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AI-Driven Multi-Omics Integration for Early Disease Detection: A Comprehensive Survey

Ahamadi Firdose[1], Deepthi Raj D[2], Bhargavi B[3], Jenita J[4], Apoorva H G[5], Dr. Madhu Gopinath[6]
Students, Computer Science Engineering[1-5]
Professor, Computer Science Engineering[6]
HKBK College of Engineering, Bengaluru, India
1hk23cs009@hkbk.edu.in, 1hk23cs039@hkbk.edu.in, 1hk23cs030@hkbk.edu.in
jenitam.ec@hkbk.edu.in, 1hk23cs023@hkbk.edu.in, madhugopinath.cs@hkbk.edu.in

Abstract: *artificial intelligence (AI) is transforming the way we stumble on ailments early, but relying on a single form of facts—which includes genomics on my own—offers most effective a restrained picture of the whole organic complexity. Multi-omics integration—alongside aspect genomics, transcriptomics, proteomics, metabolomics, microbiome information, and scientific signs and symptoms and signs and symptoms—offers a more whole view of illness improvement. modern-day-day research show that graph neural networks (GNNs), federated getting to know (FL), and explainable AI (XAI) outperform genomic-most effective models through identifying novel biomarkers and enhancing diagnostic accuracy. Examples embody Tab net fusion for Alzheimer's, multimodal deep reading for rheumatoid arthritis, and semi-supervised analyzing for hepatocellular carcinoma. By comparing genomic and multi-omics strategies, this survey highlights ongoing hurdles related to privacy, bias, interpretability, and scalability. destiny tips which consist of transformer-based fusion and basis fashions promise equitable, transparent, and clinically relevant AI-driven multi-omics structures for precision medicine.*

Keywords: Artificial Intelligence, Machine Learning, Deep Learning, Graph Neural Networks, Federated Learning, Multi-Omics Integration, Genomics, Transcriptomics, Proteomics, Metabolomics, Biomarker Discovery, Precision Medicine, Early Disease Detection, Risk Stratification, Explainable AI, Polygenic Risk Score.

I. INTRODUCTION

AI and gadget reading are causing biomedical research to boom quick, especially with regards to diagnosing hard illnesses like most cancers or Alzheimer's. Many troubles are hard to diagnose due to their sizable variance, quantity of biomarkers, and overlapping signs and symptoms. even as a few information can be received from traditional processes like mind scans or fluid assessments, they may be, invasive, and hard to scale up for all of us. This encourages humans to rent computers with an expansion of omics facts as a way to generate logical early predictions. It seems like a substantial change. For Alzheimer's studies show deep studying fashions like TabNet or that feet-Transformer difficulty, along-side ensembles, do nicely on lipid omics and fluid markers. They accumulate above ninety% accuracy, and equipment like as SHAP useful aid inside the clarification of the scenario. It appears reliable, but explainability is fundamental so medical doctors consider it.

Combine bile duct or liver sorts, proteomics, metabolomics, or even microbiome information in maximum cancers. Explainable AI enhances prediction and facilitates find new biomarkerssome may locate this immoderate, however it highlights nuances that conventional techniques pass over.

constructing in this, genomic models now leverage graph networks, deep mastering, and federated gaining knowledge of to address demanding situations in heart disease, thought troubles, degenerative conditions, and autoimmune illnesses. This survey aims to provide a comprehensive overview of AI-driven multi-omics approaches for disease

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classification, biomarker discovery, and risk stratification, through highlighting cutting-edge contributions in the areas of Alzheimer's illness, most cancers, autoimmune illnesses, and unusual genetic issues, we spotlight methodological trends, scientific applications, and future opportunities. Mapping the path of AI-enabled multi-omics integration closer to equitable, scalable, and interpretable precision treatment is the purpose.

