

# Human Heredity

## Genetic Diversity in European Populations: Evolutionary Evidence and Medical Implications

Guest Editors

**Alicia Sanchez-Mazas**, Geneva

**Guido Barbujani**, Ferrara

Summarizing the latest advances in the fight against the obesity epidemic



# Genes and the Environment in Obesity

Editors  
**John A. Dawson**  
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## Genes and the Environment in Obesity

Editors: Dawson, J.A.; Allison, D.B. (Birmingham, Ala.)  
IV + 170 p., 32 fig., 3 in color, 31 tab., 2013  
CHF 52.– / EUR 43.– / USD 61.00 (soft cover)  
CHF 62.– / EUR 52.– / USD 73.00 (online)  
Online version for institutional purchase  
Prices subject to change  
EUR price for Germany, USD price for USA and Latin America only  
ISBN 978-3-318-02477-7  
e-ISBN 978-3-318-02478-4

Special Topic Issue

**Human Heredity**  
**Vol. 75, No. 2–4 (2013)**

Included in subscription  
Listed in MEDLINE/PubMed, Current Contents/  
Life Sciences, Excerpta Medica, Biological  
Abstracts, Hum-Molgen



This publication pulls together work from more than a dozen leading obesity, energetics, and nutrition researchers working on genetic and genomic aspects of obesity. Their contributions present the latest findings from randomized controlled trials and other ongoing investigations, and direct our focus toward future genetics and epigenetic targets in our struggle with the obesity epidemic in the USA and across the globe. The topics considered range from ancestral admixture to assortative mating, from functional analysis to genome-wide association studies, and from satiety to brain anatomy. This compilation of reviews and methodological developments will help the reader to achieve a broader appreciation of the complexities of this field as well as a deeper understanding of its latest advances and important next steps.

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Continued in 1965 by M. Hauge (1965–1968)  
Changed in 1969 to 'Human Heredity'  
Continued in 1969 by M. Hauge and L. Beckman (1969–1983)  
Continued in 1984 by L. Beckman (1984–1990)  
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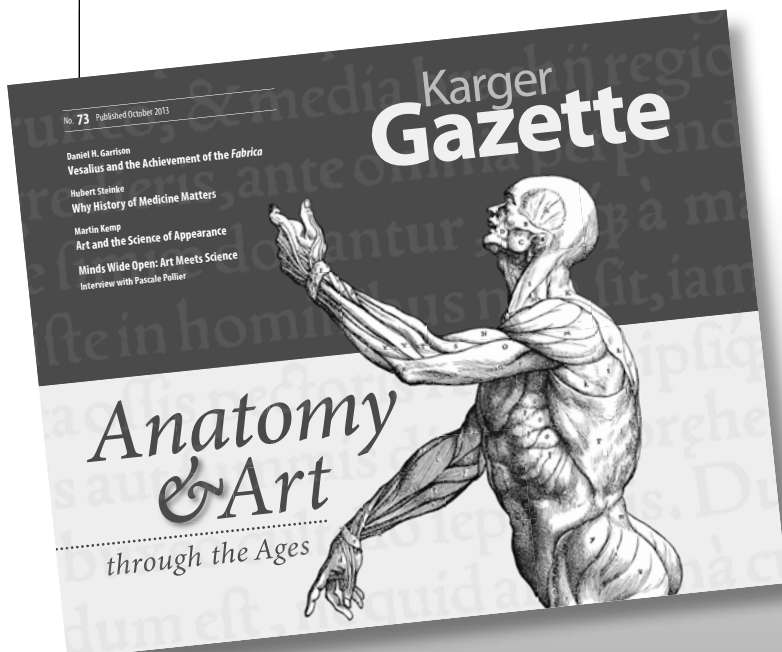
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**ISSN Print Edition:** 0001-5652  
**ISSN Online Edition:** 1423-0062

