

**ABSTRACTS COLLECTION**

## MEETING ABSTRACTS

# Abstracts from the 54<sup>th</sup> European Society of Human Genetics (ESHG) Conference

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Virtual Conference

28–31 August 2021

The ESHG conference delivered the latest findings in the field of human genetics, both basic and applied.

Additional information about the event may be found on the conference website:

<https://2021.eshg.org/>

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**ABSTRACTS FROM THE 54<sup>TH</sup> EUROPEAN SOCIETY OF HUMAN GENETICS CONFERENCE: ORAL PRESENTATIONS***Plenary sessions*

PL1 Opening Plenary  
PL2 What's New? Highlight Session  
PL4 Mendel Award Lecture

*Concurrent symposia*

S01 Machine learning methods for prioritising genetic variants  
S02 Spatial omics  
S03 Transposons  
S04 Impact of GDPR on genomic data sharing  
S05 Endogenous and exogenous mutagenesis in cancer  
S06 Comparative genomics across species and populations  
S07 Mind the gap: Translating genomic advances into clinical care  
S08 Single cell genomics in cancer  
S09 Biobanks in under-represented populations  
S10 Gene-based therapy for inherited liver diseases  
S11 Prevention, detection, and therapy in cancer

S12 Functional annotation of genomic variation  
S13 Beauty of gametogenesis  
S14 Genome architecture  
S15 Cells competing cells - mosaicism and cancer  
S16 ESHG-ASHG Building Bridges: Global genetics towards a socially just practice  
S17 Biases in genetic studies: Estimation and impact  
S18 Overgrowth syndromes, from discovery to therapy  
S20 Counselling Over Various Informatic Devices: Lessons from Covid-19  
S21 Delivering the promise of RNA therapeutics  
S22 Integrated approaches for ciliopathies

*Educational sessions*

E01 New technologies  
E02 ESHG-Y: Human organoids as genetic disease models  
E03 Translational collaborations in hereditary cancer  
E04 Dealing with uncertainty in genomic medicine  
E05 Update on imprinting disorders  
E06 Pharmacogenomics in the clinic  
E08 Variant interpretation in the clinic  
E09 What's new in preimplantation genetic testing?

- E11 Polygenic risks and me
- E12 Bayesian methods applied in clinical settings
- E13 Mapping the human body at the cellular level
- E14 DNA methylation in Mendelian diseases
- E15 Selection and population structure in biobank scale data
- E16 Advances in Mendelian randomisation
- E17 Chromosomal instability across lifetime
- E18 Introduction to statistical analysis of genome-wide association studies (GWAS)
- E19 Precision medicine in underserved populations

*Concurrent sessions*

- C01 Developmental disorders & syndromes I
- C02 Cardiovascular disorders
- C03 Bioinformatics, machine learning and statistical methods
- C04 Unraveling the complexity of neuropsychiatric genetics
- C05 Reproduction is hot!
- C06 COVID-19 Genomics
- C07 Novel insights in inherited metabolic diseases
- C08 Skeletal and connective tissue disorders
- C09 Sensory disorders: multi-omics and long-read sequencing
- C10 Genome-wide Association Studies
- C11 New technologies and better diagnostics
- C12 Counselling, communication and service delivery
- C13 Cancer susceptibility: From mechanisms to clinic
- C14 Advances in neurogenetics: From diagnosis to treatment
- C15 Pleiotropic diseases: diagnosis and mechanisms
- C16 Monogenic neurodevelopmental disorders
- C17 Population genetics and genetic epidemiology
- C18 Functional genomics and transcriptomics
- C19 ELSI in genomics
- C20 From mechanisms to therapeutic insights in cancer
- C21 Clinical immunology and novel therapies of genetic diseases
- C22 Developmental disorders & syndromes II

- C23 Internal organs - Kidney, bowel, fat
- C24 Genome variation and architecture
- C25 Using genomics to personalise medicine
- C26 Late Breaking

**ABSTRACTS FROM THE 54<sup>TH</sup> EUROPEAN SOCIETY OF HUMAN GENETICS CONFERENCE: E-POSTERS**

- P01 Reproductive Genetics/Prenatal Genetics
- P02 Sensory Disorders (Eye, Ear, Pain)
- P03 Internal Organs & Endocrinology (Lung, Kidney, Liver, Gastrointestinal)
- P04 Skeletal, Connective Tissue, Ectodermal and Skin Disorders
- P05 Cardiovascular Disorders
- P06 Metabolic and Mitochondrial Disorders
- P07 Immunology and Hematopoietic System
- P08 Intellectual Disability
- P09 Neurogenetic and Psychiatric Disorders
- P10 Neuromuscular Disorders
- P11 Multiple Malformation/Anomalies Syndromes
- P12 Cancer Genetics
- P13 Genome Variation and Architecture
- P14 Cytogenetics
- P15 New Technologies and Approaches
- P16 Diagnostic Improvements and Quality Control
- P17 Bioinformatics, Machine Learning and Statistical Methods
- P18 Personalised Medicine and Pharmacogenomics
- P19 Population Genetics and Evolutionary Genetics
- P20 Functional Genomics and Epigenomics
- P21 New Treatments for Genetic Disorders
- P22 Genetic Counselling/Services/Education
- P23 Ethical, Legal and Psychosocial Aspects in Genetics
- P24 GWAS
- P25 COVID-19