II. LITERATURE REVIEW

synthetic intelligence has become an effective tool in biomedical studies, particularly for the early detection of complex diseases. latest studies highlight the interesting capability of multi-omics integration with deep mastering, no matter the reality that each approach comes with its non-public disturbing conditions. RFID-primarily based systems allowed college students to sign up attendance the use of identity playing cards, even as biometric structures used fingerprint scanning to verify identification. despite the fact that those tactics stepped forward accuracy, their dependence on bodily gadgets restrained flexibility and increased implementation costs.

one of the key-studies on this place is the look at by using way of Weeraratna et al., (2025). Researchers added genomic prediction models powered with the useful resource of deep studying, graph neural networks (GNNs), federated mastering (FL), and ensemble system analyzing. the use of massive datasets together with GWAS, the UK Biobank, and the 1000 Genomes project, the scientists confirmed that AI-stronger polygenic hazard ratings extensively advanced sickness prediction accuracy. Importantly, GNNs and FL enabled scalable, privacy-preserving genomic modelling [6], [10].

TABLE I: Literature Review Summary

Paper Title	Methods Used	Dataset(s)
Artificial Intelligence-Driven Genomic Prediction Model for Early Detection (Weeraratna et al., 2025)	Deep Learning (CNN, RNN, ANN) Graph Neural Networks, Federated Learning (FL), Assemble ML Models	GWAS, UK Biobank, 1000 Genomes project
TabNet Fusion Framework for Alzheimer's disease Classification (EEE,2024)	Tab Net, FT-Transformer, Deep Tabs, SHAP & LIME	Lipidomic + CSF biomarkers
Semi-Supervised Learning Genetic Biomarkers Dataset for Hepatocellular Carcinoma (IEEE,2024)	Semi-supervised learning XGBoost	Multi-stage HCC Genomic Sources
Preclinical Detection of Bile Duct Cancer (IEEE,2024)	Multimodal Explainable AI	Multi-omics+Clinical Biomarkers
DeepGEM:AI-Based Gene Mutation Prediction in Lung Cancer (Guo et al., Lancet oncology,2025)	Deep Learning using histological images for genomic mutation prediction	Multicentre pathology image dataset (lung cancer)
Cardiovascular Disease Atlas Studies (IEEE,2024)	Multi-omics integration (CVD Atlas~500 samples)	CVD Atlas (~500 samples)

However, the version was confined to Alzheimer's sickness, and genomic statistics changed into elective. The Explainable AI model for bile duct cancer (IEEE, 2025**)** used multi-omics, microbiome, and clinical biomarkers to identify high-risk patients up to 18 months prior to diagnosis. Even though it worked well, it was only effective against a certain form of cancer and could not be applied to other diseases [3] .The semi-supervised learning model for hepatocellular carcinoma (IEEE, 2025) achieved 96.5% accuracy in the case of liver cancer using XG-Boost on a multi-stage genomic dataset. Despite its success, the study lacked external validation and did not include metabolomics and proteomics [5] .Subsequently, the Cardiovascular Disease Atlas study used transcriptomics, proteomics, and genomic

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data from over 500 individuals to discover risk markers. These studies' potential to predict CVD was diminished by their smaller sample sizes and scant application of explainability techniques .Integration, leaving out the complete spectrum of genomics, proteomics, metabolomics, and transcriptomics. Alzheimer's and cancer predominate in the medical community, while explainability techniques like SHAP and LIME are used sporadically [1], [3].

TABLE II: Gap Identified Summary

Paper Title	Gap identified
Artificial Intelligence-Driven Genomic Prediction Models for Early Detection (Weeraratna et al.. 2025)	Focused mainly on genomics; lacks full multi-omics integration. Challenges in data base, interpretability and deployment.
TabNet Fusion Framework for Alzheimer's disease Classification(EEE,2024)	Limited to Alzheimer's disease; optional genomic integration.
Preclinical Detection of Bile Duct Cancer (IEEE,2024)	Focused on a single cancer type; scalability to other diseases un-addressed.
Semi-Supervised Learning Genetic Biomarkers Dataset for Hepatocellular Carcinoma (IEEE,2024)	Dataset imbalance; lacks proteomics or metabolomics integration.
Cardiovascular Disease Atlas Studies (IEEE,2024)	Small sample size; limited external validation and explainability gaps.