**Journal Homepage:** [www.karger.com/hhe](http://www.karger.com/hhe)

**Publication Data:** *Human Heredity* is published 8 times a year. Volumes 77, 78, each with 4 issues, appear in 2014.

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Microbiome-genome interactions and human disease: an introduction and latest methodological developments

*Knut Rudi, Ås, Norway*

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*Martin Posch, Vienna, Austria*

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*Christoph Bock, Vienna, Austria*

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Clinical background

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QC in OMICS studies

*Bertram Müller-Myhsok, Munich, Germany*

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Study designs for predictive biomarkers

*Andreas Ziegler, Lübeck, Germany*

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Pharmacogenomics: past, present and future

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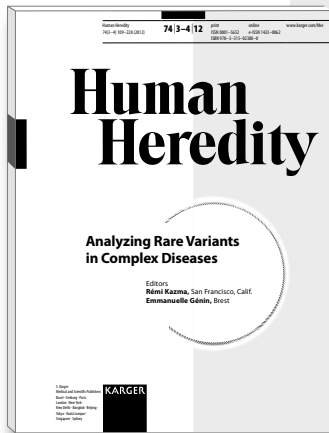
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- Incorporating Prior Biologic Information for High-Dimensional Rare Variant Association Studies: **Quintana, M.A.; Schumacher, F.R.; Casey, G.; Bernstein, J.L.; Li, L.; Conti, D.V.**
- Imputation of Rare Variants in Next-Generation Association Studies: **Asimit, J.L.; Zeggini, E.**
- Does Accounting for Gene-Environment Interactions Help Uncover Association between Rare Variants and Complex Diseases?: **Kazma, R.; Cardin, N.J.; Witte, J.S.**
- Rare Variants in Complex Traits: Novel Identification Strategies and the Role of de novo Mutations: **Jouan, L.; Gauthier, J.; Dion, P.A.; Rouleau, G.A.**
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## New discoveries and their genome-wide application to disease-oriented research

# Medical Epigenetics



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The journal's aim is to increase the distribution of scientific knowledge in the new discipline of medical epigenetics. Epigenetics, a rapidly emerging scientific discipline, seeks to define how the genome is regulated to produce distinct normal and diseased phenotypes. Epigenomics, the genome-wide application of epigenetic techniques, is at the core of systems biology extending the power of genomics, proteomics, and other high throughput techniques for the analyses of complex phenotypes.

The journal *Medical Epigenetics* seeks to catalyze discoveries and genome-wide applications in the areas of gene regulation, chromatin dynamics, and epigenetic inheritance to disease-oriented research. Articles will be focused on disseminating scientific advances and applications of this field to mapping DNA methylation, histone modifications, chromatin accessibility and small RNA transcripts in cells, tissues and organ systems frequently involved in human disease. This journal is also committed to support development, standardization and sharing of protocols, reagents and analytical tools to enable the research community to utilize, integrate and expand upon this new exciting biomedical discipline.

### **Medical Epigenetics**

2014: Vol. 2 with 4 issues

Language: English

e-ISSN: 1664-5561 (Online)

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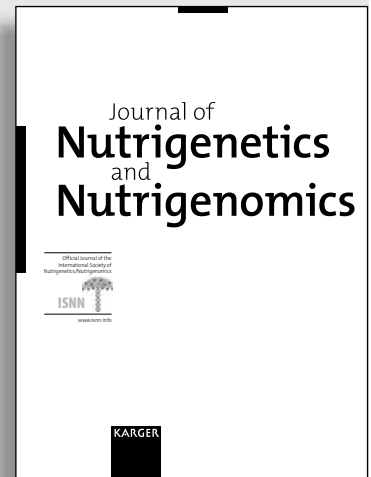
# Journal of Nutrigenetics and Nutrigenomics

Official Journal of the  
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**Journal of Nutrigenetics and  
Nutrigenomics**

2014: Volume 7

6 issues per volume

Language: English

ISSN 1661-6499 (print)

ISSN 1661-6758 (online)

