Silesian University of Technology

RESEARCH UNIVERSITY
EXCELLENCE INITIATIVE
Ministry of Science and Higher Education

Computational Oncology and Personalized Medicine
From Genome to Treatment: AI & Precision Medicine

Konferencja Onkologia Obliczeniowa i Spersonalizowana Medycyna
Od Genomu do Terapii: SI oraz Medycyna Precyzyjna

COPM2026 Conference
Book of Abstracts

Gliwice, April 29th, 2026

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Book of abstracts
Conference programme

COPM2026

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From Genome to Treatment: AI & Precision Medicine

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Od Genomu do Terapii: SI oraz Medycyna Precyzyjna

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Onkologia Obliczeniowa i Spersonalizowana Medycyna COPM2026 — Od Genomu do Terapii: SI oraz Medycyna Precyzyjna
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Preface

We are pleased to introduce the sixth event in the series of Computational Oncology and Personalized Medicine (COPM) conferences, titled COPM2026 From Genome to Treatment: AI & Precision Medicine. The online event, organized by the First Priority Research Area (POB1) of the Silesian University of Technology within the Excellence Initiative – Research University program, will be held on April 29th, 2026. The conference is organized under the honorary patronage of His Magnificence, the Rector of the Silesian University of Technology, Prof. Marek Pawełczyk, under the auspices of the Academy of Young Scientists, and several patronages and partners.
COPM2026 offers early-stage researchers, particularly young doctors and PhD students, as well as experienced scientists, the opportunity to show their work within an international and interdisciplinary forum of experts. This booklet collects the abstracts of 76 presentations, and one invited keynote lecture by our notable guest prof. Paweł Łabaj from the Małopolska Centre of Biotechnology, Jagiellonian University. The best presenters will be invited to publish in the European Journal of Pharmacology or Bio-Algorithms and Med-Systems. Moreover, we encourage all participants to publish their work in Acta Biochimica Polonica, a journal belonging to the Polish Biochemical Society and published by Frontiers, which covers fields of enzymology and metabolism, membranes and bioenergetics, gene structure and expression, structure and metabolism of proteins, nucleic acids, and carbohydrates. Also, the Best Presentation Award, funded by Elsevier, is expected. We warmly invite all interested participants to join us and make the most of this remarkable opportunity to broaden their knowledge and establish connections with fellow researchers.
We extend our sincere gratitude to all participants for contributing their valuable research, to Prof. Łabaj for kindly agreeing to deliver the keynote lecture, and to the members of the Scientific Committee for their dedicated and professional work in reviewing the submitted abstracts. We would also like to convey our warmest wishes to everyone attending the conference, and we trust that your time here will be both fruitful and inspiring, filled with stimulating discussions and productive exchanges of ideas.

Gliwice, April 2026

Michał Marczyk
Department of Data Science and Engineering
Silesian University of Technology
Head of an Organizing Committee

Joanna Polańska
Department of Data Science and Engineering
Silesian University of Technology
Coordinator of POB1: Computational Oncology and Personalized Medicine
Head of Scientific Committee

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13:15 – 14:00 **Prof. Paweł Łabaj (Małopolska Centre of Biotechnology, Jagiellonian University)**
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17:30	**Gronkowska K**, Kołacz-Milewska K, Michlewska S, Robaszkiewicz A: Expression level of TP53 and KDM5B as a promising DNA-damaging agent response biomarker in cancers	17:30	**Grzela-Fraś K**, Łucki M, Barzowska-Gogola A, Pucelik B, Tyliszczak B: The effect of incorporating various active substances on the structure and properties of hydrogels
17:40	**Nuredini A**, Corrias R, Mendelsohn D, Pini S, Garzo A, Scipioni MP, Obach M, Laporte J, Savarese M, Diaz-Manera J, Straub V, Polańska J, Santorelli FM, Schoser B, Tupler R: The CoMPaSS-NMD neuromuscular genome atlas: A new AI-based platform for advanced deep phenotyping and stratification in hereditary neuromuscular disorders	17:40	**Mazurek V**: Personalized ankle–foot orthoses: Design strategies and biomechanical considerations
17:50	**Barzowska-Gogola A**, Sułek A, Jończyk J, Klimczak J, Danel T, Pucelik B: Replication stress in hormone-dependent HER2-positive breast cancer: New perspectives for targeted treatment	17:50	**Gwóźdź K**, Michalik T, Supierz J: Mobile solution for Bliss AAC
18:00	**Gawlas G**, Nowak A, Posid D, Seget S, Jarosz-Chobot P, Matejko B: Sleep disturbances in individuals with type 1 diabetes and their caregivers - cross-sectional quantitative study	18:00	**Saad AA**, Stępień E, Moskal P: *In vivo* range monitoring in upright hadron therapy using J-PET technology: Applications to proton beams
18:10	Barzowska-Gogola A, Łucki M, Baliś A, Sułek A, Danel T, Jończyk J, Klimczak J, **Pucelik B:** From clinical evidence to redefining precision oncology: The HER2-low paradox and intelligent therapeutic design in breast cancer	18:10	**Venigalla RT**: Diffusion-based physiological imputation for robust fetal heart rate baseline estimation
18:20	**Budziaszek J**, Blat A, Barzowska-Gogola A, Łucki M, Sułek A, Jończyk J, Klimczak J, Danel T, Pucelik B: Reprogramming antibacterial therapy: AI-guided strategies for personalized treatment of chronic wound infections	18:20	**Senaweera Y**: A computational method for molecular docking analysis of bioactive phytochemicals from jackfruit seed flour (*Artocarpus heterophyllus*) against hepatocellular carcinoma (HEPG2) cell line targets
18:30	**Czerwińska G**, Wójcik L, Osewska J, Robakowska M: Determinants of delayed diagnosis of oral cavity cancer and strategies for their mitigation using AI in clinical practice	18:30	**Łucki M**, Barzowska-Gogola A, Baliś A, Pucelik B: Drug delivery platforms for personalized therapy of hormone-dependent breast cancer
18:40	Golda A, **Bosowska Z**, Śmieja J, Zębik T, Student S: Impact of atmospheric pressure variability and time of hospital admission on the incidence and clinical course of acute myocardial infarction	18:40	**Shakibania S**, Abdullah A, Tomasiak A, Knapczyk-Korczak J, Stachewicz U, Skonieczna M, Krukiewicz K: Enhanced cell adhesion on polypyrrole-modified poly(ε-caprolactone) fibers for neural tissue engineering applications

18:45	**Roy Choudhary R**: Integrative transcriptomic analysis reveals shared molecular signatures and regulatory networks in tongue squamous cell carcinoma across diverse populations	18:45	**Radfar S**, Krukiewicz K, Cosnier S: Beyond Nafion: A triple-functional electropolymerized coating for biosensing excellence
18:50	**Mazgaj P,** Hebda A, Wawrzyniak P, Sadowski D, Kijonka M, Prażmowska J, Kapek Ł, Borys D, Rembak-Szynkiewicz J: The impact of dietary supplements on brain ^{1}H-MRS spectra – new diagnostic challenges	18:50	**Lasota I**, Pilarski P, Krukiewicz K, Kappen J: Gold nanostructure modified with polypyrrole films for ultra-sensitive detection of ovarian cancer
18:55	**Rejwana N**: Abemaciclib modulates cell-cycle-regulated genes in endometrial cancer revealed by transcriptome analysis	18:55	**Dźwięga P**: Surface EMG for control applications: Methods and challenges
19:00	**Drygała B**, Dorczak J, Seget S, Jarosz-Chobot P, Polańska J: Uncovering seasonal trends in pediatric type 1 diabetes	19:00	**Wojnarowska W**: Development of a two-region geometrical model of the L5 vertebra for numerical simulations
19:05	**Cichecka K**, Suwalska A: Prediction of insulin resistance based on epidemiological data using machine learning	19:05	**Długosz K**: Synthetic data in personalized medicine and oncology: Methods, applications, and challenges
19:10	**Więckowska I**, Adamska P: Odontogenic Keratocyst (OKC): Radiological features and diagnostic challenges	19:10	**Witkowski M**: Utility of synthetic data in clinical decision support systems: A task-aware methodological evaluation using TCGA LUAD data

Plenary session (4A)

Chairman: dr hab. inż. Michał Marczyk

19:15 – 19:30 **Conference summary and closing**

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Plenary session (1A)
time 13:15 – 14:00

Chairperson:
Marek Łos

Genomic reproducibility in the bioinformatics era

Professor Paweł P. Łabaj[1]

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Keywords: genomic reproducibility, experimental design, technical variation, bioinformatics bias

Abstract

In the presentation PP Łabaj will examine the complex landscape of genomic reproducibility, emphasizing the critical interplay between experimental ”wet lab” procedures and ”dry lab” bioinformatics. Drawing on extensive data from the Sequencing Quality Control (SEQC) project, the study highlights how technical variation, ranging from library preparation inefficiencies, where as little as 0.7% of input material may remain, to the systematic biases introduced by PCR duplication and normalization algorithms, can significantly distort gene expression estimates and differential expression calls. It will be presented that these ”challenges” are not limited, however, just to the expression profiling, but similar issues are also seen in metagenomics studies. There, the results demonstrate that while taxonomic concordance is high across different sequencing platforms, functional analysis remains a challenge due to similar as before technical challenges and dependencies. Ultimately, the research advocates for a shift in experimental design: moving beyond a ”siloed” approach by integrating biostatistical consultation before experiments begin, utilizing robust replication, and collecting extensive meta-data to balance the inherent trade-offs between high-throughput sequencing costs and data quality.

Parallel session (2A)
time 14:00 – 16:50

Chairperson:
Michał Marczyk

Relevance based supervised detection of clinically actionable variants

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Keywords: variant calling, genomics, statistics, deep learning

Abstract

Conventional genomic variant calling pipelines rely on genome-wide variant callers and stringent filtering to limit false positive calls, which inherently increases the count of false negative calls. However, a small but growing number of clinically actionable variants (predictive, prognostic, or diagnostic) must not be overlooked for certain cancers. This work aims to develop highly sensitive variant calling workflows targeted at detecting specific actionable variants. Applying state-of-the-art somatic variant calling with recommended parameters to patients with multiple tumor samples and a single matched normal sample revealed numerous false negatives. In this ongoing research, we aim to address this by developing statistical and Deep Learning models that predict local sequencing error rates from genomic context and sample-specific features using negative control samples. The fitted models are then applied to tumor samples to predict error rates, and variant calls are made based on the probability of observing the actual number of alternative reads given the predicted error rate.

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Label-free prmPASEF proteomics data analysis workflow selection - benchmarking AI-based and data-driven approaches

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Keywords: label-free targeted proteomics, data analysis workflow selection, AI-based approaches, data-driven approaches, prmPASEF

Abstract

As global proteomics advance, the number of identifiable proteins increases, yet this does not guarantee best quantitative performance. While isotope-labeled targeted assays are available, label-free strategies remain cost-efficient alternatives for results validation. However, systematic evaluations of data analysis workflows for such analyses are limited. Thus, this study benchmarks multiple workflows for label-free prmPASEF proteomics.

A benchmarking dataset was generated using human proteome background (30 proteins) with three yeast proteins spiked in at varying concentrations. Following analysis using Bruker timsTOF Pro system, data was processed in Skyline and exported to R. Calibration curves were established, and only measurements in the linear range were included.

Various missing-data imputation (MDI) strategies were tested, including no imputation, k nearest neighbors, data-driven and AI-based methods, alongside consolidation approaches such as summation, best-peak selection, AI-based scaling, p-value integration and multivariate testing. Based on FDR, accuracy and other metric, data-driven MDI combined with p-value integration achieved the best performance.

Our results demonstrate that appropriate workflow selection improves quantitative outcomes compared to commonly used methods like simple summation. Proposed approach provides a robust workflow for label-free targeted validation.

Acknowledgments: This work was supported by NCN grants no. 2021/43/B/NZ7/02221 (DF, MP, AW) and no. 2024/53/B/NZ5/02780 (LM, AW) and performed using infrastructure developed under the project NEBI - National Research Center for Imaging in the Biological and Biomedical Sciences, POIR.04.02.00 00-C004/19, co-financed through the European Regional Development Fund (ERDF) in the frame of Smart Growth Operational Programme 2014-2020 (Measure 4.2 Development of modern research infrastructure of the science sector).

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Impact of optimized ToF-SIMS data preprocessing and normalization strategies on the metabolic profiling of diabetic rat liver

Michalina Kaźmierczak[1*], Magdalena Skalska[1*], Anna Kalafova[2*], Klaudia Jaszcza[3*], Dorota Wojtysiak[4*], Marcela Capcarova[2*]

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Keywords: time-of-flight secondary ion mass spectrometry, diabetes mellitus, metformin, flaxseed mucilage, normalisation strategies, regions of interest (ROI)

Abstract

Time-of-Flight Secondary Ion Mass Spectrometry (ToF-SIMS) enables high-resolution, label free analysis of the chemical composition of biological tissues at the microscale. A key challenge in quantitative analysis is to eliminate external factors such as matrix effects and variable experimental conditions. This study aimed to evaluate the impact of data preparation and normalisation strategies on the comparative metabolic profiling of liver sections from Zucker Diabetic Fatty (ZDF) rats, including untreated diabetic controls and groups receiving metformin and flaxseed mucilage. Tissue sections were analysed using a ToF-SIMS 5 instrument in positive and negative ion modes. To minimise topographical artefacts and surface charging effects, regions of interest (ROI) of 150 × 150 μm² and 82 × 82 μm²were selected. Three normalisation approaches, total ion count (TIC), primary ion dose, and normalisation to the control group, were compared against non-normalised (RAW) data. Method sensitivity was assessed through ion abundance and principal component analysis (PCA).Results indicate that TIC normalisation effectively reduces instrumental variability, although it may obscure subtle biological differences. Normalisation to the control group clearly revealed therapy-induced changes in lipid and amino acid profiles. Furthermore, ROI optimization combined with multivariate analysis significantly improves

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the separation of groups. The choice of preprocessing and normalisation strategy critically influences data interpretability and conclusions regarding pharmacological intervention.

Integrated CITE-seq analysis links leukemic differentiation to lymphoid microenvironmental signalling network

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Keywords: leukemia, tumor microenvironment, single-cell analysis, cell communication, proteogenomics

Abstract

Classification systems of Acute Myeloid Leukemia (AML) do not take into account the phenotypic plasticity of leukemic cells. We aimed to map leukemic niche interactions using an integrated multimodal approach to identify therapeutic vulnerabilities.

We integrated CITE-seq data from 4 cohorts comprising 49 patients (319,963 cells) via totalVI for joint probabilistic modeling. Malignant blasts (68,944 cells) were isolated using an XGBoost classifier trained on denoised values. Cells were annotated via Azimuth reference mapping and clustered to define distinct differentiation states. Patients were stratified based on leukemic composition. Downstream analysis employed pseudobulk aggregation differential expression, network inference (CellChat), and validation on bulk RNA-seq cohorts (TCGA, OHSU).

