

Striving for a National Service to Utilise Genomic Data in Health Care

The data obtained from the human genome will become part of health care decision-making. Combining a patient's genomic data with the information available on the current state of the patient's health enables the development of new algorithms, making it possible for a doctor to quickly select the best possible treatment and medication for the patient.



Medications have different effects due to the individual nature of a person's genome. For example, some antibiotics cause drug allergies. The body may break down the medication before it has time to take effect, or the patient may experience harmful side effects. That is why utilising genomic data in pharmacotherapy will reduce the number of incorrect prescriptions. On the other hand, if a person knows that he/she has

a digestion-related genetic trait that augments or weakens, for example, the breaking down of caffeine into energy and building materials, that knowledge may have a positive effect on his/her lifestyle. In the future, the algorithms of genetic databases linked to electronic patient record systems could automatically warn against possible adverse drug reactions and provide advice on the most effective alternative.

In Finland, CSC - IT Center for Science, the National Institute for Health and Welfare (THL) and the Institute for Molecular Medicine of the University of Helsinki are creating a secure framework for storing the genomic data produced on Finns and interpreting the data for health care purposes. The aim of the Helsinki University Hospital (HUS), which is involved in the cooperation, is to investigate the benefits of



digital health data on humans for research and care. The six-month pilot project is part of an assignment given to the Genome Center to be established in Finland, coordinated by the Ministry of Social Affairs and Health.

Cardio Compass: a tool for assessing your health risks

Storing data becomes cheaper and capacity grows year after year. An exemplary file on the data collected on the health of Finnish people is the FINRISK cohort of THL. The analyses of the data collected for decades on Finns have been developed further in the GeneRISK project studying the hereditary risk factors for cardiovascular diseases. An algorithm that calculates the risk points for an individual to suffer from cardiovascular diseases is tested at the same time. A tool called Cardio Compass provides people with their current risk level and the development of the risk over the next few decades.

Cardio Compass is tested in practice by recruiting and testing 10,000 people from the Kotka region, the customer base

of Mehiläinen and blood donors in Helsinki. The people participating in the project receive important feedback on their own health, more accurately than ever before, through the combination of genomic data. The information is collected in Cardio Compass. The participants may also talk directly with experts on the interpretations made based on the data.

Algorithms help with medication selection

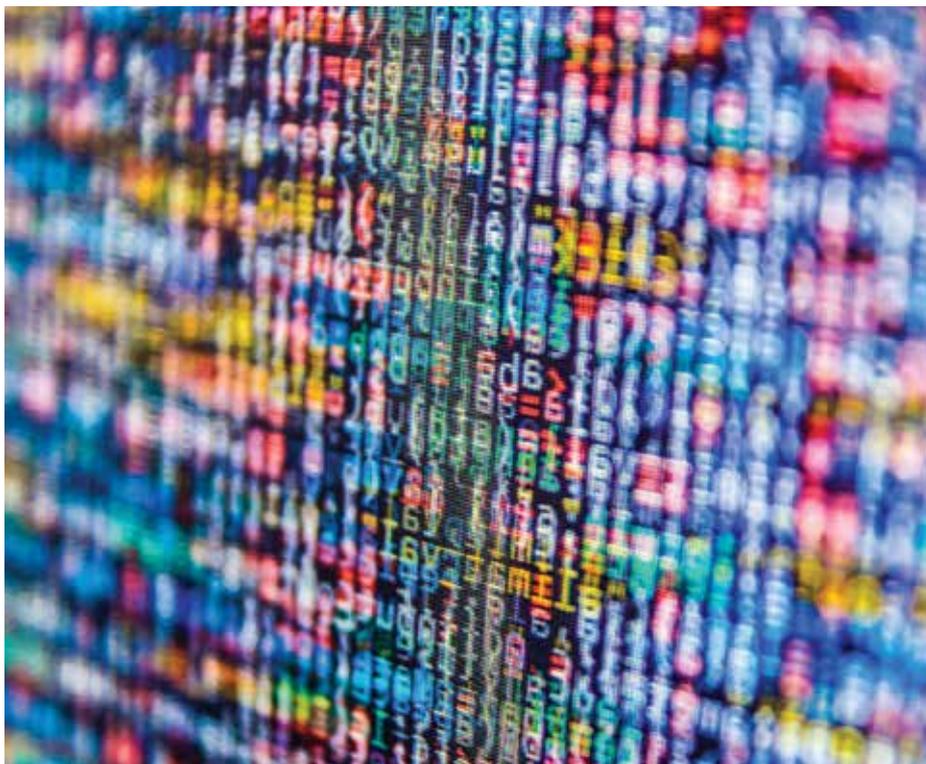
In April 2016, the Finnish Government decided to establish a Genome Center in Finland with the aim of introducing genomic data as part of health care. In order to build the functions of the Genome Center, the data already collected and stored from the Finnish population will be utilised and combined in research which, if successful, will improve the accuracy of prescriptions. It would be possible to determine suitable medications or rule out the poor ones based on the patient's genomic data. Algorithms can be developed to select a suitable drug ingredient and to optimise the amount of medication with standardised

