



TOUCHING BASE

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Mutant of the month



XY PIS^{+/+}

XX PIS^{-/-}

This month we revisit the mutation that causes the polled intersex syndrome (PIS) in goats. The identification of the genetic alteration responsible for PIS was reported in our pages in 2001 (*Nat. Genet.* 29, 453–458; 2001). This is a spontaneously occurring mutation causing polledness (absence of horns) in males and females and sex reversal in XX animals. The dominant polled phenotype (illustrated in the XX male goat at right) is considered a desirable trait, but breeders noticed abnormal sex ratios in polled herds, with an excess of sterile males; it was then recognized that the mutation underlying PIS causes recessive sex reversal in homozygous females and infertility in homozygous males. The PIS mutation does not involve alterations of the *SRY* testis-determining gene, which is another cause of XX males. Instead, this mutation is a deletion of an 11.7-kb region on goat chromosome 1. There is no evidence of a coding sequence in the deleted fragment; it is presumed to be a regulatory element governing expression of nearby genes involved in sex determination pathways. The expression of at least two linked genes, *PISRT1* and *FOXL2*, is altered by the deletion. Although the function of *PISRT1* is not known, mutations in *FOXL2* cause blepharophimosis ptosis epicanthus inversus syndrome (BPES) in humans (*Nat. Genet.* 27, 159–166; 2001). Females with this syndrome develop premature ovarian failure but are not sex reversed, suggesting that *FOXL2* is involved in ovary differentiation, whereas *PISRT1* may act to inhibit testis formation. **EN**

SNP-IT

The American Society of Human Genetics (ASHG) has launched a quarterly online newsletter called SNP-IT (Society News and Progress–IT). The first issue can be found at <http://www.ashg.org/genetics/ashg/snplit>, and it includes a variety of features and useful information for ASHG members and other interested parties. Current ASHG President Wylie Burke and immediate Past

President Steve Warren open with a letter describing the Society's activities in 2006 in the legislative, research, and educational arenas, as well as a plug for the upcoming annual meeting to be held in San Diego in October. Brief profiles of the board of directors follow, marked by answers to everyone's favorite question: if your office were on fire, what one object would you take with you? (Steve Warren provides the most original answer, saying he would grab his 1907 edition of Reginald C. Punnett's *Mendelism*.) Summaries of recent articles in the *American Journal of Human Genetics* are next, followed by a description of the ASHG's NSF-funded project to create a network of alliances between professional geneticists and secondary school teachers. The newsletter closes with a call for nominations for the Society's major awards, and a 'Dear Genie' Q & A section for trainees, complete with its very own genie logo. Society members with ideas for columns are invited to contact ASHG Director of Education Kenna Mills Shaw at kshaw@ashg.org. **AP**

New centers for translational medicine

Spotlight: Eric Topol, Scripps Genomic Medicine. Scripps Health has recently launched a new Genomic Medicine program in collaboration with The Scripps Research Institute (TSRI), all based in San Diego or La Jolla, California. Eric J. Topol is the director of the Genomic Medicine Program as well as chief academic officer for Scripps Health and professor of translational genomics at TSRI. The program involves three new initiatives: Scripps Genomic Medicine, the Scripps Translational Science Institute and Scripps Advanced Clinical Trials. These initiatives are geared towards understanding the genomic basis of health and disease and will support basic research and clinical programs. At a recent press conference, Topol explained that we are at a turning point in medicine that offers the chance to move toward individualized medicine. The Genomic Medicine program will take this opportunity to move toward genetics-based clinical trials and move from gene discovery to gene-based clinical medicine, he said. Kelly A. Frazer, formerly of Perlegen Sciences, Inc., where she was vice president of genomics, has joined as the director of genomic biology. Nicholas J. Schork, previously at the University of California San Diego, is the director of research, and Sarah Shaw Murray, the program's director of genetics, previously directed the genotyping science group at Illumina, Inc. **OB**

Spotlight: Claire Fraser, University of Maryland School of Medicine Institute of Genome Sciences. Claire M. Fraser-Liggett, currently director of the Institute for Genomic Research (TIGR) in Rockville, Maryland, announced in April that she will move to head the new Institute of Genome Sciences at the University of Maryland School of Medicine. This comes amid efforts by the University to advance its standing in the areas of applied genetics and translational medicine. Fraser-Liggett has announced plans leave her position as director of TIGR and move to the University of Maryland over the next few months and plans to bring seven to eight top scientists with her. The institute is expected to include 80–100 researchers and staff and is to be officially launched over the summer of 2007. The new institute will be located at the BioPark, adjacent to the Baltimore campus of the University. Fraser-Liggett founded TIGR together with J. Craig Venter; Venter later went on to found Celera Genomics, and subsequently the J. Craig Venter Institute (JCVI), although he remained on the board of TIGR. In the fall of 2006, TIGR became a division of JCVI. **OB**

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