NCFB individuals to elucidate mechanism and therapeutics Mentor: Neeraj Sharma, DVM PhD	Ana Topasna
2:40 PM	2:45 PM	Discussion / Question & Answer	Karen Raraigh, MGC CGC
2:45 PM	3:00 PM	Break / Refreshments	
Session 5: Genetic Treatments			Moderator: Nara Sobreira
Start	End	Title	Speaker
3:00 PM	3:10 PM	Allele-specific genome editing Mentor: Greg Newby, PhD	Johannes Dohr, PhD
3:10 PM	3:15 PM	Discussion / Question & Answer	Vasiliki Machairaki, MSc PhD
3:15 PM	3:25 PM	Hepatic targeted <i>OAT</i> normalizes plasma ornithine in gyrate atrophy mice Mentor: Jefferson Doyle, MD PhD	Julia Johansson
3:25 PM	3:30 PM	Discussion / Question & Answer	Greg Newby, PhD
3:30 PM	3:40 PM	Effects of <i>HIF1A</i> & <i>VHL</i> variants in Ollier disease and Maffucci syndrome Mentor: Nara Sobreira, MD PhD	Patricia Varela Calais, PhD
3:40 PM	3:45 PM	Discussion / Question & Answer	Julie Hoover-Fong, MD
3:45 PM	3:55 PM	Base and prime editing methods to correct GNE myopathy Mentor: Greg Newby, PhD	Noah Workman
3:55 PM	4:00 PM	Discussion / Question & Answer	David Valle, MD
4:00 PM	4:10 PM	Logic-gated CAR-T cells for the treatment of relapsed AML post-transplant Mentor: Bert Vogelstein, MD	Nikita Marcou, PhD
4:10 PM	4:15 PM	Discussion / Question & Answer	Mary Armanios, MD PhD
4:15 PM	4:25 PM	Small molecule & modulators circumvent NMD of nonsense CFTR variants Mentor: Neeraj Sharma, DVM PhD	Jhanay Davis
4:25 PM	4:30 PM	Discussion / Question & Answer	Ada Hamosh, MD
4:30 PM	4:35 PM	Closing remarks	Nara Sobreira, MD PhD
4:35 PM		Adjourn	