software methods. This is called pharmacogenetics.

In 2016, Professor Mikko Niemi from Biomedicum Helsinki was granted substantial funding by the European Research Council for a project to develop an algorithm for finding a suitable cholesterol drug for a patient. The mathematical model takes into account the patient's genome, other medication, gender, age and weight.

However, effective utilisation of algorithms requires that there is enough different data available on patients. It is important to know the quality and purpose of the data. Sufficient metadata describes the quality of the data, based on which decisions on the utilisation of the data can be made. The interpretation of the data will become easier once a functional technical distribution platform is provided for reference data, making it possible to design better interpretation algorithms for the data.

Creating interpretation algorithms for genetic data for clinical use is the long-term goal. In addition to algorithms helping doctors to, for example, determine the appropriate medication, they can even be



suitable for predicting changes in the function of proteins. The goal is that once the interpretation algorithms are ready for clinical use, they would be available in patient information systems automatically instead of as a request for information to be ordered separately.

New technological expertise to Finland

Most of the technologies exist; we just have to be able to connect them. Expertise is attracted to Finland, for example, as part of European cooperation. The Finnish node of the ELIXIR infrastructure, which operates in connection with CSC, is building the secure infrastructure necessary for the management and storage of genomic data.

In the project, information technology is applied to the sample and data files of THL's biobank. The aim of the project is to adapt genomic data so that it can be used by Finnish doctors and researchers

in the best way possible. The full genome of about 9,000 Finns (www.sisuproject.fi) has already been determined through the digitalisation of this THL resource and other important Finnish sample collections, but the genomic data of up to half a million Finns has been discussed.

The project brings together the technological expertise of THL, HUS and CSC in Finland. The future goal is that this type of data would be analysed by a large group of Finnish bio-industry experts from universities, the public sector and companies in the bio-industry. Just storing the data is not enough; a service that covers the utilisation of all biological data must be created. At present, the expertise of the parties storing and providing the data is not sufficient for all possible health applications. Thus, the implementation of the pilot will provide important guidelines on how the efficient storage and secure distribution of genomic data can be carried out in cooperation between organisations so that the

data can be fully utilised in health care, research and future innovations.

The question is largely whether Finland wants a specialised infrastructure the size of a small factory and expertise on the management and further processing of common genomic data based on which the data interpretation system would be built or if we want to outsource the data infrastructure services elsewhere.

In many countries, genomic data covering the entire country is a challenging goal. The services of the Finnish Genome Center are taking shape, and they will be created in cooperation with the parties managing the data, such as biobanks and licensing authorities. The data resources coordinated by the Genome Center are securely available for utilisation. In the future, all Finns could thus have their own health and welfare profiles that would include the data on their own genome.

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FURTHER INFORMATION:

CSC – IT Center for Science

is a non-profit, state-owned company administered by the Ministry of Education and Culture. CSC maintains and develops the state-owned, centralised IT infrastructure.

<http://www.csc.fi>

<https://research.csc.fi/cloud-computing>

ELIXIR

builds infrastructure in support of the biological sector. It brings together the leading organisations of 21 European countries and the EMBL European Molecular Biology Laboratory to form a common infrastructure for biological information. CSC – IT Center for Science is the Finnish centre within this infrastructure.

<http://www.elixir-finland.org>

<http://www.elixir-europe.org>

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