



TOUCHING BASE

QUESTIONS? THOUGHTS? IDEAS?
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Mutant of the Month

This month we are highlighting medaka *b* mutants, which have reduced melanin formation as seen in the bottom two animals in the photo. Identification of mutations in the *AIM1* gene at the B locus by Shoji Fukamachi and colleagues was the first successful positional cloning of a gene in this organism (*Nat. Genet.* 28, 381–385; 2001). *AIM1* encodes a predicted integral membrane protein with similarity to sucrose transporters. Other groups have since discovered that mutations in mammalian orthologs (also known as *MATP* or *SLC45A2*) cause pigmentation disorders: the *underwhite* mutant in the mouse, oculocutaneous albinism type 4 in humans and cream coat color in horses. Although the function of *SLC45A2* in melanocytes remains to be determined, it is clear that it has an important, evolutionarily conserved role in pigmentation. Accordingly, common variants in *SLC45A2* associate with normal skin pigmentation variation in human populations (*Hum. Mutat.* 25, 278–284; 2005). A recent genome-wide association study has identified multiple common variants associated with hair and eye pigmentation in humans (*Nat. Genet.* 39, 1443–1452; 2007); many of these variants are also in genes that, when mutated, cause pigmentation disorders in mice and humans. **EN**



Nat. Genet. 28, 381–385; 2001

Fort Lauderdale blues

There has been a great deal of commentary accompanying the publication of the sequences of ten *Drosophila* genomes. Most of this commentary has focused on the substance of the papers themselves, but tucked away in a recent piece by Michael Ashburner (*Genetics* 177, 1263–1268; 2007) is an important point about the procedures involved in publishing these papers. Ashburner notes that “it is an open secret that there has been very considerable community disquiet in the way in which the publications on these sequences have been prepared.” Community resource projects such as this one have, by general agreement, been governed by the Fort Lauderdale principles. These principles set out a “tripartite sharing of responsibility,” the linchpin of which is the responsibility of end users of the data to respect the right of the data producers to publish the first global analysis. As Ashburner rightly points out, this principle leaves open the question as to how long the community at large needs to wait before publishing analyses of the data. Although there have been occasional problems in this regard in the past, Ashburner suggests that the lack of clear guidelines has been particularly problematic in the context of work on the fly genomes, where several breaches of the policy apparently have already occurred. In

Written by Emily Niemitz, Alan Packer and Kyle Vogan

light of the enormous amount of data that will continue to be generated by these community projects, Ashburner calls for a fresh look at the conditions that may be attached to the use of data coming out of such public programs. **AP**

“This work represents a tremendous scientific milestone, the biological equivalent of the Wright brothers’ first airplane. It’s a bit like turning lead into gold. But this is not over by a long shot. It’s extremely important to temper this announcement with caution.”

- Robert Lanza, chief scientific officer of Advanced Cell Technologies, on the successful reprogramming of adult human somatic cells to pluripotent stem cells by Japanese and US researchers (as quoted by MSNBC.com).

Finding the right chemistry

If you’ve been unlucky in love, and yet are lucky enough to be living within an hour’s drive of Boston, you might be interested in a new match-making service called ScientificMatch (<http://scientificmatch.com/>). Inspired by the famous “sweaty T-shirt experiment,” which showed that variation in the MHC complex influences body odor discrimination and mate choice preferences, the company collects DNA samples from its clients and seeks to find matches on the basis of, among other things, maximizing MHC diversity between prospective partners. According to the website, benefits of this MHC-based matching process include enhanced sexual attraction, greater fidelity, increased fertility and healthier offspring. Complementing the DNA analysis is a psychological matching system and, of course, the requisite background checks for past criminal convictions, bankruptcy filings and other potential turnoffs. The price tag for this service (\$1,995.95 for a one-year membership) may seem a bit steep, but on the other hand, it’s encouraging to think that the valiant efforts of the original T-shirt-sniffing subjects are being applied to a practical end. And, as the saying goes, it’s hard to put a price on love. **KV**

Overlooked oversight of genetic testing

The personal genomics industry is rapidly expanding, with frequent announcements of companies launching individual genotyping or sequencing services offered directly to the public for the purpose of assessing risks of developing common diseases. As discussed in our December 2007 editorial titled “Risky business,” there is an urgent need for more research into how to usefully incorporate these genetic data, which offer risk assessments that most would consider provisional at best, into an individual’s disease prevention strategy. A related unresolved issue is the application of regulatory oversight of these services to ensure that accurate information on test validity, not to mention clinical utility, is available to the consumer or health-care provider. The need for increased attention to the oversight of genetic testing came to light recently at a summit on genetic testing titled “Eyes on the Prize: Truth Telling about Genetic Testing” convened by the Genetic Alliance on September 20–21 in Washington, DC. A preliminary meeting report is available on the Genetic Alliance’s website (<http://www.geneticalliance.org>). In a related development, on November 5 the Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS) issued a draft report on the oversight of genetic testing (available at <http://www4.od.nih.gov/oba/SACGHS.HTM>). The report mentions “significant gaps” in genetic testing oversight and offers numerous recommendations for enhancing the oversight of genetic testing in the US. **EN**