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## Breakthrough of the Year

THE BREAKTHROUGH OF THIS YEAR HAS TO DO WITH HUMANS, GENOMES, AND GENETICS. But it is not about THE human genome (as if there were only one!). Instead, it is about your particular genome, or mine, and what it can tell us about our backgrounds and the quality of our futures.

A number of studies in the past year have led to a new appreciation of human genetic diversity. As soon as genomes are looked at individually, important differences appear: Different single-nucleotide polymorphisms are scattered throughout, and singular combinations of particular genes forming haplotypes emerge. A flood of scans for these variations across the genome has pointed to genes involved in behavioral traits as well as to those that may foretell deferred disease liability. And more extensive structural variations, such as additions, deletions, repeat sequences, and stretches of “backwards” DNA, turn out to be more prevalent than had been recognized. These too are increasingly being associated with disease risks.

High-throughput sequencing techniques are bringing the cost of genomics down. The few “celebrity genomes” (e.g., Watson’s and Venter’s) will soon be followed by others, we hope in an order not determined by wealth but by scientific need or personal medical circumstance. Our natural interest in personal genealogy, accompanied by worries about our health, will create an incentive structure that even now is creating a sometimes dubious niche market for having one’s genome “done.”

A strong Breakthrough runner-up arrived at this year’s finish line just in time. Two new studies, one published in *Science*, showed how adult human epithelial cells could be reprogrammed, through the virally mediated introduction of just four genes, to behave like pluripotent cells; that is, able to act as embryonic stem cells do, to produce every descendent cell type. This breakthrough has produced some relief, but it also comes with some reservations. James Thompson of the University of Wisconsin, who did the first research with embryonic stem cells, has now taken a major step toward ending the “ethical” controversy over their use. But hold on: That controversy was generated by specific objections from one religion, not some universal ethic. There is every reason to continue research along the old path, with embryo-derived cells: The new methods may carry unknown liabilities, so making the case for changing Bush’s 2001 presidential order should continue.

Finally, readers will notice that we usually have a “Breakdown” of the year. That custom produced ambivalence this time around. On the strictly scientific front, progress in climate change research was spectacular. There was new information about the dynamics of the major ice sheets in Greenland and Antarctica, analyses of paleoclimates, new estimates of sea-level rise, and studies of the impacts of global warming on high-latitude ecosystems and sea ice. The Intergovernmental Panel on Climate Change delivered a summary report at year’s end emphasizing the seriousness of the risks. But on the breakdown side, continual denial by the Bush Administration added to its long history of failing to mitigate the emission of greenhouse gases.

A specimen case of the Administration’s reluctance to acknowledge climate change was added just recently when Julie Gerberding, head of the U.S. Centers for Disease Control and Prevention, was asked to present congressional testimony on the potential impacts of climate change on public health. It is surely no secret that heat spells are a health hazard, or that drought and excess rainfall can influence human susceptibility to pathogen-borne disease—just the kind of thing Congress wanted to know. Gerberding’s testimony was reviewed at the White House and soon made to disappear: Virtually all of what she said about climate change—six pages of it—was blacked out of the document filed with the Senate Environment and Public Works Committee (see <http://alt.coxnewsweb.com/ajc/pdf/gerberding.pdf>). There’s an odd behind-the-scenes story here, involving two offices that report to the president. The Office of Science and Technology Policy raised questions about particular statements and made suggestions, but then the Office of Management and Budget, apparently unwilling to work on the suggestions, simply eliminated every section about which questions had been raised. It’s worth a look just to understand what these people don’t want you to know.



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10.1126/science.1154158