

Journal of  
*Personalized  
Medicine*

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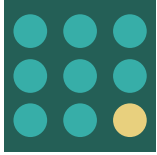
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# Section

Omics Informatics



### Editor-in-Chief

Prof. Dr. Salvatore Scacco  
Department of Basic Medical  
Sciences, Neurosciences and  
Sensory Organs, University of  
Bari "Aldo Moro", Italy

### Message from the Section Editor-in-Chief

The Section "Omics/Informatics" section of the Journal of Personalized Medicine is open to high-quality research and review articles, as well as short communications, case reports, and editorials on all aspects of omics sciences (from gene to the clinic) applied to personalized medicine. Of particular interest is original omics research, including multi-omics, genomics, proteomics, epigenomics, metabolomics, lipidomics, peptidomics, metagenomics, microbiomics, pharmacogenomics, toxicogenomics, etc., with a focus on disease etiology, prevention, diagnosis, treatment and therapy, prognosis, and monitoring.

Topics of interest with a view toward personalized medicine include (but are not limited to) the following:

- Capacity building for developing world omics;
- Applications of omics for personalized medicine (e.g., pharmacogenomics) and public health practice (e.g., vaccinomics);
- Artificial Intelligence applied to omics diagnostics and/or therapeutics in personalized medicine;
- Data standards and sharing;
- Social, legal, and ethics analysis of omics technologies and post-genomics life science innovations;
- Bioinformatics, computational biology, and biomedical informatics;
- Data standards, meta-data, data sharing, databases, biobanks, and cloud computing;
- Methodological, statistical, and algorithmic developments;

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## Selected Papers

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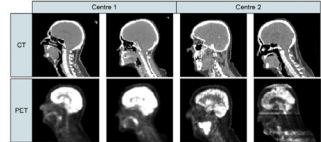
DOI:10.3390/jpm11090842

### Making Radiomics More Reproducible across Scanner and Imaging Protocol Variations: A Review of Harmonization Methods

Authors: Shruti Atul Mali, Abdalla Ibrahim, Henry C. Woodruff, Vincent Andrearczyk, Henning Müller, Sergey Primakov, Zohaib Salahuddin, Avishek Chatterjee and Philippe Lambin



Abstract: Radiomics converts medical images into mineable data via a high-throughput extraction of quantitative features used for clinical decision support. However, these radiomic features are susceptible to variation across scanners, acquisition protocols, and reconstruction settings. Various investigations have assessed the reproducibility and validation of radiomic features across these discrepancies. In this narrative review, we combine systematic keyword searches with prior domain knowledge to discuss various harmonization solutions to make the radiomic features more reproducible across various scanners and protocol settings. Different harmonization solutions are discussed and divided into two main categories: image domain and feature domain. The image domain category comprises methods such as the standardization of image acquisition, post-processing of raw sensor-level image data, data augmentation techniques, and style transfer...



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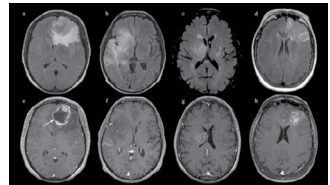
DOI:10.3390/jpm11040290

### Deep Learning Can Differentiate IDH-Mutant from IDH-Wild GBM

Authors: Luca Pasquini, Antonio Napolitano, Emanuela Tagliente, Francesco Dellepiane, Martina Lucignani, Antonello Vidiri, Giulio Ronazzi, Antonella Stoppacciaro, Giulia Moltoni, Matteo Nicolai, Andrea Romano, Alberto Di Napoli and Alessandro Bozzao



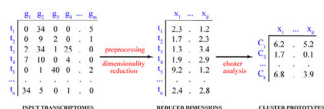
Abstract: Isocitrate dehydrogenase (IDH) mutant and wildtype glioblastoma multiforme (GBM) often show overlapping features on magnetic resonance imaging (MRI), representing a diagnostic challenge. Deep learning showed promising results for IDH identification in mixed low/high grade glioma populations; however, a GBM-specific model is still lacking in the literature. Our aim was to develop a GBM-tailored deep-learning model for IDH prediction by applying convoluted neural networks (CNN) on multiparametric MRI. We selected 100 adult patients with pathologically demonstrated WHO grade IV gliomas and IDH testing. MRI sequences included: MPRAGE, T1, T2, FLAIR, rCBV and ADC. The model consisted of a 4-block 2D CNN, applied to each MRI sequence. Probability of IDH mutation was obtained from the last dense layer of a softmax activation function. Model performance was evaluated in the test cohort considering categorical cross-entropy loss (CCEL) and accuracy. Calculated performance was: rCBV (accuracy 83%, CCEL 0.64), T1 (accuracy 77%, CCEL 1.4), FLAIR (accuracy 77%, CCEL 1.98), T2 (accuracy 67%, CCEL 2.41), MPRAGE (accuracy 66%, CCEL 2.55). Lower performance was achieved on ADC maps. We present a GBM-specific deep-learning model for IDH mutation prediction, with a maximal accuracy of 83% on rCBV maps. Highest predictivity achieved on perfusion images possibly reflects the known link between IDH and neoangiogenesis through the hypoxia inducible factor.



