OPENING THE PERSONAL GATE BETWEEN TECHNOLOGY AND HEALTH CARE

Studies in Health Technology and Informatics

Internationally, health informatics is driven by developments in biomedical technologies and medical informatics research that are advancing in parallel and form one integrated world of information and communication media and result in massive amounts of health data. These components include genomics and precision medicine, machine learning, translational informatics, intelligent systems for clinicians and patients, mobile health applications, data-driven telecommunication and rehabilitative technology, sensors, intelligent home technology, EHR and patient-controlled data, and Internet of Things.

The series Studies in Health Technology and Informatics (HTI) was started in 1990 in collaboration with EU programmes that preceded the Horizon 2020 to promote biomedical and health informatics research. It has developed into a highly visible global platform for the dissemination of original research in this field, containing more than 250 volumes of high-quality works from all over the world.

The international Editorial Board selects publications with relevance and quality for the field. All contributions to the volumes in the series are peer reviewed.

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Opening the Personal Gate between Technology and Health Care

Proceedings of MIE 2026

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Preface

The 36th Medical Informatics Europe Conference, MIE 2026, will be held from 25 to 28 May 2026 in Genoa, Italy. The Conference is co-hosted by the Italian Scientific Society of Biomedical Informatics (SIBIM) and the European Federation for Medical Informatics (EFMI). The Scientific Programme Committee is chaired by Professor Mauro Giacomini and co-chaired by Professor Jaime Delgado.

The theme of MIE 2026 is "Opening the Personal Gate between Technology and Health Care". As the editors of the proceedings of MIE 2025: Intelligent Health Systems – From Technology to Data and Knowledge, have already pointed out, artificial intelligence (AI) continues to be overestimated worldwide, even within the scientific community, and greater caution should be exercised in assessing the evolution of the technologies at our disposal. No technology can ever be a panacea for all the problems we face as we employ our tools in the fight to improve the health of humanity and all of creation. To be effective, technological solutions must be integrated into the healthcare process, providing high-quality data that is appropriately contextualised to deliver meaningful information to authorised users. But, as has always been the case in the course of human development, knowledge can only arise from an open dialogue among all those involved in the process. This is precisely the objective of this edition of EFMI MIE2026, which takes place on the 50th anniversary of the EFMI in Genoa, Italy, a city which for centuries has served as a gateway to the Mediterranean basin and all other routes of human enterprise.

These proceedings present the current trends in health and biomedical informatics. The contributions cover topics like biomedical imaging and data science; computable knowledge and decision support; ethics, explainable and trustworthy AI; generative AI and natural language processing; standardisation, interoperability, FAIR data and European health data space; information systems in healthcare; personalised medicine; telemedicine and telehealth; OneHealth; education; and human centred digital health.

The proceedings are published by Sage Publishing / IOS Press as an e-book in the open access series Studies in Health Technology and Informatics (HTI). Volumes in the HTI-series are submitted (for evaluation) for indexing by MEDLINE/PubMed; Web of Science: Conference Proceedings Citation Index – Science (CPCI-S) and Book Citation Index – Science (BKCI-S); Google Scholar; Scopus, and EMCare.

The Editors,
Mauro Giacomini, Jaime Delgado, Theodoros N. Arvanitis, Elisavet Andrikopoulou, Arriel Benis, Gabriella Balestra, Riccardo Bellazzi, Parisis Gallos, Roberto Gatta, Daniele Roberto Giacobbe, Noemi Giordano, Maria Hägglund, Lars Lindsköld, Lenka Lhotska, Sara Marceglia, Enea Parimbelli, Paolo Soda, Lăcrămioara Stoicu-Tivadar, Pierangelo Veltri, Patrizia Vizza.

Genoa, 8 April 2026

About the Conference

The Conference

The Italian Scientific Society of Biomedical Informatics (SIBIM), together with the European Federation for Medical Informatics (EFMI), organised the 36th Medical Informatics Europe Conference (MIE2026) MIE2026, which took place from 25 to 28 May 2026 in Genoa, Italy. The theme of the conference was "Opening the Personal Gate between Technology and Health Care" (https://mie2026.efmi.org/). The conference was managed by the professional conference organiser Pragma Congressi, Pavia, Italy.

Founded in 1976, EFMI is the leading organisation in medical informatics in Europe, and represents 33 countries through their respective national health informatics associations. EFMI is a not-for-profit organisation concerned with the theory and practice of information science and technology within healthcare and health sciences in a European context.

MIE is a series of medical informatics conferences that aim to promote research and development in biomedical and health informatics. Members of the global medical informatics community are invited to take part in each conference as presenters or participants. Each conference consists of scientific sessions with oral presentations of peer-reviewed full papers and short communication papers. Each conference also includes panels, workshops, demos, and tutorials, some prepared by EFMI working groups. A large exhibition of peer-reviewed posters also forms part of each conference.

