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EDITORIAL

What is the purpose of launching the *World Journal of Medical Genetics*?

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Abstract

Congratulations to the publisher, members of the editorial board of the journal, all the authors and readers for launching the *World Journal of Medical Genetics* (*WJMG*) as a new member of the World series journal family! Following the completion of the Human Genome Project, medical genetic research has seen spectacular progress over the last decade. The number of genes that have been linked to Mendelian human traits has grown exponentially and currently this process is peaking with the access to robust genome-wide sequencing power. The genomics revolution is also seen for elucidation of rare and common DNA variants that increase risk for common disorders. Given this fast progress, there is an increasing need for making the results of genetics and genomics studies rapidly and freely available to the larger community. Thus, the decision for inaugurating this new journal is a timely one. The *WJMG* is a peer-reviewed, open-access periodical centered in all aspects of medical genetics research, with multidisciplinary coverage: from human phenotype to genetic and genomic mutations and variations to the study of pathological mechanisms. If you want to share new results of your research with a link to medical genetics with your peers, you will find the *WJMG* a good media to publish your papers!

Key words: Medical genetics; Genomics; Biomedical sciences; Peer-reviewed; Open-access; Journal

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Figure 1 Editor-in-Chief of the *World Journal of Medical Genetics*. Hans van Bokhoven, Professor, PhD, Department of Human Genetics, Molecular Neurogenetics Unit, Nijmegen Centre for Molecular Life Sciences and Donders Institute for Brain, Cognition and Behaviour, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands.

INTRODUCTION

I am Hans van Bokhoven, a full professor from the Radboud University Nijmegen Medical Centre, Nijmegen, the Netherlands (Figure 1) and the Editor-in-Chief of

the *World Journal of Medical Genetics* (*World J Med Genet*, *WJMG*, online ISSN 2220-3184, DOI: 10.5496). It is my great honor to introduce the *WJMG* as a new forum for exchanging new results and sharing new hypotheses relevant to clinical genetics and to fundamental and diagnostic molecular genetics and genomics research. Congratulations to the publisher, members of editorial board of the journal, all the authors and readers for this memorable event!

I am very pleased to announce that the first issue of the *WJMG*, whose preparation was initiated on December 16, 2010, is officially published on December 27, 2011. The *WJMG* Editorial Board has now been established and consists of 103 distinguished experts from 28 countries. What is the purpose of launching the *WJMG* and what will the scope of the journal be? To start with the last question, the *WJMG* will cover new developments in all areas of medical genetics: research on the causes, the diagnosis and the management of hereditary disorders in man. Medical genetics has taken a prominent position in current medical practice and has branched into many medical specialties, such as neurology, endocrinology, oncology, psychiatry, ophthalmology, dermatology and many others.

Advances in elucidation of the etiology of the broad spectrum of disorders have been propelled by the scientific landmarks set by the Human Genome Project and various other genome projects[1,2]. In addition, technological improvements, such as microarray platforms and next generation sequencing machines, have made genome wide analyses feasible for individual laboratories. As a consequence, there are currently over 3300 disorders listed in OMIM (November 2011)[3] for which the molecular basis is known and new genes are added each day. However, there are at least an equally large number of human Mendelian disorders that are still to be resolved. In addition, genetic risk factors for common disorders are only beginning to be resolved. Thus, knowledge about the genetic contribution to human disease is still largely unexplored and the knowledge of multigenic diseases is also in progress. The high pace at which we can expect to find new genotype-phenotype connections warrants the introduction of the *WJMG* as a novel open access resource for communicating such findings to the medical genetics community. The *WJMG* will solicit contributions from all subdisciplines, which include clinical genetics, metabolic and biochemical genetics, cytogenetics, molecular genetics, DNA diagnostics, mitochondrial genetics and genetic counseling.

SCOPE

The *WJMG* aims to rapidly report the most recent results in medical diagnostics, therapeutic techniques and equipment, clinical medical research, clinical and experimental techniques and methodology. Its purpose is to provide a platform to facilitate the integration of clinical and laboratory disciplines to highlight genotype-phenotype associations at a qualitative high level, which will help to improve diagnostic accuracy and medical care, and in the longer run, therapeutic intervention. The journal publishes original articles and reviews on the following topics: (1) Laboratory research, including but not limited to techniques in DNA/RNA sequencing, whole-genome linkage analyses and association studies, copy number variation profiling, epigenetic modifications in health and disease, elucidation of molecular and cellular pathways affected by gene mutations, determination of transcription factor binding sites, protein-protein interactions, preparation and transformation of induced pluripotent stem cells, animal models of human hereditary disorders and bioinformatics; and (2) Clinical genetics research on etiology, epidemiology, pathogenesis, morphology and function, signs and symptoms.

CONTENTS OF PEER REVIEW

In order to guarantee the quality of articles published in the journal, *WJMG* usually invites three experts to comment on the submitted papers. The contents of peer review include: (1) whether the contents of the manuscript are of great importance and novelty; (2) whether the experiment is complete and described clearly; (3) whether the discussion and conclusion are justified; (4) whether the citations of references are necessary and reasonable; and (5) whether the presentation and use of tables and figures are correct and complete.

COLUMNS

The columns in the issues of the *WJMG* include: (1) Editorial: to introduce and comment on the substantial advance and its importance in the fast-developing areas; (2) Frontier: to review the most representative achievements and comment on the current research status in the important fields and propose directions for the future research; (3) Topic Highlight: this column consists of three formats, including (A) 10 invited review articles on a hot topic, (B) a commentary on common issues of this hot topic, and (C) a commentary on the 10 individual articles; (4) Observation: to update the development of old and new questions, highlight unsolved problems and provide strategies on how to solve the questions; (5) Guidelines for Clinical Practice: to provide guidelines for clinical diagnosis and treatment; (6) Review: to systematically review the most representative progress and unsolved problems in the major scientific disciplines, comment on the current research status and make suggestions on future work; (7) Original Articles: to originally report the innovative and valuable findings in medical genetics; (8) Brief Articles: to briefly report the novel and innovative findings in medical genetics; (9) Case Report: to report a rare or typical case; (10) Letters to the Editor: to discuss and make reply to the contributions published in the *WJMG*, or to introduce and comment on a controversial issue of general interest; (11) Book Reviews: to introduce

and comment on quality monographs of medical genetics; and (12) Guidelines: to introduce consensuses and guidelines reached by international and national academic authorities worldwide on the research in medical genetics.

So, if you want to share exciting novel results of your clinical, diagnostic or fundamental genetic research or your vision of new developments in the field of medical genetics research with your peers, the *WJMG* is a place you can feel at home. The staff and editorial board look forward to taking the *WJMG* to the top.

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