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Letter to the Editor

Paradigm Shifts and Medical Genetics

To the Editor:

I am prompted to write by a series of apparently unrelated articles in recent issues of the Journal [Khoury, 1997; Robin, 1997; Resta, 1997; Halliday et al., 1997]. The paradigm shift mentioned in the title of Khoury's article [1997] initially caught my eye because I have been speaking to medical geneticists about upcoming paradigm shifts for the past few years. The other articles, while not commenting on Khoury's, relate to arguments about paradigm shifts.

Khoury argues that, in order to interact successfully, medical geneticists and the public health community need to change the disease prevention paradigm and the definition of genetic disease. True enough, but I go a step further in my thinking about the subject: medical geneticists must change completely their concepts about how genetics is practiced if genetics is going to assume a meaningful position in the community of medical specialties. Medical geneticists must realize that genetics is no different than the rest of medicine and will ultimately be a treatment-oriented subspecialty. By this I mean that medical geneticists must practice sequential, interactive medicine and be prepared to offer both therapy for genetic disease and gene therapy.

Today, medical geneticists are concerned for the most part with diagnostic challenge only; not just any diagnostic challenge, but the ultimate challenge–making the diagnosis no one else has been able to make. The articles by Robin [1997] and Resta [1997] underscore this assertion. Each of these authors describes a diagnostic puzzle, mentions the fact that no one has been able to make a diagnosis, belittles his own credentials, and ultimately solves the riddle. I am not criticizing the articles or the cases described. I cite the articles to point out that homage is paid to colleagues for their problem solving skills; I just happen to feel that the homage is disproportionate to the value of the task, a feeling that is shared, I suspect, by the cited authors. More importantly, the disproportionate homage causes many contemporary medical geneticists to focus exclusively on rare and unusual cases, forsaking the rewards of dealing with the problems of people with common disorders such as Down syndrome, sickle cell disease, neurofibromatosis, and cleft lip/palate. In other words, there is no planned sequence of visits and no meaningful interaction with patients unless the case enhances respect for the medical geneticists in the general medical community.

It is time for medical geneticists to realize that managed care organizations and the architects of future health care are concerned with cost-effective plans for diagnosis and management of common disorders. It is no longer acceptable inside or outside academic institutions to focus exclusively on rare disorders and esoteric approaches to diagnosis. Furthermore, it is not necessary and may even be unconscionable to force an entire series of history recordings, anthropometrics, photometrics, physical inspections, library assisted or computer-assisted diagnostic searches, and counseling into a single visit.

When medical genetics will become a treatment subspecialty is a subject for speculation that is beyond the intent of this letter. Whenever it does, serious thought will have to be given to funding genetic services. It will no longer be appropriate to offer free, centralized genetic services, as counseling is now offered at places such as the Murdoch Institute in Melbourne [Halliday et al., 1997]. In fact, serious thought will have to be given to who is entitled to bill for genetic services. Will genetics be treated as a disease corridor, much as oncology or endocrinology are, with medical geneticists being responsible for all genetic testing and therapy? Or, will genetics be treated as a platform that crosses several disease corridors, with specialists in each corridor being allowed to order and bill for whatever genetic testing and therapy falls within the scopes of their practices?

Whatever transpires for provision and payment of genetic services, centralized registries of birth defects and genetic services should be factored out of the formula or strictly limited. There is simply too much room for abuse with such registries. Panels of ethicists have concurred that it is inappropriate to discriminate on the basis of "genetic constitution," but who's going to determine how centralized data, if it exists, is going to be used. As cause for concern, I refer to two statements in the article of Halliday et al. [1997]: 1) Anyone who has been seen by a member of the genetics team, at any one of the clinics around the state, is entered into the GF (Genetics File) system and 2) vari-

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ables required...were mother's surname, mother's first name, baby's surname, suburb, postcode, mother's date of birth, baby's date of birth, hospital of baby's birth, sex of baby, and birth defects code and descriptions. If medical records are confidential, except to protect the health of the community, what is the purpose of registries of this sort? (This is a rhetorical question, of course, and does not imply anything improper, unethical, or even questionable about the very nice paper in which the registry is described).

The fact that genetics is not being treated like other medical specialties as it develops is in part an unfortunate accident of birth: the field of medical genetics was born and is maturing at a time when our information-laden society is heading into an era of renewed individualism at the state and personal level, altered patterns of health care, political correctness, and awakened sensitivity to covert discrimination. Is it any wonder that there are unanswered questions and widespread concern about where all this is headed?

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