Researchers integrate genomics data in to electronic patient records

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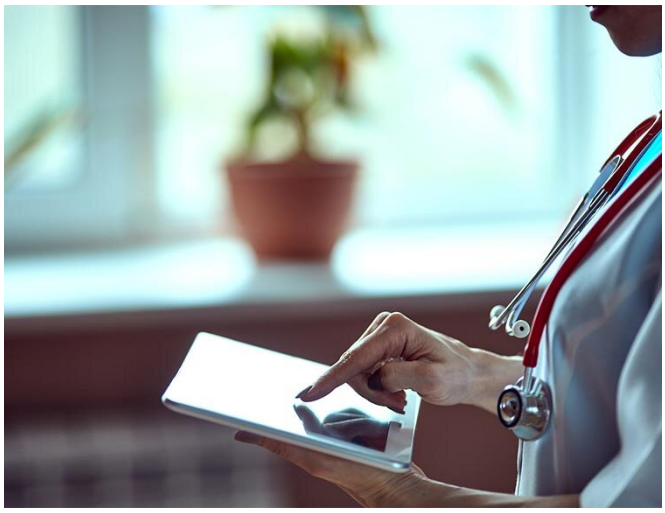

Credit: RCSI

Researchers from the HSE Epilepsy Lighthouse Project and FutureNeuro, the SFI Research Centre for Chronic and Rare Neurological Diseases hosted by RCSI, have developed a new genomics module in the Irish National Epilepsy Electronic Patient Record (EPR) system.

The work illustrates how an electronic health system can support the integration of genomic test results and new genetic knowledge into routine clinical care in the public health system. This new system will facilitate more personalised forms of medicine.

The research, funded by eHealth Ireland, the Health Service Executive (HSE) and Science Foundation Ireland, is published in the latest issue of the journal *Epilepsia*

Many adults and children with epilepsy of unknown cause now undergo genomic testing. An accurate genetic diagnosis can bring great value to the individual, their family and the clinical team. As a result of this research, many people now understand why they have epilepsy. For some, this has been a decades long journey of multiple treatments and no explanation or knowledge of the actual underlying cause for their condition.

"We now know that much of previously unexplained epilepsy is due, in part, to damaging variants in a person's genome," said Prof. Norman Delanty, Associate Professor at RCSI, FutureNeuro Investigator and Consultant Neurologist at Beaumont Hospital.

"The potential to understand the reason for a particular person's epilepsy at a molecular level, and to use this information to develop personalised therapies will become a significant advancement in the way we practice medicine."

Ireland has a world-leading national EPR system designed specifically for epilepsy. This system captures, in great depth, the subtle patient features relevant to specialist care and allows for quicker access to key clinical data to better support people with complex chronic diseases such as epilepsy. In 2015, the HSE and eHealth Ireland designated the national Epilepsy EPR as a "Lighthouse" project for the country to help build an understanding of the quality, safety, and efficiency benefits of EPRs. The Lighthouse project combined the emerging fields of genomics and EPRs to promote personalised medicine and improved healthcare for people with epilepsy.

"The epilepsy EPR system is one of the largest, most detailed collections of active epilepsy eHealth records in the world," said Mary Fitzsimons, FutureNeuro Epilepsy eHealth Lead and Director of the Epilepsy Lighthouse Project at RCSI. "To our knowledge, the epilepsy genomics module we have developed is the first such specific system in the world. We believe the combined power of genomics and electronic patient records has the capability of enhancing, and in some cases transforming, the

practice of medicine."

The new Epilepsy EPR module facilitates regular multidisciplinary meetings between clinicians, geneticists, bioinformaticians, and other team members, where they review data from genomic testing to determine if there is an identifiable genetic cause for a person's epilepsy.

"Diagnostic genomic testing is a rapidly growing area in clinical medicine, but there is much work to be done to understand the most effective way to integrate this powerful information into patient care. We hope this new eHealth technology can inform how genomics is integrated into the Irish healthcare system and act as an example for other diseases beyond epilepsy," said Gianpiero Cavalleri, FutureNeuro Deputy Director and Professor of Human Genetics at RCSI. "Having this data available in a person's secure electronic record enables multidisciplinary teams to quickly make better decisions about a person's treatment options."

More information: Norman Delanty et al, Development of a genomics module within an epilepsy?specific electronic health record: Toward genomic medicine in epilepsy care, *Epilepsia* (2019). DOI: 10.1111/epi.16278

Provided by RCSI

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