DESCRIPTION

The European Journal of Medical Genetics (EJMG) is a peer-reviewed journal that publishes articles in English on various aspects of human and medical genetics, the genetics of experimental models and developmental anomalies.

Original clinical and experimental research articles, short clinical reports and review articles are welcome on topics such as:

Clinical genetics and genomics, including neurodevelopmental disorders and single or multiple malformations syndromes and dysmorphology; Inherited predisposition to cancer: rare inherited cancer predisposition syndromes and biology and pathophysiology of inherited cancers; Fetal medicine and prenatal diagnosis, including prenatal diagnosis, prenatal screening, fetal therapies and teratology; Diagnostic methods including sequencing studies (gene level), transcriptome studies, methylome and other Omics technologies, variant interpretation and functional validation; Bioinformatics and statistical methods in genetics; Personalized Medicine and therapies, including gene therapy and gene editing, non-genetic treatment of genetic disease, pharmacogenetics and genomics, theranostics and clinical trials; Molecular biology and pathophysiology of genetic and genomic diseases: biology of cell, pathways and systems, epigenetics and gene regulation, biology and pathophysiology of specific genes and non-coding DNA; Genetics and variants in populations: carrier screening, population genetics, evolutionary genetics, genetic epidemiology of diseases, genome variation and architecture in normal population; Formal genetics: Mendelian and non-Mendelian inheritance; Polygenic/ multifactorial disorders: association studies in large cohorts and risk evaluation in polygenic disorders; Genetics and society: ethical aspects in genetics, impact of genomic medicine on public and patients, legal and reglementary aspects in genetics and clinical research, psychosocial aspects, rare diseases and genetics; Teaching and training in genetics; Genetic counselling; Internet resources in Genetics.Please note: tumor genetics and studies focusing on multifactorial diseases are not in the scope of the Journal.

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