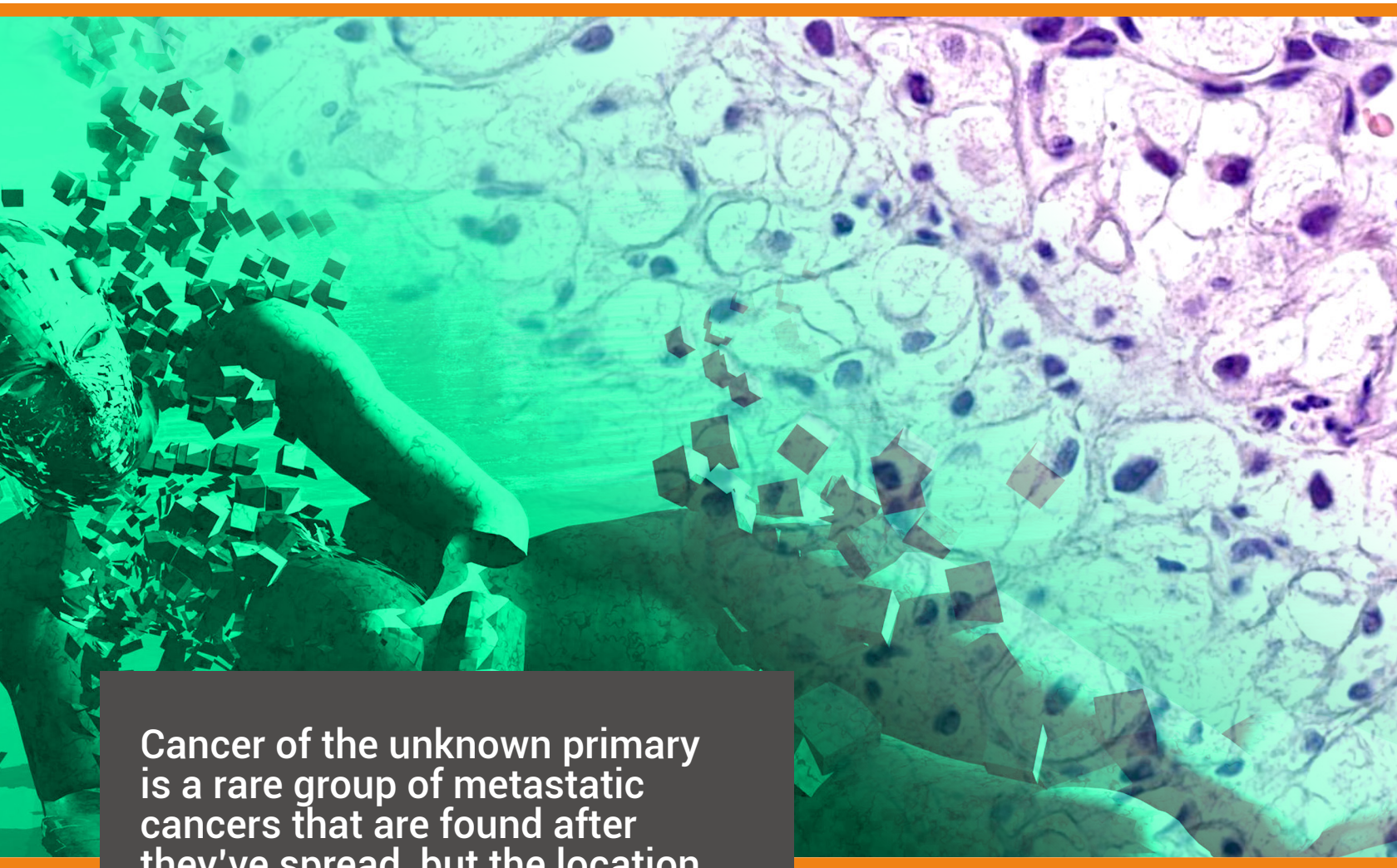
Case Study
Cancer of Unknown Primary

The value of AI paired with comprehensive RNA profiling



Cancer of the unknown primary is a rare group of metastatic cancers that are found after they've spread, but the location of the original tumor isn't clear. These cases make up about 3% to 5% of cancers diagnosed.

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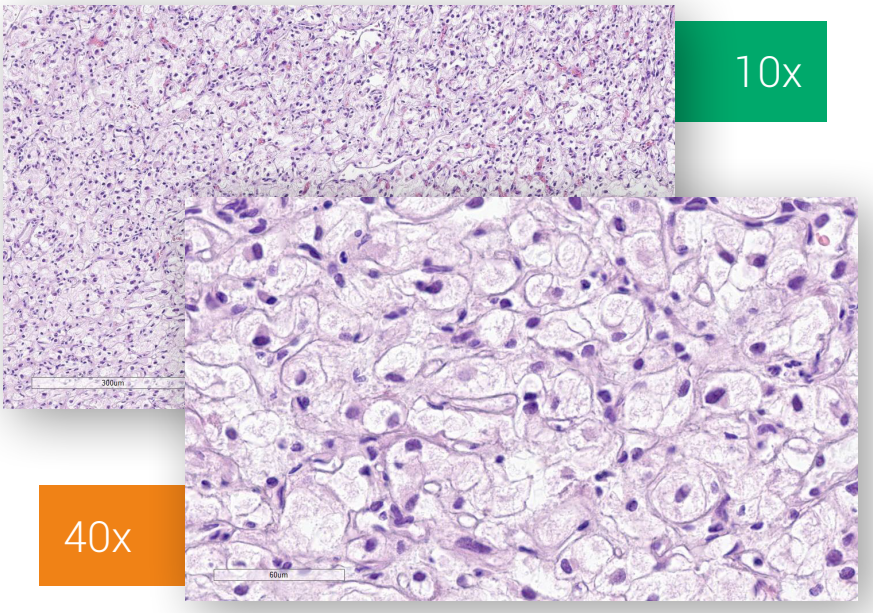
Background

Cancer of Unknown Primary (CUP) occurs in 3–5% of patients when the site of tumor origin cannot be determined using the standard histological diagnostic tests. Typically, a CUP diagnosis is treated empirically and has inferior outcomes, with median overall survival less than one year. Gene expression profiling alone has been used to identify the tissue of origin but struggles with low neoplastic percentage in metastatic sites which is where identification is often most needed. This case shows the value of AI paired with our comprehensive RNA profiling in determining Cancers of Unknown Primary (CUP)/Cell of Origin.



Clinical History

- 61-year-old male
- With metastatic cancer of unknown primary to the femur

Molecular Profiling Findings

- Expression profile consistent with renal cell carcinoma
- Mutations in VHL, KDM5C, ARID2 (2 mutations), NIN, HGF, ARID1B, NTRK1, KMT2C, EGFR(T790M) genes
- Chromosomal structural analysis shows 1q+, 3p-, 3q+, -4, 5q+ (distal), -9, 10p+, 10q-, -13, -14, and -21
- No evidence of microsatellite instability
- Tumor Mutation Burden High: 10 Mut/Mb
- Homologous recombination deficiency (HRD): Negative

Discussion

Using RNA and DNA sequencing and targeted transcriptome data coupled with machine learning; we were able to identify the kidney origin of the patient's cancer along with detecting the VHL mutation which has an FDA approved targeted therapy (Belzutifan). Our RNA profiling and artificial intelligence algorithms along with our experts interpterion of this information can provide highly useful tools for the pathologic diagnosis and classification of various cancers. The additional information that we provide such as mutation profile and clinical information can provide personalized treatment recommendations and minimize errors in pathologic diagnoses.

References

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