

## 1<sup>st</sup> European Society of Human Genetics Course on:

## Precision Medicine: A Focus on Clinical Utility

April 25<sup>th</sup> - 27<sup>th</sup> 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 25<sup>th</sup>

#### 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](mailto:eshgprecisionmedicine@gmail.com)

**70 ESHG funded scholarships to cover attendance**

## TUESDAY, APRIL 26<sup>th</sup>

### 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 27<sup>th</sup>

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 MacMathuna (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'Cearbhaill (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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