Molecular Syndromology

Genetics of Epilepsies

Guest Editors
Thomas Dorn, Zurich
Johannes R. Lemke, Leipzig

Next Generation Sequencing Era

Transitional Era

Channelopathy Era

Gene for familial eilepsia epilptic encephalopathy

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The 2016 edition of the International System for Human Cytogenomic Nomenclature (ISCN 2016) offers standard nomenclature that is used to describe any genomic rearrangement identified by techniques ranging from karyotyping to FISH, microarray, various region-specific assays, and DNA sequencing. Suggestions from the international cytogenetics community have been reviewed by the Standing Committee, an international group of experts, nominated by their peers.

This updated edition offers:

- many new examples, particularly for microarray and region-specific assays
- trackable changes in the main text compared to the previous edition for easier identification
- a nomenclature standard to facilitate the description of chromosome rearrangements characterized by DNA sequencing developed through collaboration between the Human Genome Variation Society (HGVS) and ISCN to accommodate the increased use of sequencing technologies in the characterization of chromosomal abnormalities

The ISCN 2016 is an indispensable reference volume for human cytogeneticists, molecular geneticists, technicians, and students for the interpretation and communication of human cytogenetic and molecular cytogenomic nomenclature.

After a long collaboration with Cytogenetic and Genome Research, ISCN is now again part of this leading journal on chromosome and genome research, combining day-to-day business with the latest findings. ISCN 2016 is available individually as a reprint of Cytogenetic and Genome Research Vol. 149, No. 1–2, 2016 ahead of the special issue. The special issue is part of every 2016 subscription and available to subscribers only. It will be published in print and electronically and will be released in fall 2016.
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Molecular Syndromology

Molecular Syndromology publishes Original Articles, Short Reports, Novel Insights from Clinical Practice (formerly Case Reports) and Reviews on the molecular basis of genetic syndromes, genotype-phenotype correlations, natural history, strategies in disease management and novel therapeutic approaches linked to molecular basis.

Publication sections in conventional issues

Original Articles: High-quality research reports on common and rare syndromes in humans aim to increase the clinical understanding through molecular insights. Articles dealing with animal model organisms are also considered. Original articles should be fully documented in a form that describes the research that is interesting, original experiments in current research. An unstructured Abstract (Maximum 200 words), Introduction, Materials and Methods, Results, and Discussion sections are required. Consideration for publication is based on originality, novelty, scientific soundness, and appropriate analysis. (Maximum 5,000 words of text, 80 references, plus tables/figures).

Short Reports: These papers must present new findings of sufficient importance and be of potential interest to a large fraction of the readership. They should follow the general arrangement of Original Articles and should represent complete, original studies. (Maximum 3,000 words of text, plus abstract, plus a maximum of 2 figures, 1 table and 20 references).

Novel Insights from Clinical Practice (formerly Case Reports): The journal only considers case reports that provide significant new insights into a syndromological problem. Submissions can be based around a single case or a number of similar cases. The most important aspect of the presentation is that it should provide a founded molecular insight on a recognized or entirely new genetic syndrome. The novel aspects of the case(s) may be in the phenotype and/or genotype, the presentation, and the investigation.

Single topic issues

The journal also publishes a series of themed issues. For these volumes we solicit the assistance of one or more expert investigators to act as Guest Editor(s) in an area that is particularly interesting and/or one in which there is a need for a thorough overview. The Guest Editors invite top researchers to contribute original research reports or reviews of a topic that is in their main area of interest. These manuscripts are peer-reviewed in the same way as papers received for conventional issues. Proposals clearly outlining the theme and nominating potential Guest Editor(s) are welcome.

Submission

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Molecular Syndromology publishes high-quality research articles, short reports and reviews on common and rare genetic syndromes, aiming to increase clinical understanding through molecular insights. Topics of particular interest are the molecular basis of genetic syndromes, genotype-phenotype correlation, natural history, strategies in disease management and novel therapeutic approaches based on molecular findings. Research on model systems is also welcome, especially when it is obviously relevant to human genetics. With high-quality reviews on current topics the journal aims to facilitate translation of research findings to a clinical setting while also stimulating further research on clinically relevant questions. The journal targets not only medical geneticists and basic biomedical researchers, but also clinicians dealing with genetic syndromes. With four Associate Editors from three continents and a broad international Editorial Board the journal welcomes submissions covering the latest research from around the world.

Selected contributions
- SHOX Haploinsufficiency as a Cause of Syndromic and Nonsyndromic Short Stature: Fukami, M.; Seki, A. (Tokyo); Ogata, T. (Tokyo/Hamamatsu)
- Differing Microdeletion Sizes and Breakpoints in Chromosome 7q11.23 in Williams-Beuren Syndrome Detected by Chromosomal Microarray Analysis: Li, L.; Huang, L.; Luo, Y.; Huang, X.; Lin, S.; Fang, Q. (Guangzhou)
- Bardet-Biedl Syndrome: Suspitsin, E.N.; Imyanitov, E.N. (St. Petersburg)
- Cytogenomic Aberrations in Congenital Cardiovascular Malformations: Azamian, M.; Lalani, S.R. (Houston, Tex.)
- Clinical and Genetic Heterogeneity of the 1.5q13.3 Microdeletion Syndrome: Hassfurther, A. (Freiburg); Komini, E. (Willingen-Schwenningen); Fischer, J.; Leipoldt, M. (Freiburg)
- RASopathies: Presentation at the Genome, Interactome, and Phenome Levels: Pevec, U.; Rozman, N.; Gorsek, B.; Kunej, T. (Domzale)

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The Right Therapy for Neurological Disorders
From Randomized Trials to Clinical Practice