Conference Topics included (but were not limited to):

- ARTIFICIAL INTELLIGENCE IN MEDICINE
  - Biomedical imaging in medical informatics
  - Computable knowledge and decision support
  - Data science for visualisation and analytics
  - Ethics, regulations and AI
  - Explainable and trustworthy AI
  - Generative AI, foundational models
  - Learning health systems
  - Natural language processing applications in healthcare

- INFRASTRUCTURES AND REGULATIONS
  - FAIR data and data sharing infrastructures, European Health Data Space
  - Healthcare data security
  - Information Infrastructure Integration and certification

  - Information systems in healthcare
  - Privacy and data protection methods and technologies for healthcare
  - Standardisation and interoperability issues in medical informatics

- HEALTH INFORMATICS ECOSYSTEMS
  - Automation and robotics in healthcare
  - Digital twins
  - Digitally supported Precision Medicine
  - Internet of Medical Things
  - Medical devices (including software)
  - Patient empowerment
  - Personal health records and mHealth
  - Telemedicine and telehealth

- ONEHEALTH
  - Human factors, social and organisational issues
  - Population health
  - Translational health informatics
  - Veterinary and environmental health informatics

- EDUCATION
  - Health Informatics in medical education
  - Innovative technologies and approaches for education
  - Living labs and patients' inclusion and engagement
  - Multimodal Learning in healthcare

- HUMAN CENTRED DIGITAL HEALTH

The MIE2026 conference included three keynote speeches by internationally recognised experts in medical informatics:

- **Silvana Quaglini** has an MS degree in Electronic Engineering and PhD in Bioengineering, and is a full professor of Medical Informatics at the University of Pavia, Italy. Her research focuses on decision support systems, home monitoring and care, and the economic evaluation models of healthcare interventions. The main medical areas covered by these applications include cancer, stroke, chronic diseases, and cognitive rehabilitation. The recent push towards personalised medicine has directed her latest studies toward shared decision-making and context-aware home monitoring. She has consistently conducted applied research, mainly within EU-funded projects, collaborating with local and international hospitals. She is a past-president of SIBIM (Italian Society of Biomedical Informatics, EFMI member), member of the GNB (National Bioengineering Group), and the author of approximately 350 scientific publications, with an h-index of 47 (2025, Scopus).

- **Johan Gustav Bellika** has worked at the intersection between medicine, medical research and informatics since 1992, when he joined the Department of Community Medicine at UiT The Arctic University of Norway. He has worked at the Norwegian centre for e-health research at the University Hospital of North Norway since 1997. In the period between 2007 and 2013 he joined the department of computer science to teach and research in relation to the international master programme in Telemedicine and e-health. He is now a professor in medical informatics at the department of clinical medicine, Faculty of health sciences at UiT The Arctic University of Norway and the Norwegian Centre for E-health Research at the University hospital of North Norway. His current research focus is on technology to enable privacy-preserving reuse of health data and supporting a learning healthcare system.

- **Giorgio Cangioli** is a Senior Consultant in Digital Health and Social Care and an internationally recognised expert in healthcare interoperability and standards. He actively contributes to European initiatives supporting the European Health Data Space (EHDS). He is an HL7 Fellow, Technical Lead and Board Member of HL7 Europe, and a member of the HL7 International Technical Steering Committee. He also chairs the European eHMSEG (eHealth Member States Expert Group) Semantic Task Force Architecture Working Group. With more than 25 years of experience, Giorgio specialises in ICT, standards, and business process re-engineering in health and social care. Throughout his career, he has contributed to standardisation activities within HL7, CEN, ISO, IHE, and DICOM, and has facilitated multiple European and global standardisation initiatives. He is one of the authors of the International Patient Summary (IPS) standards (ISO/EN 27269 and HL7 IPS FHIR Implementation Guide).

Scientific Programme Committee

- Mauro Giacomini: Department of Informatics, Bioengineering, Robotics and System Engineering, University of Genoa, Italy
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- Elisavet Andrikopoulou, EFMI Publications Officer, EFMI Exec Board Member, Faculty of Technology, School of Computing, University of Portsmouth, UK.
- Parisis Gallos, Past EFMI Publications Officer; ICU Follow up – Care Research Lab, Department of Nursing, University of West Attica, Greece.

Peer Review Process

We received 884 submissions from 57 countries. A thorough review process was conducted with valuable support from 385 active reviewers. Almost all submissions were reviewed by at least three reviewers and assessed by one SPC co-chair. Based on their recommendations, final decisions were made by SPC members during a three-day virtual meeting. Papers requiring major revision underwent a further review by SPC members.

Finally, from among the 582 full papers, 103 short communication papers, 80 posters, 34 demonstrations, 25 panels, 54 workshops, and 6 tutorials submitted, 415 full papers (acceptance rate of 71%), 91 short communication papers (conversion of 29 full papers), 117 posters (conversion of 50 full papers and 15 short communications), 29 demonstrations, 26 panels (conversion of 3 workshops), and 26 workshops were accepted. All accepted full papers, short communication papers, and posters are included in these proceedings. Notably, this collection encompasses contributions originating not only from Europe (the primary geographic focus of Medical Informatics Europe 2026) but also from various other continents. This international participation highlights the global significance of sharing knowledge and practical experiences related to scientific and implementation challenges in healthcare informatics. We firmly believe that such global exchange provides valuable insights and meaningful learning opportunities for the European community and beyond.