Stratification defined HSC-like, Monocyte-like, and Heterogeneous subgroups. Communication analysis revealed that differentiation status is strongly associated with distinct patterns of microenvironmental remodeling. Primitive cell interactions are characterized by CD99 and LGALS9 axes, whereas mature cells exhibit enriched signaling through specific integrins and nicotinamide phosphoribosyltransferase (NAMPT). Transcriptomic validation linked these drivers to survival and distinct ex vivo drug sensitivity.

The AML microenvironment is shaped by different leukemic differentiation states. Our computational framework defines tumor architectures, highlighting vulnerabilities for precision therapies.

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Longitudinal multi-omics analysis of FRG1-mice identifies Cdkn1a as a therapeutic target for FSHD

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Keywords: FSHD, FRG1, longitudinal multi-omics analysis, Cdkn1a

Abstract

FRG1 gene encodes a ubiquitous RNA binding protein with an unknown molecular function. Its overexpression in mouse skeletal muscle induces defects that resemble human facioscapulohumeral muscular dystrophy (FSHD), the third most common inherited myopathy, including progressive muscle wasting, abnormal spinal curvature, reduced body weight, and impaired muscle maturation.

As a complex disease, FSHD is influenced by a combination of multiple factors that occur over time. To obtain a comprehensive overview of disease progression and early pathogenic events, not achievable with single-omic analyses alone, we applied a longitudinal multi-omics approach integrating transcriptomic and proteomic data from skeletal muscles collected at different time points, postnatal weeks 1 to 4, prior to the onset of overt disease. We identified coordinated and progressive alterations, including an early impairment of postnatal muscle maturation and a reduced metabolic efficiency, followed by activation of stress pathways, including p53 signaling, along with progressive inflammation and fibrosis.

Among the molecules identified by multi-omics analysis, we focused on Cdkn1a, whose expression is increased in FRG1 mice muscles and FSHD patients. Genetic ablation of Cdkn1a in FRG1 mice ameliorates muscle architecture and reduces chronic inflammation, suggesting Cdkn1a as a key effector of dystrophic progression and identifying it as a potential novel druggable target for FSHD and other related neuromuscular disorders.

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RNA-Seq library preparation bias affects long transcript detection

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Keywords: RNA-seq, RNA, ribosomal RNA, mRNA, transcriptome, splice junction, clinical diagnostics, variant interpretation, human tissue

Abstract

RNA sequencing (RNA-seq) plays an important role in both basic biology and precision medicine by supporting biomarker discovery, aberrant splicing detection, and variant interpretation. However, the type of RNA captured depends strongly on the library preparation step. Poly(A)+ RNA-seq enriches for polyadenylated transcripts, whereas rRNA depletion captures a broader range of RNAs, including non-polyadenylated and partially processed transcripts.

In this study, we compared poly(A)+ selection and rRNA depletion using human and blood RNA cohorts to determine how these techniques differ in transcript coverage, splice junction detection, and representation of long transcripts. Using RNA-seq analysis tools, we found that rRNA depletion consistently detected more splice junctions and more novel splicing junctions than poly(A)+ libraries. In contrast, poly(A)+ libraries showed a stronger 3' bias, with reduced coverage across long transcripts.

Most importantly, rRNA depletion performed better for long genes and transcripts with complex splicing patterns, which are often clinically relevant but difficult to capture with poly(A)+ selection.

These findings highlight that RNA-seq library preparation is not only a technical consideration, but also an important factor influencing which disease associated transcripts can be detected. For studies focused on long transcripts, alternative splicing, or degraded clinical RNA samples, rRNA depletion may provide a more complete and clinically informative view of the transcriptome.

Acknowledgments: We would like to thank Marco Savarese and Ali Oghabian for their guidance and support throughout this project, and CSC - IT Center for Science and the University of Helsinki IT Center for computational resources.

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Performance evaluation of different machine learning algorithms for classifying type 1 diabetes on genomic data

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Keywords: type 1 diabetes, machine learning, bioinformatics, genomics

Abstract

Type 1 diabetes (T1D) is an incurable, chronic autoimmune disease that is difficult to classify due to its unknown etiological causes and the rise in cases among adults, which contradicts traditional diagnostic criteria. Therefore, the selection of appropriate classification methods capable of capturing complex relationships in genetic data is crucial.

In this study, a comparative analysis of WES data was conducted, comprising a total of 3,437 T1D cases and 5,018 controls from the Ukrainian population. Three classification approaches were used to distinguish patients from controls: logistic regression, XGBoost, and a multilayer perceptron.

The results of the experiments indicate that the XGBoost model achieves significantly higher classification performance (Accuracy=86.67%, AUC=94.46%, Specificity=87.96%, Sensitivity=84.77%) compared to logistic regression (62.38%, 65.85%, 70.48%, 50.37%, respectively) and MLP (57.54%, 61.59%, 51.63%, 66.33%, respectively). The results emphasize that simple linear models may be insufficient to capture the complex structure of genomic data. In contrast, training more advanced architectures, such as deep neural networks, most likely requires much larger datasets to achieve satisfactory classification results. In practice, the optimal choice of classifier should balance model complexity, biological interpretability, and the characteristics of the dataset being analyzed.

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Evaluation of the cutoff significance for genetic targets of selected miRNAs in pathway enrichment analysis

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Keywords: miRNA, pathway enrichment analysis, signaling pathways

Abstract

Pathway enrichment analysis is one of the key bioinformatics tools, enabling the identification of biological processes and signalling pathways. To interpret the results, an initial selection of genes or proteins based on specific cutoff thresholds is required. The choice of an appropriate threshold determines which genes or proteins are included in the analysis, thereby influencing the results obtained and their subsequent biological interpretation. The aim of this study is to assess the impact of the choice of cutoff threshold, target selection parameters, and parameters defining the minimum size of pathways on the results of signalling pathway enrichment analysis.
The analysis was conducted for two different miRNAs - has-miR-19b-3p and has-miR-30a-3p- using three independent tools, such as STRING, ShinyGO0.82 and one implemented in-house - CATNAP . The analysis focused on two annotation databases: KEGG and Gene Ontology. The targets were identified using the miRDB tool.
The study demonstrated that the parameters determining the number of targets submitted for analysis have the most significant impact on the results. This trend was consistent across the results obtained from both annotation databases and for both tested miRNAs. The results obtained may serve as a practical recommendation regarding the selection of enrichment analysis parameters.

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Semantic similarity of LLMs and overrepresentation results in pathway enrichment analysis

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Keywords: large language models, over-representation analysis, gene sets

Abstract

Large language models (LLMs) have gained significant popularity and are now being applied across various fields. This includes analysis of results from molecular biology experiments. Here one of classic method for studying a list of statistically significant genes is overrepresentation analysis (ORA), which identifies disturbed signalling pathways. Similarly, LLM requires a list of genes as input data and returns a list of pathways.

The aim of this study was to investigate the potentials and capabilities of widely available untrained LLMs as well as trained LLMs, such as GSAI and llm2geneset, in analysing pathway enrichment compared to the ORA algorithm.

None of the untrained LLMs return the same number and pathways as ORA. The best coverage was achieved by the GPT-5o with a score of 19.2% and Gemini 2.5 Pro with a score of 8.9%. Yet, the biological pathway returned by LLMs are more general compared to ORA. In terms of semantic similarity (cosine distance on MedCPT embeddings) detailed prompts give very similar results between each other (0.97). However, the similarity to ORA is 0.9. In terms of trained LLMs, they reach high semantic similarity between their own results (over 0.95) regardless of input granulite. The similarity to ORA is 0.9 for GSAI, similar to untrained model. Yet, llm2geneset in best option shows even 0.95 to standard method. These findings show that LLMs are able to assign genes to specific pathways and provide correct labels. LLMs can be helpful in analysis, but they have certain of limitations.

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Towards intelligent risk assessment in infertility diagnosis: preliminary results of a machine learning-based approach

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Keywords: risk assessment, clinical data, infertility treatment, reproductive medicine, random forest

Abstract

Infertility diagnostics is inherently multidimensional, but many decision-support models are still based on a limited set of clinical variables. This project addresses that gap by developing a machine learning framework to support diagnostic planning using data from four infertility domains: genetics, cytogenetics, microbiology/cytology, and immunology. The aim is to improve risk assessment and help prioritize diagnostic pathways from the earliest stages of infertility evaluation.

The data consisted of selected infertility diagnostic tests that help to predict pregnancy. We coded the descriptions of the test results as follows: abnormal – 0; inconclusive – 1; normal – 2. For each domain, data were filtered, and missing values were imputed using the k-nearest neighbors algorithm (k=10). Next, a Random Forest classifier was trained using class balancing. Model performance was assessed with balanced accuracy, precision, and recall. The decision threshold was optimized on the validation set using F1 score.

The current work established a pipeline to integrate complex infertility-related diagnostic data and evaluate predictive performance across distinct research domains. Preliminary results indicate that the model performs best in predicting pregnancy outcomes in the field of immunology testing. Future work will focus on comparing results across the study groups and translating the system into a clinically useful tool for personalized infertility diagnostics.

Acknowledgments: The project was conducted in cooperation with the Genetic Laboratory of the Gyncentrum Sp. z o.o. in Sosnowiec. This work was supported by the Ministry of Science and Higher Education 'Implementation Doctorate' grant number DWD/7/0396/2023.

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Building a Polish medical language corpus for LLMs training

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Keywords: Polish medical corpus, clinical text mining, large language models, domain-adaptive pretraining, Polish-DistilRoBERTa

Abstract

Large Language Models (LLMs) are usually trained on large general-domain corpora that represent common language. In many tasks, their performance can be improved by further training on domain-specific corpora, enriched with specialised terminology. In the Polish medical domain, the availability of resources is limited, which slows down the development of Natural Language Processing (NLP) models in this field. This study aimed to construct a Polish medical language corpus focused on infertility, genetics, and gynaecology fields, intended for domain adaptation of a language model. A medical dataset was collected with over 4 million words, pre-processed, and three corpus construction methods were applied, including two publicly available tools and a novel approach. Subsequently, the initial corpus file was unified through post-processing. The resulting corpus was used for continuous domain-adaptive pretraining of the Polish-DistilRoBERTa model using the Masked Language Modelling (MLM) method. To evaluate the usefulness of the developed corpus, an MLM loss function was used to determine the accuracy of predicting masked tokens.

The evaluation of the developed corpus confirmed an improved training process and the suitability of the developed Polish medical language corpus. In future work, the corpus will be expanded and made publicly available. Additionally, the developed model will be applied to clinical data classification, enabling more efficient NLP analysis of clinical data.

Acknowledgments: The project was conducted in cooperation with the Genetic Laboratory of the Gyncentrum Sp. z o.o. in Sosnowiec. This work was supported by the Ministry of Science and Higher Education 'Implementation Doctorate' grant number DWD/7/0396/2023.

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Improvement of the C-Mixup method for effective reduction of batch effects in prediction models based on whole-exome sequencing data

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Keywords: data augmentation, batch effect reduction, whole exome sequencing, C-Mixup

Abstract

Whole-exome sequencing (WES) studies are prone to non-biological variation, especially in multi-center settings, which may reduce the generalizability of deep learning models. These effects can arise from differences in laboratory protocols or bioinformatics pipelines. Existing methods for mitigating batch effects in variant data remain limited.

To address this issue, we propose the following modification of the C-Mixup algorithm: (i) computation of label-based similarity via kernel density estimation; (ii) construction of a penalty matrix using clustering to reduce mixing of samples within the same cluster; (iii) dataset balancing by sampling underrepresented observations; (iv) generation of new samples as weighted combinations of two selected instances. The method was evaluated on a multicenter dataset using UMAP visualizations and the Adjusted Rand Index (ARI) computed between the original and augmented datasets. The impact on predictive performance was assessed using a TabNet model, evaluated with Lin's Concordance Correlation Coefficient (CCC).

Results showed a significant reduction in ARI to 0.15, indicating reduced batch effect. No negative impact related to patient age was observed. Test performance improved with the proposed method (CCC = 0.41) compared to models trained on original data (CCC = 0.35) and using vanilla C-Mixup (CCC = 0.31). The proposed modification will be tested on different models and tasks.

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Enhancing variant detection accuracy in FFPE Samples: Comparative evaluation of computational and enzymatic correction approaches

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Keywords: FFPE, formalin fixation, artifact correction, next generation sequencing

Abstract

Formalin-fixed, paraffin-embedded (FFPE) tissues are widely used in molecular studies, but formalin fixation causes DNA damage, especially cytosine deamination leading to C>T changes, that can affect variant detection. To reduce these artifacts, both computational tools (SOBDetector, Ideafix, MicroSEC, FFPolish, DeepOmics FFPE/FFPE-Plus, FFPErase) and enzymatic repair methods such as NEBNext® FFPE DNA Repair Mix v2 are used. However, their performance has not been consistently compared. In this study we evaluate these approaches using three independent datasets, including whole genome (CGCI-BL) and whole exome sequencing data (TCGA-PC and SUT-LUAD, the latter including enzymatically repaired samples), with matched fresh frozen (FF) tissues as reference. Among the computational methods, FFPErase showed the most balanced performance, achieving the best trade-off between sensitivity and precision in variant calling relative to FF references. Enzymatic repair with NEBNext® FFPE DNA Repair v2 Module provided the most consistent recovery of native variant profiles, outperforming all computational approaches in overall data quality. Combining enzymatic repair with computational filtering did not improve performance.

In summary, genomic analysis of FFPE samples requires either enzymatic DNA repair or appropriate bioinformatic correction tools to effectively reduce artifacts and improve the reliability of somatic mutations in cancer studies.

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Early excess methylation at disease-associated CpGs in endometrial cancer inferred from bulk tissue

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Keywords: DNA methylation, cell type deconvolution, linear mixed modelling, CpG-specific modelling

Abstract

Endometrial carcinoma is an epithelial malignancy, but DNA methylation is usually measured in bulk tissue, where epithelial, fibroblast, and immune signals are mixed. This makes it difficult to determine whether disease-associated methylation changes arise specifically from the epithelial compartment. We asked whether epithelial-associated DNA methylation can be inferred from bulk tissue data at CpG resolution and when excess methylation at disease-associated loci emerges during endometrial disease development.

We analysed matched tumour and normal-adjacent TCGA UCEC samples using cell type deconvolution and CpG-specific linear mixed modelling. For each CpG, we derived an adjusted expected tumour methylation value from estimated cell type proportions and compared it with observed tumour methylation. This identified CpGs with higher observed methylation than expected in tumour samples, including loci in GYPC and ZSCAN12. In cancer-free hyperplasia, highlighted CpGs already showed elevated methylation relative to normal-adjacent tissue, whereas metastatic samples showed little evidence of widespread additional methylation gain beyond that expected from matched primary tumours.

These results show that bulk tissue DNA methylation data can be used to assess epithelial-associated methylation change at CpG resolution and suggest that excess methylation at disease-associated CpGs is acquired early in endometrial disease development.

