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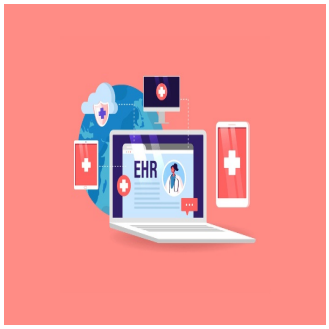
EHR Signatures Help Diagnose Patients With Common Variable Immunodeficiency



Common Variable Immunodeficiency (CVID) is a complex and rare inborn error of immunity characterised by antibody deficiency and impaired B cell responses. Due to its heterogeneous presentation, CVID often eludes timely diagnosis, resulting in delayed treatment and prolonged patient suffering. However, a groundbreaking development from the University of California, Los Angeles (UCLA) Health offers hope through the innovative application of machine learning technology to expedite the diagnostic process.

Addressing the Diagnostic Challenge of CVID with Machine Learning: The Development of PheNet

CVID is one of many inborn errors of immunity (IEI), a group encompassing approximately 500 inherited conditions that cause immune system dysfunction. Affecting about one in 25,000 individuals, CVID presents a diagnostic challenge due to its diverse symptomatology and overlap with more common diseases. Patients with CVID may suffer from frequent infections, autoimmune disorders, and inflammatory conditions, often receiving treatment from various specialists without a definitive diagnosis for years. This fragmentation of care exacerbates delays in diagnosis and treatment, driving excessive healthcare costs and impacting patient outcomes. To address this challenge, a team led by Dr. Manish Butte, a professor in pediatrics, human genetics, and microbiology/immunology at UCLA, developed PheNet, a machine learning model designed to identify patients likely to have CVID from their electronic health records (EHRs). The study, published in Science Translational Medicine, outlines how PheNet learns phenotypic patterns from verified CVID cases and uses this knowledge to rank patients based on their likelihood of having the disease.

Leveraging Machine Learning for Early CVID Detection

PheNet represents a significant advancement in the early detection of CVID. Traditional clinical methods often fail to diagnose CVID promptly due to the absence of a single causal mechanism and the condition's rarity. Unlike genetic tests that cannot definitively diagnose CVID, PheNet analyses extensive EHR data to identify subtle phenotypic signatures indicative of the disease. By doing so, the model can highlight patients "hiding in the medical system" who are prime candidates for referral to immunology specialists. The retrospective analysis conducted by Johnson et al. demonstrated that PheNet could have diagnosed more than half of the CVID patients in their cohort one or more years earlier than conventional methods. This early diagnosis is crucial, as timely treatment can significantly improve health outcomes and reduce healthcare costs. The model's effectiveness was further validated using over six million records from disparate medical systems in California and Tennessee, reinforcing its robustness and generalizability.

Practical Application and Broader Potential in AI-Driven Healthcare

In practice, PheNet operates by analysing patterns derived from the records of patients known to have CVID, as well as data from medical literature. It generates a numerical score, ranking patients based on their likelihood of having the condition. The model's initial application at UCLA Health involved a blinded chart review of the top 100 patients flagged by PheNet, of which 74% were deemed highly probable to have CVID. This high success rate underscores the model's potential to revolutionise the diagnostic approach for CVID and similar rare diseases. The implications of PheNet's success extend beyond CVID. Dr. Bogdan Pasaniuc, a professor of computational medicine at UCLA, highlighted the broader potential of AI-driven tools in healthcare, stating, "We show that artificial intelligence algorithms such as PheNet can offer clinical benefits by expediting the diagnosis of CVID, and we expect this to apply to other rare diseases, as well." The ongoing implementation of PheNet across the University of California medical centers is already making a tangible impact, with efforts underway to refine the model's precision and expand its application to other diseases.

The development of PheNet marks a significant leap forward in the diagnosis of CVID. By leveraging machine learning to parse extensive EHR data, this innovative tool promises to reduce diagnostic delays, enhance patient care, and alleviate the burden on healthcare systems. As AI continues to integrate into medical practice, models like PheNet exemplify the transformative potential of technology in improving health outcomes for patients with rare and elusive diseases.

Source: Science Translational Medicine

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