APPLICATION OF ARTIFICIAL INTELLIGENCE AND MACHINE LEARNING IN GENOMIC DATA ANALYSIS: OPPORTUNITIES AND LIMITATIONS IN PERSONALIZED MEDICINE

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Annotatsiya: Mazkur maqolada sun’iy intellekt (SI) va mashinaviy o‘rganish (MO) texnologiyalarining genom ma’lumotlarini tahlil qilishdagi ahamiyati va ularning shaxsiylashtirilgan tibbiyotdagi qo‘llanilish imkoniyatlari ko‘rib chiqiladi. SI va MO vositalari orqali bemorlarning genetik ma’lumotlariga asoslangan individual tashxis va davolash uslublari ishlab chiqilmoqda. Shu bilan birga, maqolada mavjud texnik cheklovlar, ma’lumotlar maxfiyligi, etik muammolar va amaliyotdagi qiyinchiliklar ham yoritilgan.

Kalit so‘zlar: Sun’iy intellekt, mashinaviy o‘rganish, genomika, shaxsiylashtirilgan tibbiyot, bioinformatika, genetik tahlil, tibbiy texnologiyalar

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Аннотация: В статье рассматривается значимость искусственного интеллекта (ИИ) и машинного обучения (МО) в анализе геномных данных и их применение в персонализированной медицине. Использование ИИ и МО позволяет разрабатывать индивидуальные подходы к диагностике и лечению на основе генетических данных пациентов. Также обсуждаются технические ограничения, вопросы конфиденциальности данных, этические аспекты и сложности практического внедрения данных технологий в медицине.

Ключевые слова: Искусственный интеллект, машинное обучение, геномика, персонализированная медицина, биоинформатика, генетический анализ, медицинские технологии

Abstract: This article discusses the significance of artificial intelligence (AI) and machine learning (ML) in the analysis of genomic data and their application in personalized medicine. AI and ML technologies enable the development of individualized diagnostic and treatment approaches based on patients' genetic information. The article also highlights technical limitations, data privacy concerns, ethical issues, and challenges in implementing these technologies in clinical practice.

Keywords: Artificial intelligence, machine learning, genomics, personalized medicine, bioinformatics, genetic analysis, medical technologies

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Last ten in years genomics in the field sequencing technologies (NGS next -generation sequencing) human genome complete and half complete information cheap and fast in a way to take opportunity created by a patient genetic code organization 3 billion nucleotides that make up gigabytes at the level raw information to be, statistic methods with again Even if it works, it is a complex gene- -gene interaction. effects, epigenetics modifications and population heterogeneity because of complete analysis to do difficult. Especially , the disease predisposition detection, pharmacogenomics recommendations to give or oncological therapy for targeted medicine choice such as in tasks such in size information automated , accuracy high without tools again work almost impossible

That's it in the situation artificial intelligence (SI) and mechanical learning (ML) methods to learn genetic information automatic accordingly analysis to make patterns determination and predictions in the making important to the tool SI /ML approaches not only information size and complexity overcome to pass opportunity gives, maybe new biomarkers to find genetic risk factors prophecy help to do gives. With this together, personal concept of precision medicine genetic profile, organized health status and vital to factors based individual treatment strategies

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working exit goal that he did because genomics and SI/ML integration modern medicine inseparable to the part rotation condition;

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1. Genomics your information Features: Genomics data, that is genetic information, human all genetic code own inside. This information is size and complexity them traditional methods with analysis to do very makes it difficult. Human in the genome about 3 billion nucleotides this is information analysis to do for high effective technologies necessary.

- ***Size:*** Human genome complete sequence consists of approximately 3 GB information organization it will, this and systems for to be a big burden possible. Like this of information again processing and analysis to be done , information storage and processing in giving high effective requires technologies .
- ***Complex dependencies:*** Genetic changes only in individual genes not, maybe genes between each other also occurs in the effect Genetic mutations epigenetic changes with each other related to be it is possible, this and analysis further complicates. Example for, epigenetic modifications (DNA methylation, histone modifications) gene expression to change take comes and disease in development important role plays.
- ***Heterogeneity:*** Genomics information not only every one human for individual, maybe diseases are also different genetic to options have to be possible. Therefore, for analysis in doing high level **heterogeneity** Genetic problems determination for similar information bases and to algorithms there is a need. With this together, information quality storage and analysis automation for artificial intellect and mechanical study from algorithms use this the complexities in overcoming help gives.