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Multi-modal deep learning predicts cell-type-specific chromatin accessibility

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Keywords: ATAC-seq, multimodel deep learning, personalised therapy

Abstract

Dysregulation of chromatin accessibility is a hallmark of oncogenesis, yet traditional predictive models often rely on static DNA sequences, failing to account for the dynamic trans-regulatory environments of specific tumors. I present an AI-driven multi-modal framework for the continuous regression of log2 fold-enrichment ATAC-seq signals, bridging the gap between genome-wide data and precision therapeutics.

Methodology

My ”Dual-Stream” architecture utilizes a DNA sequence stream to process 1,000-bp segments via a deep 1D CNN integrated with Residual connections and Squeeze-and-Excitation (SE) attention. This enables the recalibration of sequence-motif features based on their regulatory potential. Simultaneously, the model captures cellular context through an expression stream incorporating RNA-seq data. A critical innovation is the high-resolution ”Multi-Gene Context” vector, which evaluates the transcriptional activity of the nearest genes within a ±100kb window. These streams are fused into a joint representation to predict accessibility in held-out cellular conditions, demonstrating high performance in capturing quantitative gradients.

Conclusion

This dynamic, multi-modal mapping of the regulome provides a scalable tool for discovering novel oncogenic drivers and predicting the consequences of non-coding variations, paving the way for advanced personalized cancer interventions.

Acknowledgments: I would like to thank Dr. Ravi Shankar, Principal Scientist at CSIR-IHBT, and Mr. Umesh Bhati for giving me this opportunity and the resources to work on this project, as well as for their invaluable guidance and support.

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From single markers to bacterial synergies: MultiDimensional Feature Selection reveals conserved microbiome signatures for personalized medicine

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Keywords: bioinformatics, metagenomics, microbiome, synergies

Abstract

Metagenomic studies of disease-associated microbiome states have relied on univariate approaches evaluating microbial features in isolation, missing combinatorial signals from co-occurring taxa and functions. We present a framework leveraging the MultiDimensional Feature Selection (MDFS) algorithm to identify microbial feature pairs whose joint information gain substantially exceeds that of either constituent alone. Using a colorectal cancer (CRC) meta-analysis of 12 cohorts in a leave-one-cohort-out cross-validation framework, we show that synergistic pairs match state-of-the-art classification benchmarks (AUC=0.85) while uncovering interactions undetectable by conventional methods. High-stability pairs maintained discriminatory power across independent cohorts under stringent per-cohort effect size testing. Synthetic features encoding log-ratio and geometric mean transformations captured biologically interpretable relationships consistent with the Driver-Passenger model of CRC carcinogenesis. Extending the framework to 20 additional cohorts spanning IBD, type 2 diabetes, liver cirrhosis, and cardiovascular disease, we identified thousands of synergistic interactions and 21 conserved cross-cohort markers. These results establish pairwise feature synergy as a reproducible, biologically informative axis of disease-associated variation, offering a principled computational approach for interaction-based biomarker discovery in precision medicine.

Acknowledgments: We would like to thank dr Krzysztof Mnich for his contributions to the MDFS algorithm and dr Tomasz Kościołek for his valuable feedback.

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What omics modalities reveal about lung cancer: A machine learning study of subtype classification and immune stratification in LUSC

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Keywords: lung squamous cell carcinoma, multi-omics, machine learning

Abstract

Molecular subtypes of lung squamous cell carcinoma (LUSC) carry distinct prognostic implications, yet it remains unclear which omics modalities faithfully encode subtype identity and what signals different machine learning architectures capture from each. We address this by training five classifiers- logistic regression, elastic net, SVM, random forest, and XGBoost - across five TCGA-LUSC platforms (gene expression, DNA methylation, miRNA expression, copy number variation, and reverse phase protein array), utilising feature importance and SHAP-based analysis to characterise the biological signals each model prioritises. Differential expression analysis of top-ranked features contextualises findings within established LUSC biology and highlights candidate biomarkers. We further examine whether architecturally distinct models converge on shared biological signals or exploit complementary feature sets.

To extend beyond expression-derived subtype labels, we identify immune subtypes of LUSC through consensus clustering, validated against survival outcomes and immune infiltration estimates. The best-performing classifier is applied to this immune stratification and evaluated on an independent cohort (GSE30219) assessing transferability across platforms and institutions. Together, these analyses provide a modality-level account of what machine learning encodes about LUSC biology, with practical implications for biomarker prioritisation and clinically transferrable classification.

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Exploring clinical heterogeneity among patients diagnosed with Type 1 diabetes

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Keywords: type 1 diabetes, clustering analysis, pediatric diabetes

Abstract

Type 1 diabetes (T1D) is a chronic autoimmune disease with considerable variation in the course of the disease among patients. The aim of this study is to investigate whether unsupervised machine learning (ML) methods can assist clinicians in predicting the metabolic control (MC) pattern.

The study population comprised 50 children diagnosed with T1D who were undergoing treatment with an AI-controlled insulin pump and four years of follow-up. Several clustering scenarios were applied to identify subgroups of patients exhibiting diverse patterns over time.

The BMI can serve as an indirect MC indicator. The study identified three distinct, stable over the four years, subgroups of patients (around age-specific mean BMI, below and above the mean). The higher BMI group demonstrated lowest time spent in a hypoglicemic state, but heightened results in hiperglicemic ones.

When comparing the patient subgroups stratified according to the disease duration, it was observed that initial differences in clinical indicators (e.g. daily insulin doses, $p = 0.0060$) became less pronounced over time. Finally, a patient stratification in a domain spanned over all clinical variables at the beginning timepoint revealed three distinctive subgroups, which differed significantly in their clinical profile, but the differences vanished within four years of observations.

The preliminary results confirmed the potential of ML tools to predict the course of T1D among newly diagnosed patients.

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Analysis of genetic variants in trio WES sequencing in support of the diagnosis of neuromuscular diseases – preliminary analysis

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Keywords: whole exome sequencing, GATK, ANNOVAR, Slivar, Mendelian error

Abstract

Identification of rare and damaging variants contributing to complex and Mendelian disorders is a fundamental aspect of precision medicine. Despite great advances in genetic testing, numerous patients remain undiagnosed. The project focuses on the analysis of genetic variants in trio WES sequencing in support of the diagnosis of neuromuscular diseases (NMDs).

WES data from one family trio was processed using a GATK-ANNOVAR-Slivar pipeline. Inclusive Slivar filtering was applied to retain low-confidence and missing parental genotypes, prioritizing sensitivity over specificity. Initial analysis yielded Mendelian violations for 0.31% records, and SNP Ti/Tv ratio around 2.29. Further analysis was conducted for all variants tagged by inclusive Slivar (89 386). In the first stage, only variants marked as PASS (23 270) or LowGQ (1 640) were scored using a composite pathogenicity score integrating functional consequence, population frequency, inheritance model prior, and technical quality. Tiering was performed based on this scoring. 326 variants absent from gnomAD and 740 rare variants (AF<0.001) were retained as candidate causal variants for further evaluation. Candidate genes associated with muscular dystrophy were identified: TTN, PLEC, ANO5. Further analysis is required, including evaluation of variants marked as LowDP.

The described pipeline shows promise as a structured framework for candidate variant prioritization in trio-based WES analysis, but needs further validation.

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Integrative classification of loss of function variants in ovarian cancer genomes

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Keywords: loss of function, SNVs, SVs, CNV, ovarian cancer, MMEJ

Abstract

Most pathogenic mutations occur within protein-coding regions, affecting protein structure and function. However accurately identifying Loss of Function (LoF) variants remains a major challenge, since such alterations arise from diverse genomic events that extend beyond typical approaches focused on missense mutations. Therefore classification of these changes requires an integrative approach that considers diverse types of genomic events.

The aim of this study was to develop a LoF classification algorithm, based on the integration of somatic single-nucleotide variants (SNVs), structural variants (SVs), and copy number variation (CNV). LoF status was defined binarily, based on four criteria: the impact of an SNV, partial or complete gene loss associated with CNV, and the presence of SV that disrupt gene structure. A gene was classified as LoF if at least one of those features was identified.

The algorithm was tested using whole genome sequencing data from ovarian cancer samples, with particular consideration of deletions associated with the microhomology-mediated end joining (MMEJ) repair mechanism. The analysis demonstrated that CNVs are the primary drivers of LoF classification in this context, although SVs and SNVs provide additional, complementary contributions. Integrating these data provides a more comprehensive understanding of gene inactivation mechanisms in cancer, extending beyond conventional analysis focused primarily on coding SNVs.

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Parallel session (2B)
time 14:00 – 16:50

Chairperson:
Marek Socha

Imaging-based assessment of porous anatomical models of the L5 vertebra using micro-CT for applications in personalized medicine

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Keywords: micro-computed tomography, phantom models, porosity assessment, volumetric analysis, spinal implants, L5 vertebra, non-destructive testing, surgical planning tools, clinical workflow

Abstract

The development of personalized spinal implants and AI-assisted surgical planning tools requires reliable anatomical models that accurately reflect the structural properties of real bone. This study evaluates the porosity of three variants of the Sawbones® L5 vertebra phantom model differing in cancellous bone fill, using micro-computed tomography (micro-CT) imaging performed on a Nikon XT H 225 ST 2x scanner, with volumetric analysis carried out in VG Studio MAX 2025.4.

The aim of this work is to demonstrate the full assessment workflow using phantom models, a process identical to what would be applied to real human bone. Phantoms provide a controlled and reproducible environment for validating the methodology, while the workflow itself remains directly transferable to patient-specific cases. While this work is conducted on standardized phantoms, the methodology directly translates to personalized medicine applications. Patient-specific anatomical models can be derived from individual CT scans, enabling the creation of implants tailored to a particular person's geometry and bone structure. Micro-CT imaging can then serve as a non-invasive quality control step verifying the structural and geometric fidelity of a custom implant before it is introduced into the patient's body, reducing risk and improving surgical outcomes.

This study demonstrates the potential of micro-CT as a precise, non-destructive assessment tool within a broader personalized healthcare workflow, bridging the gap between phantom-based research and real-world clinical application.

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Prediction of 2-Hydroxyglutarate (2HG) based on nuclear magnetic resonance spectroscopy, using a GLX3/4 ratio between tumorous and healthy regions

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Keywords: spectroscopy analysis, 2-Hydroxyglutarate (2HG) prediction, signal analysis, early glioma

Abstract

Introduction:
Early non-invasive diagnosis of glioma is critical for optimizing treatment strategies. The presence of an oncometabolite, 2-Hydroxyglutarate (2HG), is a major indicator of IDH mutations and is difficult to detect by magnetic resonance spectroscopy (MRS). The study presents a nonparametric method for signal processing and final prediction.
Materials and Methods:
The study cohort comprised 51 patients with brain tumors, including 21 patients with confirmed glioma with IDH1 mutation. MRS samples were acquired from two locations per patient: the lesion core, and the contralateral normal-appearing brain tissue. The prediction method is based on the relation between the estimated areas under the curves of GLX3 and GLX4 metabolites. Since the resonant frequency of 2HG overlaps with the GLX3 range, discrepancy between signals acros regions increases significantly in the presence of 2HG.
Results:
The method was initially developed on 20 patients, achieving a balanced accuracy of 0.76. Subsequent validation in an independent cohort of 31 patients yielded balanced accuracy of 0.74. When evaluated across the entire dataset, the method maintained an overall balanced accuracy of 0.75.
Conclusions:
While the proposed method requires further refinement before clinical implementation, these preliminary results outperform other existing MRS-based diagnostic techniques. Current limitations in accuracy are primarily attributed to low metabolite

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level and inherent signal distortion in the spectroscopic data.

Acknowledgments: The present work is a part of broader research project supported by the Medical Research Agency number KPOD.07.07-IW.07-0197/24 titled Radiogenomics as a tool to determine the genotype of the primary brain tumors with multiparametric magnetic resonance imaging with particular usage of spectroscopy (1H-MRS) targeting 2-hydroxyglutarate (2-HG) as an oncometabolite.

MRI image analysis for Alzheimer's disease diagnosis using machine learning methods

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Keywords: Alzheimer's disease, MRI, machine learning, convolutional neural networks, ResNet-18

Abstract

The thesis explores in detail the potential of machine learning techniques in the context of diagnosing one of the most common neurodegenerative diseases, Alzheimer's disease. The disease, a leading cause of dementia in the elderly, is a growing challenge for healthcare systems worldwide, and its early diagnosis is crucial for effective treatment and slowing its progression. This paper focuses on magnetic resonance imaging (MRI), allowing detailed analysis of brain structures. Traditional diagnostic approaches, based on visual assessment of MRI images by radiologists, are often not sensitive enough, especially in identifying subtle changes in the early stages of the disease, justifying the need for more sophisticated tools. Therefore, the aim of this study was to investigate and evaluate the effectiveness of different machine learning methods on a pre-selected dataset. In this study, a convolutional neural network model, adapted to the specificities of brain MRI images, was developed to identify pathological features that could indicate the presence of disease. This model was compared with the ResNet-18 model and traditional machine learning methods, allowing an in-depth evaluation of its performance. The results of the experiments show that the use of convolutional neural networks allows for a significant increase in the accuracy and efficiency of Alzheimer's disease diagnosis, outperforming classical approaches in many cases.

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Comparison of deep learning methods for coronary vessel segmentation from angiographic images

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Keywords: biomedical images, neural networks, image analysis, deep learning

Abstract
The development of computational technologies enables advanced tools for data analysis, particularly in medicine, where their main goal is to support diagnostics.
The project aims to build U-Net models, train them on images with different preprocessing, and select the most effective one. The training data were obtained from two datasets: Invasive Coronary Angiography (ICA), which contains 616 images and Database X-ray Coronary Angiograms (DCA1) which contains 134 images.
Three U-Net–based architectures were applied. The first was a classical variant of the U-Net neural network. The second architecture is a Dual-Branch U-Net, a model with two encoders whose features are merged after encoding stage. Last architecture is Attention-based U-Net, which uses attention to filter skip connections and improve vessel segmentation.
Next, the effect of input image preprocessing on prediction performance was evaluated, using Laplacian of Gaussian gradient localization, CLAHE contrast enhancement, and rolling ball background removal.
Training was evaluated with Focal Loss. Prediction performance was assessed using Intersection over Union (IoU), Dice coefficient, Recall, Precision, and clDice.
Based on the evaluation of metrics Dice, IoU, Recall, Precision, and clDice, the most effective U-Net model was selected, which segments vessels most efficiently and accurately in the analyzed datasets. This model demonstrates significant potential for future clinical applications.
Acknowledgments: Calculations were carried out using the infrastructure of the Ziemowit computer cluster (www.ziemowit.hpc.polsl.pl) in the Laboratory of Bioinformatics and Computational Biology, The Biotechnology, Bioengineering and Bioinformatics Centre Silesian BIO-FARMA. Funding: Project-Based Learning (Excellence Initiative – Research University Program), in accordance with Regulations No. 54/2020 and 55/2020 of the Rector of the Silesian University of Technology dated March 13, 2020.

