UC San Diego Health

Cancer-associated Mutations are Common in Patients with Unexplained Low Blood Counts

Researchers use genetic tools to propose a new diagnostic category

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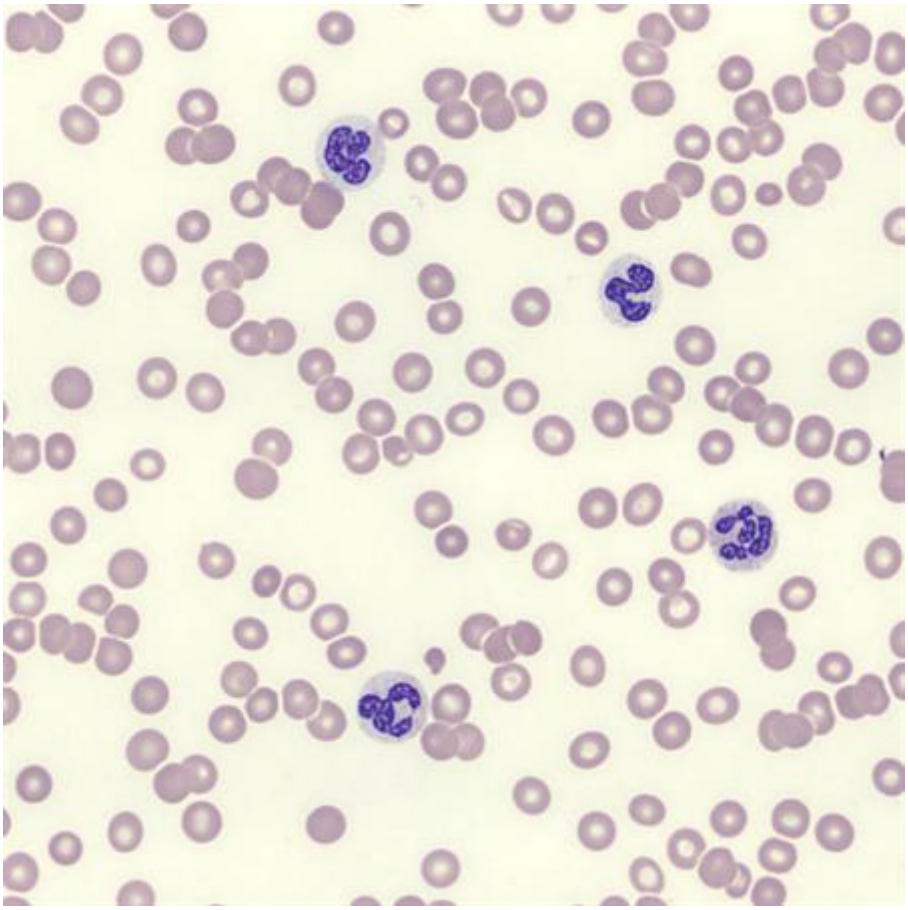
Patients with unexplained low blood counts and abnormally mutated cells who do not fit the diagnostic criteria for recognized blood cancers should be described as having clonal cytopenias of undetermined significance (CCUS), suggest University of California, San Diego School of Medicine researchers in a recent paper published in the journal *Blood*. The researchers found the condition surprisingly common in older patients with low blood counts.

Diagnosing the cause of anemia and other low blood counts, called cytopenias, can be challenging. The data show that a significant number of patients with unexplained cytopenias have genetic mutations in their blood and bone marrow cells that are typical of blood cancers, such as myelodysplastic syndromes (MDS) or acute myelogenous leukemia (AML). Despite a bone marrow biopsy, many patients will not be diagnosed with any recognized disorder, said Rafael Bejar, MD, PhD, assistant professor of medicine and senior author of the study.

"We don't know to what extent patients who have low blood counts and mutations are at increased risk of developing an overt malignancy," said Bejar, a hematologist specializing in blood cancers at Moores Cancer Center at UC San Diego Health. "We hope that by defining CCUS, future studies will follow these patients to learn what these mutations mean for their future as their genetically abnormal cells may represent early stages of subsequent blood cancers."

Bejar describes the mutated cells in CCUS patients as "polyps" of the bone marrow. Just as polyps detected in colonoscopies represent growths capable of evolving into cancers, the mutated blood cells in CCUS patients may be the precursors to cancers of the blood like MDS and AML.

Presently, patients with unexplained cytopenias who do not meet the criteria for a MDS diagnosis, are described as having idiopathic cytopenias of undetermined significance (ICUS). Bone marrow samples analyzed by Genoptix Medical Laboratory revealed that 35 percent of ICUS patients carry MDS-associated somatic mutations and can be described as having CCUS. Knowing that CCUS



Abnormal Blood Cells

and MDS patients share similar mutations may have diagnostic utility once outcomes in CCUS patients are better understood.

Researchers found that CCUS incidence among patients with cytopenias is more common than MDS. However, Bejar suggests that larger, carefully controlled studies will be needed to confirm the findings.

Co-authors include Brian Kwok, Jeff M. Hall, Yin Xu, Prashanti Reddy, Keming Lin, Rachel Flamholz, Bashar Dabbas, Aine Yung, Jenan Al Hafidh, Emily Balmert, Christine Vaupel, Carlos El Hader, Matthew J. McGinniss, Shareef A. Nahas, Julie Kines, Genoptix Medical Laboratory; John S. Witte of UC San Francisco.

Disclosures: Hall, Kwok, Reddy, Lin, Flamholz, Yung, Xu, Hafidh, Balmert, Vaupel, Hader, McGinniss, Nahas, Kines, and Dabbas are employees of Genoptix. Witte has served as a consultant for Genoptix. Bejar has served both as a consultant and member of the Scientific Advisory Boards for Genoptix and Celgene, he sits on a Celgene steering committee, has intellectual property licensed by Genoptix and has been paid to speak at a Genoptix event.

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