New method to analyze function and genetic variation in cells in leukemias and other cancer diseases

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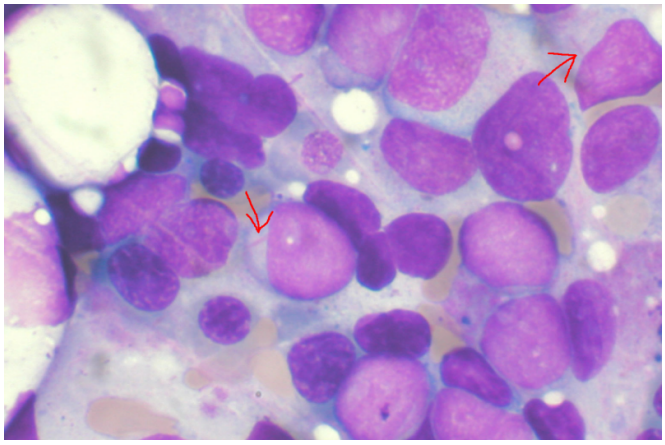

Bone marrow aspirate showing acute myeloid leukemia. Several blasts have Auer rods. Credit: Wikipedia

Martin Enge's research group at the department of Oncology-Pathology has developed a new method for joint analysis of a cell's state and accumulated genetic variation in single cells, applied to childhood leukemias and other cancers. The article is published in the journal *Molecular Cell*.

The method enables studies of how these genetic changes alter transcription, and thereby a cell's function. The study shows that it is technically feasible and cost-efficient to do such analyses in thousands of single cells, including cells from biobanked material routinely sampled from children with leukemia.

We know today that in most cancers, every malignant cell in a tumor is not identical. An individual's leukemia or tumor is often composed of many subgroups of cells, or clones, that may differ slightly in important ways and affect response to treatment.

Conventional molecular genetic techniques are excellent for describing the average of thousands or millions of tumor cells. But within that average, there may be smaller groups of cells whose properties only become apparent when analyzed independently.

To expose this underlying heterogeneity is important, both for the development of more targeted cancer therapies, but also to understand how different clones interact to sometimes evade current treatments.

The new method is broadly applicable to study any cancer type, and hopefully it will help understand how cancer cells evolve within the tumor and sometimes escape treatment.

More information: Vasilios Zachariadis et al. A Highly Scalable Method for Joint Whole-Genome Sequencing and Gene-Expression Profiling of Single Cells, *Molecular Cell* (2020). DOI: 10.1016/j.molcel.2020.09.025

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