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Application of artificial intelligence in musculoskeletal X-ray analysis for fracture detection

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Keywords: artificial intelligence, deep learning, convolutional neural networks, medical image analysis, X-ray imaging, fracture detection, computer-aided diagnosis

Abstract

This study presents the application of artificial intelligence for detecting fractures in musculoskeletal X-ray images. The aim was to develop and evaluate a convolutional neural network (CNN) model for automatic classification of fractures. The dataset, obtained from a public Kaggle source, included labeled X-ray images of fractured and non-fractured cases.

The methodology involved image preprocessing, including normalization, resizing, and division into training, validation, and test sets. A CNN model was implemented and optimized using selected hyperparameters. Model performance was evaluated using accuracy, precision, recall, F1-score, and confusion matrix.

The results demonstrate that the model effectively detects fractures; however, a tendency to over-detect fractures was observed. The study confirms the potential of AI in supporting medical diagnostics, while highlighting the need for further optimization to improve reliability.

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Augmentation influence on classification of selected tumor types in MRI images

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Keywords: machine learning, brain tumors, MRI, image classification, data augmentation, medical image

Abstract

Accurate diagnosis is a critical step in effective patient treatment. With the development of machine learning methods, image classification models are increasingly used to support medical decision-making by detecting subtle abnormalities in MRI scans. However, the performance of such models strongly depends on the size and diversity of the training dataset, which is often limited in medical applications.

In this study, I investigated the impact of data augmentation techniques on the classification of selected tumor types in MRI images. Due to the limited size of available medical datasets, augmentation was applied to artificially increase data diversity. The tested methods included Gaussian noise, rotation, brightness adjustment, width and height shifts, shear transformations, zooming, horizontal and vertical flipping.

The aim of this work was to evaluate which augmentation techniques improve model performance the most. I trained and tested models using differently augmented datasets and compared their classification accuracy. The results show that data augmentation can significantly improve model performance, although its effectiveness depends on the type and intensity of transformation. Some techniques enhanced generalization, while others had little or negative impact. These findings confirm that careful selection of augmentation strategies is essential in medical image classification tasks.

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Machine learning-based classification of neuromuscular disorder patients using H&E-stained muscle biopsies

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Keywords: neuromuscular diseases, histopathology, machine learning, automated diagnosis, muscle biopsy

Abstract

Histological analysis of skeletal muscle is essential for diagnosing neuromuscular disorders; however, manual evaluation of muscle biopsies is labor-intensive and prone to subjective grading by pathologists. To address this, we present an automated image processing pipeline designed for the quantitative characterization of muscle tissue Whole Slide Images (WSIs). Our approach utilizes machine learning-based cell detection to extract 1613 distinct morphological and textural features. These include cell size, circularity, and percentage of fatty deposits, as well as mathematical features like fractal dimension. To ensure robustness against staining variability a custom color quantization method was used, alongside a tissue detection algorithm. WSIs were split into tissue regions and subsequently into 600 000 patches of size 2000x2000 pixels. Using this data we developed two classifiers: the first to distinguish pathological from healthy tissue with an AUC of 0.89, and the second to identify five specific disease subtypes with an AUC of 0.73. Validated on a large-scale dataset of 1711 clinical patients, our pipeline demonstrates high diagnostic potential and offers a scalable solution for standardized, automated neuromuscular disorder screening.

Acknowledgments: This research was supported by the CoMPaSS-NMD grant.

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MRI-based deep radiomic phenotyping of neuromuscular disorders: A topology-driven classification

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Keywords: radiomics, neuromuscular diseases, graph theory, GMM, tissue segmentation, classification

Abstract

Quantitative assessment of muscle MRI is crucial for monitoring neuromuscular disorders. This preliminary study introduces an automated radiomic pipeline based on 32 original features engineered across four domains: morphological, spatial distribution, structural complexity, and graph-based topology. Using 842 MRI images from the CoMPaSS-NMD project, we developed an adaptive segmentation framework using GMM optimized via BIC to decompose tissues into three phases: muscle and two different fats. The innovation lies in the graph-based skeletonization of fat infiltrates to quantify architectural changes. Statistical Kruskal-Wallis analysis confirmed the high discriminative power of these novel descriptors across the genetic hierarchy, notably Fat_CoM_Y (eta-squared=0.36) and Fat_Skel_Nodes (eta-squared=0.19). To ensure a robust diagnostic tool, an optimized 15-feature signature was selected through 100-iteration Monte Carlo ranking to maximize feature stability. Random Forest classification was optimized using Grid Search and evaluated by 10-fold CV (with and without SMOTE). The best-performing model achieved a global accuracy of 80.4% and a weighted F1-score of 0.81. It showed exceptional performance for specific phenotypes, reaching a precision of 0.95 for Myotonic Dystrophies and a recall of 0.89 for Limb-Girdle Dystrophies. These novel topological features represent a promising class of biomarkers. This framework introduces a methodology for topology-based radiomics, offering new avenues for objective structural feature engineering in neuromuscular diagnostics.

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CoMPaSS-NMD: Directional radiomics as a segmentation-free approach for muscular dystrophy characterisation on T1-weighted MRI

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Keywords: radiomics, MRI, muscular dystrophy, statistical analysis

Abstract

Fatty infiltration in Muscular Dystrophy (MD) often prevents accurate muscle segmentation. We propose a framework using directional radiomics to characterise disease patterns, bypassing the need for individual muscle delineation.

The original dataset consisted of 2,017 T1-weighted MRI scans. A set of 651 thigh subvolumes was automatically established from the dataset using the SARA framework. The dataset was split into training, validation, and hold-out subsets containing 385, 136, and 130 scans, respectively. The femoral region was partitioned into five radial segments for radiomic descriptors extraction. The obtained features were aggregated over both legs and the axial cross-sections using 10th, 25th, 50th, 75th, and 90th percentiles. Statistical analysis employed the Common Language Effect Size (CLES) for pairwise and One-versus-Rest (OvR) comparisons among 5 MD types: LGMD, DD, DM, FSHD and DMD.

The analysis identified a substantial number of features (pairwise 72.8% and OvR 52.1% of 4,550) exhibiting a large effect size (CLES >= 0.71) for at least one comparison. The OvR analysis demonstrated a correspondence between the segments with the largest CLES features and established clinical knowledge regarding muscle involvement. Directional radiomics provides a segmentation-free alternative for the characterisation of the MD. This constructed feature space shows high potential for prediction and quantification of the disease involvement.

Acknowledgments: We express our deepest gratitude to the MYO-Guide Consortium and all the clinicians worldwide who have contributed to this initiative by sharing muscle MRIs of patients. This work was financed by CoMPaSS-NMD, Computational Models for New Patients Stratification Strategies of Neuromuscular Disorders, HORIZON RIA, Tools and Technologies for Healthy Society, ID: GAP-101080874.

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State Space Models for precise 3D glioma segmentation

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Keywords: glioma segmentation, mamba, state space models, deep learning, MRI

Abstract

Accurate glioma segmentation is a significant challenge due to the complex MRI appearance of gliomas. Manual tracing takes hours and varies between experts. While CNNs are reliable, they often lack global context. 3D Vision Transformers capture this context, but at a high quadratic cost.

To address this problem, we explore State Space Models (SSMs), specifically the Mamba architecture. By linearizing 3D MRI data into 1D sequences, a novel class of selective SSMs (Mamba) captures global context with linear complexity. We integrated Mamba 1.0 and 2.0 blocks into a U-Net backbone and tested different placements using the BraTS 2021 dataset (T1, T1ce, T2, FLAIR). We evaluated our approach against a 3D Swin Transformer and the nnU-Net gold standard using Dice scores.

Our findings show Mamba 2.0 consistently outperforms the Swin Transformer baseline. The best configuration achieved Dice scores of 0.938 (WT), 0.932 (TC), and 0.873 (ET). These results closely match the nnU-Net gold standard (WT: 0.944, TC: 0.937, ET: 0.900) while offering superior linear efficiency. Notably, Mamba 2.0's multi-head structured state duality led to a significant +0.007 improvement in Tumor Core (TC) compared to Mamba 1.0, highlighting its ability to handle nested tumor structures.

Mamba 2.0 provides a scalable, highly competitive alternative to current architectures, serving as a robust foundation for automated radiomics.

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Dual-stage segmentation of DNA plasmids from atomic force microscopy images

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Keywords: atomic force microscopy (AFM), image segmentation, plasmid DNA, YOLO, automated analysis

Abstract

Analyzing radiation-induced DNA damage in plasmids frequently relies on Atomic Force Microscopy (AFM). This enables the investigation of the impact of radiotherapy on DNA. However, manual evaluation is time-consuming. Automating plasmid segmentation is essential, but severe AFM instrumental noise and surface artifacts render classical, single-step processing methods insufficient.

Images of pUC19 plasmids, irradiated with 0–50 Gy doses, were acquired via AFM located at the Jagiellonian University (Cracow, Poland). Scans varied in physical size (1x1 to 3x3 µm). A dual-stage segmentation pipeline was developed. First, a YOLO model was trained to detect and crop individual plasmids. Second, pixel-level segmentation of the crops was performed using Frangi vesselness filtering followed by Li's minimum cross-entropy binarization.

The quality of the proposed pipeline was evaluated using standard quantitative metrics. For the initial detection stage, the optimal YOLO architecture was selected based on achieving high values of recall, precision, and mean Average Precision (mAP). For the second segmentation stage, the generated automated masks were compared against manually annotated ground truth data. Intersection over Union (IoU) and Dice similarity coefficients were used to quantify spatial agreement with manual annotations.

The proposed solution successfully automated plasmid segmentation despite noisy AFM backgrounds.

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Chest X-Ray–predicted age as a biomarker for short- and long-term mortality: Development and validation of a deep learning model

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Keywords: biological age, chest radiograph, deep learning, CXR-age gap, mortality prediction, convolutional neural network, DenseNet-121, medical imaging biomarker, risk stratification, cardiopulmonary aging, radiological age estimation, all-cause mortality, prognostic imaging, biological age estimation, chest X-ray

Abstract

Background: Chronological age is an imperfect proxy for physiological health. Biological age estimated from medical imaging may more accurately reflect an individual's true health status and mortality risk. This study developed a deep learning model to estimate age from chest radiographs (CXR) and evaluated whether the discrepancy between predicted "CXR-age" and chronological age predicts all-cause mortality.

Methods: We collected 404,812 frontal CXRs from five databases (MIMIC-CXR, CheXpert Plus, PadChest, RexGradient, CASIA CXR). A DenseNet-121 CNN was trained on 80% of images, with a dual-input architecture incorporating sex and race/ethnicity. The CXR-age gap (predicted minus chronological age) was assessed as a predictor of 30-day, 90-day, and 1-year all-cause mortality using multivariable logistic regression and Cox proportional hazards models. *Results:* The model predicted age with MAE 3.6 years and r=0.85. Mean CXR-age gap was −0.2 years (SD 4.6). Each 5-year increase in CXR-age gap was independently associated with higher mortality at 30 days (aOR 1.22, 95% CI 1.19–1.25), 90 days (aOR 1.25, 95% CI 1.22–1.28), and 1 year (aHR 1.18, 95% CI 1.16–1.20). Adding the gap to a baseline clinical model improved 1-year mortality discrimination (AUC 0.79→0.83). Saliency maps highlighted the heart, mediastinum, and pulmonary vasculature. *Conclusions:* The CXR-age gap is an independent, incremental predictor of short- and long-term mortality. This accessible imaging biomarker could meaningfully support clinical risk stratification and decision-making.

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Emphysema as a significant risk factor for lung cancer: preliminary results from the Pilot National Lung Cancer Screening in Poland

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Keywords: lung cancer screening, emphysema, low-dose computed tomography (LDCT), risk factors, odds

Abstract

Lung cancer is the leading cause of cancer-related mortality globally. Low-dose computed tomography (LDCT)-based screening programs, including the Polish Pilot National Lung Cancer Screening Program, enable early detection of precancerous changes. This study investigates whether emphysema represents a significant lung cancer risk factor based on findings from this program.

The study included participants who joined the program, underwent an LDCT chest scan, and attended a follow-up appointment to discuss the results. The LDCT scans were reviewed and annotated by qualified radiologists. The risk of developing emphysema was calculated for the groups, and the odds ratio (OR) was assessed using data from the Northern Macroregion.

The final assessment included 1438 women (48.4%) and 1536 men. The mean age across the cohort was 63.3±6.0 years (median 63.7). Women and men averaged 36.2 and 36.4 pack-years respectively (overall mean 36.3±16.6, median 35.0). T-tests confirmed no significant difference between the sexes in either age or smoking history.

The group diagnosed with cancer consisted of 30 women (52.6%) and 27 men. The odds ratio of emphysema as a risk factor for lung cancer was significant and equal to 2.85 (small effect size), with a value of 2.28 for women and 3.87 for men.

The findings indicate a statistically significant correlation between the presence of emphysema and the diagnosis of lung cancer within the screening program.

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Age-related differences in paediatric brain morphology: A comparative analysis of global tissue volumes and subcortical structures

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Keywords: MRI, brain segmentation, paediatric brain volumetry, anatomical altases

Abstract

Introduction:
Brain maturation involves region-specific structural changes across development. This study was aimed to assess age-related differences of global brain tissue volumes and subcortical structures.
Materials and Methods:
The study included 147 healthy children (6-18 y), divided into three equinumerous age groups representing different stages of development. MRI-based volumetric analysis included grey matter (GM), white matter (WM), cerebrospinal fluid (CSF) and 14 subcortical structures. Global tissues segmentation was performed in SPM (MATLAB) using ICBM template, while subcortical structures in FSL. Group differences were assessed using one-way ANOVA with Tukey post-hoc tests. False discovery rate correction (Benjamin-Hochberg) was applied for multiple comparisons across the subcortical structures, while global tissue measures were analysed separately without additional correction.
Results:
A significant age effect was observed for WM, with lower values in the youngest group, compared to older groups, while no significant differences were found for GM and CSF. After FDR, only bilateral pallidum and right amygdala remained significant, with differences driven by contrasts between the youngest and older groups.
Conclusions:
Age-related brain changes are selective rather than global, with the strongest effects observed in WM, pallidum, and right amygdala, highlighting necessity to consider alternative approaches to age modelling, such as use of age as a continuous variable.

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Evaluation of image quality and mean glandular dose in grid and non-grid mammography techniques

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Keywords: mammography techniques, mean glandular dose, prime technology, grid in technique

Abstract

Background and Aims:

Virtual grid techniques are becoming more common in clinical imaging. Siemens' PRIME (Progressive Reconstruction Intelligently Minimizing Exposure) aims to replace physical anti-scatter grids with software-based scatter subtraction in digital mammography to reduce radiation doses. This study evaluates the impact of physical versus virtual grids on image quality and mean glandular dose (MGD).

Materials and Methods:

Experiments were performed using Siemens Revelation and B.Brilliant mammography units. Image quality was evaluated through Contrast-to-Noise Ratio (CNR) with a 0.2mm aluminum square placed in PMMA. The obtained CNR values were corrected by the threshold contrast value and compared to European breast screening guidelines. MGD was calculated using Dance dosimetry model, both with and without physical anti-scatter grids.

