The integration of artificial intelligence into personalized medicine

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Priya Hays, CEO and Science Writer at Hays Documentation Specialists, LLC, discusses the integration of Artificial Intelligence (AI) into personalized medicine (PM), highlighting its potential to enhance healthcare, particularly in genomic medicine and precision oncology

When personalized medicine (PM) emerged as a field with the growth and implementation of omics, such as genomics and proteomics, stakeholders were already becoming aware of the potential of Big Data and Artificial Intelligence to further enhance PM innovation and technologies. Genomic medicine grew concomitantly with the development of next-generation sequencing and vast streams of data that were required for storage and portability. Artificial Intelligence, of which Big Data is a foundation, involves the analysis of huge reservoirs of data to reveal information about patterns in the data, and is increasingly being used in healthcare settings. In precision oncology, AI methods have found fruitful applications in imaging and pathology, improving accuracy, sensitivity, and specificity in the analysis of radiographic images and histologic slides. In pathology, through algorithmic and statistical modeling, patterns in the specimens can be identified and aid in clinical decision making through image analysis, whereby abnormal features can be detected, leading to more efficient decision making and accurate and personalized diagnosis. The relationships between Machine Learning (ML) and Deep Learning (DL) methods can be summarized as follows: ML is a subset of AI, and DL, a subset of ML, utilizes artificial networks, such as convolutional neural networks and artificial neural networks, to enable more complex learning processes, particularly in cancer diagnosis. (1)

AI methods in precision medicine

Deep Learning methods in precision medicine were evident in the application of convolutional learning networks, or CNN, in the Prostate, Lung, Colorectal and Ovarian screening trial enrolling 41,856 patients, demonstrating that patterns revealed in CT scans led to the prediction of 12-year incidence of lung cancer with better accuracy than routine screening methods, with further validation of the CNN model from the National Lung Screening Trial. Characteristics such as gender, age, and smoking status in EHR records were utilized in data analysis to create patterns with improved ability to identify high-risk patients, particularly smokers, for lung cancer, and compared with positive radiographic screening and standard data sets provided by the CMS. Despite different follow-up times, cancer occurrence rates were similar in the PLCO group (3.7%; 207 of 5615) and the NLST (3.8%; 206 of 4593), taking into account variability in response to smoking and patients recalling smoking history, as reported in the Annals of Internal Medicine. [2]

AI methods have also been applied to precision medicine in terms of predictive analytics, with an example being their ability to detect breast cancer spread with 90% accuracy. This is achieved by creating a patterned library using a Deep Learning model programmed with micrograph images of breast carcinoma from 70 patients, based on their clinical history. The model had a 90% accuracy rate, predicting which breast cancers would spread after surgery, determining prognosis and therapy choice for 70 patients with cancer recurrence. The data, with a 4% false positivity rate, are encouraging when compared with a 70% accuracy rate, as elicited from viewing images. With a higher level of prediction for detecting up to one million pixels in a standard image, compared to the human eye, which does not readily recognize patterns in the image, this AI method forms a powerful adjunct to pathology. Validation of this method is ongoing in further studies. The findings were reported in the American Journal of Physiology-Cell Physiology. [3]

Improved identification of biomarkers and genomic variations

A framework for AI in personalized medicine and the next generation of precision oncology is revealed through the ability and potential of AI for imaging processing and analyzing genomic information. Numerous cancers are routinely characterized and diagnosed through the screening of images, both radiologic and histologic, and oncology heavily relies on the analysis and interpretation of these images, with a shift toward digital pathology and digital radiology. Powerful computational AI methods can be applied to improve upon subjective assessments and enable high-throughput analysis as well. The breakthrough came with the CNN AlexNet in 2012, which is capable of yielding image-based biomarkers that predict response to treatment. AI applications in cancer genomics were made possible by the advent of next-generation sequencing, which has enabled the analysis of genomic information such as whole-genome, whole-transcriptome, and whole-exome analysis. This advancement allows for large-scale analysis of genomic variation, with the potential for personalized care. Machine Learning can also prove useful for early cancer detection, wherein DL models can engage in defining clinically actionable molecular alterations and molecular profiling for developing individualized cancer treatment. [4,5]

Advancing AI in personalized medicine

The following areas of advancement for AI in personalized medicine may be in drug development and the utilization of large language models for analyzing electronic health records and patient- reported outcomes. However, this field is in its early growth stages, and proof-of-concept studies are ongoing. With the success of AI, ML, and DL models in answering many of the hypothesis-generating questions in personalized medicine, progress remains strong and holds great potential for enhancing clinical medicine.

References

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