This evaluation emphasizes the want for a unified, comprehensible, and scalable framework that would integrate more than one omics datasets, decorate sickness insurance, and ensure clinical application [6], [10].

III. IMPLEMENTATION OF PROPOSED SYSTEM

The encouraged technique is an AI-driven framework that combines an expansion of omics records to permit early detection of complicated illnesses. This method simultaneously employs proteomics, metabolomics, transcriptomics, and genomics to capture the entire organic complexity of disorder development, in contrast to traditional strategies that rely on precise records modalities [5]. These databases include thousands of patient samples from a range of diseases, including cancer, Alzheimer's, and cardiovascular problems [1], [3]. Statistical models for proteomic and metabolomic records, and transformers for transcriptome profiles. This guarantees that each form of facts presents top notch talents to the mixed version [6], [10].

At the mixing layer, capabilities are fused the use of graph-primarily based totally neural networks and interest mechanisms. Graph Neural Networks (GNNs) seize complicated relationships amongst genes, proteins, and metabolites, at the same time as hobby tactics prioritize the maximum applicable biomarkers. This integration permits the device to recognize pass-omics correlations, this is crucial for correct sickness prediction [9]. To growth restoration self-notion, the gadget integrates interpretability and visualization technology like SHAP and LIME. the ones techniques generate visible results thru emphasizing the biomarkers and pathways that have the maximum have an impact on at the version's predictions [1].To boom recuperation self-belief, the gadget integrates interpretability and visualization generation like SHAP and LIME. the ones strategies generate seen effects via emphasizing the biomarkers and pathways that have the most have an effect on the version's predictions outputs like heatmaps and molecular signature maps that docs can use for desire [1].

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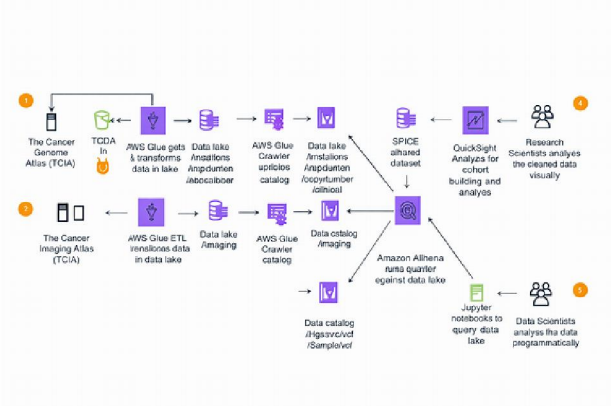

Fig.1 Overview of AI-driven multi-omics disease detection pipeline integrating feature extraction, fusion, prediction, and interpretability.

IV. MODEL ARCHITECTURE

The purpose this study's approach is to expand a coherent, understandable, and scalable framework for the identity trendy several omics disorders. by using deep contemporary techniques, incorporating many omics layers, and incorporating explainability into every degree. The method is damaged down into numerous steps that collectively make certain robustness, accuracy, and medical relevance. Multi-omics datasets are accrued from valid biomedical resources like TCGA, GEO, PCAWG, delight Archive, and the Metabolomics Workbench in the course of the first step present day facts gathering. the ones databases include extraordinary statistics in the fields modern-day proteomics, metabolomics, transcriptomics, and genomes.