Results:

MGD showed a positive correlation with breast thickness across all devices and techniques. The PRIME option resulted in a 16% dose reduction at 20mm thickness on both systems. The dose benefits from virtual grid technique decreased as breast thickness increased. At PRIME upper limits (70mm for Revelation; 40mm for B.Brilliant), it showed no dose reduction. Additionally, a slight decline in CNR values was observed across the entire range of breast thicknesses.

Conclusions:

The clinical advantages of the PRIME technique seem limited in routine mammography. This is due to the low prevalence of 20 mm breast thicknesses in clinical practice and the lack of significant dose savings at the upper limits of its typical use.

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Patient-specific lung CT quantification

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Keywords: LDCT, lung cancer, emphysema, lung diseases, machine learning

Abstract

Quantitative lung CT usually reports disease burden as a percentage of reference parenchyma derived from lung-healthy panels, rather than from each patient's true structural capacity. Literature-based reference panels typically report mean CT-derived inspiratory lung volumes of roughly 4.5–4.7 l in women and 4.6–6.2 l in men, with sex-, height- and age-specific prediction equations defining broad "normal" ranges. Such panel-based denominators, obtained in healthy cohorts, are then applied unchanged to high-risk populations, which can bias percentage disease scores for emphysema, scarring, fibrosis, or nodules.

This work trains a sex-stratified linear model on low-dose CT scans from two high-risk cohorts (DUKE and Moltest 1; 1298 men, 1445 women), using patient's LDCT-derived lung volume to estimate their parenchyma volume directly. In this population, mean lung volumes were 6.28 l (95% CI 6.22–6.34) in men and 4.68 l (95% CI 4.64–4.73) in women, and learned relationships were: for men, parenchyma = −72.74 + 0.9713·lung_volume; for women, parenchyma = −56.77 + 0.9710·lung_volume. Resulting patient-specific estimates can be combined with volumes of pathological changes from external tools to obtain percentage disease scores that better reflect parenchymal capacity. Model also yields tight uncertainty bounds, with maximum 95% interval widths of 19.3 ml in males and 15.5 ml in females, offering narrower and more informative confidence intervals than panel-based normalization.

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Segment Anything Model evaluated on medical segmentation dataset

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Keywords: medical image segmentation, breast cancer, segment anything model

Abstract

Segment Anything Model is a foundation model released by Meta, which aims to generalize the task of image segmentation and achieve good results without the need for fine-tuning. While many dedicated models exist for precise tumor segmentation, this study aims to verify the viability of SAM as a tool for automatic tumor delineation on MRI images.

The study uses images from the BraTS 2020 dataset, which contains 1251 cases across 4 modalities: T1, T1ce, T2, FLAIR. It evaluates different prompting choices required to use SAM for automatic segmentation. It examines the point, bounding box, and mask prompts, as well as the model's automatic segmentation mode.

The results show that bounding-box prompting is highly sensitive to precise contour definition. It has been shown that, although the model supports a mask prompt, it is incompatible with a standardized binary mask format (1 for ROI, 0 for background). Choice of a larger backbone did not yield significantly better results in the simulated sample. The relationship between the point prompt number and the Dice index has been tested; it suggests that the model's performance might be affected not only by the prompt content but also by the order in which the points are provided.

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Automatic detection of stenosis in coronary vessels from angiographic images

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Keywords: stenosis, coronary angiography, automatic stenosis detection

Abstract

Detection of stenosis, which is abnormal narrowing of blood vessels, plays an important role in the assessment of cardiovascular conditions. This research proposes a method for automatic stenosis detection that is based on skeletonization, distance transform analysis and both local and global comparison algorithms. The data on which experiments have been performed were obtained from two datasets (ICA and DCA1), which consisted of 751 images.

The approach was based on geometrical analysis of vessel masks. It folllows a general pipeline consisting of skeletonization, division of vessels into smaller parts, filtering them by their thickness, and calculating distance transforms, enabling estimation of vessel diameter. In the next step local and global strategies were applied to ensure reduced number of false stenosis detection.

Obtained results indicate that the proposed method is capable of detecting stenosis in a consistent way. The algorithm detects both very wide and smaller regions of stenosis with keeping a low number of false positives in most cases. Such results demostrates a potential to use this approach as a supportive tool in angiography image analysis.

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Influence of time-point selection on organ dose assessment in patients treated with ^{177}Lu-DOTATATE

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Keywords: ^{177}Lu-DOTATATE treatment, SPECT/CT dosimetry, time points optimization

Abstract

Introduction

The implementation of individual dosimetry in radioisotope therapy is crucial for optimizing treatment. This study aimed to evaluate the impact of the number of time points (TP) in SPECT/CT imaging on the accuracy of absorbed dose estimation in organs at risk for patients treated with ^{177}Lu-DOTATATE.

Materials and Methods

The analysis used the SNMMI ^{177}Lu Dosimetry Challenge 2021 dataset, which includes raw and processed imaging data from two patients. The dataset provides serial planar and SPECT/CT images acquired at four TP after radiopharmaceutical administration. Absorbed dose calculations were performed using MIM Version 7.4.3, exploring various configurations with one to four TP and different fitting models for time–activity curves.

Results

In the results, differences relative to the full set and other TP combinations were observed, indicating slight variability in estimates of absorbed dose for critical organs (the liver, kidneys, and spleen) across different TP numbers. Reducing the number of measurements to 2–3 results in a max. 10% deviation in the dose estimates. The largest deviation 20% was observed at a single TP (24 h).

Conclusion

This study shows that reducing imaging from 4 to 3 or even 2 TP is justified. This will reduce the time and cost of patient care and minimize the patient's discomfort associated with the imaging procedure. Increasing the number of clinical cases will enable verification of whether the observed findings are confirmed by a statistical analysis.

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Implant landmark detection and implant-aware bone segmentation in postoperative hip AP radiographs

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Keywords: implant landmark detection, bone segmentation, deep learning, artificial intelligence, medical image analysis, hip arthroplasty, pelvis X-ray

Abstract

Hip arthroplasty is one of the most common orthopedic procedures, and postoperative radiographic follow-up remains challenging because implants alter image appearance and obscure adjacent anatomy. This study evaluated an initial AI-based approach for implant landmark detection and bone segmentation in postoperative hip radiographs.
This study used postoperative AP pelvis radiographs and two open-source datasets: one containing implant crops for aseptic loosening assessment and one containing AP pelvis radiographs with annotated bone structures. For implant landmark detection, U-Net and HRNet were tested, while bone segmentation models used ResNet and EfficientNet encoders.
Landmark detection achieved acceptable measurable errors, and bone segmentation reached satisfactory overlap on non-implant data, while postoperative cases with real implants showed visually useful results and supported further geometric analysis.
These findings suggest that combining implant landmark detection with implant-aware bone segmentation is feasible and may support future automated and more comprehensive postoperative hip radiograph analysis.
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Coupled application of GANs and YOLO models for data augmentation and automated detection of DNA plasmids in AFM imagery

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Keywords: atomic force microscopy (AFM), generative adversarial network (GAN), data augmentation, YOLO, plasmid DNA

Abstract

The analysis of DNA plasmid morphology based on atomic force microscopy (AFM) images provides crucial structural information reflecting the state of individual biomolecules. However, AFM images are typically characterized by high noise levels and uneven backgrounds, which hinders automatic image analysis, particularly when employing classical computer vision methods. To address these limitations, deep learning object detection models, such as YOLO, offer a promising alternative for automating the extraction of individual plasmids. Training such models, however, requires the prior preparation of an extensive training dataset - specifically, the manual annotation of bounding boxes for each plasmid - which is a tedious and time-consuming task. To improve YOLO performance, this study focuses on advanced generative data augmentation using Generative Adversarial Networks (GANs).

The analyzed dataset included AFM images of pUC19 plasmids deposited at mica and acquired using Peak Force Tapping mode. The effectiveness of different approaches was evaluated using standard quantitative object detection metrics, including precision, recall and mean average precision (mAP), as well as by a qualitative visual assessment of the generated images.

Expanding the training dataset by a few percent through the addition of synthetic plasmids resulted in an improvement in model performance, particularly enhancing

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recall and mAP. Ultimately, the study outlines the advantages of GAN-based augmentation.

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Validation of automatic segmentation of subcortical structures using multiple filtering methods compared to manual delineation in multi-parametric MRI analysis

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Keywords: magnetic resonance imaging, brain segmentation, automatic segmentation, subcortical structures

Abstract

Magnetic resonance imaging (MRI) is a non-invasive technique widely used to assess morphological changes in the brain. Given the critical role of subcortical structures in motor control and learning, their automated segmentation remains a key research priority.

This study aimed to evaluate differences between manual delineation and automated segmentation of subcortical structures in brain MRI images subjected to various filtering techniques.

The dataset consisted of T1-weighted MRI scans from 38 healthy volunteers, acquired at MSC-NRIO in Gliwice, Poland. To establish a ground truth, three specialists independently delineated the thalamus, putamen, and the caudate nucleus. Final masks were generated by combining individual annotations using a majority voting scheme. Before segmentation, ten different filtering algorithms were applied to each subject's images. Automatic segmentation was then conducted on the pre-processed images

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using FSL software. The Dice similarity coefficient was employed to quantify the overlap between automatic and manual segmentations.
Automatic segmentation methods employing various filtering algorithms produced statistically significant differences in the volumes of general and subcortical structures. However, statistical tests did not identify a single optimal filtering technique. Quality metrics, including the Dice index and Intersection over Union (IoU) enabled a performance-based ranking to identify the most effective approaches. Results were also compared using the BRISQUE non-reference metric.
Acknowledgments: This work was supported by XIV Project Based Learning (PBL) (Initiative of Excellence - Research University Program), in accordance with regulations no. 251/2024 issued by the Rector of the Silesian University of Technology on December 18, 2024. Calculations were carried out using the computer cluster Ziemowit funded by the Silesian BIO-FARMA project No. POIG.02.01.00-00-166/08 in the Biotechnology Centre in the Silesian University of Technology.

Parallel session (3A)
time 17:00 – 19:15

Chairperson:
Aleksandra Suwalska

Alternative splicing landscapes across heart and skeletal muscle reveal mechanisms of development and disease

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Keywords: neuromuscular diseases, muscle development, muscle ageing, alternative splicing

Abstract

RNA splicing is the process by which introns are removed and exons are joined to generate mature mRNA transcripts. Alternative splicing enables the production of diverse mRNA isoforms through mechanisms such as exon skipping, alternative 3' and/or 5' splice site usage, mutually exclusive exons, and intron retention.

We have demonstrated that investigating splicing regulation in muscle function–associated genes, including TTN and OBSCN, across skeletal and cardiac muscle development provides critical insights into both normal muscle biology and disease mechanisms. To this end, we analyzed alternative splicing and gene expression across 75 pre- and postnatal skeletal and cardiac muscle RNA-seq samples.

Our findings reveal dynamic isoform remodeling of key sarcomeric genes, such as TTN and OBSCN, during muscle development and ageing. This isoform-level characterization improves interpretation of clinical phenotypes in muscle disorders, including predicting disease onset in titinopathy patients. Furthermore, our analysis enables estimation of the population frequency of rare splicing variants, exemplified by a pathogenic TNNT1 splicing variant identified in a patient with severe nemaline myopathy.

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Autophagy acts as a molecular switch controlling fibroblast phenotypic reprogramming

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Keywords: autophagy, epithelial–mesenchymal transition (EMT) phenoconversion, wound healing, human foreskin fibroblasts

Abstract

Fibroblast plasticity critically regulates tissue repair and fibrotic remodeling by enabling transitions between quiescent, activated, and mesenchymal-like states that control extracellular matrix (ECM) deposition and tissue architecture. Although autophagy is a key regulator of cellular homeostasis and stress adaptation, its role in fibroblast phenotypic transitions remains unclear. We investigated the effect of autophagy modulation on fibroblast plasticity using human dermal fibroblasts (Hs27, Hs68). Autophagy was inhibited with Bafilomycin A1 (5 nM) and activated with Rapamycin (500 nM). Autophagic activity was assessed through LC3B-II and p62 dynamics, while phenotypic reprogramming was evaluated via morphological analysis and expression of key epithelial–mesenchymal transition (EMT)-associated markers. Autophagy inhibition resulted in LC3B-II and p62 accumulation, confirming impaired flux, and pronounced a mesenchymal-like phenotype with increased Vimentin and reduced E-cadherin expression, consistent with a pro-fibrotic state. Conversely, autophagy activation mitigated mesenchymal markers and promoted a contractile phenotype, suggesting a regulatory role in fibroblast activation. Collectively, autophagy acts as a regulator of fibroblast state transitions by limiting mesenchymal reprogramming and supporting alternative differentiation states. These findings provide an in vitro platform for studying fibroblast dynamics in wound healing and fibrosis and highlight autophagy modulation as a potential therapeutic strategy to control pathological tissue remodeling.

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Integrative multi-omics reveals FRG2A lncRNA-driven nuclear reorganization in facioscapulohumeral muscular dystrophy

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Keywords: lncRNA, nucleolus, genome architecture

Abstract

Facioscapulohumeral muscular dystrophy (FSHD) is associated with contractions of the D4Z4 macrosatellite at chromosome 4q35, leading to chromatin alteration and dysregulation of nearby genes. Among these, we discovered that FRG2A is part of a family of long non-coding RNAs (lncRNAs) expressed in skeletal muscle, with levels varying among patients. FRG2A localizes to the nucleolus and associates with repetitive DNA at rDNA loci and centromeres, suggesting a role in higher-order genome organization. FRG2A overexpression in FSHD cells alters heterochromatin organization at the nucleolar periphery, reducing rDNA transcription, translation rates, and protein synthesis during myogenic differentiation.

To assess genome-wide effects, we performed ATAC-seq in primary myoblasts and differentiated myotubes, identifying a stage-specific chromatin accessibility state in proliferating FSHD myoblasts with over 12,000 regions remodeled and attenuated upon differentiation. Changes mainly affect intronic and distal intergenic regions enriched in non-coding and repeat-proximal sequences, clustered within heterochromatin- and nucleolus-associated domains, indicating coordinated nuclear architectural reorganization rather than a canonical transcriptional program.

Together, our results support a disease mechanistic model in which a D4Z4-derived lncRNA contributes to FSHD pathogenesis by reshaping nuclear organization and modulating non-coding chromatin landscapes, ultimately impairing protein synthesis.

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Expression level of TP53 and KDM5B as a promising DNA-damaging agent response biomarker in cancers

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Keywords: DNA damage response, cancer chemotherapy, drug resistance, epigenetics

Abstract

The tumour suppressor p53 is widely recognized as a transcriptional activator and the mechanisms underlying its repressive function remain incompletely defined. Here, we uncover a chromatin-based mechanism by which p53 directly suppresses a subset of E2F1-driven promoters despite co-occupancy with the co-activator p300. These promoters exhibit a distinct repressive chromatin architecture marked by low H3K27 acetylation, and depletion of H3K4me3. We show that p53 enforces this state by sustaining promoter-associated KDM5B, thereby promoting H3K4me3 demethylation and transcriptional silencing. Activation of the ATM/ATR–Chk1/2–p53 pathway by cisplatin disrupts this axis, triggering p53 and KDM5B eviction, chromatin relaxation, and rapid acquisition of transcription-permissive histone modifications. This switch enables p300 enrichment and transcriptional activation of genes promoting cancer cell survivalence. Notably, the p53–KDM5B regulatory axis is conserved across diverse cancer cell types and functionally linked to chemotherapy response. In summary, our findings define p53-mediated repression as a dynamic process regulated by chemotherapy, which could be exploited clinically. Expression level of TP53 and KDM5B may be a candidate biomarker of chemotherapy sensitivity, with potential clinical implications.

