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Similar outcomes seen in colorectal cancer of differing genetic origin

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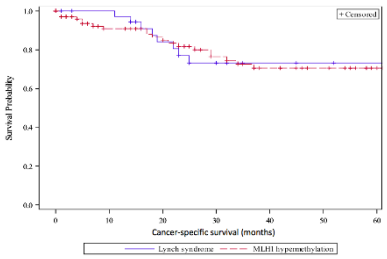

Defects in DNA mismatch repair (MMR) account for about one in seven cases of colorectal cancer. However, this umbrella grouping includes both the inherited disorder Lynch syndrome (secondary to mutations in several different genes) and sporadic gene inactivation (most typically through hypermethylation of the *MLH1* gene promoter (*MLH1-hm*)). Measurable molecular differences in the two types of tumors include *BRAF* mutations, which occur in 60–70% of sporadic tumors but very rarely in LS-associated tumors. Yet the effect of these distinct origins of MMR deficiency on tumor behavior and patient outcome has not been systematically explored. Now Haraldsdottir *et al*. report a study in which, despite differing molecular origins of the MMR deficiency among tumors, there were no statistically significant differences in pathologic features or overall patient outcome. The research team retrospectively explored differences in clinical presentation and outcomes in 189 consecutive patients accrued between May 1998 and May 2012 at The Ohio State University Comprehensive Cancer Center, Columbus, Ohio. Both categories of patients had exceptionally good cancer-related survival for both stage I (5-year, 100%) and stage II disease (5-year, 90%). Based on their findings, the researchers conclude that the prognosis for colorectal cancer patients whose tumors contain defects in MMR genes is the same regardless of whether a mutation existed in the germ line or the deficiency occurred sporadically. *—Karyn Hede, News Editor*

Direct-to-consumer genetic testing fills in health risk gaps for adoptees

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Atomic Imagery/©Getty Images

The uniquely human desire to understand our origins motivates nearly all adoptees who choose to purchase a direct-to-consumer (DTC) genomic test, according to a new study presented in this issue. The Impact of Personal Genomics (PGen) Study, a collaboration between academic researchers and industry scientists from 23andMe and Pathway Genomics, has been evaluating the motivations behind consumer interest in DTC genomic tests. For individuals not biologically related to their parents, family health history may be scant or nonexistent. The current study showed that lack of access to knowledge about their genetic makeup often motivated a personal search for genetic information through DTC tests. The exploratory analysis compared baseline analyses and six-month follow-up of 1,607 participants, including 80 adoptees and 1,527 nonadoptees, who used personal genomic testing (PGT). Participants self-reported demographic information, psychosocial characteristics, family health history, and motivations for seeking PGT. Compared with nonadoptees, adopted individuals were more motivated by their limited knowledge of their family health history and desire to learn their personal disease risk. Half of adoptees and nonadoptees factored perceived actionability of results into their purchasing decision. Both groups were more interested in learning about their own ancestry and disease risk than in their carrier status. Investigators concluded that concerns that adoptees might place too much weight on test results seem unfounded, but they emphasized the need for further study of the long-term health impact on adoptees who receive genetic information. *—Karyn Hede, News Editor*

NEWS BRIEFS

Machine learning to crunch genomic data in search of cancer cures for veterans


Martha Hoelzer

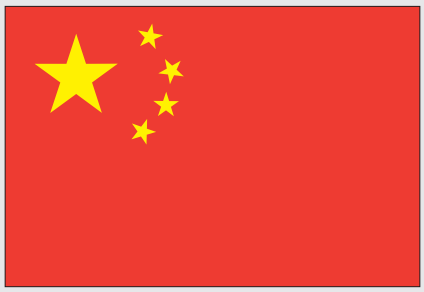
After two years of accumulating data from top-tier cancer research institutions, the IBM Watson for Genomics machine learning system is being put to the test solving complex cancer cases for US veterans. IBM is donating access to Watson for Genomics to Veterans Affairs (VA) hospitals nationwide, as part of Vice President Joe Biden's Cancer Moonshot program. The VA estimated that it would be able to increase the number of patients who receive targeted therapy by 30-fold using the technology. The increase in caseload is possible because Watson can review an individual's genomic data and incorporate the latest findings from clinical reports and medical literature within a few minutes, producing evidence-based recommendations on treatments that may be more likely to work with the individual's unique DNA profile. The system is designed to learn continuously, to understand complex questions, and to respond in natural language, as demonstrated by its performance on the TV show *Jeopardy!* several years ago. Over the next two years, it is hoped that 10,000 veterans could be treated with targeted cancer therapies based on the insights provided by the Watson for Genomics system. Further, the collaboration could allow patients who do not live near major academic medical centers access to the

NEWS BRIEFS *(continued)*

insights and experience of physicians in those institutions to guide their treatment. *—Karyn Hede, News Editor*

China investing in genomic medicine on an industrial scale

While the National Institutes of Health was busy touting its Precision Medicine Initiative (PMI), China threw down the gauntlet with its identically named initiative, announced in March 2016. Whereas the United States is investing $215 million in funding for the first year, China announced an investment of US $9.2 billion over 15 years for its PMI. Working in parallel with the Chinese government, DNA-sequencing powerhouse Beijing Genomics Institute is expected to inaugurate the China National Genebank in Shenzhen by September, according to reporting in a special June 2016 issue of the journal *Nature* devoted to science in China. The Genebank will house samples and data not only of an expected 1 million human genomes but also genomes of plants, animals, and microbes. The government initiative includes investments not only in DNA sequencing technologies but, perhaps just as importantly, in information technology (IT) infrastructure. The China Precision Medicine Cloud will provide the IT infrastructure for organizing, mining, and sharing genomics big data throughout China and beyond. Far from being cloistered away, the Cloud meets international data standards for information exchange and is designed to be interoperable with technology used by the US Food and Drug Administration. The China initiative includes plans to sequence more



than 1 million human genomes and then use that information, along with clinical and health data, to develop new drugs and diagnostics. Academic and industry partnerships, backed by massive government funding, could accelerate China's ascendance to a world power in genomic research and its application to medicine. *—Karyn Hede, News Editor*