Artificial intelligence tracks down leukemia

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Artificial intelligence can detect one of the most common forms of blood cancer—acute myeloid leukemia (AML)—with high reliability. Researchers at the German Center for Neurodegenerative Diseases (DZNE) and the University of Bonn have now shown this in a proof-of-concept study. Their approach is based on the analysis of the gene activity of cells found in the blood. Used in practice, this approach could support conventional diagnostics and possibly accelerate the beginning

of therapy. The research results have been published in the journal *iScience*.

Artificial intelligence is a much-discussed topic in medicine, especially in the field of diagnostics. "We aimed to investigate the potential on the basis of a specific example," explains Prof. Joachim Schultze, a research group leader at the DZNE and head of the Department for Genomics and Immunoregulation at the LIMES Institute of the University of Bonn. "Because this requires large amounts of data, we evaluated data on the gene activity of blood cells. Numerous studies have been carried out on this topic and the results are available through databases. Thus, there is an enormous data pool. We have collected virtually everything that is currently available."

Fingerprint of Gene Activity

Schultze and his colleagues focused on the "transcriptome," which is a kind of fingerprint of gene activity. In each and every cell, depending on its condition, only certain genes are actually "switched on," which is reflected in their profiles of gene activity. Exactly such data—derived from cells in blood samples and spanning many thousands of genes—were analysed in the current study. "The transcriptome holds important information about the condition of cells. However, classical diagnostics is based on different data. We therefore wanted to find out what an analysis of the transcriptome can achieve using artificial intelligence, that is to say trainable algorithms," said Schultze, who is member of the Bonn-based "ImmunoSensation" cluster of excellence. "In the long term, we intend to apply this approach to further topics, in particular in the field of dementia."

The current study focused on AML. Without adequate treatment, this form of leukemia leads to death within weeks. AML is associated with the proliferation of pathologically altered bone marrow cells, which can

eventually enter the bloodstream. Ultimately both healthy cells and tumor cells drift in the blood. All these cells exhibit typical gene activity patterns, which were all considered in the analysis. Data from more than 12,000 blood samples—these came from 105 different studies—were taken into account: the largest dataset to date for a metastudy on AML. Approximately 4,100 of these blood samples derived from individuals diagnosed with AML, the remaining ones had been taken from individuals with other diseases or from healthy individuals.

High Hit Rate

The scientists fed their algorithms parts of this data set. The input included information about whether a sample came from an AML patient or not. "The algorithms then searched the transcriptome for disease-specific patterns. This is a largely automated process. It's called machine learning," said Schultze. Based on this pattern recognition, further data were analysed and classified by the algorithms, i.e. categorized into samples with AML and without AML. "Of course, we knew the classification as it was listed in the original data, but the software did not. We then checked the hit rate. It was above 99 percent for some of the applied methods. In fact, we tested various methods from the repertoire of machine learning and artificial intelligence. There was actually one algorithm that was particularly good, but the others were close behind."

Application in Practice?

Put into application, this method could support conventional diagnostics and help save costs, said Schultze. "In principle, a blood sample taken by the family doctor and sent to a laboratory for analysis could suffice. I guess that the cost would be less than 50 euros." Classical AML diagnostics includes a variety of methods. Some of these cost a few

hundred euros per run, Schultze noted. "However, we have not yet developed a workable test. We have only shown that the approach works in principle. So we have laid the groundwork for developing a test."

Schultze emphasised that the diagnosis of AML will continue to require specialised physicians in the future. "The aim is to provide the experts with a tool that supports them in their diagnosis. In addition, many patients go through a real odyssey until they finally end up with a specialist and get a diagnosis." Because in the early stages the symptoms of AML can resemble those of a bad cold. However, AML is a life-threatening disease that should be treated as quickly as possible. "With a blood test, as it seems possible on the basis of our study, it is conceivable that the family doctor would already clarify a suspicion of AML. And when the suspicion is confirmed, the patient is referred to a specialist. Possibly, the diagnosis would then happen earlier than it does now and therapy could start earlier."

More information: Stefanie Warnat-Herresthal et al, Scalable prediction of acute myeloid leukemia using high-dimensional machine learning and blood transcriptomics, *iScience* (2019). DOI: 10.1016/j.isci.2019.100780

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