Computational biologists simplify diagnosis for hereditary diseases

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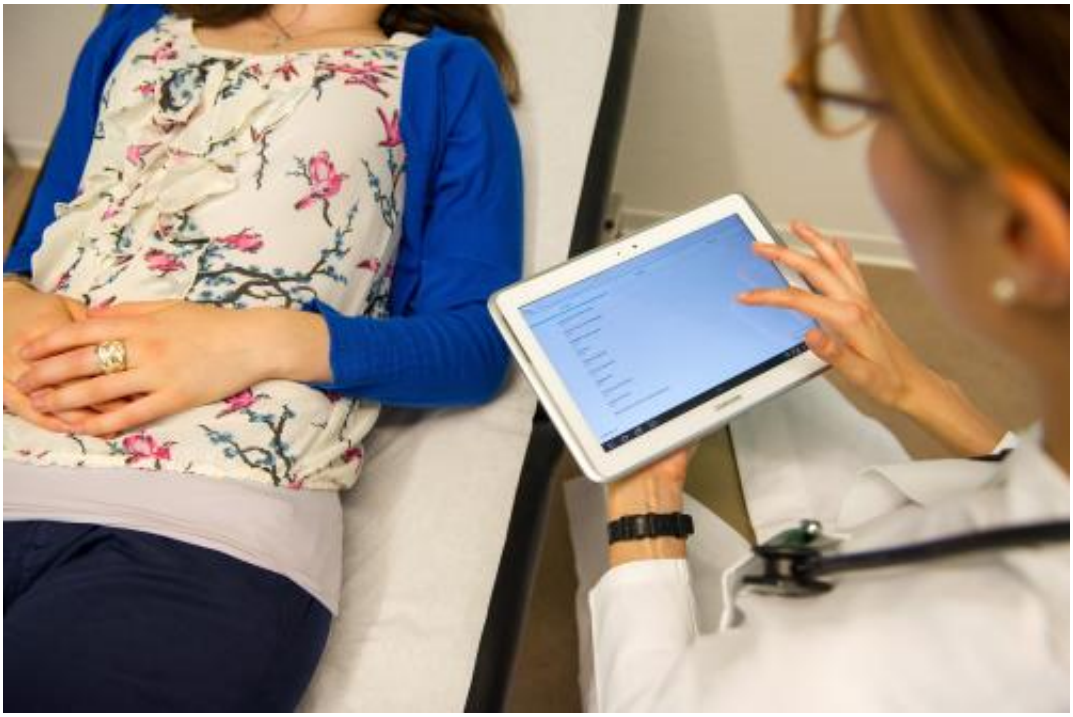

The programm Phenomizer as app. Credit: Oliver Dietze

In the case of a cough or a sore throat, the doctor can usually diagnose a common cold immediately. However, the diagnosis of hereditary illnesses like cystic fibrosis, which affects the metabolism, or Huntington's disease, which leads to cognitive decline, is much more complex. A patient may suffer from a multitude of symptoms, pointing to several different diseases. This can now be remediated using a

program developed by bioinformatics experts from Saarbrücken, which is now also available as an app. With the aid of this application, physicians can discover patients' afflictions quickly and without great research effort. The computing method that the program is based on compares different patterns of hereditary diseases from an extensive online database and weights them by their likelihood.

Diseases like diabetes, epilepsy, a heart defect or deafness can themselves be symptoms of a range of hereditary diseases. "That makes it so difficult for medical specialists to diagnose someone with the correct disease from the beginning", says Marcel Schulz, who is leader of the research group " High-throughput Genomics & Systems Biology" at the Max Planck Institute for Informatics and also a researcher at the Cluster of Excellence "Multimodal Computing and Interaction". "Additionally, each disease appears with different characteristics in different patients". In the case of a heart defect, for instance, the patient may not only be afflicted by the defect itself, but could also be suffering from the Miller-Dieker syndrome or Cat eye syndrome, depending on the patient's other symptoms.

Together with physicians and computational biologists from the working group of Professor Dr. Peter Robinson at the Charité clinical center in Berlin, Schulz has developed the program "Phenomizer", which can be used by doctors to discover what the patient is afflicted with. This approach can be used for various hereditary illnesses like trisomy 21, Morbus Wilson or the Marfan syndrome. "We are using an extensive online data base developed at Charité, called the 'Human Phenotype Ontology',,which lists more than 10 000 disease characteristics structurally and assigns them to 7500 diseases", explains Schulz. The computing method scans, compares and weights the data related to the symptoms the user provided, and then assigns these characteristics to certain diseases. Within seconds, the doctor receives a list with the most probable results. The advantage of the program is clear to Schulz: "The

doctors no longer have to research in databases or books for several hours. The list supports them in detecting the disease more quickly. Moreover, doctors can ask patients about their symptoms in greater detail. This makes it easier to assess which aspects they need to pay attention to.

The Phenomizer program has recently been made available online as an Android version for smartphones and tablets. It can be downloaded for free from the "Google Play" platform. "We developed the app together with six different computer scientists from Saarbrücken", explains Schulz. The students created the app within the context of a software engineering course at Saarland University.

The Phenomizer app is available for free on Google Play.

More information: Sebastian Köhler, Marcel H. Schulz, Peter Krawitz, Sebastian Bauer, Sandra Dölken, Claus E. Ott, Christine Mundlos, Denise Horn, Stefan Mundlos, Peter N. Robinson, "Clinical Diagnostics in Human Genetics with Semantic Similarity Searches in Ontologies," *The American Journal of Human Genetics*, Volume 85, Issue 4, 9 October 2009, Pages 457-464, ISSN 0002-9297, DOI: 10.1016/j.ajhg.2009.09.003.

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