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The CoMPaSS-NMD neuromuscular genome atlas: A new AI-based platform for advanced deep phenotyping and stratification in hereditary neuromuscular disorders

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Keywords: neuromuscular diseases, neurology, histology, genetics, myology

Abstract

Hereditary neuromuscular disorders (HNMDs) are characterized by marked clinical and molecular heterogeneity, which limits accurate diagnosis, prognosis, and patient stratification. Addressing these challenges requires large-scale, harmonized, and interoperable integration of multimodal data across centers. The CoMPaSS-NMD Neuromuscular Genome Atlas (NMDGA) aims at establishing a federated, GDPR-compliant infrastructure to integrate multimodal data and enable ontology-driven and AI-ready patient stratification. The Atlas integrates collected clinical, genetic, histopathological, and MRI data from 500 undiagnosed patients and deploys AI-based analytical tools developed and trained using large retrospective cohorts. Clinical phenotypes collected through a structured electronic case report form (>200 parameters) were systematically mapped to the Human Phenotype Ontology (HPO), resulting in the integration of 203 HPO terms and the proposal of eight novel neuromuscular-specific terms. This semantic layer enables hierarchical phenotype navigation, ontology-aware filtering, and cross-domain queries combining phenotypic, genetic, imaging, and histopathological features. The NMDGA provides a shared, web-accessible resource for multimodal neuromuscular data, integrating harmonized phenotypes with

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computational analysis and visualization tools. The deep phenotyping enables precise cohort definition, improved genotype-phenotype correlation, and robust patient stratification, supporting translational research and future clinical trials.

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Replication stress in hormone-dependent HER2-positive breast cancer: New perspectives for targeted treatment

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Keywords: replication stress, breast cancer, targeted therapy, AI-assisted drug discovery

Abstract

Replication stress is a key driver of genomic instability, cancer progression, and treatment resistance, yet its role in hormone-dependent HER2-positive breast cancer remains insufficiently characterized. This clinically challenging subtype, despite benefiting from endocrine and HER2-targeted therapies, still shows substantial relapse rates and limited durable responses. In this study, we explored replication stress pathways as potential therapeutic targets in hormone-dependent HER2-positive breast cancer. Candidate compounds were selected using AI-assisted screening and evaluated in malignant and non-malignant breast cell models. Their biological effects were assessed using flow cytometry, RT-PCR, Western blot, and ELISA, with particular focus on molecular markers associated with replication stress response. The identified compounds promoted selective cancer cell death accompanied by enhanced replication stress signaling, indicating previously unrecognized therapeutic opportunities in this breast cancer subtype. Moreover, selected agents showed promising combinational activity with standard chemotherapeutics, supporting their translational relevance. Our findings suggest that replication stress modulation may provide a new direction for targeted therapy in hormone-dependent HER2-positive breast cancer and highlight the value of integrating computational approaches with experimental oncology in the development of precision treatment strategies.

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Sleep disturbances in individuals with type 1 diabetes and their caregivers - cross-sectional quantitative study

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Keywords: sleep disturbances, type 1 diabetes, insomnia, caregivers, insulin therapy

Abstract

Sleep disturbances are common in type 1 diabetes (T1D) due to nocturnal glycemic variability, treatment burden, and psychological stress, affecting both patients and caregivers.

Aim: To assess sleep quality and consequences of sleep deprivation in individuals with T1D and caregivers, in relation to intensive insulin therapy modalities.

Methods: A cross-sectional online survey was completed by 167 participants (93 individuals with T1D, 74 caregivers). Sleep quality was assessed using the Athens Insomnia Scale (AIS) and sleep deprivation burden using the CHICa scale. Associations with intensive insulin therapy modalities (multiple daily injections, continuous subcutaneous insulin infusion (CSII), including standard insulin pump therapy and automated insulin delivery (AID)) were analyzed.

Results: Probable clinical insomnia was significantly more prevalent among caregivers than individuals with T1D (58.9% vs 23.7%; $p < 0.001$). Caregivers also reported higher CHICa scores (34.0 ± 13.3 vs 31.2 ± 14.5). No significant associations were found between insulin therapy modality and sleep outcomes. A trend toward greater insomnia severity was observed in caregivers of children using "standard" insulin pumps compared to those with AID function. Fear of nocturnal hypoglycemia and frequent nighttime monitoring were associated with poorer sleep.

Conclusions: Sleep disturbances are a substantial burden in T1D, particularly among caregivers. Advanced technologies alone may not improve sleep, highlighting the need for interventions targeting nocturnal anxiety and caregiving burden.

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From clinical evidence to redefining precision oncology: The HER2-low paradox and intelligent therapeutic design in breast cancer

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Keywords: HER2 expression, hormone-dependent breast cancer, bystander effect, bioconjugates, (bio)polymeric nanoparticles, patient-derived organoids

Abstract

Recent results from the DESTINY-Breast04 clinical trials have fundamentally challenged the target-centric paradigm of precision oncology. In HER2-low breast cancer, clinically meaningful responses to antibody–drug conjugates (ADCs) such as trastuzumab deruxtecan have been observed despite minimal target expression, revealing a striking paradox: therapeutic efficacy can be achieved even when the classical molecular target expression is negligble. This observation suggests that treatment outcomes are increasingly governed not by target abundance, but by the design of drug delivery systems and their interaction with tumor biology. However, current ADCs remain limited by i.e. systemic toxicity and suboptimal patient stratification. Addressing these challenges requires a shift toward more intelligent, integrative therapeutic design. Here, we propose a next-generation framework combining AI-driven optimization of drug combinations, nanocarrier-based delivery systems, and patient-derived organoids as high-fidelity functional models. Within this approach, organoids enable patient-specific validation of therapeutic response, while AI supports the identification of predictive biomarkers and synergistic treatment strategies. Together, these technologies define a new direction for precision oncology — one that moves beyond static targets toward dynamically designed, patient-adapted therapeutic systems.

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Reprogramming antibacterial therapy: AI-guided strategies for personalized treatment of chronic wound infections

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Keywords: chronic wound infections, antimicrobial therapy, artificial intelligence, drug delivery

Abstract

Chronic wound infections, particularly diabetic foot infections, represent a major clinical challenge due to their heterogeneity, polymicrobial composition, and increasing antimicrobial resistance. Standard treatment strategies often fail to account for patient-specific factors, including microbiome variability, biofilm formation, and impaired tissue regeneration, highlighting the need for personalized therapeutic approaches.

This work presents a multidisciplinary framework for developing personalized antibacterial therapies by integrating artificial intelligence (AI), nanotechnology, and human-relevant experimental models. Computational methods are employed to identify and prioritize small-molecule compounds active against Gram-positive pathogens and to predict effective combination therapies tailored to specific infection profiles.

To better reflect the clinical complexity of diabetic wounds, advanced in vitro models are utilized, including 3D human skin equivalents and wound-mimicking systems. These platforms enable evaluation of antibacterial activity, biofilm disruption, and tissue compatibility under physiologically relevant conditions. In parallel, nanotechnology-based delivery systems are explored to enhance local drug concentration, improve tissue penetration, and increase treatment efficacy.

This integrative approach aims to bridge computational prediction with experimental validation, providing a foundation for precision antimicrobial therapy tailored to individual patients and specific wound environments.

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Determinants of delayed diagnosis of oral cavity cancer and strategies for their mitigation using AI in clinical practice

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Keywords: AI, oral cavity cancer, diagnosis delay

Abstract

Introduction: Oral cavity cancer is the sixteenth most common malignancy worldwide. Despite a moderate incidence rate (5.6/100,000), it has a high mortality rate, reflected in a mortality-to-incidence ratio of 46%. Although well recognized, diagnostic intervals often exceed five months. Artificial Intelligence (AI) offers potential to reduce time-to-treatment.

Aim: This study aims to identify and analyze the clinical and systemic determinants of diagnostic delays in oral cavity cancer, while exploring strategies to reduce time-to-diagnosis interval focusing on opportunities offered by AI-supported approaches.

Materials and methods: An extensive literature review of the literature was performed, synthesizing evidence from systematic reviews and meta-analyses to evaluate factors contributing to diagnostic delay, their impact on prognosis, and AI-assisted interventions to reduce the time-to-treatment interval.

Conclusion: Despite existing clinical initiatives, oral cavity cancer diagnostic delays remain suboptimal. This underscores the need for multidimensional strategies addressing modifiable determinants of delay through education, optimized referral pathways, and heightened oncological vigilance. Furthermore, integrating validated Artificial Intelligence (AI) tools is essential to improve diagnostic sensitivity and specificity, ultimately reducing delay and enhancing patient survival.

Acknowledgments: Department of Public Health and Social Medicine, Student Scientific Circle Interdisciplinary Healthcare Management

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Impact of atmospheric pressure variability and time of hospital admission on the incidence and clinical course of acute myocardial infarction

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Keywords: acute myocardial infarction, meteorological factors, hospital admission time, clinical parameters, mortality risk

Abstract

Acute myocardial infarction (AMI) remains a leading cause of morbidity and mortality worldwide. In addition to established cardiovascular risk factors, environmental and clinical variables may influence the incidence and outcomes of AMI. The aim was to assess the impact of meteorological factors, hospital admission time, and selected clinical parameters on AMI incidence and survival in patients from Gliwice County. 3778 patients with AMI were analyzed and stratified into STEMI and NSTEMI groups. Meteorological and clinical data were integrated. Survival analysis was performed using the Kaplan–Meier method with log-rank testing, along with statistical comparisons. In the NSTEMI group, decreases in atmospheric pressure exceeding 5hPa were associated with a significant increase in AMI incidence, whereas increases exceeding 5hPa were associated with a reduction in incidence. Men over 60 demonstrated increased susceptibility to pressure decreases. Daytime AMI onset was associated with better renal function and more stable electrolyte profiles, whereas nighttime onset correlated with elevated inflammatory markers. Survival outcomes differed significantly according to sex, age, and selected clinical variables. Reduced LVEF and impaired renal function were independently associated with worse prognosis. Rapid changes in atmospheric pressure may represent non-classical risk factors for AMI in NSTEMI patients. Survival is influenced by multiple factors, including sex, LVEF, and renal function, highlighting the importance of integrated prognostic models in the management of AMI.

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Integrative transcriptomic analysis reveals shared molecular signatures and regulatory networks in tongue squamous cell carcinoma across diverse populations

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Keywords: tongue cancer, biomarkers, bioinformatics

Abstract

Background: Tongue cancer is a prevalent oral malignancy with significant global health burden. Identifying molecular signatures across populations is essential for developing diagnostic and therapeutic strategies.

Objective: To identify common differentially expressed genes (DEGs) in tongue cancer across China, USA, and Australia, and elucidate their functional roles through integrative bioinformatic analysis.

Methods: Transcriptomic datasets were retrieved from GEO database. Common DEGs were identified via Venn analysis, followed by functional enrichment using GO, KEGG, and DO databases. PPI and gene–miRNA networks were constructed using NetworkAnalyst.

Results: Analysis identified 133 common DEGs (104 protein-coding; 29 non-coding). Key pathways included extracellular matrix disassembly, collagen catabolism, and IL-17/HPV signaling. Hub genes were MMP10, MMP3, COL1A1, CDKN2A, CCNA1, SERPINE1, STC2, and CXCL11. Non-coding RNAs (TENM3-AS1, LINC00491, DUXAP10) were associated with IL-4/IL-13 immune signaling.

Conclusion: This analysis identifies biomarkers and therapeutic targets in tongue cancer regulating extracellular matrix remodeling and immune pathways, warranting experimental validation.

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The impact of dietary supplements on brain ^{1}H-MRS Spectra – new diagnostic challenges

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Keywords: ^{1}H-MRS, MRS, MRI, brain, methylsulfonylmethane, MSM

Abstract

Proton magnetic resonance spectroscopy (^{1}H-MRS) is a non-invasive method that enables the assessment of tissue biochemical composition by quantifying the concentrations of selected metabolites. In neuroimaging studies, analysis primarily focuses on compounds such as N-acetylaspartate (NAA), creatine (Cr), choline (Cho), myo-inositol (mIns), and the glutamate–glutamine complex (Glx).

This study aims to present the effect of methylsulfonylmethane (MSM) supplementation on brain ^{1}H-MRS spectra. The examinations were performed at the Department of Radiology and Diagnostic Imaging of the Maria Skłodowska-Curie National Research Institute of Oncology in Gliwice, using 3T magnetic resonance scanners in patients undergoing MSM supplementation.

Methylsulfonylmethane can cross the blood–brain barrier and accumulate in brain tissue, generating a detectable signal at approximately 3.15 ppm. Compounds present in dietary supplements may lead to additional peaks in spectroscopic spectra. These signals may overlap with those of key metabolites, making their correct interpretation more difficult and potentially affecting the assessment of results.

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Abemaciclib modulates cell-cycle-regulated genes in endometrial cancer revealed by transcriptome analysis

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Keywords: endometrial cancer, abemaciclib, transcriptome, cell cycle checkpoint

Abstract

Endometrial cancer remains a clinically significant malignancy with limited molecularly characterized responses to targeted therapies. While CDK4/6 inhibitors such as Abemaciclib are well established in breast cancer, their transcriptomic impact in endometrial cancer is not fully understood. In this study, we analyzed publicly available RNA-seq data (GSE247791) to investigate gene expression changes induced by Abemaciclib treatment.

Differential expression analysis identified a distinct set of genes associated with treatment response, including MKI67, CCNA2, CDKN3, and E2F2, which are central regulators of cell cycle progression. In contrast, only a small subset of genes, such as IL1A and SLC6A13, showed reduced expression. Notably, the transcriptional response was characterized by a pronounced enrichment of proliferation- and mitosis-related pathways, alongside an asymmetric pattern favoring gene upregulation.

These findings highlight a context-dependent transcriptional signature of CDK4/6 inhibition in endometrial cancer cells and provide a gene-level framework for understanding therapeutic response. The results suggest potential molecular markers for future investigation and emphasize the importance of cancer-type-specific transcriptomic analysis in precision oncology.

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Uncovering seasonal trends in pediatric type 1 diabetes

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Keywords: type 1 diabetes, seasonality, epidemiology

Abstract

Type 1 diabetes is an autoimmune disease with great increase in incidence among children. This study aimed to investigate trends in incidence and assess any seasonal patterns in type 1 diabetes. Data were obtained from the Silesian region between 1989 and 2025. For each of 4289 patient date of birth, date of first insulin injection and sex were extracted.

In first part of the study, each of years have been treated separately. The values were examined both quantitatively and normalized to percentages to allow comparison between different years. In the second part of experiment all datasets were combined into single unified database in order to observe general seasonal trends of incidence in type 1 diabetes. Additionally, the counts have been standardized with respect to the distribution of births to take structure of underlying population into account.

Obtained results indicated that the number of type 1 diabetes incidences has increased in last 30 years. After careful inspection of the combined dataset, notable seasonal pattern has been observed, with higher incidence rate noticed in winter months compared to the summer period.

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Prediction of insulin resistance based on epidemiological data using machine learning

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Keywords: machine learning, insulin resistance, diet, unsupervised learning

Abstract

Introduction: Insulin resistance is a major risk factor for type 2 diabetes and other metabolic disorders, making its early detection important in preventive and personalized medicine. The aim of this study was to predict insulin resistance using epidemiological data and machine learning methods, and to evaluate the importance of selected health-related and dietary factors.

Methods: The analysis was based on NHANES 2021-2023 data, including 3,158 adult participants and 30 demographic, anthropometric, laboratory, and behavioral variables. Insulin resistance was defined as a binary variable using the HOMA-IR index. Missing values were imputed using the k-nearest neighbors method, and features were standardized. Three models were applied: logistic regression, support vector machine, and random forest. Model performance was evaluated using k-fold cross-validation with balanced accuracy, sensitivity, and specificity.

Results: The random forest model achieved the best performance, obtaining the highest balanced accuracy and showing a good balance between sensitivity and specificity. SHAP analysis showed that anthropometric and metabolic variables, such as BMI, waist circumference, and lipid parameters, had the greatest impact on predictions. Dietary factors had a smaller but still noticeable influence.

Conclusions: The results confirm the usefulness of machine learning methods, particularly ensemble models, in predicting insulin resistance. At the same time, they highlight the limitations of observational data and the need for further research.

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Odontogenic keratocyst (OKC): Radiological features and diagnostic challenges

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Keywords: odontogenic keratocyst, odontogenic cyst, mandible, radiological appearance, diagnostic imaging

Abstract

Odontogenic keratocyst (OKC) is the third most common odontogenic cyst of the jawbones and originates from remnants of the dental lamina. It is more frequently observed in Caucasian males, with peak incidence occurring in the second and third decades of life. This cyst is most commonly located in the body and angle of the mandible. In most cases, OKC develops without noticeable clinical symptoms; however, as it enlarges, manifestations such as bone expansion, pain, swelling, or displacement of adjacent teeth may occur. The aim of this study was to summarize knowledge about OKC and case studies. The radiological appearance of OKC is not pathognomonic and may be easily mistaken for other intraosseous lesions, such as ameloblastoma (AM), even by experienced clinicians. Nevertheless, advances in radiological techniques and the application of newer imaging modalities based on conventional methods have improved diagnostic accuracy. The management of OKC remains challenging due to its relatively high recurrence rate. Accurate diagnosis is essential for appropriate treatment planning, and histopathological examination remains the gold standard for definitive diagnosis. This study summarizes current knowledge regarding the diagnostic imaging of OKC, with the aim of improving diagnostic precision and facilitating appropriate therapeutic management.

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Parallel session (3B)
time 17:00 – 19:15

Chairperson:
Joanna Tobiasz

Redox-active PEDOT antibacterial coatings for bacteria-triggered drug delivery

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Keywords: antibacterial coatings drug release, ceftazidime, conducting polymers, medical devices

Abstract

In this study, we developed a redox-active antibacterial coating based on ceftazidime-loaded PEDOT (PEDOT@CAZ) and evaluated its electrochemical performance, bacteria-triggered drug release, and antibiofilm activity. The coating retained the redox functionality of PEDOT after drug incorporation. PEDOT@CAZ exhibited a charge-storage capacity of 14.6 ± 3.4 mC cm^{-2}, compared with 11.2 ± 2.8 mC cm^{-2} for pristine PEDOT, while the charge-transfer resistance increased from 494 ± 38 Ω to 10.7 ± 0.5 kΩ, consistent with successful antibiotic incorporation into the polymer matrix. Passive ceftazidime release remained low (1.23 ± 0.07 µg cm^{-2} after 60 min), whereas exposure to electroactive bacteria induced substantially higher release, reaching 44.4 ± 9.0 µg cm^{-2} for *Shewanella oneidensis* and 62.2 ± 2.8 µg cm^{-2} for *Pseudomonas aeruginosa*. These findings indicate that bacterial activity can alter the redox state of PEDOT and stimulate localized antibiotic delivery. The PEDOT@CAZ coating also showed strong antibacterial performance at the material interface over an 8-day period, reducing bacterial viability on day 1 to 13% for *S. oneidensis* and 20% for *P. aeruginosa*, while significantly suppressing biofilm formation relative to unmodified controls throughout the study period. Overall, these results demonstrate that redox-active PEDOT-based coatings provide a promising platform for bacteria-responsive antibacterial surfaces with potential applications in biomedical devices.

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Nanostructurization of electrode surface as a crucial role in biosensor fabrication

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Keywords: biosensors, cancer, PEDOT, structurization, microcavities, electrochemistry

Abstract

Cancer is the third leading cause of death worldwide, and early diagnosis is crucial for patient survival. The development of accessible devices for rapid detection of cancer biomarkers could significantly improve treatment outcomes.

Biosensors are an intensively studied area with growing industrial relevance, with the global market estimated to reach 27.5 billion dollars in 2024. Among various materials, poly(3,4-ethylenedioxythiophene) (PEDOT) has attracted significant attention; however, conventional flat PEDOT coatings often suffer from limited sensitivity.

Here, PEDOT coatings with a microcavity architecture were fabricated using a template-assisted method and extensively characterized, including redox mediator response. A functionalized derivative, azido-PEDOT (PEDOT–N_3), was then used to construct a biosensor for thyroglobulin detection via click chemistry.

The microcavity structure increased electrochemical capacity and enabled independent modification of the polymer and substrate, allowing development of a ratiometric sensor. Additionally, the honeycomb-like arrangement of cavities resulted in tunable optical properties, highlighting potential for optical biosensing.

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Quinone-fused diazepine π-conjugated carbon black: A chemically induced, topological-defect-engineered platform for ultrastable redox activity and ultrasensitive electrocatalytic sensing of ascorbic acid

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Keywords: diazepine–quinone redox system, ascorbic acid detection, defect-engineered carbon, nanostructured sensing platforms, surface-confined redox systems

Abstract

Redox-active molecular species functionalized on carbon nanomaterials via covalent or non-covalent (π–π) interactions are widely used in electrochemical applications but often suffer from fouling, low conductivity, and poor stability. Herein, we report a diazepine–quinone (DZ-HQ) system immobilized onto carbon black (CB)-modified glassy carbon electrodes (GCE) via strong π–π interactions (GCE/CB@DZ-HQ). This platform exhibits enhanced structural stability, conductivity, and redox activity, enabling efficient electrocatalytic oxidation and sensitive detection of ascorbic acid (AA). In contrast, direct adsorption on bare GCE showed negligible performance, highlighting the critical role of CB. Characterization (TEM, Raman, FTIR, EIS, EQCM, SECM) confirms robust π–π stacking and defect formation. The modified electrode shows excellent electrocatalytic activity at 0 V vs. Ag/AgCl. Electrochemical studies (CV, amperometry, FIA, BIA) reveal high sensitivity (1 $\mu A \cdot \mu M^{-1} \cdot cm^{-2}$) and a low detection limit (10 nM), with strong selectivity. The sensor was validated in real samples, including serum, fruits, and vegetables. A disposable screen-printed version retained 100% performance over five years, outperforming conventional systems. This CB@DZ-HQ interface provides a robust platform for next-generation electrochemical sensing.

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The effect of incorporating various active substances on the structure and properties of hydrogels

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Keywords: hydrogels, drug delivery systems, natural biopolymers

Abstract

In recent years, hydrogels based on natural polymers have emerged as a promising class of biomaterials for advanced applications in drug delivery. They are characterized by biocompatibility, biodegradability, low toxicity, and structural similarity to the extracellular matrix; therefore, these polysaccharide-based systems provide a favorable microenvironment for the incorporation and controlled release of therapeutic agents. Electrostatic interactions between cationic and anionic polymers enable the formation of polyelectrolyte complexes under mild conditions, leading to the creation of three-dimensional networks with tunable physicochemical properties. In this study, alginate or carrageenan with chitosan hydrogels were designed as matrices for incorporating nanoparticle drug carriers containing hydrophobic active compounds or free drug molecules. The hydrogel network serves as an additional delivery platform, enabling localized administration, improved retention at the target site, and sustained release. The structural characteristics of hydrogels—including chemical composition, swelling capacity, crosslinking density, mechanical stability, and toxicity—are among the key parameters that were evaluated. This study presents the structural and physicochemical differences observed in hydrogels depending on the incorporated substances. Overall, hydrogels offer a versatile and environmentally friendly platform for the development of drug delivery systems. These systems have significant potential to improve therapeutic efficacy while reducing systemic exposure and side effects.

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Personalized ankle–foot orthoses: Design strategies and biomechanical considerations

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Keywords: ankle-foot orthoses, design strategies, personalization methods, biomechanical function

Abstract

Ankle-foot orthoses (AFO) are widely prescribed to improve patient's quality of life. However, studies show that up to 27% of prescribed orthotic devices are discontinued after initial use due to their lack of comfort and poor user perception. Traditional AFO are most commonly manufactured using thermoplastic vacuum forming, but a method called additive manufacturing (AM) aims to solve the usual problems of AFO manufacturing.

The aim of this study was to analyze the design process of personalized AFO and evaluate factors influencing their biomechanical function. A review of selected open access scholarly articles lead to identifying two key aspects: design methods and orthotic parameters. Their impact on movement biomechanics, customization, and accessibility was assessed.

The review revealed that personalization methods in AFO design significantly affect comfort of use, adherence, and consequently the effectiveness of rehabilitation. Off-the-shelf AFO, although inexpensive and readily available are not tailored to individual patients, which limits their functional benefit. Understanding advanced customization strategies provides insights into optimizing orthotic performance offers the potential to enhance patient comfort and daily functioning.

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Mobile solution for Bliss AAC

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Keywords: Augmentative and Alternative Communication, inclusive healthcare technologies, mobile platform, Bliss

Abstract

Augmentative and Alternative Communication (AAC) systems provide essential support therapy and treatment of non-speaking individuals. Among many options, the Blissymbols system shows favorable outcomes for rehabilitation and speech development in autistic communities. Yet despite its modular, component-based design, there are no widely available tools that utilize its structure as symbols linked together to create meaning.

The purpose of this study is to empirically evaluate the novel Blissymbols input and organization method based on their internal components and assess its impact on communication effectiveness. The study aims to determine whether the component-based input and learning approach facilitates faster symbol retrieval, better comprehension, and more effective interaction between the user and the clinician or therapist compared to conventional AAC symbol organization methods. Additionally, the compositional nature of Blissymbols lends itself to a learning paradigm inspired by systems in which complex structures are constructed from simpler elements, analogous to mechanics known from so-called “alchemy”-style games.

To enable controlled experimentation, a prototype mobile tool was developed as a research instrument, supporting bidirectional mapping between natural language, Blissymbols, and speech output. It could prove to be a convenient alternative to dedicated devices or speaking aids, giving nonverbal people a proficient voice in everyday context.

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In vivo range monitoring in upright hadron therapy using J-PET technology: applications to proton peams

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Keywords: hadron therapy, upright proton radiotherapy, Bragg peak, proton range verification, positron-emitting isotopes, Jagiellonian Positron Emission Tomography (J-PET) scanner

Abstract

Hadron therapy, particularly proton beam therapy, is a radiotherapy that utilizes charged particles such as protons and carbon ions to target and treat cancer cells with high precision. The strength of hadron therapy lies in the unique physical and radiobiological properties of these particles that can penetrate the tissues with little diffusion and deposit the maximum energy just before stopping, at a peak shape called Bragg peak. Bragg peak which allows for the delivery of a lethal dose to the tumor while significantly sparing healthy tissue distal to the tumor site. With the use of hadrons, the tumor can be irradiated while the damage to healthy tissues is less than with the conventional photon beams.

Proton interactions in tissue involve both electromagnetic processes, responsible for energy deposition, and nuclear interactions that lead to the production of positron-emitting isotopes such as ^{11}C, ^{15}O, and ^{13}N. These isotopes form the basis for positron emission-based range monitoring techniques. By detecting annihilation photons resulting from β^+ decay and leveraging the principle of modular Jagiellonian Positron Emission Tomography (J-PET) technology based on plastic scintillators, as a cost-effective tool, one can indirectly verify the range of proton beam within the patient. Our proposed approach allows for assessing whether the dose will be delivered exactly where intended, compensating for anatomical variations or setup uncertainties.

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Diffusion-based physiological imputation for robust fetal heart rate baseline estimation

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Keywords: cardiotocography, diffusion, generative AI, fetal health

Abstract

Clinical cardiotocography (CTG) recordings often contain missing data caused by fetal or maternal movement, leading to discontinuities that affect fetal heart rate (FHR) baseline estimation. Traditional interpolation and autoregressive gap-filling methods create oversmoothed and physiologically implausible reconstructions. This study proposes a Conditional Diffusion Imputer (CDI) based on 1D denoising diffusion models to regenerate missing FHR segments using contextual information from uterine contractions (UC). The diffusion imputer is expected to enhance the accuracy and morphological fidelity of baseline estimation algorithms such as ARDSIAsLS and WMFB.

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A computational method for molecular docking analysis of bioactive phytochemicals from jackfruit seed flour (*Artocarpus heterophyllus*) against hepatocellular carcinoma (HEPG2) cell line targets

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Keywords: hepatocellular carcinoma, jackfruit seed, molecular docking, anticancer activity

Abstract

This research employs a comprehensive computational approach that includes molecular docking, drug-likeness evaluation, ADMET profiling, explore the anti-cancer potential of key bioactive phytochemicals derived from jackfruit seed flour (JSF) against confirmed Hepatocellular carcinoma molecular targets. A panel of 12 phytochemicals, including artocarpin, cyclomorusin, morusin, gallic acid, kaempferol, and norartocarpin, was screened against four major cancer-related targets expressed in HepG2 cells: epidermal growth factor receptor (EGFR), vascular endothelial growth factor receptor-2 (VEGFR-2), Bcl-2 anti-apoptotic protein, and phosphatidylinositol 3-kinase (PI3K). Molecular docking simulations using PyRx Virtual Screening Software with the AutoDock Vina engine revealed that artocarpin exhibited the strongest binding affinity for EGFR (-9.4 kcal/mol) and PI3K (-8.9 kcal/mol), surpassing the reference drug erlotinib (-8.2 kcal/mol) in both cases. These results provide a compelling computational basis for considering JSF-derived phytomedicinals as new hepatoprotective and anticancer agents.

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Drug delivery platforms for personalized therapy of hormone-dependent breast cancer

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Keywords: nanotechnology, drug delivery, hormone-dependent breast cancer, precision medicine, nanocarriers

Abstract

Hormone-dependent breast cancer remains a major clinical challenge due to tumor heterogeneity, systemic toxicity of therapy, and the development of therapeutic resistance. Precision medicine approaches require not only the identification of effective drug combinations, but also advanced delivery systems capable of improving selectivity, stability, controlled drug release, and reducing systemic cytotoxicity. In this context, nanotechnology offers new opportunities for the development of multifunctional therapeutic platforms integrating endocrine and chemotherapeutic agents. Our research focuses on the design of nano-enabled hormone-chemotherapeutic systems for targeted treatment of hormone-dependent breast cancer. The proposed strategy combines rational drug selection with the development of polymeric nanocarriers incorporating cyclodextrin-based inclusion complexes to enhance drug accumulation in tumor tissue and reduce off-target toxicity. In addition, surface functionalization with HER2-targeting antibodies will be explored to promote selective delivery to HER2-overexpressing tumor cells. Particular emphasis is placed on optimizing drug loading, controlling release profiles, and evaluating biological activity in cellular models of hormone-dependent breast cancer.

This approach integrates nanotechnology with modern anticancer therapy design and highlights the potential of advanced drug delivery systems to improve therapeutic selectivity and overcome limitations of current breast cancer treatments.

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Enhanced cell adhesion on polypyrrole-modified poly(ϵ-caprolactone) fibers for neural tissue engineering applications

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Keywords: neural tissue engineering, electrospinning, cell adhesion

Abstract

Poly(ϵ-caprolactone) (PCL) has shown a significant potential in neural and spinal cord tissue engineering. However, its clinical translation is limited by inherently poor cell adhesion, which is an essential factor for maintaining tissue integrity and enabling effective tissue repair.

In this study, we have introduced a simple, single-step strategy to enhance the biointerface properties of PCL by incorporating polypyrrole (PPy) into electrospun fibers. The PPy-modified scaffolds exhibited a less negative surface potential at physiological pH (−13.57 ± 0.81 mV) compared to pristine PCL (−21.80 ± 0.32 mV), suggesting reduced electrostatic repulsion and improved conditions for cell attachment. Additionally, incorporation of PPy led to decreased surface roughness and enhanced wettability, which resulted in creating a more favorable surface for cell adhesion. The biological performance of the scaffolds was evaluated using the neuroblastoma (SH-SY5Y) cell line. Scanning electron microscopy analysis revealed significantly improved cell spreading and surface coverage on PPy-modified fibers. Cells occupied a significantly larger surface area (6126 ± 4907 μm^2) in the PPy-modified sample than in pristine PCL fibers (944 ± 480 μm^2). Moreover, a higher number of focal adhesion points was formed on the PPy-modified scaffolds compared to pristine PCL.

These findings demonstrate that PPy incorporation effectively enhances the physicochemical and biological properties of PCL fibers, creating a more supportive microenvironment for cell attachment and proliferation.

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Beyond Nafion: A triple-functional electropolymerized coating for biosensing excellence

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Keywords: conductive polymer coating, poly(pyrrole-NHS) film, MXene–Pt nanozyme, label-free immunosensor

Abstract

Hydrogen peroxide is a widely used electroactive substrate in nanozyme-based immunosensors, where both catalytic efficiency and surface engineering critically determine analytical performance. Herein, we report a multifunctional electrode design based on MXene decorated with platinum nanoparticles (MXene-Pt), integrated with an electropolymerized poly(pyrrole-N-hydroxysuccinimide) (PPy-NHS) coating.

The PPy-NHS layer is introduced as an alternative to conventional Nafion films, aiming to overcome their limitations in conductivity, sensitivity, and analyte permeability. Unlike Nafion, PPy-NHS forms a uniform and conductive coating that simultaneously stabilizes MXene-Pt on the electrode surface, preserves hydrogen peroxide accessibility to catalytic sites, and provides reactive NHS groups for direct covalent antibody immobilization.

Electrochemical studies reveal that PPy-NHS not only maintains but enhances catalytic activity toward hydrogen peroxide through a synergistic interaction with MXene-Pt, resulting in improved current response and signal stability compared to both Nafion-coated and unmodified MXene-Pt electrodes. Moreover, SEM imaging confirms effective surface coverage without blocking catalytic sites.

Finally, the platform was applied to immobilize anti-thyroglobulin antibodies, enabling the development of a reliable label-free immunosensor for thyroglobulin detection. This work shows that PPy-NHS bridges material design and biosensing performance for advanced diagnostics.

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Gold nanostructure modified with polypyrrole films for ultra-sensitive detection of ovarian cancer

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Keywords: electropolymerization, pyrrole, gold nanostructures, graphene quantum dots, cancer antigen 125

Abstract

Ovarian cancer, labeled a ”silent killer,” remains among the deadliest gynecological malignancies globally, due to the absence of readily identifiable signs, symptoms, and effective screening methods. A prominent biomarker for diagnosing ovarian cancer is the cancer antigen 125 (CA-125). Electropolymers fabricated via direct electropolymerization can provide stable, electroactive interfaces as well as ensure uniform surface coverage and strong electrode adhesion, making them ideal components for the development of high-performance electrochemical biosensors.

This study presents graphene quantum dots GQDs/AuNPs/polymer-film-based electrochemical biosensor for ultra-sensitive detection for cancer antigen 125 (CA-125). GQDs coated glassy carbon electrodes supported the templated fabrication of AuNPs directly on the electrode surface from the mixture of auric chloride, ascorbic acid and dodecyltrimethylammonium bromide to tune the morphology. The formation of AuNPs was confirmed by SEM, EDS and cyclic voltammetry. Subsequently, pyrrole-3-carboxylic acid was electropolymerized with en optimized number of cycles to provide a stable interface with -COOH groups. After electrode fabrication, EDC/NHS chemistry was applied to form covalent grafting of anti-CA-125 antibody on the electrode surface through amide linkages.The modified electrodes were used to detect the presence of CA-125 antigen with different concentrations by following electrochemical impedance spectroscopy (EIS).

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Surface EMG for control applications: Methods and challenges

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Keywords: surface electromyography, human-machine interaction, prosthetic control systems

Abstract

Surface electromyography (sEMG) is widely used as a control input for prosthetic and robotic systems by measuring muscle activity. A typical EMG-based system consists of signal acquisition, preprocessing, feature extraction, and decoding. Preprocessing methods, such as EMG envelope extraction and wavelet transforms, reduce noise but increase computational cost and latency.

This work presents a literature review on the use of sEMG for controlling prosthetic and robotic systems, comparing classical, machine learning, and deep learning approaches in terms of accuracy, computational cost, and real-time applicability.

Classical methods are simple and computationally efficient, but are susceptible to noise and provide limited functionality. Machine learning approaches improve accuracy and allow more complex movement, but require manual feature engineering, and may not meet real-time constraints. Deep learning methods enable automatic feature extraction and higher accuracy, but have high computational demands and are often limited to offline usage.

EMG signals are inherently noisy, unstable, and dependent on electrode placement, lacking long-term reliability and standardized methodology. Most systems are limited to basic movement patterns. Despite recent advances, EMG-based systems remain limited by signal variability, lack of robustness, and real-time challenges. These findings highlight the need for more robust and standardized approaches to achieve reliable real-time control of prosthetic and robotic systems.

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Development of a two-region geometrical model of the L5 vertebra for numerical simulations

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Keywords: computational biomechanics, L5 vertebra, finite element method, geometrical modeling, cortical bone, cancellous bone, micro-computed tomography, Sawbones model

Abstract

Accurate numerical simulations of vertebral mechanics require anatomically realistic geometry and representation of material heterogeneity. This study aimed to develop a three-dimensional geometrical model of the L5 vertebra with anatomically accurate partitioning into compact (cortical) and trabecular regions for finite element analyses. The model was based on a commercial Sawbones® anatomical model and supplemented with micro-computed tomography (micro-CT) imaging to capture internal structures. Micro-CT data were used to define the boundaries of cortical and trabecular regions and determine geometrical parameters such as cortical wall thickness, ensuring a realistic internal structure for numerical simulations.

This two-region model provides a foundation for studies investigating the influence of material heterogeneity on vertebral mechanical response. Preliminary assessments indicate that distinguishing dense and porous regions improves the representation of stress distribution and deformation patterns under compressive loading compared to homogeneous models. The model will be used in future finite element analyses to quantify the effects of bone heterogeneity and anisotropy on vertebral stiffness and local stress concentrations.

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Synthetic data in personalized medicine and oncology: Methods, applications, and challenges

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Keywords: synthetic data, personalized medicine, advanced methods

Abstract

Synthetic data has emerged as a key solution to challenges in healthcare, particularly in personalized medicine and oncology, where access to high-quality and privacy-compliant data is limited. The aim of this study is to analyze how synthetic data can support clinical applications, focusing on generation methods, practical use, and associated ethical, technical, and data security challenges. To achieve this, a literature review of recent open-access studies was conducted, examining approaches to data generation, application, and evaluation.

Advanced methods, including generative adversarial networks (GANs), variational autoencoders (VAEs), and diffusion models are widely used to generate medical images, electronic health records, and other clinical data. In oncology, synthetic data supports tumor detection, diagnosis, and treatment planning, while in personalized medicine it enables predictive modeling and patient-specific treatment strategies. It also addresses data scarcity, especially for rare diseases, and facilitates secure data sharing across institutions.

Despite these benefits, significant challenges remain, including bias propagation, lack of standardized evaluation methods, and difficulties in ensuring realistic and clinically valid data. Ethical considerations and high computational requirements further limit widespread adoption.

Overall, synthetic data has considerable potential to advance AI-driven healthcare, but its effective use requires robust validation, standardization, and careful consideration of ethical issues.

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Utility of synthetic data in clinical decision support systems: A task-aware methodological evaluation using TCGA LUAD data

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Keywords: synthetic data, clinical decision support systems, task-aware evaluation, computational oncology, algorithms, classification, data processing, TCGA LUAD

Abstract

Limited access to high-quality clinical data remains a major challenge in the development of clinical decision support systems. In recent years, synthetic data generated using statistical and machine learning methods have gained increasing attention as a potential solution; however, their practical usefulness for clinical prediction tasks is still insufficiently understood.

The aim of this study was to evaluate whether classifiers trained exclusively on synthetic clinical data retain prognostic utility when applied to real oncology patients. Publicly available clinical data from The Cancer Genome Atlas (TCGA) were used, focusing on lung adenocarcinoma (LUAD). One-year overall survival was formulated as a binary classification task. Synthetic clinical datasets were generated using the SDV framework with a Gaussian Copula synthesizer trained on real patient data. Logistic regression models were trained either on real or synthetic data and evaluated on a held-out real test set using ROC-AUC and balanced accuracy.

The model trained on real data achieved a ROC-AUC of 0.62, while the model trained on synthetic data achieved a ROC-AUC of 0.56 when tested on real patients. These results indicate a moderate performance degradation but preserve non-random discriminative capability. The findings suggest that synthetic clinical data may be useful for methodological studies, preliminary model development, and benchmarking, but should not fully replace real patient data in high-risk clinical applications.

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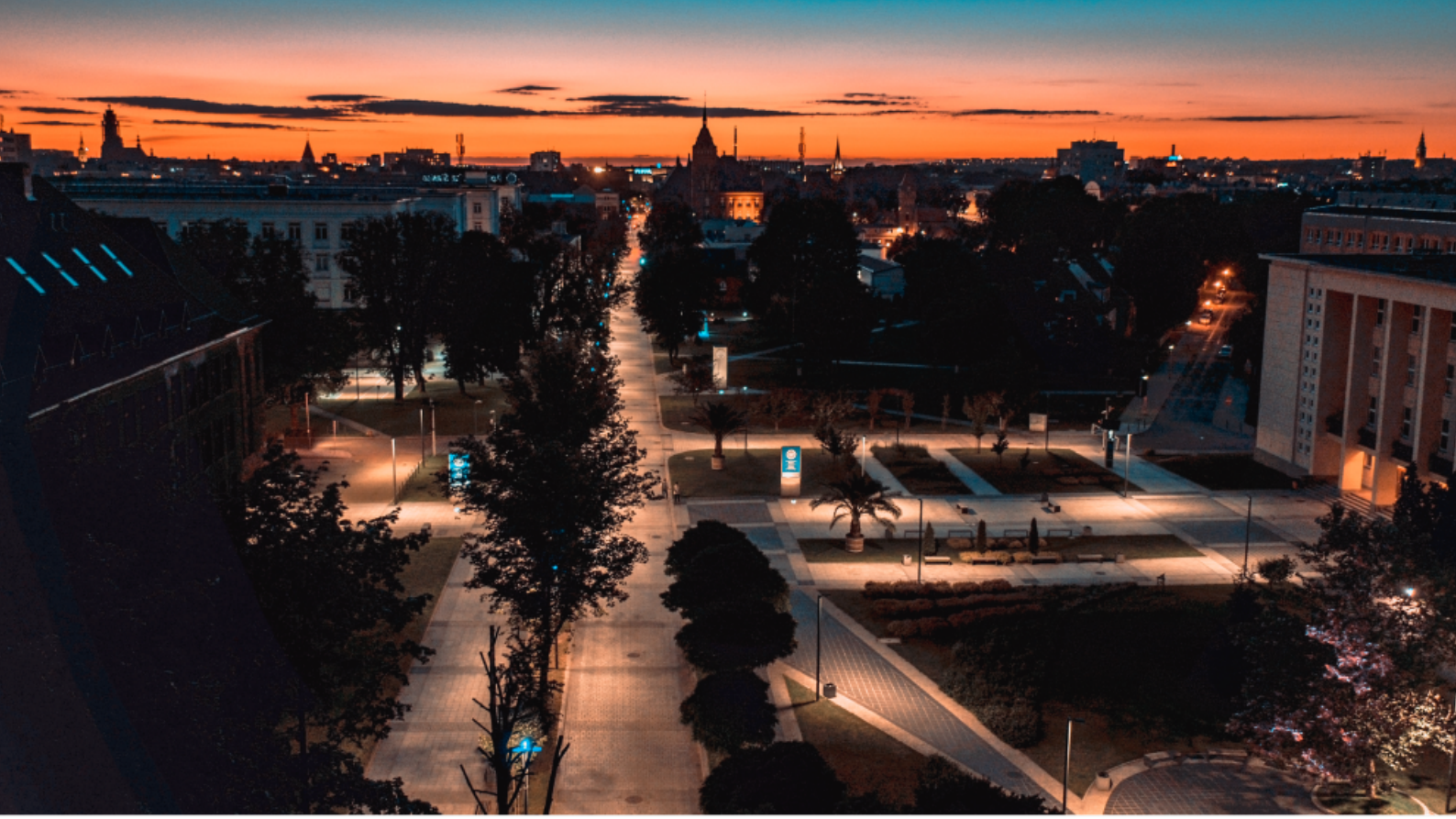

Priority Research Area #1 (POB1)

Computational Oncology and Personalized Medicine

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