

ABSTRACTS COLLECTION



MEETING ABSTRACTS

Abstracts from the 56th European Society of Human Genetics (ESHG) Conference

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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://2023.eshg.org/>

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ABSTRACTS FROM THE 56TH EUROPEAN SOCIETY OF HUMAN GENETICS CONFERENCE: ORAL PRESENTATIONS

Plenary Sessions

PL1 ESHG Award Lecture
 PL2 Opening Plenary
 PL3 What’s New? Highlight Session
 PL6 ELPAG Award Lecture

Concurrent Symposia

S01 Translational metabolism: From inborn errors of metabolism to multifactorial diseases
 S02 Sex differences in neurodevelopmental disorders
 S03 Application of GWAS in social sciences
 S04 Honoring diversity: Toward culturally competent genetic counseling
 S05 Oligogenics in rare disease
 S06 Treatment for interferonopathies
 S07 Understanding pre-cancer: Molecular analysis to clinical cancer prevention
 S08 Exploiting new approaches in GWAS
 S09 Data Altruism and future of data sharing in genomics
 S10 Somatic mutations in immune disorders

S11 Complete genomes
 S13 Leveraging genetic studies for drug discovery
 S14 Precision medicine in renal cancer
 S15 Placenta: The forgotten organ
 S16 High-throughput functional analysis of variants
 S17 Long reads for solving old problems
 S18 Co-creating research with participants
 S19 Addressing diversity in the genomic world
 S21 Gene-disease relationships
 S22 Diagnostics in prenatal setting - the present and the future
 S23 Beyond coding point mutations: New mutation types in cancer
 S24 Health Economics in Genomic Medicine

Educational Sessions

E01 New Technologies: Recent advances in sequencing technologies
 E02 Not only DNA editing: Gentler ways to tweak genes
 E03 Let’s get up to date on blood cancer genetics and genomics
 E04 Harnessing pleiotropy
 E05 Therapeutic Gene Editing in Europe: What will it take to make it happen?
 E06 Spatial transcriptomics/cell lineages
 E08 Should we be routinely returning results from genomic research?

E09 Epilepsy: Genetics & Treatment
 E10 Spinal Muscular Atrophy Treatments
 E11 Genetics Care for Trans and Gender Diverse Patients: From Medicalization to Empowerment
 E13 New mutations and evolution
 E14 3D genomes explained
 E16 PGS: How close is it to clinical applicability

Concurrent Sessions

C01 RNA and epigenetics in intellectual disability
 C02 New treatments and clinical approaches
 C03 Multiple congenital anomalies
 C04 Clinical genetic counseling
 C05 Internal medicine genetics
 C06 Inherited metabolic disorders
 C07 Evolutionary genetics & genetics of diverse populations
 C08 Reproductive genetics
 C09 Genome variation: from small to large
 C10 Neurogenetics: an update
 C11 Genetic epidemiology of molecular traits
 C12 Cancer genetic services
 C13 Developmental anomalies
 C14 Eye genetics
 C16 Prenatal genetics
 C17 Functional genomics
 C18 Neurodevelopmental disorders: new genes
 C19 Innovative methods in statistical genetics
 C20 High- and low-penetrance inherited cancer risk
 C21 Late Breaking Abstracts
 C22 Cardiovascular and muscular genomics
 C24 Intellectual disability: an update
 C25 Diagnostics: large cohorts and novel omics
 C26 GWAS
 C27 New technologies in cancer genetics
 C28 Innovative genomic services
 C29 Skin and skeletal disorders: New genes and pathways

ABSTRACTS FROM THE 56TH EUROPEAN SOCIETY OF HUMAN GENETICS CONFERENCE: E-POSTERS

P01 Reproductive Genetics
 P02 Prenatal Genetics
 P03 Sensory Disorders (Eye, Ear, Pain)
 P04 Internal Organs & Endocrinology (Lung, Kidney, Liver, Gastrointestinal)
 P05 Skeletal, Connective Tissue, Ectodermal and Skin Disorders
 P06 Cardiovascular Disorders
 P07 Metabolic and Mitochondrial Disorders

P08 Immunology and Hematopoietic System
 P09 Intellectual Disability
 P10 Neurogenetic and Psychiatric Disorders
 P11 Neuromuscular Disorders
 P12 Multiple Malformation/Anomalies Syndromes
 P13 Cancer Genetics
 P14 Genome Variation and Architecture
 P15 Cytogenetics
 P16 New Technologies and Approaches
 P17 Diagnostic Improvements and Quality Control
 P18 Bioinformatics, Machine Learning and Statistical Methods
 P19 Personalized Medicine and Pharmacogenomics
 P20 Population Genetics and Evolutionary Genetics
 P21 Functional Genomics and Epigenomics
 P22 New Treatments for Genetic Disorders
 P23 Genetic Counselling/Services/Education
 P24 Ethical, Legal and Psychosocial Aspects in Genetics
 P25 GWAS

ABSTRACTS FROM THE 56TH EUROPEAN SOCIETY OF HUMAN GENETICS CONFERENCE: HYBRID POSTERS

P01 Reproductive Genetics
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