New mathematical model provides 'disease causation index'

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MedUni Vienna researchers are able to establish whether a complex disease has a genetic or environmental cause . Credit: Medical University of Vienna

Patients with complex diseases have a higher risk of developing others. Multi-morbidity represents a huge problem in everyday clinical practice, because it makes it more difficult to provide successful treatment. By analysing data from all over Austria, Peter Klimek and Stefan Thurner have developed a mathematical model that can be used to distinguish whether a disease has a genetic or environmental cause.

It is well known in medical research today that a range of diseases are genetic, while others are caused by environmental factors, or it might be a combination of both. One of the main objectives within medicine is to gain an exact understanding of the main causes of complex multifactorial diseases. Such diseases include diabetes, COPD and asthma, for example. With a new set of phenotypic data of diseases in combination with molecular biological data and modern complex system mathematics, it is possible to understand the interaction between genetic and environmental disorders in the development of complex diseases.

The science of complex systems aims, amongst other things, to identify relevant information within the "big data ocean" that leads to clinically relevant knowledge and hence to better treatment options. At MedUni Vienna's Section for Science of Complex Systems, data networking is being used to develop mathematical models and network theories covering a broad spectrum, from communication pathways in cell systems to analysis of the public healthcare system.

In a study recently published in *Scientific Reports*, Klimek and Thurner succeeded for the first time in developing a new method that can be used to establish whether various complex diseases are caused by genetic or environmental factors. By comparing molecular networks with networks of the common diseases that occur in the Austrian population, they were able to calculate a so-called Geneticity Index. If a disease has a high geneticity index, there is a high probability that it is of genetic origin. On the other hand, if this index is low, environmental influences can be further investigated in order to tell, for example, whether the disease is caused by chemical toxins or problems with molecular signalling pathways. Says Stefan Thurner: "The main value of the study lies in the fact that we are able to match practically all available molecular data with the data of the actual diseases so that we can see the causes of the disease. And this is possible without having genetic data for the patient."

One of the main findings of the study is that diseases are usually caused by entirely genetic or entirely environmental factors. It is rare for a disease to be caused by both mechanisms at the same time. "A better understanding of the causes of the disease provides us with a much better starting point for any treatment," explains Peter Klimek. The new Disease Causation Index might also help to improve diagnosis.

The study was conducted in collaboration with the Federation of Austrian Social Insurance Institutions, which helped the researchers to analyse the underlying anonymised research datasets for around eight million patients over a period of two years. Currently, the researchers are working on applying complexity research methods to pharmaceuticals, in order to systematically identify any potential unexpected compound actions in millions of possible combinations. Only recently, they were able to show that the combined use of insulin and statins can drastically reduce the cancer risk of diabetics on insulin treatment. The long-term goal is to use highly innovative data science to advance personalised medicine.

More information: Peter Klimek et al. Disentangling genetic and environmental risk factors for individual diseases from multiplex comorbidity networks, *Scientific Reports* (2016). DOI: 10.1038/srep39658

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