AI Tool May Improve Diagnosis of Metastatic Cancers

New research published in the journal Nature describes an artificial intelligence (AI) tool that could improve the diagnosis for patients with complex metastatic cancer. In 1 to 2 percent of these cases, the primary site of tumour origin cannot be determined, and patients often have to undergo extensive diagnostic procedures, which can delay treatment.

In a bid to improve diagnosis for patients with complex metastatic cancers, especially those in low-resource settings, researchers from the Mahmood Lab at the Brigham and Women's Hospital developed an artificial intelligence (AI) system that uses routinely acquired histology slides to accurately find the origins of metastatic tumours while generating a "differential diagnosis," for CUP (cancer of unknown primary) patients.

"Almost every patient that has a cancer diagnosis has a histology slide, which has been the diagnostic standard for over a hundred years", explained Faisal Mahmood, PhD, of the Division of Computational Pathology at the Brigham and an assistant professor at Harvard Medical School. "Our work provides a way to leverage universally acquired data and the power of artificial intelligence to improve diagnosis for these complicated cases that typically require extensive diagnostic work-ups."

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The algorithm developed by the researchers, called Tumour Origin Assessment via Deep Learning (TOAD), simultaneously identifies the tumour as primary or metastatic and predicts its site of origin. The researchers trained their model with gigapixel pathology whole-slide images of tumours from more than 22,000 cancer cases, and then tested TOAD in about 6,500 cases with known primaries and analysed increasingly complicated metastatic cancers to establish utility of the AI model on CUPs.

For tumours with known primary origins, the model correctly identified the cancer 83% of the time and listed the diagnosis among its top three predictions 96% of the time. Researchers then tested the model on 317 CUP cases for which a differential diagnosis was assigned, finding that TOAD's diagnosis agreed with pathologists' reports 61 percent of the time and top-three agreement in 82 percent of cases.

"The top predictions from the model can accelerate diagnosis and subsequent treatment by reducing the number of ancillary tests that need to be ordered, reducing additional tissue sampling, and the overall time required to diagnose patients, which can be long and stressful," Mahmood said. "Top-three predictions can be used to guide pathologists next steps, and in low-resource settings where pathology expertise may not be available the top prediction could potentially be used to assign a differential diagnosis. This is only the first step in using whole-slide images for AI-assisted cancer origin prediction, and it's a very exciting area with the potential to standardise and improve the diagnostic process."

Source: Brigham and Women's Hospital

Reference: Lu, M.Y., Chen, T.Y., Williamson, D.F.K. *et al.* AI-based pathology predicts origins for cancers of unknown primary. *Nature*, 2021; DOI: 10.1038/s41586-021-03512-4

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