Optical genome mapping provides more precise information on types of leukemia

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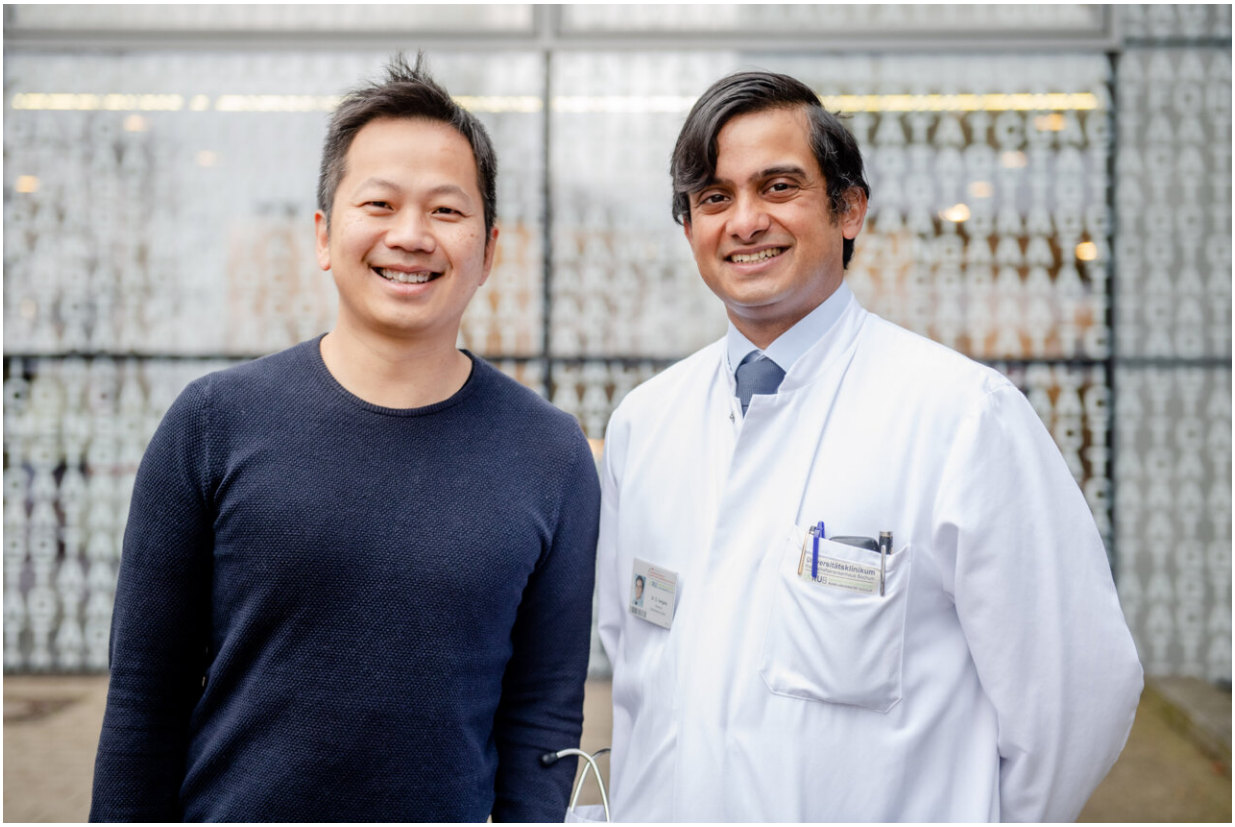

Chair Huu Phuc Nguyen (left) and Deepak Vangala from the Miners' Hospital ensured close cooperation between their departments. Credit: RUB, Marquard

Optical genome mapping involves the extraction of very long DNA molecules, for example routinely collected blood samples or bone

marrow material from patients. These long DNA molecules are labeled with dye molecules at more than half a million different positions in the entire human genome and are then moving through ultrathin nanochannels on a special chip. As the DNA molecules move through the nanochannels, a laser is used to make them visible and they are photographed using a fluorescence microscope. The images of the entire genome are then analyzed using bioinformatic analyses. "The aim is to identify and interpret changes in genetic regions that are relevant for the development of cancer," explains Dr. Wanda Gerding from the Bochum Department of Human Genetics.

Optical genome mapping thus facilitates genome-wide analysis of regions that are important for the classification and therapy of leukemias using one methodology. Furthermore, it also allows the identification of new relevant genomic regions and new genes.

Reliable and additional results

In the current study, the team compared the methodology to current standard diagnostics in patients with acute myeloid leukemia as well as myelodysplastic syndromes. The researchers showed that the results obtained by optical genome mapping methodology were concordant in 93 percent of samples compared toa conventional methodology, the so-called cytogenetic karyogram, where whole chromosomes are visualized. In 67 percent of the samples, it was even possible to obtain additional genetic information.

The methodology can thus not only detect structural changes in the genome more accurately, but also has the potential to become an important component of routine diagnostics for patients with leukemia. "As a further benefit, genome research can provide data and new insights for further research work in the field of tumor biology," says Wanda Gerding.

More information: Wanda M. Gerding et al, Optical genome mapping reveals additional prognostic information compared to conventional cytogenetics in AML / MDS patients, *International Journal of Cancer* (2022). DOI: 10.1002/ijc.33942