TOUCHING BASE

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Mutant of the month

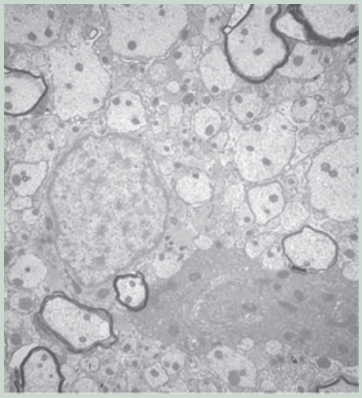

Image courtesy of Klaus Nave and Ian Griffiths

This month we highlight the *jimpy* mouse mutant, which carries a point mutation in the X-linked gene *Plp1*, encoding the myelin proteolipid protein. The *jimpy* mutation arose spontaneously at the Institute of Animal Genetics in Edinburgh and was first reported in the literature in 1954. Male *jimpy* mice have severely deficient myelination of the central nervous system; they develop tremors and convulsions and die prematurely. The image above shows a cross section of optic nerves from a *jimpy* mouse; most axons are unmyelinated, and the occasional myelin sheaths that are present (identified by dark rings) are thin. There are several animal proteolipid protein mutants, including the *rumpshaker* mouse, the *myelin-deficient* rat and the *shaking pup* dog, and mutations of *PLP1* in humans cause Pelizaeus-Merzbacher disease. Interestingly, mice entirely lacking Plp develop morphologically normal oligodendrocytes and myelination and have only late-onset neurological dysfunction due to axonal changes. This not only implicates point mutations of *PLP1* in toxic gain-of-function effects but also indicates that oligodendrocytes have a myelin-independent role in maintaining axonal integrity. *EN*

Profiling test wins FDA approval

MammaPrint, a test that uses expression profiling to predict breast cancer recurrence risk, has been cleared for marketing by the US Food and Drug Administration, marking the first time that such a multivariate prognostic test has won FDA regulatory approval. The test, developed by the Amsterdam-based company Agendia, profiles expression of 70 markers in primary tumor samples and classifies cancers according to their likelihood of recurrence. Clinically, the test seems to be particularly useful for identifying individuals at low risk for recurrence, which could help inform decisions regarding follow-up therapies in which the potential benefits of treatment are weighed against the risks of recurrence and the established side effects of current treatment regimens. Although similar diagnostic tests without FDA approval are currently available in the US as so-called 'home brew' tests, the FDA has signaled that it intends to bring at least a subset of these tests under regulatory control. These developments mark a shift in the FDA's stance toward this category of molecular diagnostics and is likely to accelerate the acceptance and widespread use of such signature-based tests in clinical practice. *KV*

Preprints and slow boats

The National Library of Medicine has added a selection of the papers of Rosalind Franklin to its 'Profiles in Science' website. The papers come from several different periods of Franklin's career, shedding light on topics ranging from the technical aspects of her crystallographic work to her personal relationships with her mentors, contemporaries and students. Correspondence with James Watson, Francis Crick, Donald Caspar, J.D. Bernal, Alexander Rich, Aaron Klug, Sydney Brenner and others is included, all in the form of that endangered species, the type-written letter. One can read letters dealing with Franklin's struggle for an adequate salary and adequate equipment, as well as advice and constructive criticism of drafts and preprints of her manuscripts that she sent to colleagues for comment. The letters inevitably evoke the slower pace of a different scientific era, with one letter from Rich to Franklin standing out in this regard. Rich wrote to Franklin in May 1955: "Something amusing happened in that a few days ago I received a very formal letter from the Editors at NATURE complaining about the fact that I had not sent my proof in, and asking whether I had received it. I answered no, I had not received it, and a few days later it arrived with a three halfpenny stamp on it. They thought they were sending it airmail and instead it took a tramp steamer across the ocean getting here. However, it should be out pretty soon I imagine". *AP*

"If you have 1 million fewer nucleotides than your buddy, shouldn't you get a break on your golf handicap?"
—Steve Scherer, as quoted in ***Bio IT World***

Genomic Science Education

The National Center for Research Resources (NCRR) recently announced that it is granting $1.35 million to the University of Utah's Genetic Science Learning Center, which is directed by Louisa Stark, for the development of an educational program entitled 'Genome Science for Health: Web-Based Curricula for Biology'. The grant falls under the auspices of the NCRR's Science Education Partnership Award Program. The Genome Science for Health program will develop web-based curriculum modules on cell biology, developmental biology, molecular genetics and clinical trials in order to illustrate how medical treatments are developed. The target audience for the program will be high school students and teachers. The website of the Genetic Science Learning Center (http://learn.genetics.utah.edu/) currently has a variety of educational information on genetics and genome science as well as feature stories on topics varying from transgenics in farming to telomeres in cancer. *EN*

Touching Base written by Emily Niemitz, Alan Packer and Kyle Vogan.