TOUCHING BASE

QUESTIONS? THOUGHTS? IDEAS?
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Mutant of the Month

Photo credit: János Szabad

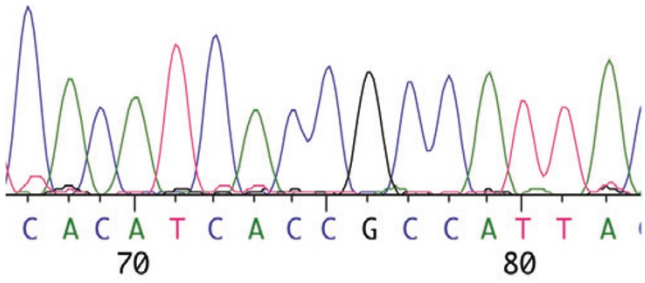
Named for extinct aristocracy, János Szabad's dominant female sterile mutations of *Drosophila* hold court at the pinnacle of sophistication in advanced transmission genetics. Like extinct aristocracy, they are somewhat refined in their breeding, requiring both a balancer and a dominant male sterile or translocation to maintain them as a true-breeding stock. *Fs(3) Horka* bears the noble surname denoting a military judge among the ninth-century Magyar. The mutant females have meiotic defects, and their embryos arrest during the initial cleavage divisions; this maternal-effect sterility provides selection for recessive loss-of-function revertants. The fly depicted here is half male and half female, with the boundary right down the middle. This XX/XO gynandromorph results from the dominant paternal effect of *Fs(3) Horka*. The X chromosome and autosomes become unstable during spermatogenesis, and they, but not the Y chromosome, may then be lost from the early embryo. This mutation has been used as a reliable way to generate gynanders at frequencies of 2–20%, depending on genetic background. The paternal effect also results in equational nondisjunction during spermiogenesis. ***MA***

CDC to test newborn testing

Approximately 1 in 800 babies is born with a severe or potentially lethal inborn error of metabolism or other genetic condition that is readily detectable and treatable (see editorial at http://www.nature.com/ng/journal/v36/n11/full/ng1104-1127.html). We estimate from the American College of Medical Genetics (ACMG) report (http://www.mchb.hrsa.gov/screening/) that of the infants born in the US with one of the 29 genetic conditions for which the ACMG says infants should be tested, about one-third (or about 1,600 per year) are not detected owing to state-by-state variation. Some states test for as few as three conditions, while others test for up to 43. In those states where testing is mandated, detection, follow-up and treatment may not occur as intended because of errors in record-keeping, misunderstandings between doctor and parents, births in rural areas and the remarkable mobility of the US population. The number of missed cases has not been assessed since 1986, so it is encouraging that the US Centers for Disease Control (CDC) and the National Newborn Screening and Genetics Resource Center have now agreed to undertake the sensitive task of estimating the proportion of children left untreated. We hope that the study will lead to better application of existing genetic knowledge to the health of those babies born with genetic conditions. Those interested in helping with this project should contact Brad Therrell at http://genes-r-us.uthscsa.edu/contact.htm. *MA*

Publishing sequence traces

Following our request for tips for publishing high-quality figures incorporating sequence traces, Tom Schwei and Anne Stover at DNASTAR pointed out that researchers using either a Mac or a PC can cut and paste from their Lasergene SeqMan sequence alignment software into Adobe Illustrator and get very high quality EPS files for publication. This particular sequence viewer allows stretching and zooming in on the sequence traces and as a consequence it is important initially to paste the clip into a vector graphics program like Illustrator and not a photo editing program like Adobe Photoshop. We have determined that there are also open-source and freely available trace viewers and vector graphics programs for those who want to work out how to use them. We would be pleased to continue the discussion of publishing optimal traces on our blog (http://blogs.nature.com/ng/freeassociation/). *MA*



FINDbase provides mutation frequency data

Providing a centralized gathering place for National and Ethnic Mutation Databases (NEMDBs), FINDbase (http://www.findbase.org), operating under the coordination of George Patrinos, is up and running. NEMDBs, usually maintained by curators who are experts in the field, compile mutation frequency data on inherited disorders in specific populations. The data comes from publications, conference proceedings and communications with researchers. These databases fill the gap left by locus-specific databases that do not provide researchers with the distribution of mutation frequencies in various ethnic or national groups. NEMDBs aim to assist genetic diagnostic services by providing reference information for the interpretation of diagnostic test results. Also, since the history of a population is linked to the history of its genetic variants, NEMDBs can help elucidate the origins and migration patterns of populations. FINDbase links together NEMDBs operating through an upgraded version of the ETHNOS software and provides mutation frequency data in various populations worldwide. At the time that *Nature Genetics* went to press, FINDbase contained frequency data for 17 disorders and 18 genes within 86 populations, covering 1,220 mutations. *EN*

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http://www.nature.com/ng/journal/v38/n6/full/ng0606-613.html