LETTERS

Nucleic Acid Sequences: Data Bank

The National Institute of General Medical Science of the National Institutes of Health (NIH) sponsored a meeting of scientists on 14–15 July 1980 to evaluate the need for a nucleic acid sequence data bank. A similar meeting was sponsored by the European Molecular Biology Laboratory (EMBL) at Heidelberg on 24–25 April 1980. As a result of the recommendations of these workshops, NIH in collaboration with the EMBL has taken steps toward the establishment of such a bank. While the details are being worked out, an interim sequence collecting effort is being carried out informally. Sequence collections compiled by individuals are being merged and made available to interested scientists. Anyone wishing to obtain the minutes of the meeting held at NIH, contribute sequences, or use the interim data bank arrangements should contact me at the address below or Greg Hamm, European Molecular Biology Laboratory, Postfach 10 2209, 6900 Heidelberg, West Germany, for information.

ELKE JORDAN

Genetics Program, National Institute of General Medical Sciences, National Institutes of Health, Bethesda, Maryland 20205

Cancer Risk Counseling

The article "Testing for cancer risk" by Gina Bari Kolata (Research News, 29 Feb., p. 967) provided a sensationalized and inaccurate account of the counseling about cancer risk I gave 40 parents of ataxia telangiectasia (AT) patients in 1974 and 1975. The meetings with the parents followed our study of the incidence of cancer and other diseases in 27 families of AT patients (*1*). The actual event differed in many important (and unimportant) details from the dramatic story she told.

AT is an autosomal recessive syndrome in which the affected homozygotes are unusually likely to develop a malignant neoplasm. A homozygote receives a single copy of the AT gene from each parent. Because of the high frequency of heterozygotes in the population, it is important to know whether AT heterozygotes are also predisposed to cancer. I have been systematically studying cancer incidence in families of persons with cancer-associated recessive syndromes and estimating the cancer risk of heterozygotes for specific genes from these family data. For example, from the AT family data we estimated that the risk to an AT heterozygote of dying from cancer before age 45 was greater than five times the general population risk. The general significance of this inference derives from the estimate that AT heterozygotes comprise approximately 1 percent of the U.S. population.

These findings have particular significance to parents of AT patients, who are, at the present time, the only heterozygotes who can be identified reliably. Because of the potential impact of discussions of cancer risk and the tentative nature of the conclusions derived from our study, I decided to visit each parent who wanted a report approximately 1 year after data analysis was complete. (This was dramatized as: ". . . immediate action . . ." ". . . so concerned that he flew around the country visiting relatives of AT patients . . . .") I had, in any event, promised the parents of the patients a report at the conclusion of the study. Because the benefit to more distant blood relatives in our study was less clear-cut and my relationship to them in the course of the study more remote, my post-visit letter of summary (which I had provided to Kolata, along with the pertinent reprint), stated explicitly that I left it to the parents to decide which of our findings, if any, they wished to pass along to other relatives. A few siblings (probability of heterozygosity is .67) of the probands sat in when I talked with their parents.

Kolata criticized my actions in three respects: First are the statements "Even more worrisome is Swift's inability to tell the relatives of AT patients whether they carry the AT gene . . ." and "Even though Swift says he wants to tell people of their genetic makeup, the fact is that he can only tell them of their relatives' genetic makeup." Throughout the article Kolata used the terms "parents," "relatives," and "AT heterozygotes" interchangeably and often incorrectly. The parents whom I visited and counseled are, according to contemporary genetic principles, likely to be heterozygous carriers of the AT gene with an extremely high probability. Tests for heterozygotes are in general less reliable than the inference that a parent of a homozygous proband is an obligatory heterozygote.

Kolata's second criticism was that the cancers which appeared to be associated with heterozygosity for the AT gene were those for which there was no evidence that enhanced alertness and early diagnosis might improve survival. Of the eight cancer types for which there was evidence of an association with AT gene, only five were listed in the article, although the eight were clearly stated in my summary letter and reprint. Omitted were colon and cervical cancer—two cancers for which there is good evidence of the benefit of surveillance for high-risk individuals—as well as gastric cancer, for which there is only preliminary evidence. There may well be health benefits from enhanced awareness and early diagnosis of the cancers Kolata named; there is simply no evidence yet.

Finally, although Kolata expressed concern over psychic damage to the AT parents I visited ("No matter how carefully Swift words his message of cancer risk, it is likely that some AT relatives will misunderstand what he says and be convinced that they will soon die of cancer"), she was not interested in my offer to obtain permission from the parents for her to contact them, an offer made in response to her question about the impact of the counseling on the relatives. Speculation was published; the facts were available.

The research and recommendations for counseling were published more than 4 years ago, and there has been ample opportunity for clinicians, geneticists, or cancer epidemiologists to go on record with criticisms of my work or the provision of information to families at the end of each study. No one has done so. Apart from a homily about genetic counseling credited to Park Gerald, attributed statements in Kolata's critique were conspicuous by their absence. Thus when Kolata wrote, "What Swift did is highly controversial" or "The controversy over Swift's actions . . ." or "Swift defends . . .", she was only giving her opinion or that of an anonymous source. Anonymity makes it difficult to distinguish responsible scientific or ethical criticism from comments arising from petty motives.

Since the conclusion of the AT family study in 1973, I have stayed in touch with many of the parents of the affected children. The unfortunate and remarkable incidence of cancer among these parents has provided new evidence that AT heterozygotes have a substantial cancer risk and that persons at risk who are vigilant may improve their chances of surviving the cancer.

MICHAEL SWIFT

Biological Sciences Research Center, University of North Carolina, Chapel Hill 27514

References

1. M. Swift, L. Sholman, M. Perry, C. Chase, *Cancer Res.* **36**, 209 (1976).