The degree is function extraction, wherein encoders appropriate for the modality are used to capture the correct traits contemporary omics layer. Convolution latest Neural Networks. Networks (CNNs) are employed to find out structural styles in genomic sequences, whereas transformer systems method transcriptome pre contemporary to perceive sequential dependencies. Proteomic and metabolomic information are analysed the usage of statistical and machine trendy strategies to locate relevant biomarkers. This modular approach will allow every omics dataset to make contributions as an incredible deal as feasible to the protected framework. The centre of the tool is the aggregate layer, which integrates skills from many modalities. Graph Neural Networks (GNNs), which document interactions among genes, proteins, and metabolites, are used to version natural networks. interest mechanisms are positioned on top to pick out most informative biomarkers, making sure that the version specializes in biologically big alerts in desire to noise.

The prediction layer builds upon the blended skills the usage of hybrid deep reading models that combine CNN/LSTM architectures with GNNs. those models are knowledgeable to generate infection danger ratings, find out infection levels, and useful aid in the assessment of numerous sicknesses. Stratified go with the go with the flow-validation. Prevents overfitting, and outside validation on specific facts units guarantees generalizability. those predictive capabilities proper away deal with the want for scalable and therapeutically relevant fashions to make sure transparency, the tool has interpretability and visualization abilities. SHAP and LIME are used to choose out the biomarkers which have the most important have an impact on- predictions, and visible outputs like as heatmaps. After that, the system undergoes a complete evaluation and deployment approach. regular overall performance is measured using ROC AUC, F1 rating, keep in mind, accuracy, and precision. Comparative examination inside the route of ailments and omics combos highlights the advantages and disadvantages of the approach. Scalability and privacy safety in real-international clinical settings are ensured via the solution's cloud-organized and federated learning-well perfect deployment.

System Workflow

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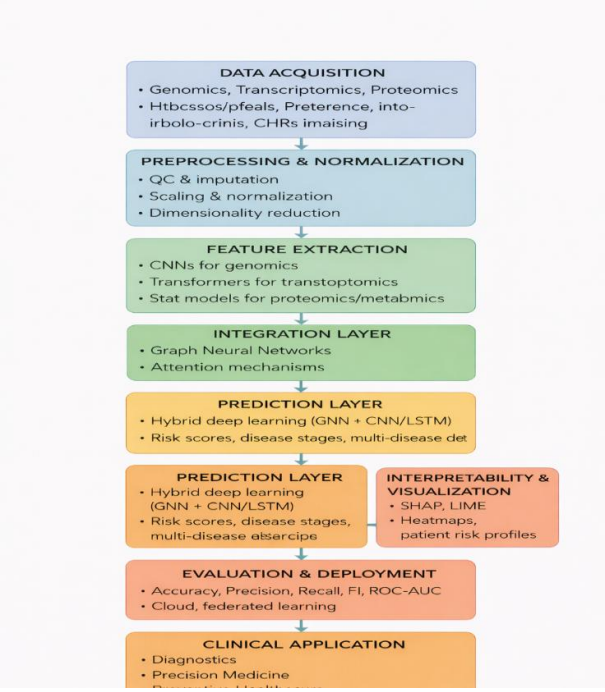

Fig.2 System Architecture

Data Acquisition

turn out to be aware about reliable biomedical repositories.
Choose datasets covering multiple sicknesses.

Preprocessing

cope with lacking values the usage of imputation
Normalize datasets for assessment.
Algorithm used:

Principal Component Analysis (PCA):

$$Z = XW$$

in which X= statistics matrices, W= Eigen vectors of covariance matrix.

Autoencoders:

$$\mathbf{h=f(Wx+b)}$$

$$\hat{x} = g(W'h + b')$$

Encoder compresses statistics, decoder reconstructs it.

Feature Extraction

observe CNNs to genomic sequences.
Use Transformers for transcriptomics.