We want to thank the Editors and the Editorial Committee, and all the reviewers for their invaluable contributions to MIE2026.

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The MIE 2026 conference is hosted at the Congress Center “Magazzini del Cotone”, Genoa, Italy.

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Section 10. Education

Opening the Personal Gate between Technology and Health Care
M. Giacomini et al. (Eds.)

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Sub-Phenotyping of Pediatric Celiac Disease with Topological Data Analysis

Daniele Pala[a,1], Giuseppe Albi[b], Valentina Brembilla[a], Erika Lenzi[a], Emanuele Medolago[a], Chiara Sirtoli[a], and Arianna Dagliati[b]
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Abstract. Celiac Disease (CD) is an autoimmune disorder triggered by gluten ingestion in genetically susceptible individuals. Its heterogeneous presentation, especially in pediatric patients, limits the effectiveness of current symptom-based classifications such as the Oslo definitions. This study proposes a framework based on Topological Data Analysis (TDA) to identify new, clinically meaningful sub-phenotypes of pediatric CD. We used a multicentric dataset of over 3,000 children. Compared with standard clustering (DBSCAN, Agglomerative, K-Medoids), our TDA Mapper revealed stable, interpretable communities reflecting serological and clinical patterns. Results indicate that serology, histology, and comorbidities jointly define distinct pediatric phenotypes, supporting data-driven approaches for precision medicine in CD.

Keywords. Topological Data Analysis, Computational Phenotyping, Celiac Disease

1. Introduction

Celiac Disease (CD) affects about 1% of the global population, with higher prevalence among females. It results from an autoimmune response to gluten, mediated by HLA-DQ2/DQ8 genotypes and antibodies such as tissue transglutaminase (tTG-IgA) and anti-endomysium (EMA). CD is diagnostically challenging due to variable gastrointestinal and extraintestinal manifestations, especially in children. Current classifications, particularly the Oslo definitions, group patients based on symptoms into classical, non-classical, and subclinical categories [1]. However, clinical experience shows significant overlap and heterogeneity, suggesting the need for new, data-driven phenotypes, or sub-phenotypes, that integrate laboratory, histological, and comorbidity information. In this study, we use a TDA Mapper pipeline [2] for the sub-phenotyping of pediatric CD. TDA Mapper [3] is a topological-based algorithm which has already proved to be able to reveal hidden structures in biomedical data [4], and has already been successfully employed for the computational phenotyping of different diseases.

2. Dataset and Methods

We used a multicentric dataset of 3,241 pediatric CD patients (0–18 years); after preprocessing and imputation, 2,922 cases and 39 features were retained. Variables

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included demographics, serology (IgA, EMA, HLA), histology (Corazza), symptoms, and comorbidities. Missing data were handled through clinically informed imputation, and categorical variables were encoded for analysis.

To handle mixed data types, Gower distance was used as the distance measure. Dimensionality reduction was performed with Principal Component Analysis (PCA), Multidimensional Scaling (MDS), and t-distributed stochastic neighbor embedding (t-SNE). We compared the results obtained with the TDA Mapper pipeline implemented in pheTDA [2] to four clustering techniques—K-Medoids, Agglomerative Hierarchical clustering, Hierarchical Density-Based Spatial Clustering of Applications with Noise (HDBSCAN) and DBSCAN, by using the Silhouette coefficient. Logistic regression was implemented for feature ranking, highlighting the most predictive clinical markers.

3. Results and Conclusion

Traditional clustering methods produced limited results. DBSCAN and HDBSCAN either merged most patients into a single group or produced numerous micro-clusters. Hierarchical and K-Medoids clustering yielded low silhouette values (<0.5) and clinically unbalanced groups. After dimensionality reduction, PCA improved separability, revealing two dominant data regions corresponding to serological status. Patients with high IgA and positive EMA clustered distinctly from those with negative serology.

TDA Mapper produced a more clinically informative structure, identifying seven groups divided into the same two large branches found by the other algorithms, but with sub-branches that captured patterns such as greater histological atrophy and complication risk, growth delay, anemia, autoimmune, and neurological comorbidities. Sankey analysis showed that many “Silent” or “Minor” Oslo categories re-clustered within serology-positive communities, revealing underappreciated clinical risk. Logistic regression confirmed IgA, EMA, and HLA positivity as dominant features, while growth delay and autoimmune diseases were secondary but discriminative.

Overall, TDA outperformed traditional clustering, enabling precision phenotyping and showing that this framework could support clinicians in differentiating silent from high-risk patients, optimizing diagnostic processes and improving patients monitoring.

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