NEWS BRIEFS

New gene variant linked to malaria protection

A gene variant that has been hiding in plain sight may provide protection against malaria in much the same way that the sickle cell trait does, according to new research published 22 March 2018 in the journal *Cell*. The novel human gain-of-function allele PIEZO1, E756del, turns out to be present in a third of the African population and in many African Americans. A similar variant produces hereditary xerocytosis, which dehydrates red blood cells. Shang Ma, a research associate at The Scripps Research Institute (TSRI) and first author of the study, explained in an accompanying news release that the team uncovered the new variant while investigating the underlying cause of this disease. Follow-up studies in a mouse model of the disorder showed that the newly discovered variant, a deletion of one codon, impedes growth of malaria parasites in red blood cells. Genomic studies confirmed that it is prevalent in regions where malaria is endemic. Like sickle cell, the E756del PIEZO1 variant produces partially dehydrated red blood cells; however, the health effects for individuals carrying two copies of the variant are unclear. The variant is rarely found in non-African populations and had not previously been studied in a population study. "This study is a good example of a host/pathogen arms race playing out in real-time—this time with the host a likely winner," said Kristian Andersen, assistant professor at TSRI and director of Infectious Disease Genomics at the Scripps Translational Science Institute. She and Ardem Patapoutian of TSRI led the study, which was conducted by an international team of scientists.

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—Karyn Hede, News Editor

Medicare patients with cancer gain access to genomic testing

In recognition that many forms of personalized cancer treatment available today require genomic tumor testing to match tumors with biologic treatment, the Centers for Medicare & Medicaid Services (CMS) will cover next-generation sequencing (NGS) companion diagnostic testing. The coverage will apply to patients with advanced recurrent, metastatic, relapsed, or refractory cancer. The unprecedented coverage decision was explained as reflecting a recognition that genomic testing is a necessary companion to identify genetic mutations that are targets of biologic cancer treatment. Without the tests, oncologists would be making treatment decisions without all the necessary information. Specifically, the decision followed parallel review with the approval by the US Food and Drug Administration (FDA) of the FoundationOne CDx test, which matches mutations with 15 targeted therapies for solid tumors and can detect genetic mutations in 324 additional genes. The FDA approved the assay (marketed by Foundation Medicine, Cambridge, MA) for the newly established Breakthrough Device Program, which was created specifically to accommodate advanced diagnostic tests such as NGS. "These tests can help doctors consult with patients about more targeted care or enrollment in a clinical trial," said Kate Goodrich, CMS's chief medical officer. CMS will also cover FDA-approved or cleared companion diagnostics for any test with a clear match to an FDA-approved treatment for advanced cancer. In addition, CMS provided a clear signal that it wants to see investigators publish the results of clinical studies in this arena, especially on the endpoints of overall survival, progression-free survival, objective response, and patient-reported outcomes relevant to the quality of life for Medicare beneficiaries.

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