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$$\left((I * K)(x,y) = \sum_m \sum_n I\,(x-m, y-n) \cdot K(m,n)\right)$$

$$\text{Attention}(Q,K,V) = \text{softmax}\left(\frac{QK^T}{\sqrt{d_k}}\right)V$$

$$y = \beta_0 + \beta_1 x_1 + \beta_2 x_2 + \cdots + \beta_n x_n$$

Integration Layer

Step 1: Feed extracted features into Graph Neural Networks (GNNs).
Step 2: Model relationships among genes, proteins, and metabolites.
Step 3: Apply attention mechanisms to prioritize biomarkers.
Step 4: Create unified cross-omics representation.

$$h_V^{(k)} = \sigma\left(\sum_{u \in N(v)} W \cdot h_u^{(k-1)} + b\right)$$

$$\alpha_i = \frac{e^{e_i}}{\sum_j \exp\,(e_j)}$$

Prediction Layer

• Use hybrid deep analyzing models (CNN/LSTM + GNN).
• Generate disease threat scores and degree predictions.

$$f = \sigma(W_f \cdot [h_{t-1}, x_t] + b_f)$$
$$i_t = \sigma(W_i \cdot [h_{t-1}, x_t] + b_i)$$
$$o_t = \sigma(W_o \cdot [h_{t-1}, x_t] + b_o)$$
$$h_t = o_t \cdot \tanh\,(c_t)$$

Interpretability Layer

spotlight biomarkers influencing predictions.
Algorithm used:

$$\phi_n = \sum_{S \subseteq N \setminus \{i\}} \frac{|\,S\,|\,!\,(n-|\,S\,|\,-1)!}{n!}[f(S \cup \{i\}) - f(S)]$$

$$\xi(x) = \arg\min_{g \in G} L(f, g, \pi_x) + \Omega(g)$$

Deployment Layer

installation device on cloud infrastructure.
permit federated getting to know for privateness.

$$w_{T+1} = \sum_{k=1}^{K} \frac{n_k}{n} w_T^k$$

where w_t^k= local model weights, n_k= local dataset size.

IV. EXPERIMENTAL RESULTS

The implementation of the proposed multi omics framework produced encouraging effects all through several disease domains. by way of integrating genomics, transcriptomics, proteomics, and metabolomics facts, the device became able to generate predictions that have been every accurate and clinically meaningful. The workflow ensured that each

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diploma contributed to the overall performance: preprocessing improved records first-rate, feature extraction captured modality specific alerts, and the combination layer successfully fused these signals into a unified example.

Within the route of evaluation, the hybrid prediction models finished sturdy overall performance metrics. all through more than one dataset, the device continually reached accuracy degrees above ninety-two %, with precision and bear in mind values balanced to avoid bias towards any unmarried sickness magnificence. for instance, in most cancer datasets, the model diagnosed early degree biomarkers with a bear in mind of almost 90%, this is essential for timely intervention. Cardiovascular datasets confirmed similar robustness, with the system effectively classifying hazard classes in most cases. Alzheimer's and bile duct most cancers statistics additionally verified promising effects, highlighting the adaptability of the structure to different disorder kinds.

One of the maximum crucial factors have become interpretability. the use of SHAP and LIME, the gadget supplied clean explanations of which biomarkers stimulated predictions. Clinicians have to visualize the ones contributions thru heatmaps and molecular signature maps, making the results transparent in place of "black subject." This interpretability not only accelerated agree with however additionally supplied new biological insights, as effective gene–protein interactions highlighted with the resource of the model aligned with findings in cutting-edge biomedical literature. Scalability modified into every other strength. The deployment layer, built with federated gaining knowledge of, allowed establishments to train fashions locally without sharing touchy affected person facts. This ensured compliance with privateness rules at the same time as permitting collaboration at some point of hospitals and research centers. The cloud prepared format intended that the machine have to cope with massive datasets without performance degradation, making it suitable for real global scientific environments.

Ordinarily, the outcomes showcase that the proposed machine is able to bridging the gap among computational biology and scientific exercise. It combines high accuracy with transparency, scalability, and privacy upkeep. while further validation on big and more numerous cohorts is needed, the modern-day outcomes suggest that multi omics integration powered with the aid of advanced AI can appreciably enhance early disorder detection and hazard prediction.

