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The Cystic Fibrosis Gene Story

In this issue of *Science*, there is a story that does not begin at the beginning or end at the end, but has a very happy middle. The beginning is the basic research that made it possible to search for a genetic needle in a haystack of DNA bases. The end is a cure for a fatal disease. The middle is the finding of the cystic fibrosis gene, a milestone of major importance.

The middle of the story, the three papers published in this issue on the cystic fibrosis gene (pp. 1059–1080), is an event of great scientific achievement that brings credit to the investigators whose dedication and ingenuity made it possible (see the news story by Jean Marx, *Science*, 1 September, p. 923). Until now cystic fibrosis could not be studied in animals, and clues to the actual defect are circumstantial. The discovery of the gene makes possible its manipulation and insertion into experimental systems, thus bringing the day of therapy and cure much closer. This advance immediately increases the accuracy of diagnosis, both in the born and the unborn. It also has provided strategies that will be useful in searching for other disease-causing genes.

The beginning of the story explains why scientists believe in the importance of basic research. The tools that made this finding possible arose from a background of basic research into such apparently esoteric and academic subjects as the understanding of the genetic code, the recognition that enzymes from soil bacteria are able to cut DNA at specific locations, a solid familiarity with the structure of chromosomes, classical genetics, and the use of statistical probability. Much of the early basic research did not seem relevant to the cystic fibrosis problem, and was pursued in the quest for extended knowledge, not practical application. At times, legislators get impatient with scientists who emphasize such research, implying that while scientists may prefer it, society does not need it. Scientists have learned, however, that basic research often turns out to be practical, but the time scale for its application differs from that of applied research. There is a time when the search for basic knowledge is essential because there are no tools available for a direct application. Once the tools have been obtained, often by investigations that were primarily directed toward another goal, the clever and prepared investigator will apply them to the problem at hand. Thus the apparently arcane interests of ivory-tower scientists are essential and inexorable steps along the path to the triumphs of today.

Another happy feature of the present story is the combination of private philanthropy and public funding that made this research possible. The thousands of individuals who contributed to the Cystic Fibrosis Foundation, the brilliant and eccentric man who created the Howard Hughes Foundation, the supporters of the Hospital for Sick Children in Canada, the sensible managers of those charities, and the farsighted government officials who also provided funds, all deserve credit. In a world in which terrorists, embezzlers, prevaricators, and self-indulgers seem to grab the daily headlines, a story in which thousands of people directly or indirectly provided funds to attack a disease that would not affect them personally provides a warm glow.

Finally, the story illustrates another angle on that frequently maligned phenomenon, competition. There is a great deal of discussion these days that the “publish or perish” pressure creates the occasion for misconduct, and certainly the race for the cystic fibrosis gene had elements of competitive pressure. What those standing on the sidelines, who sometimes seem to want a competition-free world, fail to say is that the same competition that attracts Olympic sprinters and mountain climbers impels scientists to work with intensity and dedication. The personal satisfaction of knowing one has contributed to the solution of a difficult problem, the excitement of doing one’s best in an all-out effort, and the lure of fame and fortune, in whatever proportion, all combine to ensure that the jobs of science get done faster than would be accomplished in a leisurely 9 to 5 schedule. In the case of cystic fibrosis, honorable competition led to cooperation, when three of the laboratories racing toward the same goal decided to join forces and share information. That cooperation and intensity means that the one in 2000 children born each year with a fatal defect now has a greater chance for a happy future.—DANIEL E. KOSHLAND, JR.