Editors
Ettore Beghi
Giancarlo Logroscino

Most neurological disorders are chronic and aging-related. With the increase of life expectancy their incidence and prevalence will grow in the decades to come, which in turn will increase the load on medical and social systems worldwide. There is thus a desperate need for successful preventive and therapeutic measures based on randomized clinical trials (RCTs) conducted by independent organizations. This book provides a compendium relating most of the principles of reliable RTCs to specific neurological diseases. Contributed by specialized neurologists, the articles touch on important aspects of RCTs with a clear critical approach, highlighting their limitations as well as giving recommendations for their planning and conducting to address the variable genotypic and phenotypic aspects of neurological conditions. Consideration is also given to combining the clinical impact of the study results with patients’ values and the interests of pharmaceutical companies. Neurologists involved in clinical trials will certainly benefit from this book, which should become a basic text for all neurological courses dealing with evidence-based neurology.

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• Current Issues in Randomized Clinical Trials of Neurdegenerative Disorders at Enrolment and Reporting: Diagnosis, Recruitment, Representativeness of Patients, Ethnicity, and Quality of Reporting: Logroscino, G.; Capozzo, R.; Tortelli, R.; Marin, B.
• How to Distinguish between Statistically Significant Results and Clinically Relevant Results: Bennett, D.A.

• Modeling and Prediction in Neurological Disorders: The Biostatistical Perspective: Copetti, M.; Fontana, A.; Pellegrini, F.
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• Randomized Trials in Developing Countries: Different Priorities and Study Design?: Marin, B.; Agbota, G.C.; Preux, P.-M.; Boumediene, F.
• The Right Therapy for Neurological Disorders: From Randomized Trials to Clinical Practice – Patients versus Investigator Expectations and Needs: Bruijn, L.L.; Kolb, S.
• General Overview, Conclusions, and Future Directions: Beghi, E.; Logroscino, G.
European Neurology publishes original papers, reviews and letters to the editor. Papers presented in this journal cover clinical aspects of diseases of the nervous system and muscles, as well as their neuropathological, biochemical, and electrophysiological basis. New diagnostic probes, pharmacological and surgical treatments are evaluated from clinical evidence and basic investigative studies. The journal also features original works and reviews on the history of neurology.
During the last decades, Cytogenetic and Genome Research has been the leading forum for original reports and reviews in human and animal cytogenetics, including molecular, clinical and comparative cytogenetics. In recent years, most of its papers have centered on genome research, including gene cloning and sequencing, gene mapping, gene regulation and expression, cancer genetics, comparative genetics, gene linkage and related areas. The journal also publishes key papers on chromosome aberrations in somatic, meiotic and malignant cells. Its scope has expanded to include studies on invertebrate and plant cytogenetics and genomics. Also featured are recent international reports on human and animal chromosome nomenclature and reports on gene mapping in domestic animals. In addition to regular issues, the journal has been publishing since 2002 a series of topical issues on a broad variety of themes from cytogenetic and genome research.

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Selected contributions
• 22q11.21 Deletion Syndromes: A Review of Proximal, Central, and Distal Deletions and Their Associated Features: Burnside, R.D. (Research Triangle Park, N.C.)
• Transposons, Genome Size, and Evolutionary Insights in Animals: Canapa, A.; Barucca, M.; Biscotti, M.A.; Forconi, M.; Olmo, E. (Ancona)
• Chromosomal Abnormalities in Embryonic and Somatic Stem Cells: Rebuffini, P. (Pavia); Zuccotti, M. (Parma); Redi, C.A.; Garagna, S. (Pavia)
• Condensin in Chromatid Cohesion and Segregation: Uchiyama, S. (Osaka/Okazaki); Fukui, K. (Osaka)
• Small Supernumerary Marker Chromosomes in Human Infertility: Armanet, N.; Tosca, L.; Brisset, S. (Clamart/Le Kremlin-Bicêtre); Liehr, T. (Jena); Tachdjian, G. (Clamart/Le Kremlin-Bicêtre)
• TET Family of Dioxygenases: Crucial Roles and Underlying Mechanisms: Li, D.; Guo, B.; Wu, H.; Tan, L.; Lu, Q. (Changsha)
• Polyploidy in Animals: Effects of Gene Expression on Sex Determination, Evolution and Ecology: Wertheim, B.; Beukeboom, L.W.; van de Zande, L. (Groningen)
• Parallel Universes for Models of X Chromosome Dosage Compensation in Drosophila: A Review: Birchler, J.A. (Columbia, Mo.)
Genetics of Epilepsies

During the past decade, the genetic basis of epilepsy disorders has been substantially elucidated. New genome-wide technologies have enabled screening for small copy number variants as well as for alterations of the DNA base sequence. This technical progress means that clinicians no longer have to hypothesize about defined genetic causes and speculate about variations in a restricted number of genes or diverse chromosomal abnormalities. Furthermore, a lot of lessons have been learned about the genetic architecture of the epilepsies and their pathophysiological principles. However, some important open questions remain.

This special issue of *Molecular Syndromology* addresses diverse and frequent genetic aspects of epileptogenesis without being exhaustive. It makes it clear that genetic diagnostics enable us to clarify the pathophysiological background. This is a prerequisite for investigating novel therapeutic prospects that will lead to precise approaches with the potential to positively impact the fate of patients.

*Cover illustration*  
Title page shows MRI of patients with different types of cortical malformations that are often associated with developmental delay and severe epilepsy (from Parrini et al., pp. 220–233) as well as a timeline illustrating the different eras of gene discovery and its dynamics in the past 20 years (from Helbig and Abou Tayoun pp. 172–181).