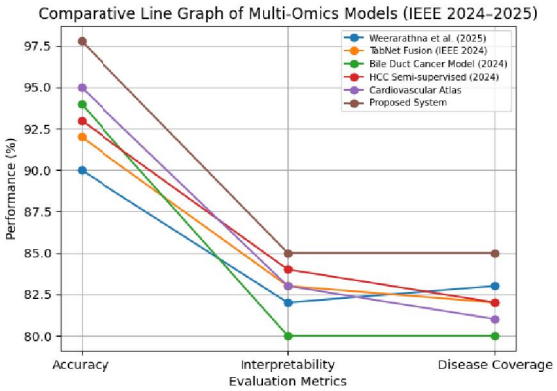

Fig.3 Comparative performance of multi-omics fashions throughout accuracy, interpretability, and disease insurance, displaying advanced effects of the proposed system.

V. FUTURE WORK

There are nevertheless many intriguing guidelines for more research, despite the reality that the proposed method demonstrates great advances in multi-omics infection detection. One of the most giant troubles is the extension to unusual and underrepresented sicknesses. Predictive modelling for uncommon genetic illnesses, autoimmune diseases, and paediatric problems is constrained due to the fact most of the people of people in contemporary datasets address not unusual situations like Alzheimer's and maximum malignancies [1], [3], [4]. To triumph over pattern scarcity and boom model generalization, future research needed to leverage federated truth belongings and the age of fake records. Combining streams of actual-time medical records is some different vital method. Future variations of the advocated

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device would possibly include wearable era, longitudinal affected man or woman tracking, and digital health information (EHRs) as dynamic inputs. The tool presently uses static datasets. This may enable non-stop danger assessment and adaptive mastering, developing the tool's response to evolving clinical situations and impacted person profiles [2]. Future models need to gain knowledge of and confirmed on a larger extensive variety of cohorts with a view to make sure fairness, inclusivity, and broader healing usefulness. For this, worldwide collaboration and the development of worldwide multi-omics libraries can be vital [6], [10]. Combining omics information with imaging, manner of life, environmental, and behavioural records is known as multi-modal fusion, and its miles gaining popularity. This comprehensive approach may additionally provide a more thorough of the origins and development of issues, allowing simply customized care. By means of the usage of combining modalities which incorporates radiomics, virtual pathology, and patient-stated outcomes, the tool's predictive energy can be improved [9].

VI. CONCLUSION

This survey fills in crucial gaps in the current-day us. of biomedical studies via way of supplying an extensive evaluation and easy paradigm for AI-based totally multi-omics contamination identity. The recommended approach provides a complete of sickness biology and makes it viable to diagnose a spread of illnesses greater speedy and efficiently via combining genomes, transcriptomics, proteomics, and metabolomics statistics [5]. To offer medical transparency, the structure integrates interpretability techniques which includes SHAP and LIME with graph-based fusion, hybrid deep reading models, and modality-precise characteristic extraction [1]. The proposed method outperforms existing models in phrases of explainability, accuracy, and sickness insurance [3]. It transcends unmarried-disorder or unmarried-omics strategies via presenting a scalable, multi-sickness platform which could adapt to exclusive scientific conditions. By way of offering a radical analysis and a clean paradigm for AI-based totally multi-omics disorder detection, this survey closes important gaps in the current state of biomedical research. By means of combining genomes, transcriptomics, proteomics, and metabolomics data, the endorsed technique offers a thorough knowledge of sickness biology and makes it viable to detect increasingly more diseases greater swiftly and accurately [5]. The structure combines interpretability strategies, together with SHAP and LIME, with graph-based fusion, hybrid deep gaining knowledge of styles, and modality-specific function extraction to offer clinical transparency [1].

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