



**27<sup>th</sup> ANNUAL SCIENTIFIC MEETING**  
**19<sup>th</sup> of September 2024**  
**Royal College of Physicians of Ireland, Dublin**



**MORNING SESSION**

- 09:00 Registration, Tea & Coffee, Poster Viewing and Sponsor Visits**
- 09.50 Welcome from ISHG chairperson**  
Dr Deirdre Donnelly
- 10:00 Keynote I The enigma of the mind: the interface of psychiatry and genomics**  
Prof Aiden Corvin, Trinity College Dublin
- 10:45 Oral presentations (Basic) Plenary I** (Each presentation 8 minutes + 2 minutes Q&A)  
Chairs: Dr Edmund Gilbert and Dr Brónagh Ó hCí
- OP01 Characterisation of diverse global ancestries within participants of the UK Biobank**  
Ms Fiona Pantring, Prof Gianpeiro Cavalleri, Dr Edmund Gilbert, Royal College of Surgeons, Dublin
- OP02 Genomic Insights into an Irish Hunter-Gatherer Community**  
Mr Corey Alwell, Dr Marion Dowd, Dr Linda Fibiger, Dr Lara Cassidy, Trinity College Dublin
- OP03 Analysis of blood-based DNA Methylation signatures and disease progression in Inflammatory bowel diseases patients**  
Mr Trevor Doherty, Dr Edel McDermott, Prof Hugh Mulcahy, Prof Sarah Jane Delaney, Dr Therese Murphy, Technological University, Dublin
- OP04 FOXP1 Dysregulation and its Association with Schizophrenia and Cognitive Function**  
Ms Deema Ali, Prof Derek Morris, University of Galway
- OP05 Non-invasive testing of hospitalised patients identifies novel actionable targets against SARS-CoV-2**  
Ms Laura Freeman, Dr Joseph McLaughlin, Dr Darren McDaid, Dr Seodhna Lynch, Dr Andrew English, Mr Jonathon McLaughlin, Dr Maurice O’Kane, Dr Martin Kelly, Dr Manav Bhavsar, Dr Victoria McGilligan, Dr Priyank Shukla, Dr Shu-Dong Zhang, Dr Elaine Murray, Dr Taranjit Singh, Prof Anthony Bjourson, Dr David Gibson, Dr Rachelle Irwin, Prof Colum Walsh, Ulster University
- 11:45 Tea & Coffee, Poster Viewing and Sponsor Visits**
- 12.10 Keynote II: From Gene Hunting to New Treatments – The Complex Genetics of Amyotrophic Lateral Sclerosis**  
Prof Orla Hardiman, Trinity College Dublin
- 12:55 Student Summer Scholarship talks** (Each presentation 8 minutes + 2 minutes Q&A)  
Chairs: Dr Therese Murphy and Ms Jacqueline Turner
- OP06 Exploring the limits of short-read WGS by investigating samples from three population cohorts**  
Ms Ruth McNulty, P.I. Dr Niamh Ryan, Trinity College Dublin
- OP07 Comparing the Rare disease diagnostic journey in the Republic of Ireland to the rest of Europe, how do we compare?**  
Ms Ceri Arnott, P.I. Prof Sally Ann Lynch, Children’s Health Ireland at Crumlin, Dublin
- OP08 Using CRISPR to create ciliopathy models in C. Elegans**  
Ms Glazier Ventura, P.I. Dr Karen Lange, University College Dublin
- 13:25 Lunch, Poster Viewing and Sponsor Visits**
- 14:25 ISHG Annual General Meeting & Chairperson Report**

## AFTERNOON SESSION

- 14:40 Keynote III: Genomic insights into mechanisms and consequences of reproductive ageing and menopause**  
Prof Anna Murray, University of Exeter
- 15:25 Oral presentations (Clinical) Plenary II** (Each presentation 8 minutes + 2 minutes Q&A)  
Chairs: Dr James O'Byrne and Dr Lisa Bradley
- OP09 A clinical evaluation of GLA variants of uncertain significance**  
Ms Cliona Carroll, Dr Laoi Shakerdi, Dr Robert O'Byrne, Ms Alison Sheerin, Ms Jessica Ivory, Mr Kevin Jon Ilagan, Ms Aya Ibrahim, Dr James O'Byrne, Mater Misericordiae Hospital, Dublin
- OP10 Catalogue of inherited autosomal recessive disorders found amongst the Roma population of Europe**  
Dr Shauna Quinn, Dr Ioana Streata, Dr Aoibhinn Walsh, Dr Kathleen Gorman, Dr Ellen Crushell, Prof Andrew Green, Dr Janna Kenny, Dr Anca Lelia Riza, Prof Sally Ann Lynch, Children's Health Ireland at Crumlin, Dublin
- OP11 Direct to Consumer Genetic testing; the experience of two Clinical Genetics services in the Republic of Ireland**  
Ms Emma O'Donoghue, Ms Cliona Carroll, Ms Rebecca Redmond, Ms Claire Giffney, Dr James O'Byrne, Dr Robert O'Byrne, Dr Laoi Shakerdi, Prof Sally Ann Lynch, Prof Andrew Green, Children's Health Ireland at Crumlin, Dublin
- OP12 DRAGENv4.0 increases the accuracy and utility of the Genomics England Rare Disease Bioinformatics Pipeline applied in the NHS Genomic Medicine Service**  
Mr Gabriel Aldam, Dr Mark Doherty, Dr Liam Abrahams, Mr Kevin Savage, Dr Susan Walker, Dr Javier Lopez, Dr Eva Serra, Dr Catherine Whibley, Dr Rachael Mein, Dr Augusto Rendon, Dr Ellen Thomas, Dr Dalia Kasperaviciute, Dr Jamie Ellingford, Genomics England
- OP13 Genotype-phenotype correlations in IFT140-related retinal dystrophy in a cohort of Irish patients**  
Ms Anna Ridgeway, Dr Laura Finnegan, Dr Matthew Carrigan, Dr Laura Whelan, Dr Adrian Dockery, Ms Giuliana Silvestri, Prof David Keegan, Ms Emma Duignan, Dr Paul Kenna, Prof Jane Farrar, Trinity College Dublin
- 16:25 Tea & Coffee, Poster Viewing and Sponsor Visits**
- 16:50 Keynote address IV: What can zebrafish teach us about human genetics?**  
Prof David Lyons, University of Edinburgh
- 17:35 Honorary Membership Presentation**  
Prof Michael Gill, Trinity College Dublin
- 17:50 Presentation of Prizes followed by Wine & cake reception**
- 19:00 Meeting Closes**

## SUPPORTED BY:

