Review Article

University J Dent Scie 2025; Vol. 11, Issue 2

Bioinformatics in Dentistry: A Review

Abstract:
Bioinformatics has revolutionized the field of dentistry by enabling the analysis and interpretation of complex biological data, offering new insights into oral health and disease mechanisms. This review explores the application of bioinformatics tools in the diagnosis, treatment, and prevention of dental and periodontal diseases. Key areas include the use of genomic data for identifying genetic markers linked to oral diseases, microbiome analysis in periodontitis, and the role of proteomics and metabolomics in understanding disease progression. Furthermore, bioinformatics aids in the development of personalized treatment strategies by integrating clinical, genetic, and environmental data. The review also highlights advancements in data-driven technologies, such as machine learning, that assist in predicting disease outcomes and improving patient care. By fostering interdisciplinary collaboration, bioinformatics has the potential to significantly advance clinical practices and contribute to the future of precision dentistry and periodontology.

Key-words: Bioinformatics, Dental informatics, Genomics, Epigenetics, Salivaomics, Oral disease diagnosis

Introduction:

Informatics is an applied information science focused on designing data systems that secure, organize, and respond to user needs.[1] Health informatics merges information science, computer science, and cognitive science to manage healthcare information, encompassing areas like bioinformatics, neuro-informatics, clinical informatics, public health informatics, and dental informatics.[2,3]

Bioinformatics, a rapidly evolving field, uses molecular data to address biological problems, significantly advancing healthcare research.[4] It aids in drug design, early cancer prediction, and patient care database development, leveraging machine learning and AI to enhance diagnostics and treatment.[5] Recent studies have integrated bioinformatics with dentistry, leading to terms like "oral genomics" and enhancing our understanding of oral diseases at the genomic level.[6,7]

High-throughput technologies, including DNA microarrays, are vital for analyzing large datasets generated by projects like the Human Genome Project.[7] These tools enable better clinical decision-making by identifying key genetic segments. Bioinformatics also serves various health disciplines, integrating methodologies from multiple fields to improve healthcare delivery.[5]

Dental informatics is an emerging field aimed at enhancing clinical practice, diagnosis, and management in dentistry.[7,8] Techniques like microarray analysis allow for the examination of gene expression profiles, facilitating a deeper understanding of disease mechanisms at the genetic level.[9,10] While microarrays cannot assess protein levels, they can illuminate pathways of malignancy and support predictive modeling in oncology.[11]

Access this article online	
Website: www.ujds.in	Quick Response Code
DOI: https://doi.org/10.21276/ujds.2025.v11.i2.15	

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Received : 24 July, 2024, **Published :** 31 Dec., 2024

How to cite this article: Vijay, D. S., Dad, D. J., Saini, D. N., Kapoor, D. A., & Jayakumar, D. P. (2025). Bioinformatics in Dentistry- A Review. UNIVERSITY JOURNAL OF DENTAL SCIENCES, 11(2).

This review offers an overview of bioinformatics and its implications in dentistry, highlighting its potential to revolutionize the field.

Heath Informatics and Bioinformatics:

Health informatics is the logic of healthcare, paralleling how physiology studies life and pathology examines disease. It focuses on how clinical information is created, shared, and utilized to enhance healthcare system organization and operations. Key aspects include understanding information systems, developing interventions, implementing effective methods, and evaluating the impact of these interventions.[8]

While dental informatics has existed since 1986, it is less cited than medical informatics. It supports Evidence-Based Dentistry (EBD) by integrating systematic assessments of clinical evidence to improve oral healthcare. In the current research landscape, health informatics is crucial for maintaining ethical integrity and effectively communicating findings to society.

Bioinformatics combines computer science and biology, focusing on technology for storing, retrieving, and analyzing data about biological macromolecules like DNA and proteins. It is essential for managing complex genomic data tasks, differing from computational biology, which encompasses a broader range of biological computations

History:

Computers and specialized software have become indispensable tools in modern biology, especially with the rise of next-generation sequencing (NGS), which revolutionized fields like population genetics and microbial ecology. The journey began in the 1950s, when the role of DNA as the genetic information carrier was established, primarily through the work of Avery, Hershey, and Chase.[12,13] Initial research focused on protein analysis, with Margaret Dayhoff emerging as a pioneer in bioinformatics by developing early computational methods for protein sequencing.[14]

By the 1970s, attention shifted from proteins to DNA, leading to advancements like the Maxam–Gilbert sequencing method and the first software for analyzing sequencing data.[15] The 1980s saw the rise of personal computers and the free software movement, facilitating bioinformatics research.[16] The 1990s marked the dawn of genomics, highlighted by the complete sequencing of *Haemophilusinfluenzae*.[17] The advent of NGS in the 2000s further democratized DNA sequencing, while recent years have seen the emergence of specialized bioinformaticians and a shift towards systems biology, emphasizing a holistic view of biological processes.[18]

Goals, Scope And Limitations:

Bioinformatics aims to deepen our understanding of cellular functions at the molecular level by analyzing genetic data, following the central dogma of biology (DNA → RNA → proteins). It includes two main areas: developing computational tools and applying them to generate biological knowledge, particularly in sequence, structure, and functional analysis.

The goals of dental informatics focus on improving patient outcomes and supporting evidence-based clinical practices. Despite its potential, bioinformatics has limitations, such as reliance on data quality and algorithm accuracy. Predictions are not definitive proofs and should be complemented by experimental validation. Caution is needed when interpreting results, and using multiple tools can enhance accuracy.[19]

Tools and Techniques In Bioinformatics:

Bioinformatics utilizes databases and analytical tools to process data from high-throughput methods like mass spectrometry and sequencing.[20] Key software applications aid in nucleotide sequence analysis, genetic variation identification, and sequence alignment, while also predicting enzyme sites, open reading frames (ORFs), and analyzing gene expression and protein structures.[21]

Data and Retrieval Systems:

Understanding genomic data encompasses concepts like transcriptome, proteome, interactome, and miRNome. Sequence data is typically formatted in ASCII, with FASTA and PHYLIP being common formats.[22,23] Primary databases like GenBank store raw data, while secondary databases like NCBI RefSeq provide curated sequences.[24] Retrieval systems like Entrez facilitate efficient searches across multiple databases.

Sequence Alignment:

Sequence alignment is vital for exploring evolutionary relationships. Key methods include global alignment (Needleman-Wunsch algorithm) and local alignment (Smith-Waterman algorithm).[25] Tools like BLAST and FASTA help search for related sequences, with BLAST being notably faster.

Nucleic Acid Sequencing:

Genome sequencing can be done through primer walking or shotgun methods, with genome annotation predicting functional components.[26] The Open Reading Frame Finder identifies potential ORFs, and microarray analysis assesses gene expression.[27]

Phylogenetic Analysis:

Phylogeny outlines species' evolutionary history, with phylogenetic trees visually representing relationships.[28] Key steps include selecting molecular markers, aligning sequences, and assessing tree reliability, leading to classifications such as monophyly, polyphyly, and paraphyly. Monophyletic groups provide accurate evolutionary insights.[29]

Application In Dentistry:

Application In Periodontology:

Periodontitis is a chronic inflammatory disease influenced by genetic and epigenetic factors, primarily triggered by oral bacteria in dental plaque. Progression is affected by genetics, lifestyle, and systemic health. If untreated, it can cause alveolar bone destruction, loss of periodontal attachment, and increased tooth mobility.[30]

The oral microbiome, with over 500 bacterial species, is crucial in periodontal disease, with specific complexes indicating health or disease.[31] The host's inflammatory response to these pathogens drives tissue destruction, varying among individuals due to genetic and epigenetic differences.[32]

Recent research emphasizes epigenetics in understanding periodontal disease. Environmental factors can induce changes that affect gene expression without altering DNA. Mechanisms like DNA methylation and histone modifications play significant roles in immune regulation and disease susceptibility.[33] Genome-wide analyses reveal distinct methylation patterns in diseased tissues, influenced by lifestyle factors such as smoking.[34] Histone modifications and long non-coding RNAs (lncRNAs) also warrant further study, as they may influence inflammatory processes.[35]

Periodontal disease is linked to non-communicable diseases (NCDs) like cardiovascular diseases and diabetes, emphasizing the need for integrated health strategies addressing shared risk factors.[36-38] Understanding the interplay between microbial factors, host responses, and epigenetics opens new avenues for improved diagnosis, treatment, and prevention of periodontal and systemic diseases.

Detection of Oral Cancer:

Bioinformatics is crucial for managing and analyzing biological data, enabling the extraction of genetic information that informs clinical decision-making and the development of new treatments. This field integrates health sciences with computer science and biostatistics to enhance healthcare outcomes.

Oral cancer represents a significant global health threat, causing approximately 10 million deaths in 2020. A majority of patients present with metastasis or symptoms at the time of diagnosis, which complicates prognosis and highlights the importance of early detection for improving treatment outcomes and quality of life.

However, visual detection of oral cancer can be challenging, as distinguishing between premalignant and malignant lesions is often difficult. This can result in missed diagnoses, emphasizing the need for more effective identification methods.

Advancements like the OralCDx® system offer promising solutions for early detection. This system utilizes computer-aided image analysis of oral brush biopsies to identify atypical cells, assisting pathologists in making more accurate diagnoses. By improving diagnostic accuracy, OralCDx® ultimately contributes to better treatment outcomes for patients with oral cancer.[39]

Microarray Technology:

Microarray technology has been vital in cancer research and diagnosis for over a decade, overcoming the limitations of traditional gene analysis methods. Using oligonucleotide and cDNA chips, microarrays analyze gene expression, SNPs, and mutations, revealing genetic and epigenetic profiles of cancer cells. They help differentiate tumors that look similar morphologically, impacting disease progression.[40]

In oral health, conditions like oral cancer and precancerous lesions have a genetic basis, with microarrays identifying genetic alterations before morphological changes occur. This capability aids drug discovery and targeted therapies. Traditional visual inspection often misses early-stage cancers, but the OralCDx® system enhances detection using computer-assisted image analysis of oral brush biopsies, streamlining the identification of abnormal cells and reducing manual workload for pathologists.[41]

Patient Care Databases:

Patient care databases are online systems that store essential information related to patient diagnoses, procedures, and prescriptions, allowing easy access to rare case data. By applying machine learning algorithms to this data, healthcare providers can deliver effective, cost-efficient treatments. A well-designed hospital database can gather up-to-date patient information, facilitating quick access and seamless information exchange among different healthcare facilities. These databases enhance the monitoring and improvement of healthcare services, assist with documentation and billing, and help reduce operational costs by minimizing paperwork. Health Database Organizations (HDOs) focus on two key aspects: comprehensiveness, which ensures complete records of patient care events, and inclusiveness, which aims to cover a wide population within a geographic area.[42]

Image Analysis:

Computational analysis of medical images enables the identification of disease patterns and correlations among patients, improving diagnosis and treatment strategies. Machine learning and deep learning have revolutionized medical imaging modalities, including X-ray, MRI, PET, CT, and SPECT, allowing for accurate and efficient diagnoses. The use of advanced imaging technologies has enhanced data acquisition, while bioinformatics tools aid in detecting abnormalities and monitoring their progression.

Bioimages, encompassing all medical imaging data related to biological samples, have become increasingly complex. Techniques like CT and MRI have distinct applications, with CT excelling in detecting cancers and cerebral hemorrhages, while MRIs provide clearer images of brain tumors.

The DICOM (Digital Imaging and Communication in Medicine) standard has emerged to streamline the storage and sharing of medical images, accommodating diverse imaging technologies. Each DICOM image instance contains metadata and pixel data, ensuring efficient handling and transmission of medical information while maintaining data integrity.[43]

Salivaomics Knowledge Base:

Saliva is increasingly recognized as a valuable biofluid for non-invasive disease diagnostics. The National Institute of Dental and Craniofacial Research (NIDCR) supports advancing saliva research, which is facilitated by the Salivaomics Knowledge Base (SKB). SKB integrates saliva biomarker discovery with the Saliva Ontology (SALO) developed through collaboration with various experts.

The SKB serves as a comprehensive data repository for salivary proteomics, transcriptomics, miRNA, metabolomics, and microbiome research. It features a three-tier architecture consisting of a Data Layer for storage, an Ontology Layer for mapping data to controlled vocabularies, and an Interface Layer for user interaction.

SALO, optimized for both clinical diagnostics and omics research, provides a structured vocabulary for saliva-related terms. It includes links to existing ontologies, such as Gene Ontology and Protein Ontology, and offers public access to definitions and relevant literature. Overall, SKB and SALO aim to enhance understanding and utilization of human saliva in research and diagnostics.[44]

BioMart:

BioMart is a free, open-source federated database system compatible with various relational databases like MySQL and PostgreSQL. It allows easy integration and collaboration across geographically dispersed databases, leading to the creation of the BioMart Central Portal—a single access point for numerous biological databases. This portal simplifies cross-database searches and provides integrated tools for tasks like ID conversion and sequence retrieval.

SDxMart is a specialized BioMart portal focused on salivaomics, hosting salivary proteomic, transcriptomic, metabolomic, and microRNA data. It facilitates saliva biomarker discovery through complex queries that integrate genomic, clinical, and functional information. SDxMart includes data from various oral and systemic diseases, such as oral cancer and Sjögren's syndrome, and continuously expands its datasets, incorporating resources like the Ensembl genome database.

Future Recommendations:

To enhance accessibility and effectiveness in dental informatics, several recommendations are proposed:

1. Establish a global community of dental informaticians.
2. Increase the number of biomedical informaticians focused on dental issues.
3. Develop career-oriented pathways in dental informatics to provide job opportunities.

4. Ensure dental informatics addresses public health challenges at the community level.

Additionally, a robust surveillance system must be implemented to protect confidentiality, along with comprehensive ICT education and training for dental professionals.[45-47]

Conclusion:

Research in dental sciences is set to transform through high-throughput techniques and vast data generation, enabling detailed analysis of molecular sequences. This will enhance our understanding of genotypes and their clinical phenotypes, revolutionizing dentistry by integrating genomic information into diagnostics, therapeutics, and prognostics. Bioinformatics and data mining will play crucial roles in identifying risk factors and therapeutic targets, especially in oncology. The ongoing development of the Saliva Ontology aims to improve data retrieval and integration across research fields, leading to a new gold standard for studying oral diseases through combined experimental and theoretical approaches.

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