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## Automated assistance with copy-number variant calls

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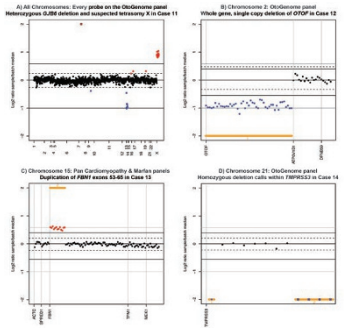

Software to assist clinical genomic laboratories in interpretation of complex diagnostic results is becoming essential as the volume and pace of testing continue to accelerate. Here, investigators from the Laboratory for Molecular Medicine, Partners HealthCare Personalized Medicine, Boston, report they have developed a software program to detect and visualize germ-line copy-number variants (CNVs), which can then be confirmed by a trained technician. The open-source software, called VisCap, and accompanying training documents are freely available at http://www.github.com/pughlab/viscap. Designed for use by clinical laboratories, the software allows the setting of laboratory-defined thresholds and standardized procedures. The developers validated the product in their own clinical laboratory by using the tool to conduct CNV analysis on more than 4,000 patients. In addition to data normalization and identification of candidate CNVs, the software provides visualization tools to facilitate quality control through manual review of results. As part of their validation process, trained technicians blinded to results scored VisCap plots for 27 candidate CNVs as either true-positive or false-positive calls. All four technicians correctly identified all 10 verified CNVs. But two of four technicians flagged what were false positives for follow-up, highlighting the need for training and experienced review of data. *—Karyn Hede, News Editor*

## Better understanding of cystic fibrosis needed in Africa

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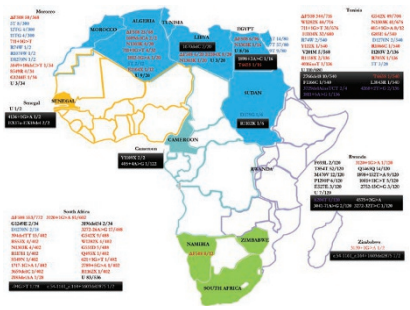

Cystic fibrosis (CF) is commonly thought of as a disease affecting primarily Caucasians, but of course it also occurs in people of a wide variety of other ethnic heritages. Yet, although mutations leading to CF are distributed globally, in many places a detailed understanding of the disease's incidence and molecular epidemiology is absent. In this issue, Stewart and Pepper, of the University of Pretoria, South Africa, review the literature on molecular analysis of CF in Africa. The disease has been recognized among Africans only since the late 1960s, and the extent of underlying genetic variants, particularly in non-Caucasian populations, remains unknown. Therefore, the use of a standard genetic test for diagnosis of CF would not be practical or realistic in Africa. Developing an effective diagnostic test, the authors point out, would require sequencing the entire *CFTR* gene in people suspected of having CF and using aggregate data to identify the prevalence of each variant. To provide a better understanding of the current state of knowledge about CF in Africa, the team summarizes mutational analysis in patients suspected to have CF reported from 12 of the 54 African states. Of the mutations reported, 39 are known to cause CF, others are of uncertain significance, and 21 are unique to Africa. The authors propose a systematic investigation of the nature and extent of the disease to ensure that "the public health needs of African CF patients—both those in Africa and those of African descent living elsewhere—are met." *—Karyn Hede, News Editor*

# NEWS BRIEFS

## Woman in famous Wyeth painting suffered from genetic disorder

Its gripping imagery has captivated art lovers for decades, but Andrew Wyeth's almost universally recognizable painting *Christina's World* still has a story to tell. The painting's subject, Wyeth's friend and neighbor Christina Olson, suffered from a progressive neurodegenerative disorder that, although undiagnosed, had been attributed to the effects of polio. Now a modern medical investigation suggests that she actually had a form of Charcot-Marie-Tooth disease (CMT), which would explain the progressive difficulty with movement that eventually made her unable to walk or use her hands. After closely examining the evidence pertaining to her condition—including close scrutiny of the painting itself, which was executed with photorealistic precision—Marc Patterson, a professor of neurology, pediatrics, and medical genetics at the Mayo Clinic in Rochester, Minnesota, made the diagnosis of early-onset CMT. He presented his findings at the 23nd annual Historical Clinicopathological Conference, held May 2016 at the University of Maryland School of Medicine. "This was a fascinating case," Patterson stated in a news release issued by the meeting organizers. "This painting has long been a favorite of mine, and the question of Christina's ailment was an intriguing medical mystery. I think her case best fits the profile of this disease." *—Karyn Hede, News Editor*