

Researchers develop new software for unlocking cancer's ancestry

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Could knowing where your ancestors came from be the key to better cancer treatments? Maybe, but where would that key fit? How can we trace cancer's ancestral roots to modern-day solutions? For Cold Spring

Harbor Laboratory (CSHL) Research Professor Alexander Krasnitz, the answers may lie deep within vast databases and hospital archives containing hundreds of thousands of tumor samples.

Krasnitz and CSHL Postdoctoral Fellow Pascal Belleau are working to reveal the genealogical connections between [cancer](#) and race or ethnicity. They've developed [new software](#) that accurately infers continental [ancestry](#) from tumor DNA and RNA. Their latest study is published in *Cancer Research*, and their work may help clinicians develop new strategies for early cancer detection and personalized treatments.

"Why do people of different races and ethnicities get sick at different rates with different types of cancer? They have different habits, living conditions, exposures—all kinds of social and [environmental factors](#). But there may be a [genetic component](#) as well," Krasnitz says.

Krasnitz's team trained their software tools using hybrid DNA profiles. They created these profiles from cancerous and unrelated cancer-free genomes of a known background. They then tested the software's performance against pancreatic, ovarian, breast, and blood cancer specimens from patients with known ancestry. The team found the software matched their hybrid profiles to continental populations with over 95% accuracy.

"We have a good model to build on," Krasnitz says. "But very few individuals come from a single ancestry. We're all mixed to some extent. So now we're working to look deeper, test tumor samples of unknown ancestry, reveal ancestral mixtures, and achieve more regional specificity." How specific? For now, think West Africa as opposed to East Africa.

Krasnitz and Belleau recently joined a colorectal cancer study in collaboration with Northwell Health and SUNY Downstate Medical

Center. The study allows them to explore how [colorectal cancer](#) mutates genes in different ways depending on specific races or ethnicities. They hope to further refine their software to infer ancestry of not only whole genomes but every individual sequence therein.

"If we can identify more localized ancestries that are susceptible to different cancers or other aggressive diseases, it could help us pinpoint the specific part of the genome responsible and target it for treatment," Belleau says.

Right now, a simple DNA swab can tell you where you came from and which diseases you stand to inherit. In the future, it might give you the means to beat them too.

More information: Pascal Belleau et al, Genetic Ancestry Inference from Cancer-Derived Molecular Data across Genomic and Transcriptomic Platforms, *Cancer Research* (2022). [DOI: 10.1158/0008-5472.CAN-22-0682](#)

Provided by Cold Spring Harbor Laboratory

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