

1st European Society of Human Genetics Course on:

Precision Medicine: A Focus on Clinical Utility

April 25th - 27th 2022

Mater Misericordiae University Hospital, Dublin (Virtual for 2022)

Organising Committee: Prof (Assoc) James J. O'Byrne (Director), Prof Sally Ann Lynch, Dr Janna Kenny, Dr Shane McKee, Prof G Jane Farrar, Prof Peter Doran

MONDAY, APRIL 25th

9.00 – 9.10 Welcome Comments

- Course Director: Prof James J. O'Byrne
- MMUH Representative : Prof Jim Egan

9.10 – 11.00

Session 1: Precision Medicine: Introduction, Overview & Patient Perspective
9.10 – 9.25: The Patient Experience of Genetics in Ireland
Avril Daly (Retina International)
9.25 – 9.50: History and Fundamentals of Precision Medicine
Prof Ross McManus (TCD)
9.50 – 10.15: Gene Replacement, Gene Augmentation and Gene Editing: From Bench to Bedside
Prof Gwyneth Jane Farrar (TCD)
10.15 – 10.40: Is Precision Medicine Relevant in the Age of COVID-19?
Prof Paul S. Appelbaum (Columbia University)
10.40 – 11.00: GROUP DISCUSSION

11.00 – 11.20: BREAK

11.00 – 12.40

Session 2 : Clinical Applications of Precision Medicine I: Germline Disorders
11.20 – 11.32: Biochemical Genetics/ Inherited Metabolic Disorders (IMDs)
Prof James J. O'Byrne (MMUH/TCD/UCD); Prof A. A. Monavari (CHI/UCD) (*"Precision Medicine in Children with IMDs"*)
11.32 – 11.44: Cardiac Disorders
Dr Joseph Galvin (MMUH); Dr Terence Prendiville (CHI/NUIG) (*"Modelling Inherited Heart Disease for functional validation and therapeutics development"*)
11.44 – 11.56: Retinal Dystrophies
Prof David Keegan (MMUH/UCD/CHI/RetinaScreen/ERN-EYE)
11.56 – 12.08: Treatment of Renal Disease
Prof Peter J. Conlon (Beaumont/ RCSI)
12.08 – 12.20: Rare Bone Disorders
Dr Rachel Crowley (SVUH/UCD)
12.20 – 12.40: GROUP DISCUSSION
12.40 – 13.20: LUNCH BREAK

13.20 – 16.45

Session 3: Precision Medicine: Economics, Access, Public Health & Ethics
13.20 – 13.55: Health Economics of Precision Medicine
Dr James O'Mahony (TCD)
13.55 – 14.30: Public Health and the Precision Medicine Role
Dr Abigail Collins (Director of Public Health, HSE)
14.30 – 15.05: Rare Diseases and Access to Precision Medicines in Europe
Prof Eileen Treacy (National Rare Diseases Office Dublin/TCD/UCD)
15.05 – 15.40: Ethical Considerations in Precision Medicine
Dr Janna Kenny (CHI/RCSI)
15.40 – 16:15: Rapid Whole Genome Sequencing & Precision Medicine in the Critically Ill ICU Patient
Prof Stephen Kingsmore (Rady Children's Institute for Genomic Medicine)
16.15 – 16.45: GROUP DISCUSSION

END OF DAY

To register go to:

<http://reg.eshgprecisionmedicine.com/>

For enquires contact:

eshgprecisionmedicine@gmail.com

70 ESHG funded scholarships to cover attendance

TUESDAY, APRIL 26th

9.00 – 9.05 Welcome to Day 2

- TCD Representative: Prof Michael Gill

9.05 – 10.55 **Session 4: Clinical Applications of Precision Medicine II: Germline Disorders**

9.05 – 9.35: Pharmacogenetic Testing in Clinical Practice

Prof Jesse J. Swen (Leiden University Medical Center, The Netherlands)

Clinical Application of Precision Medicine in:

9.35 – 9.47: Neuromuscular Disease

Prof Declan O'Rourke (CHI/UCD/TCD/ERN-NMD)

9.47 – 9.59: Neurocutaneous Disorders

Dr Deirdre Donnelly (Belfast Health and Social Care Trust)

9.59 – 10.11: Epilepsy

Dr Kathleen Gorman (CHI/UCD)

10.11-10.23 Cystic Fibrosis

Prof Paul McNally (CHI/RCSI)

10.23 – 10.35: Diagnosis and Treatment of Sickle Cell Disease

Prof Corrina McMahon (UCD/TCD/CHI)

10.35 – 10.55: **GROUP DISCUSSION**

10.55 – 11.10: **BREAK**

10.55 – 13.30 **Session 5: Clinical Applications of Precision Medicine III: Focus on Mitochondrial/ Prenatal and Lipid Disorders**

11.10 – 11.35: Precision Medicine in Mitochondrial Disease Service and Mitochondrial Donation

Prof Grainne Gorman (Newcastle upon Tyne Hospitals NHS Foundation Trust/Newcastle University)

11.35 – 11.55: Acute Genomics in the Prenatal Setting – Improving Outcomes for Families

Dr Samantha Doyle (NMH/UCD)

11.55 – 12.10: Application of Precision Medicine to Dyslipidaemia

Dr Kathryn Ryan (South Eastern HSC Trust/ Chair - NI regional Familial Hypercholesterolaemia Group)

12.10 – 12.30: Evolution of Genetic Counselling in the era of Precision Medicine

Dr Alana Ward (National Rare Disease Office, Dublin)

