Genetic testing cost effective for newly diagnosed gastrointestinal stromal tumors
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Because gastrointestinal stromal tumors (GIST) are sensitive to the targeted small molecule therapy imatinib, oncologists tend to treat all patients with metastatic GIST with this drug. However, because this rare type of cancer is caused by different genetic mutations, imatinib does not help all patients equally.

To determine whose cancer may be most responsive, the National Comprehensive Cancer Network suggests that patients undergo genetic testing to identify each individuals' tumor mutations. And yet, only 30 percent of patients have genetic testing at the time of diagnosis, likely due to concerns over cost and utility of testing, said Jason Sicklick, MD, professor of surgery in the Division of Surgical Oncology at University of California San Diego School of Medicine.

"We recommend that all patients with a new diagnosis of metastatic GIST undergo genetic testing prior to the initiation of first-line chemotherapy," said Sicklick, surgical oncologist and co-leader of the Sarcoma Disease Team at Moores Cancer Center at UC San Diego Health. "In doing so, those who are unlikely to benefit from imatinib can be given a treatment better suited for their individual tumor."

In a paper published online on September 29, 2020 in the journal JAMA Network Open, Sicklick and colleagues reported that genetic testing is cost-effective and beneficial for newly diagnosed patients with metastatic GIST, a type of soft tissue sarcoma that develops in specialized nerve cells in the wall of the digestive system, most often occurring in the stomach or small intestine.

The team developed a model to compare the cost effectiveness of targeted gene testing and personalized therapy to patients with metastatic GIST who were prescribed imatinib (marketed as Gleevec). Data analyses were conducted October 2019 to January 2020.

"Genetic testing is cost-effective as it allows clinicians to prescribe chemotherapy in a tumor-specific manner. Patients who would not benefit from imatinib, because of primary tumor resistance, are given alternative therapy," said first author Sudeep Banerjee, MD, who did the research while in the Sicklick lab at Moores Cancer Center and who is now chief resident at David Geffen Medical School at UCLA. "Avoiding ineffective treatment and reduced rates of disease progression are the reasons why genetic testing is cost-effective."

Although the most common sarcoma, GIST is rare with an estimated annual incidence of 6.8 cases per million people in the United States. Eventually this cancer becomes highly resistant to existing drug therapies.

Clinicians fight the growth with progressively aggressive drugs, the downside being that each line of therapy has diminishing effectiveness and higher toxicity for patients. More than 95 percent of
patients eventually succumb to drug-resistant GIST, underscoring the importance of starting patients on the most effective available drug not only because it is most cost-effective, but crucial for maintaining and improving quality of life in these patients, said Sicklick, co-corresponding author on the paper.

"The treatment of cancer is becoming an increasingly personalized process," said Banerjee. "There is a rapidly expanding body of research around gene-specific and even mutation-specific therapies that can be effective independent of the site of origin of a given tumor. Genetic testing provides the necessary information for patients to potentially benefit from those novel therapies."


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