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
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 IMCs”)
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: GROUP DISCUSSION

16.15 – 16.45: LUNCH BREAK

END OF DAY

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

For enquiries 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**

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<tr>
<th>Time</th>
<th>Session 7: Clinical Applications of Precision Medicine IV: Focus on Cancer &amp; Somatic Disorders</th>
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| 9.00 – 9.05 | Welcome to Day 3  
- UCD Representative: Prof Peter Doran                                                   |
| 9.10 – 11.10 |  
- **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) |
| 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 Padraic MacMathuna (MMUH/UCD) |
| 10.38 – 11.00 | **GROUP DISCUSSION** |

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<tr>
<th>Time</th>
<th>Session 8: Precision Medicine: Next Generation Technologies</th>
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- **Session 8:** Precision Medicine: Next Generation Technologies  |
| 11.15 – 12.05 | MultiOmics and Precision Medicine: Focus on Epigenomics  
- Dr Therese Murphy (TU Dublin) |
| 12.05 – 12.30 | The Role of Gene Delivery in the Clinical Translation of Gene-Based Therapeutics  
- Prof Sally-Ann Cryan (RCSI) |
| 12.30 – 12.55 | Artificial Intelligence & Machine Learning in the era of Precision Medicine  
- Dr Sudipto Das (RCSI) |
| 12.55 – 13.10 | Deep learning and single cell RNA-seq: understanding and predicting response to TIL therapies  
- Dr Pilib Ó Broin (NUIG) |
| 13.10 – 13.30 | **GROUP DISCUSSION** |

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<tr>
<th>Time</th>
<th>Session 9: Precision Medicine: New Frontiers</th>
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<td>13.30 – 14.00</td>
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- **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** |

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<th>Time</th>
<th>Closing Comments</th>
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| 16.25 – 16.30 | Course Director: Prof James J. O’Byrne  
- **END OF COURSE** |
COURSE ACCREDITATION

An Application has been made to the UEMS
European Accreditation Council for Continuing Medical Education (EACCME)
for CME accreditation of this event

COURSE REGISTRATION

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

For enquires contact: eshgprecisionmedicine@gmail.com

70 ESHG funded scholarships to cover attendance

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
Industry Talk (April 27th 13.30-14.00)

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

Prof John M. Greally

Albert Einstein College of Medicine and Montefiore Medical Center, NY