2. AI/ML methods and architecture: Genomics information analysis in doing artificial intelligence (AI) and mechanical machine learning (ML) methods wide These methods are used. genetic analyses optimization, diseases early identification , and individual treatment plans in creation important to the tool has become . The following approaches genomics in the analysis effective is being used:

- ***Supervised learning learning):*** In this way to the model in advance known was based on data (input and output) study opportunity is given. Example for, genetic mutations classification for genetic features and diseases about information is used. Example: Random Forest, SVM (Support Vector Machine), logistic regression from models is used.

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- ***Unsupervised learning study):*** Uncontrolled study algorithms genetic in the data hidden structures determination for This method is used. using genes between similarities, genetic options groups (clustering) are determined. Example: K-means, Hierarchical Clustering and PCA (Principal Component Analysis).
- ***Deep Learning learning): Deep*** Learning algorithms genomics in the analysis the most progressive technology They are big. in size information processing in giving high to efficiency have For example, Convolutional Neural Networks (CNNs) are genetic mutations in determining and genome analysis in doing is used.
  Example : CNN, Recurrent Neural Networks (RNN), Generative Adversarial Networks (GAN).
- ***Reinforcement Learning learning)***: ***This*** method genomic analysis optimization for used. Changes and new genetic options through awards to take possible was situations analysis in doing effective. Example: Repeat work and information collect in the process awards based on algorithm improvement. Each approach genomics information analysis in doing known to the advantage has to be, their right selection to the results big impact shows.

3. Personal in medicine Usage: Personal medicine is every one the patient's genetic characteristics, life style and other to factors based individual treatment approach Genomics and AI/ML technologies personal in medicine application clinical analysis and treatment further more precisely to do opportunity is creating:

- ***Pharmacogenomics:*** Pharmacogenomics genetic to options based of medicines efficiency analysis For example, some patients genetic to the characteristics depending on, known to drugs good answer not giving possible. Using ML such mutations determination Example: Warfarin drug doses in marking genetic analyses using individual recommendations.
- ***Disease predisposition:*** using AI heart diseases, diabetes, cancer such as to diseases was predispositions Genetic tests, life style and biomarkers based on analysis to do through of the disease development prophecy to do possible.
  Example: Personal genome based on cholesterol level and sugar amount control to do
- ***Oncology:*** Genetic profile based on to the patient targeted therapy Tumor genetics using AI mutations determination and to him/her against effective

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medicines choice can. Example: Personalized cancer therapy — cancer genomics, where AI selects appropriate treatment plans for cancer patients based on their genetic profiles.

4. Successful practical Examples: Genomics and artificial of intellect successful to be used examples very many. The following examples to bring possible :

- ***Google DeepVariant:*** By Google working issued DeepVariant model from sequencing obtained NGS (next-generation sequencing) data analysis in doing high accuracy This model provides a small changes in determining human mistakes reduces and 50% better results gives.
- ***IBM Watson for Genomics:*** IBM Watson oncology according to genomic analysis in doing mutations determines and personal therapy plan in the making help This system is 95% accurate. results presented it will, that's it with together of patients treatment efficiency noticeable at the level increases.
- ***23 andMe:*** 23andMe Genomics and artificial intellect using to users own genetic predispositions and to your health impact doer features determination opportunity gives.

5. Technical and boot limitations: AI and genomics technologies to be used against some technician and boot problems available. To them the following input possible:

- ***Model transparency:*** Deep study algorithms, especially the " black hole" of deep learning models " box " nature analysis of the results understandable to be absent take Genetic analysis how in the basics done is being increased understanding for the model explanation necessary.
- ***Information safety :*** Genetic information very secret and personal is considered . Their made laws (e.g. GDPR) and boot to the rules suitable accordingly preservation and processing to be given necessary.
- ***Bias:*** AI algorithms working on the way out information Risk of bias in the aggregates available. Example for some genetic information sets only known one ethnic from the group was to patients justification it is possible, this and of the results wrong to be take arrival possible.

Conclusion

Genomics information analysis SI and ML algorithms in making personal medicine concept to practice implementation in the process of solution doer role They are playing. They are playing. high in accuracy again performance, complex gene- gene interaction effects to determine, and pharmacogenomic recommendations in giving efficiency increases. Example for, DeepVariant and

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like Watson for Genomics platforms clinical-genomic analysis in the processes mistakes reduce, treat the results is improving. With this together, model interpretability, information privacy and regulator compatibility such as issues complete solution immature Therefore, SI/ML methods wide current to grow still limited. In the future interpretive AI approaches, information exchange consortiums and health storage systems with close cooperation through this restrictions overcome He wished to die. As a result genomics based on personal medicine new stepmother The patient is getting up. safety, treatment efficiency and resources saving in terms of noticeable to achievements achieves.

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