12.30 – 12.50: **GROUP DISCUSSION**

12.50 – 13.30: **LUNCH BREAK**

13.30 – 16.40 **Session 6: Challenges in Precision Medicine: Data/ Interpretation/ Literacy**

13.30 – 14.10: What's baby, what's bathwater? Pulling genomic data and health records together to deliver diagnostics in rare disorders
Dr Shane McKee (Belfast HSC Trust)

14.10 – 14.50: Variant Interpretation/Reinterpretation And implications in Precision Medicine
Dr Sinéad Howard (NGS Lab, MMUH)

14.50 – 15.30: Clinical Pitfalls in Genomic/ Precision Medicine
Prof Sally Ann Lynch (CHI/UCD)

15.30 – 16.10: Increasing the Literacy in Genomic/ Precision Medicine: The role of European Society of Human Genetics
Prof William Newman (Manchester University NHS Foundation Trust/University of Manchester)

16.10 – 16.40: **GROUP DISCUSSION**

END OF DAY

WEDNESDAY, APRIL 27th

9.00 – 9.05 **Welcome to Day 3**

- UCD Representative: Prof Peter Doran

9.10 – 11.00 **Session 7: Clinical Applications of Precision Medicine IV: Focus on Cancer & Somatic Disorders**

9.05 – 9.50: Insights Offered by Cancer Whole Genome Sequencing

Prof Serena Nik-Zainal (University of Cambridge)

Clinical Application of Precision Medicine in:

9.50 – 10.02: Breast Cancer

Prof David Gallagher (St James's Hospital/ TCD)

10.02 – 10.14: Ovarian Cancer

Prof Karen Cadoo (St James's Hospital/ TCD)

10.14 – 10.26: Endometrial Cancer

Prof Donal Brennan (MMUH/SVUH/ UCD)

10.26 – 10.38: Familial Colorectal Cancer: Emerging Clinical Recognition and Insights in Precision Medicine

Prof Pádraic MacMathúna (MMUH/UCD)

10.38 – 11.00: GROUP DISCUSSION

11.15 - 13.30 **Session 8: Precision Medicine: Next Generation Technologies**

11.15 – 11.40: MultiOmics and Precision Medicine: Focus on Epigenomics

Dr Therese Murphy (TU Dublin)

11.40 – 12.05: The Role of Gene Delivery in the Clinical Translation of Gene-Based Therapeutics

Prof Sally-Ann Cryan (RCSI)

12.05 – 12.30: Artificial Intelligence & Machine Learning in the era of Precision Medicine

Dr Sudipto Das (RCSI)

12.30 – 12.55: Deep learning and single cell RNA-seq: understanding and predicting response to TIL therapies

Dr Pilib Ó Broin (NUIG)

12.55 – 13.10: CAR T Cell Therapies Focus

Dr Roisin O'Ceirbhail (Memorial Sloan Kettering Cancer Center/ Weill Cornell Medical College)

13.10 – 13.30: GROUP DISCUSSION

13.30 – 14.00: LUNCH BREAK

Lunch Time Industry Talk:

“Precision Medicine 2025: Making Medical Genomics Easy for all Clinicians”

Prof John M. Greally (Albert Einstein College of Medicine and Montefiore Medical Center, NY)

14.00 – 16.30

Session 9: Precision Medicine: New Frontiers

14.00 – 14.15: Precision Medicine in Amyotrophic Lateral Sclerosis: Are we there yet?

Prof Orla Hardiman (Beaumont Hospital/TCD)

14.15 – 14.40: New Education and Careers Required: How to start to populate the Precision Medicine landscape

Prof Ute Moog (Chair - European Board Medical Genetics/Heidelberg University Hospital)

14.40 – 15.05: Increasing Literacy in Precision Medicine: The role of the European Reference Networks for Rare Diseases

Prof Franz Schaefer (Heidelberg University Center for Pediatrics and Adolescent Medicine, and Center for Rare Diseases/ERKNet)

15.05 – 15.30: Precision Diagnostics & Therapeutics in Mosaic Overgrowth

Dr Leslie Biesecker (Centre for Precision Health Research NIH/USA)

15.30 – 15.55: Precision Medicine: The European context and the next steps

Prof Maurizio Scarpa Director, Regional Coordinating Centre for Rare Diseases, Udine University Hospital, Italy) & ERN Coordinator For Hereditary Metabolic Diseases (MetabERN)

15.55 – 16.25: GROUP DISCUSSION

16.25 – 16.30: CLOSING COMMENTS

Course Director : Prof James J. O'Byrne

END OF COURSE

Key for Abbreviations:

CHI = *Children's Health Ireland*
ERKNet = *European Reference Network for Rare Kidney Diseases*
ERN = *European Reference Network*
HSC = Health and Social Care Trust
HSE = Health Service Executive
MMUH = *Mater Misericordiae University Hospital*
NHS = *National Health Service*
NMH = *National Maternity Hospital*
NUIG = *National University of Ireland Galway*
RCSI = *Royal College of Surgeons Ireland*
SVUH = *St Vincent's University Hospital*
TCD = *Trinity College Dublin*
TU Dublin = *Technical University Dublin*
UCD = *University College Dublin*

COURSE ACCREDITATION

Accreditation for CPD/ CME is being sourced through the
European Accreditation Council for Continuing Medical Education (EACCME)

COURSE REGISTRATION

To register go to: <http://reg.eshgprecisionmedicine.com/>

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