Listed in *MEDLINE*

### Selected contributions

- The Emerging Role of MicroRNAs in the Regulation of Gene Expression by Nutrients: **García-Segura, L.; Pérez-Andrade, M.; Miranda-Ríos, J.** (México)
- Associations between Polymorphisms in Genes Involved in Fatty Acid Metabolism and Dietary Fat Intakes: **Bouchard-Mercier, A.; Paradis, A.-M.; Pérusse, L.; Vohl, M.-C.** (Quebec, Que.)
- Identification of Metabolic Biomarkers for Personalized Nutrition: **Kang, J.X.** (Boston, Mass.)
- Rat Chromosome 8 Confers Protection against Dyslipidemia Caused by a High-Fat/Low-Carbohydrate Diet: **Solberg Woods, L.C.** (Milwaukee, Wisc.); **Woods, B.C.** (Whitewater, Wisc.); **Leitschuh, C.M.; Laurie, S.J.** (Beloit, Wisc.); **Jacob, H.J.** (Milwaukee, Wisc.)
- Vitamin E Alters Inflammatory Gene Expression in Alcoholic Chronic Pancreatitis: **Monteiro, T.H.; Siqueira Silva, C.; Cordeiro Simões Ambrosio, L.M.; Zucoloto, S.; Vannucchi, H.** (Ribeirão Preto)
- Copy Number Polymorphism of the Salivary Amylase Gene: Implications in Human Nutrition Research: **Santos, J.L.** (Santiago); **Saus, E.** (Barcelona); **Smalley, S.V.; Cataldo, L.R.; Alberti, G.** (Santiago); **Parada, J.** (Valdivia); **Gratacòs, M.; Estivill, X.** (Barcelona)
- Cannabinoid Type 1 Receptor Gene Polymorphism and Macronutrient Intake: **Caruso, M.G.** (Castellana Grotte); **Gazzerro, P.** (Fisciano); **Notarnicola, M.; Cisternino, A.M.; Guerra, V.; Misciagna, G.** (Castellana Grotte); **Laezza, C.** (Naples); **Bifulco, M.** (Fisciano)
- The Influence of Vitamin A Supplementation on Foxp3 and TGF- $\beta$  Gene Expression in Atherosclerotic Patients: **Mottaghi, A.; Salehi, E.; Keshvarz, A.; Sezavar, H.; Saboor-Yaraghi, A.-A.** (Tehran)

The emerging field of nutrigenetics and nutrigenomics is rapidly gaining importance, and this new international journal has been established to meet the needs of the investigators for a high-quality platform for their research. Endorsed by the recently founded 'International Society of Nutrigenetics/Nutrigenomics' (ISNN), the *Journal of Nutrigenetics and Nutrigenomics* welcomes contributions not only investigating the role of genetic variation in response to diet and that of nutrients in the regulation of gene expression, but is also open for articles covering all aspects of gene-environment interactions in the determination of health and disease. Original papers and reviews cover the genetic basis for the variable responses to diet and lifestyle factors in chronic conditions (e.g. cardiovascular disease, obesity, diabetes, cancer), methods to assess gene-environment interactions and other related relevant topics, with research drawing from both human and animal studies.

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# Human Heredity

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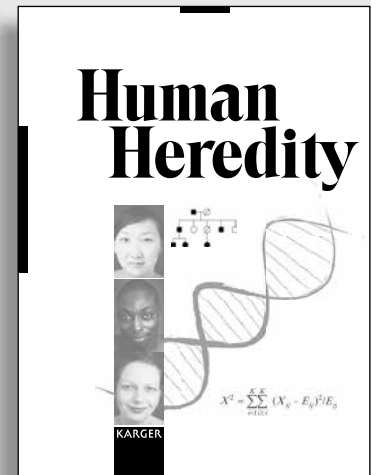
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**Human Heredity**  
2014: Volumes 77, 78  
4 issues per volume  
Language: English  
ISSN 0001-5652 (print)  
ISSN 1423-0062 (online)

