ABSTRACTS COLLECTION

MEETING ABSTRACTS

Abstracts from the 55th 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://2022.eshg.org/

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ABSTRACTS FROM THE 55TH EUROPEAN SOCIETY OF HUMAN GENETICS CONFERENCE: ORAL PRESENTATIONS

Plenary sessions

PL1 Opening Plenary
PL2 What's New? Highlight Session
PL3 ELPAG Award Lecture
PL5 ESHG Award Lecture

Concurrent symposia

S01 Reproductive carrier screening in 2022
S02 Methods for admixed populations
S03 Personalized care in breast cancer patients
S04 How do we deliver genomics education for all?
S05 Chromothripsis and complex rearrangements
S06 Disorders of lysosomal biogenesis and autophagy
S07 Cell lineages and organoids
S09 Mutation signatures
S10 Left/Right patterning/heterotaxy
S11 Peopling the world: One archipelago at a time
S12 The genetics of omics and beyond
S13 Unstable heritable genomic variations and cancer development
S14 Predictive genetic counselling for neurodegenerative conditions
S15 Genetic architecture of the human face
S18 The importance of somatic variation
S19 Polygenic scores: from methods to applications
S20 Cross-cultural communication and counselling
S21 Translational genetics of bone
S23 Non-coding RNAs in biology and disease
S25 Multiomics for diagnostics
S26 Federation of genomic medicine databases

Educational sessions

E02 Genetics, 200 years after the births of Mendel and Galton
E04 ESHG-Y: Filling the gaps by publishing negative results in genetics
E05 Mosaicism
E06 Genetic discrimination: Surveying the ethico-legal landscape
E07 Progress of imputation
E09 Pharmacogenomics for personalized drug treatment
E11 New treatments for congenital disorders
E12 Inherited metabolic disorders with acute presentations
E14 Precision medicine in the digital health era

Concurrent sessions

C01 New genes in NDDs
C02 Hereditary cancer syndromes and risk assessment
C03 Cellular differentiation and regulation
C04 Unravelling causes and mechanisms of multiple congenital anomalies
C05 Population and Evolutionary Genetics
C06 Counselling, Education and Service Delivery
C07 Metabolic and mitochondrial diseases
C08 New findings for old NDD genes
C09 Cardiac and neuromuscular genetics
C10 Internal organs and Immunology
C11 New diagnostics and treatments
C12 GWAS
C13 Patient Views on Clinical Genetics
C14 Late Breaking Abstracts
C16 Prenatal Genetics
C17 Clinical impact of molecular tumor profiling
C18 Machine learning, bioinformatics and biostatistics
C19 New gene defects and pathways in syndromic conditions
C20 Non-coding genome variation in Mendelian diseases
C21 Different strategies to unravel the genetic basis of NDDs
C22 Innovation in genetic risk assessment and diagnostics
C24 Molecular mechanism in cancer
C25 Reproductive Genetics
C26 New genes in multiple congenital anomalies
C27 New approaches and large datasets to unravel human traits
C28 Novel neurogenetic disorders
C29 Eye Genetics

ABSTRACTS FROM THE 55TH 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
P26 COVID-19

ABSTRACTS FROM THE 55TH EUROPEAN SOCIETY OF HUMAN GENETICS CONFERENCE: HYBRID 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
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