

Scripps physicians call for change in cancer tissue handling

January 4 2013

Genetic sequencing technology is altering the way cancer is diagnosed and treated, but traditional specimen handling methods threaten to slow that progress.

That's the message delivered this week in a column appearing in the <u>Journal of the American Medical Association</u> (*JAMA*) by Scripps Clinic physicians Eric Topol, Kelly Bethel and Laura Goetz.

Dr. Topol is a cardiologist who serves as chief academic officer of Scripps Health and director of the Scripps Translational Science Institute (STSI), leading Scripps' <u>genomic medicine</u> research efforts. Dr. Bethel is a pathologist, and Dr. Goetz is a general surgeon and a researcher at STSI.

"Deciding how best to obtain (tumor) samples and how best to process them for whole genome or exome sequencing is a pivotal yet unresolved issue with several layers of complexity," the doctors wrote. "As the new clinical applicability of genomics emerges at a fairly rapid rate, the field of pathology will arrive at a tipping point for a fundamental change in how <u>cancer specimens</u> are handled."

Currently, tumor tissue obtained through a biopsy is fixed in formalin, a mixture of formaldehyde and water, and embedded in paraffin for microscopic viewing. However, because the <u>chemical mixture</u> damages DNA, sequencing tissue processed in this way can be difficult, if not impossible.



A better alternative is to also routinely freeze a portion of the specimen, which retains the tissue's genetic coding while preserving it for future analysis. In order to have enough tissue to freeze, larger or additional biopsy samples may be required, especially when using minimally invasive <u>needle biopsy</u> procedures.

"We need to completely rethink the way we have collected and stored <u>cancer tissue</u> samples for decades," said Dr. Topol, "It's becoming increasingly clear that obtaining an accurate map of a tumor's DNA can be the key to determining the specific mutations that are driving a person's cancer, how best to treat it and how likely it is to recur."

Even though complete genetic evaluations of tumors might require higher sample-storage costs and a more invasive biopsy procedure, most patients would likely agree to that option if it translates into a better diagnosis and possible treatment, the authors wrote.

Evidence of such benefit must come from randomized clinical trials that compare detailed genetic evaluation of tumor tissue with the current standard of care for cancer patients, they said.

"Drs. Goetz, Bethel and Topol's editorial acknowledges kindly the critical role pathologists play in patient care," said Dr. Stanley Robboy, president of the College of American Pathologists. "This type of change will require discussion about new operative standards, which will need the cooperation of surgeons, pathologists, ethicists and, of course, appropriate patient consents. It's these types of implications we will need to consider and incorporate as a progressive healthcare agenda is moved forward."

Patients and their advocates have a direct role to play in this transformation and should raise questions about tumor specimen storage and genetic testing with their doctor, said Dr. Topol, who outlines a



consumer-led digital health revolution in his book "The Creative Destruction of Medicine."

Provided by Scripps Health

Citation: Scripps physicians call for change in cancer tissue handling (2013, January 4) retrieved 3 May 2024 from https://medicalxpress.com/news/2013-01-scripps-physicians-cancer-tissue.html

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