Listed in bibliographic services, including  
*Current Contents®/Life Sciences*, *MEDLINE*,  
*Biological Abstracts*, *EMBASE/Excerpta Medica*

## Selected contributions

- Performance of Genotype Imputations Using Data from the 1000 Genomes Project: **Sung, Y.J.**; **Wang, L.** (St. Louis, Mo.); **Rankinen, T.**; **Bouchard, C.** (Baton Rouge, La.); **Rao, D.C.** (St. Louis, Mo.)
- Cumulative Meta-Analysis for Genetic Association: When Is a New Study Worthwhile? **Rotondi, M.A.**; **Bull, S.B.** (Toronto, Ont.)
- Regression Modeling of Allele Frequencies and Testing Hardy Weinberg Equilibrium: **Schaid, D.J.**; **Sinnwell, J.P.**; **Jenkins, G.D.** (Rochester, Minn.)
- Joint Analysis for Integrating Two Related Studies of Different Data Types and Different Study Designs Using Hierarchical Modeling Approaches: **Li, R.** (Los Angeles, Calif./Cambridge, Mass.); **Conti, D.V.** (Los Angeles, Calif.); **Diaz-Sanchez, D.** (Chapel Hill, N.C.); **Gilliland, F.**; **Thomas, D.C.** (Los Angeles, Calif.)
- A Novel Kernel for Correcting Size Bias in the Logistic Kernel Machine Test with an Application to Rheumatoid Arthritis: **Freytag, S.**; **Bickeböllner, H.** (Göttingen); **Amos, C.I.** (Lebanon, N.H.); **Kneib, T.** (Göttingen); **Schlather, M.** (Mannheim)
- Challenging False Discovery Rate: A Partition Test Based on p Values in Human Case-Control Association Studies: **Ott, J.** (Beijing); **Liu, Z.** (Chicago, Ill); **Shen, Y.** (Boston, Mass.)
- Interface Between Pharmacotherapy and Genes in Human Obesity: **O'Connor, A.**; **Swick, A.G.** (Kannapolis, N.C.)
- Epigenetics and Obesity: A Relationship Waiting to be Explained: **Symonds M.E.**; **Budge, H.** (Nottingham); **Frazier-Wood, A.C.** (Houston, Tex.)

Gathering original research reports and short communications from all over the world, *Human Heredity* is devoted to methodological and applied research on the genetics of human populations, association and linkage analysis, genetic mechanisms of disease, and new methods for statistical genetics, for example, analysis of rare variants and results from next generation sequencing. The value of this information to many branches of medicine is shown by the number of citations the journal receives in fields ranging from immunology and hematology to epidemiology and public health planning, and the fact that at least 50% of all *Human Heredity* papers are still cited more than 8 years after publication (according to ISI Journal Citation Reports). Special issues on methodological topics (such as analysis of rare variants in 2012) or reviews of advances in a particular field (the genetics of obesity in 2013) are published every year. Renowned experts in the field are invited to contribute to these special issues.

# Human Heredit

## Genetic Diversity in European Populations: Evolutionary Evidence and Medical Implications

Although it is the continent with the lowest genomic diversity, Europe embraces a significant degree of variation, shaped by complex demographic events and locus-specific selective factors linked to environmental conditions. The European genetic diversity observed today has also important clinical and epidemiological implications. Different questions related to these topics are addressed in this special topic issue of eight excellent papers written by specialized European teams. The papers document the remarkable progress achieved in the last years in exploring human genomic diversity, both in past and present populations, in reconstructing complex scenarios of European peopling history using sophisticated data analyses and computer simulations, in identifying signatures of adaptive selection in milk digestion- and immune-related genes, and in applying this knowledge to crucial health issues, from tissue transplantation to disease associations.

*Genetic Diversity in European Populations: Evolutionary Evidence and Medical Implications* is of special interest to researchers and students in biological and medical sciences and clinicians, who wish to update their knowledge in the field of European genetic diversity for basic research and medical applications.