TOUCHING BASE

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Mutant of the Month

Images courtesy of Andrea Gallavotti and Robert Schmidt

This month we highlight *barren stalk1*, a spontaneous maize mutant identified in 1928. Homozygous *barren stalk1* plants have defective lateral meristems, the groups of stem cells that initiate growth of lateral structures. They don't produce vegetative branches, female inflorescences (ears) or normal male inflorescences (tassels). Tassels from a *barren stalk1* plant (left) and a wild-type plant (right) are shown above. Andrea Gallavotti and Robert Schmidt and colleagues determined that the *barren stalk1* mutation is the result of a helitron insertion in the *ba-1* gene, which encodes a basic helix-loop-helix transcription factor that presumably has a role in regulating the expression of genes controlling meristem formation (*Nature* 432, 630–635; 2004). Because plant architecture is an important agricultural trait, *barren stalk1* may bring insight into the process by which early farmers turned wild plants into domesticated crops. Like *teosinte branched1*, another gene involved in the control of maize architecture, there is some evidence that *barren stalk1* variants were under selection during the domestication of maize from its wild progenitor, teosinte. ***EN***

Array-CGH diagnostics

Quest Diagnostics has introduced the first commercial test based on array comparative genome hybridization (CGH), designed to detect chromosomal aberrations, including submicroscopic losses and gains, underlying common forms of idiopathic mental retardation and developmental delay. The test, called ClariSure, uses an array composed of 1,300 bacterial artificial chromosomes (BACs), with probes arrayed in triplicate and enriched at subtelomeric and pericentromeric regions and at regions corresponding to known dosage-abnormality syndromes. The array-CGH analysis is complemented with classical Giemsa-banded chromosome analysis to detect larger chromosomal aberrations, and positive results are verified using specific fluorescence *in situ* hybridization (FISH) assays. The company is also developing similar tests designed to identify chromosomal abnormalities associated with hematological malignancies such as leukemia, which they expect to make available by the end of the year. The introduction of these tests, which offer substantially higher resolution and efficiency than traditional cytogenetic methods, marks an important shift in the use of array-CGH–based technologies in the clinical diagnostic setting. *KV*

Written by Orli Bahcall, Emily Niemitz, Alan Packer & Kyle Vogan

"The individual whose genome is described in this report is J. Craig Venter, who was born on 14 October 1946, a self-identified Caucasian male. The DNA donor gave full consent to provide his DNA for study via sequencing methods and to disclose publicly his genomic data in totality."

– Levy, S. *et al. PloS Biology* **5**, e254 (2007).

Genomics goes personal

In a venture toward the anticipated 'personal genomics' era, J. Craig Venter and colleagues recently published the diploid genome sequence of an individual human (*PloS Biol.* **5**, e254; 2007). In addition to the paper describing the challenges faced in sequencing and assembly and reporting summary analyses, an interactive poster of his complete genome is available on the journal's website. Overall, they found 99.5% similarity between the two chromosomal copies of this individual genome, termed HuRef. The publication also highlights characterization of genetic variation in the genome, through comparisons to a reference sequence (NCBI 36). They found 3.1 million SNPs, with non-SNP variation accounting for only 22% of events detected, but 74% of variant bases. This included a high proportion of insertion/deletion events, as well as large-scale structural variation. In May of this year, another individual genome, that of James Watson, was reported at a press conference, as part of a collaboration with the Baylor College of Medicine Human Genome Sequencing Center and 454 Life Sciences. These first reports of individual genome sequences provide important guidance in terms of the technological challenges faced in sequencing and additional reference sequences to guide future individual genome sequencing. In addition to the ethical, legal and social questions individual genome sequencing raises, we are left asking: who will be the next volunteer? *OB*

News you can use

The Genetic Alliance has announced the launch of the National Consumer Center for Genetics Resources and Services (NCCGRS), a five-year, $500,000-per-year project funded by the US Department of Health and Human Services. The NCCGRS is essentially an open-access searchable website offering a variety of resources related to genetic disease, genetic testing, newborn screening, advocacy, policy debates and related topics. The press release notes that "the diagnosis of a genetic disorder is a life-altering event and the difficulty of accessing information adds to the stress of coping with a diagnosis or managing a chronic genetic condition." The NCCGRS will "mitigate the substantial information and resource deficit for consumers of genetic services." There will also be two 'Wiki'-type resources offered. WikiGenetics (wikigenetics.org) will serve as a user-generated, up-to-date encyclopedia of human genetics that should be accessible to individuals with no scientific background. WikiAdvocacy (wikiadvocacy.org) offers an online, interactive community where members of disease-specific advocacy organizations can share their experiences and offer advice. Even in these early stages, each is already an impressive synthesis of links and authoritative information. Visit the Center at http://www.geneticalliance.org. *AP*