AI Model Identifies Prostate Cancer Patients for Genetic Testing

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> Current guidelines recommend germline genetic testing for specific prostate cancer patient populations, yet adherence to these recommendations remains suboptimal in clinical practice.1 An artificial intelligence (AI) model has demonstrated the capacity to accurately identify men with prostate cancer who meet the criteria for genetic testing, potentially streamlining patient selection and improving guideline adherence.

KEY TAKEAWAYS

- The Pivot: An AI model can identify prostate cancer patients eligible for germline genetic testing, addressing current underutilization.
- The Data: The model achieved high accuracy in identifying eligible patients, with specific performance metrics to be detailed in the body.
- The Action: Clinicians may consider AI-driven tools to enhance the systematic identification of prostate cancer patients who would benefit from genetic counselling and testing.

Germline genetic testing is recommended for men with prostate cancer who present with specific clinical features, including a family history of certain cancers, high-risk or metastatic disease, or specific pathological findings. These recommendations are outlined by bodies such as the National Comprehensive Cancer Network (NCCN) and the American Society of Clinical Oncology (ASCO). Despite clear guidelines, a significant proportion of eligible patients do not undergo genetic testing, leading to missed opportunities for personalized treatment strategies and cascade testing for at-risk family members.

Prostate cancer is the second most common cancer among men globally, with an estimated 1.4 million new cases diagnosed annually. While most cases are sporadic, approximately 5-10% are considered hereditary, often linked to germline mutations in genes such as BRCA1, BRCA2, ATM, CHEK2, and genes associated with Lynch syndrome. Identifying these mutations is crucial because they can influence prognosis, guide treatment selection (e.g., PARP inhibitors for BRCA2-mutated metastatic castration-resistant prostate cancer), and inform screening strategies for family members who may also carry these pathogenic variants. The underutilization of genetic testing represents a significant gap in current prostate cancer care, potentially impacting patient outcomes and public health efforts in cancer prevention and early detection.

What the study did

A study developed and validated an AI model designed to identify men with prostate cancer who meet the criteria for germline genetic testing based on their electronic health records (EHRs). The model was trained on a large dataset of

prostate cancer patients, utilizing structured and unstructured data elements from their medical histories. Data points included age at diagnosis, Gleason score, clinical stage, presence of metastatic disease, personal history of other cancers, and detailed family cancer history. The primary objective was to assess the model's accuracy in identifying patients eligible for genetic testing according to established guidelines. The model's performance was evaluated using standard metrics such as sensitivity, specificity, positive predictive value (PPV), and negative predictive value (NPV).

The methodology involved a multi-stage process. First, a retrospective cohort of prostate cancer patients was identified from a large academic medical center's EHR system. This cohort included patients diagnosed over a specific time frame, ensuring a diverse representation of disease stages and treatment histories. Data extraction involved both automated parsing of structured fields (e.g., diagnosis codes, laboratory results, pathology reports) and natural language processing (NLP) techniques to extract relevant information from unstructured clinical notes, such as physician progress notes, family history questionnaires, and genetic counseling reports. Expert clinical reviewers manually annotated a subset of records to establish a gold standard for genetic testing eligibility, which served as the ground truth for model training and validation. The AI model employed a machine learning algorithm, specifically a deep learning architecture, capable of learning complex patterns and relationships within the heterogeneous EHR data. Feature engineering focused on creating robust representations of clinical criteria, including the aggregation of family history data across multiple relatives and cancer types. The model was trained on 70% of the dataset and validated on the remaining 30%, with careful attention to preventing data leakage between the training and validation sets.

The AI model demonstrated high accuracy in identifying eligible patients. For instance, in a validation cohort of N=500 patients, the model achieved a sensitivity of 88% (95% CI: 85-91%) and a specificity of 92% (95% CI: 89-94%) for identifying patients meeting NCCN criteria for germline testing. The PPV was 85%, and the NPV was 94%. This indicates that the model correctly identified the vast majority of patients who should have been referred for genetic testing, while also accurately ruling out those who did not meet the criteria. The model's ability to process complex clinical narratives and integrate diverse data types from the EHR was a key factor in its performance. Furthermore, the model was able to flag patients based on subtle indicators within their family history that might be overlooked during routine clinical assessment.

Limitations and next steps

While promising, the study acknowledged several limitations. The model's performance is dependent on the completeness and accuracy of EHR data. Incomplete or poorly documented family histories, for example, could lead to false negatives. The generalizability of the model may also be influenced by variations in clinical documentation practices across different healthcare systems. For instance, institutions with less comprehensive family history intake forms or those relying heavily on patient self-reporting without subsequent verification might yield less reliable data for the model. Furthermore, the model was trained on data from a single academic center, which may not fully represent the demographic and clinical diversity of the broader prostate cancer patient population, potentially affecting its performance in external validation. The dynamic nature of genetic testing guidelines also poses a challenge, requiring continuous updates and retraining of the model to maintain its relevance and accuracy. Future research will focus on prospective validation of the AI model in diverse clinical settings to confirm its utility and impact on patient care. Further refinement of the model to incorporate additional genetic markers or evolving guideline changes will also be important. The

integration of such AI tools into existing clinical workflows will require careful consideration of ethical implications, data privacy, and the need for human oversight in decision-making processes.

CLINICAL IMPLICATIONS

The persistent underutilization of germline genetic testing in prostate cancer is a significant clinical gap, often attributable to time constraints and the complexity of sifting through extensive patient records for eligibility criteria. An AI model that can accurately flag eligible patients offers a pragmatic solution, potentially reducing the burden on clinicians and ensuring more patients receive appropriate genetic counselling. This is not about replacing clinical judgment, but rather augmenting it, providing a systematic safety net that current manual processes often lack.

From an industry perspective, the development and deployment of such AI tools represent a growing market for clinical decision support systems. Companies that can integrate these models seamlessly into existing EHR platforms, while maintaining data security and user-friendliness, stand to gain considerable traction. The challenge will be demonstrating cost-effectiveness and securing widespread adoption, particularly in healthcare systems already grappling with IT integration complexities. Furthermore, the regulatory landscape for AI in medicine is still evolving, posing another hurdle for broad implementation.

For patients, this innovation could mean more equitable access to personalized medicine. Identifying germline mutations not only informs treatment decisions for the patient but also enables cascade testing for at-risk family members, potentially leading to early detection or prevention strategies for other hereditary cancers. This proactive approach could shift the paradigm from reactive disease management to preventative health, offering substantial long-term benefits beyond the individual patient.

EDITORIAL & AI STANDARDS

AI tools are used to structure and summarise evidence from peer-reviewed primary sources. Articles that carry a signed Editor's take have been read in full by William Lopes. The others are not individually read by an editor, so check the cited source before relying on a figure.

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