Machine learning comes of age in cystic fibrosis

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"Clubbing" of the fingers is a classic features of Cystic Fibrosis, although not present in many patients. Credit: Jerry Nick, M.D./ Wikipedia

World-leading AI technology developed by the Cambridge Centre for AI in Medicine and their colleagues offers a glimpse of the future of precision medicine, and unprecedented predictive power to clinicians caring for individuals with cystic fibrosis.

Accurately predicting how an individual's chronic illness is going to progress is critical to delivering better-personalized, precision medicine. Only with such insight can a clinician and patient plan optimal treatment strategies for intervention and mitigation. Yet there is an enormous challenge in accurately predicting the clinical trajectories of people for chronic health conditions such as cystic fibrosis (CF), cancer, cardiovascular disease and Alzheimer's disease.

"Prediction problems in healthcare are fiendishly complex," said Professor Mihaela van der Schaar, Director of the Cambridge Centre for AI in Medicine (CCAIM). "Even machine learning approaches, which deal in complexity, struggle to deliver meaningful benefits to patients and clinicians, and to medical science more broadly. Off-the-shelf machine learning solutions, so useful in many areas, simply do not cut it in predictive medicine."

Unlock this complexity, however, and enormous healthcare gains await. That is why several teams led by Professor van der Schaar and CCAIM Co-Director Andres Floto, Professor of Respiratory Biology at the University of Cambridge and Research Director of the Cambridge Centre for Lung Infection at Royal Papworth Hospital, have developed a rapidly evolving suite of world-class machine learning (ML) approaches and tools that have successfully overcome many of the challenges.

In just two years, the researchers have developed technology that has moved from producing ML-based predictions of lung failure in CF patients using a snapshot of patient data—itself a remarkable improvement on the previous state of the art—to dynamic predictions of individual disease trajectories, predictions of competing health risks and comorbidities, 'temporal clustering' with past patients, and much more.

The researchers presented three of their new ML technologies recently at the North American Cystic Fibrosis Conference 2020. In-depth details

of the technologies and their potential implications are available on the CCAIM website.

The tools developed by the Cambridge researchers represent astonishing progress in a very short time, and reveal the power of ML methods to tackle the remaining mysteries of common chronic illnesses and provide highly precise predictions of patient-specific health outcomes of unprecedented accuracy. What's more, such techniques can be readily applied to other chronic diseases.

Applying new ML techniques in cystic fibrosis

"Cystic fibrosis is an excellent example of a hard-to-treat, chronic condition," said Professor Floto. "It is often unclear how the disease will progress in a given individual over time, and there are multiple, competing complications that need preventative or mitigating interventions."

CF is a genetic condition that affects a number of organs, but primarily the lungs, where it leads to progressive respiratory failure and premature death. In 2019, the median age of the 114 people with CF who died in the UK was 31. Only about half of the people born in the UK with CF in 2019 are likely to live to the age of 50.

Cystic fibrosis is also a fertile ground to explore ML methods, in part because of the UK Cystic Fibrosis Registry, an extensive database that covers 99% of the UK's CF population which is managed by the UK Cystic Fibrosis Trust. The Registry holds both static and time-series data for each CF patient, including demographic information, CFTR genotype, disease-related measures including infection data, comorbidities and complications, lung function, weight, intravenous antibiotics usage, medications, transplantations and deaths.

"Almost everyone with cystic fibrosis in the UK entrusts the Registry to hold their patient data, which is then used to ensure the best care for all people with the condition," said Dr. Janet Allen, Director of Strategic Innovation at the Cystic Fibrosis Trust. "What's exciting is that the approaches developed by Professor van der Schaar take this to a completely new level, developing tools to harness the complexity of the CF data. Turning such data into medical understanding is a key priority for the future of personalized healthcare."

Looking to the future

The suite of new tools offers tremendous potential benefit to everyone in the CF ecosystem, from patients to clinicians and medical researchers. "Our medical ML technology has matured rapidly, and it is ready to be deployed," said Professor van der Schaar. "The time has come to bring its clear benefits to the individuals who need it most—in this case, the people living with cystic fibrosis. This means collaborating further with clinicians and increasing our engagement with wider healthcare systems and with data guardians beyond the UK."

Machine learning technologies have proven to be adept at predicting the clinical trajectories of people with long-term health conditions, and innovation will continue at pace. The patient-centered revolution in precision healthcare will enable and empower both clinicians and researchers to extract greater value from the growing availability of healthcare data.

The challenge ahead is to realize the potential of these tools by making them available to clinicians and hospitals around the world, where they can help improve and save the lives of people living with chronic illness. This is one of the goals of the Cambridge Centre for AI in Medicine.