## Multi-Objective Genetic Algorithm for Cluster Analysis of Single-Cell Transcriptomes

Authors: Konghao Zhao, Jason M. Grayson and Natalia Khuri

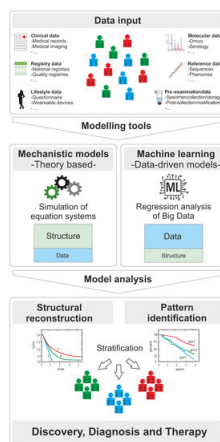
**Abstract:** Cells are the basic building blocks of human organisms, and the identification of their types and states in transcriptomic data is an important and challenging task. Many of the existing approaches to cell-type prediction are based on clustering methods that optimize only one criterion. In this paper, a multi-objective Genetic Algorithm for cluster analysis is proposed, implemented, and systematically validated on 48 experimental and 60 synthetic datasets. The results demonstrate that the performance and the accuracy of the proposed algorithm are reproducible, stable, and better than those of single-objective clustering methods. Computational run times of multi-objective clustering of large datasets were studied and used in supervised machine learning to accurately predict the execution times of clustering of new single-cell transcriptomes.



## Pharmacogenetics of Direct Oral Anticoagulants: A Systematic Review

Authors: Johanna Raymond, Laurent Imbert, Thibault Cousin, Thomas Dufлот, Rémi Varin, Julien Wils and Fabien Lamoureux

**Abstract:** Dabigatran, rivaroxaban, apixaban, edoxaban, and betrixaban are direct oral anticoagulants (DOACs). Their inter-individual variability in pharmacodynamics and pharmacokinetics (transport and metabolism) is high, and could result from genetic polymorphisms. As recommended by the French Network of Pharmacogenetics (RNPgX), the management of some treatments in cardiovascular diseases (as antiplatelet agents, oral vitamin K antagonists, and statins) can rely on genetic testing in order to improve healthcare by reducing therapeutic resistance or toxicity. This paper is a review of association studies between single nucleotide polymorphisms (SNPs) and systemic exposure variation of DOACs. Most of the results presented here have a lot to do with some SNPs of *CES1* (rs2244613, rs8192935, and rs71647871) and *ABCB1* (rs1128503, rs2032582, rs1045642, and rs4148738) genes, and dabigatran, rivaroxaban, and apixaban. Regarding edoxaban and betrixaban, as well as SNPs in the *CYP3A4* and *CYP3A5* genes, literature is scarce, and further studies are needed.



## Special Issues Open for Submission

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### Mathematical Models of Personalized Medicine

Guest Editors: Prof. Dr. Paolo Castorina and Dr. Alberto D'Onofrio  
Deadline: 20 February 2024



### Selected Papers from the pHealth 2022 Conference, Oslo, Norway, 8–10 November 2022

Guest Editors: Prof. Dr. Bernd Blobel, Dr. Mauro Giacomini and Dr. Bian Yang  
Deadline: 29 February 2024



### The Development of Radiomics in Personalized Diagnosis

Guest Editors: Dr. Giulia A. Zamboni and Dr. Sofia Gourtsoyianni  
Deadline: 10 March 2024



### Recent Medical Imaging Developments in the Era of Precision Medicine

Guest Editors: Dr. Nicolò Brandi and Dr. Matteo Renzulli  
Deadline: 15 April 2024



### Bioinformatics and Big Data Challenges in Personalized Medicine

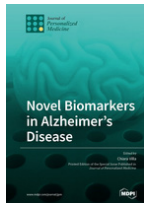
Guest Editors: Dr. Amir Hajjam El Hassani and Dr. Younes Jabrane  
Deadline: 26 April 2024



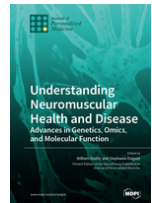
## Special Issue Books

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### Novel Biomarkers in Alzheimer's Disease



### Understanding Neuromuscular Health and Disease: Advances in Genetics, Omics, and Molecular Function



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