lic, the insufficiency of primary health care, or both that motivated the public to seek help in hospital emergency rooms and outpatient clinics as alternatives to community primary care centers? Did these factors cause the shift away from community primary care centers or were disappointed consumers and insufficient care the result of changing the patterns of spending for health care? Table 4 sheds some light on changes in the national health expenditure by type of service.

Perspective for Tomorrow

Newly emerging needs, repeated economic restrictions, and skyrocketing costs call for critical reevaluation of both existing programs and how services are delivered. An alternative strategy for making health decisions should be problem- and population-oriented. It should identify needs, define goals clearly, set priorities, allocate resources differentially in a "cold economic climate," delineate responsibilities clearly, and coordinate multidisciplinary and multi-institutional efforts.

The strategy that consists of these components should relate health hazards to their determinants. By analyzing cause and effect, we may weigh the relative contribution of each variable and set priorities accordingly.

Formulators of health policy must make better use of quantitative tools in order to balance alternative uses of resources and to measure the economic benefit for decisionmaking.

Furthermore, the health care system of tomorrow will deal less with overall costs of intervention and treatment and more with costs per case. We will have to analyze the performance of health services and link costs to defined "products," with much more involvement of quantitative tools.

These proposed changes will require a corporate effort and vigorous perseverance to prevent setbacks and crises and to open new horizons for the coming years.

Problems in Medical Genetic Services as Viewed from Israel

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This paper is based on the author's presentation at the Second Binational Symposium: United States–Israel, held at Bethesda, Md., October 17–19, 1983. Data on Tay-Sachs disease and certain aspects of prenatal diagnosis were supplied by Prof. B. Padeh, Dr. R. Navon, and Ms. E. Akstein.

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Synopsis

Problems in genetic services in Israel are discussed from four vantage points: genetic screening, prenatal diagnosis, genetics and the mentally retarded child, and genetic counseling. Emphasis is given to issues unique to Israel, and recommendations for the improvement of genetic services are presented. A central, recurring theme is the continuous and concerted effort that must be made by those working in genetics to keep both the medical community and the lay community adequately informed.

THE PURPOSE OF THIS REPORT is threefold: (*a*) to give a capsule view of some of the major problems that we in Israel face regarding genetic aspects of health services, (*b*) to suggest possible solutions to these problems, and (*c*) to raise the issue of cost benefits of certain of these services. Although some of our problems are universal, others are unique and reflect the multifaceted medical, cultural, religious, and political milieu of Israel.

The four major areas that I wish to discuss include genetic screening for Tay-Sachs disease, prenatal diagnosis, genetics and the mentally retarded child, and genetic counseling. Most of what I relate will be based on experience gleaned over a 15-year period at the largest medical center in Israel (Chaim Sheba Medical Center), which also has the largest medical genetic unit in the country.

Background

Israeli medicine for the most part is socialized, and medical genetic services certainly fall in this category. All centers for the practice of medical genetics are located in major teaching hospitals. Jerusalem has one center; the Tel-Aviv area, five; Rehovot, one; Haifa, one; and Beersheva, one. Referrals to genetic centers come both from within the hospital setting and from the hundreds of medical outpatient clinics located throughout the country. For the most part these outpatient clinics are staffed by physicians who did not receive their medical education in Israel but were trained in countries where little or no emphasis was given to the teaching of medical genetics.

Concerning postgraduate medical training in medical genetics, no official programs are offered in Israel because the Israel Board of Medical Specialties does not recognize the specialty of medical genetics. Two universities (Tel-Aviv and Hebrew) offer advanced degrees (MS and PhD) in human genetics, but only the Hebrew University at present awards the MS degree in genetic counseling. Medical genetics is taught as a distinct course in two of our four medical schools: Tel-Aviv and Jerusalem, both of which have departments of human genetics. In the other two medical schools (Haifa and Beersheva), matters pertaining to medical genetics are dispersed throughout other courses.

As Childs and coworkers (*1*) have shown, this scattered educational exposure to the teaching of genetics in medicine is not unique to Israel but is the rule in the United States and in most other parts of the world. As a result, many practitioners of medicine tend to view medical genetics as an esoteric subject not relevant to the more common problems in medicine. Such a myopic view stunts the capabilities of medical genetics and also diminishes the scope of care given many patients and families in need.

To complete the picture of the medical genetic setting in Israel, one other aspect must be mentioned. The Israeli Jewish population of approximately 3.6 million is composed of three major communities: Sephardim and Oriental Jews (53 percent) and Ashkenazim (47 percent). These communities, with their subgroups, come from more than 100 different countries. In many instances each major community, along with its component groups, can be characterized genetically by the high frequency of certain genetic disorders found within these varying Jewish ethnic divisions (*2*). Thus, in Israel it becomes impossible to ignore the relevance of medical genetics among our various populations.

These key factors that I have briefly touched upon will come up again as we begin to look at selected problems related to provision of medical genetic services in Israel.

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Genetic Screening for Tay-Sachs Disease

Without question, genetic screening for Tay-Sachs disease (TSD) among the Ashkenazi Jewish communities of the world has been a most worthwhile measure in the prevention of this disorder (*3*). However, my purpose here is not to laud the success of these programs but rather to point out some of the problems that have arisen in Israel as a result of our Tay-Sachs screening efforts.

Early in 1973, a group at the Chaim Sheba Medical Center headed by Professor B. Padeh embarked on a voluntary premarital screening program for Ashkenazi couples. This project had to be curtailed because of the Yom Kippur war, but from the start it became clear that there were problems in screening such couples premaritally (*4*). These problems were the following:

1. Persons identified as carriers of the TSD gene did not always understand the meaning and implication of being a carrier.
2. For some, the psychological burden of being a carrier for a lethal disease like TSD was considerable.
3. In a few instances, when both partners were told they were carriers, they decided to break their engagement.

In 1974, the program was restarted but redesigned to screen only already married Ashkenazi couples. Over the years, approximately 20,000 persons have been tested. The heterozygote frequency in the Ashkenazi community in Israel has been found to be 1:29.5, very close to the heterozygote frequency in the Ashkenazi population overall—1:29.7.

By the end of 1980, 18 couples at risk had been identified, 29 pregnancies had been monitored, and 6 TSD fetuses had been identified. (Only 4 of these TSD fetuses were aborted. One family elected to continue the pregnancy on religious grounds, and the other affected fetus was diagnosed too late in pregnancy to be aborted.)

In families with a previously affected child, 133 pregnancies had been monitored, and 30 TSD fetuses had been identified. All 30 of these fetuses were aborted and the diagnosis was confirmed.

On the basis of current demographic information, it is estimated that about 20,000 babies are born per year in Israel to couples in which both parents are of Ashkenazi origin. This means that one would expect about 6 new cases of TSD per year; however, only 2–3 cases per year are being diagnosed. With this discrepancy before us, it is now appropriate to ask, What's going on?

1. It has become apparent that physicians, primarily obstetricians, are no longer referring as many of their patients or patients' husbands to our laboratory for TSD screening. It is as if we screened once and the gene has gone away. Since most physicians lack an adequate understanding of genetics, they tend to forget about the importance of continuous screening in Ashkenazi couples if not reminded or urged.
2. The previous comment is further substantiated by the fact that we are seeing couples who have had one or more healthy children, have never been screened, and now have a child with TSD. Within a 3-month period, I saw four such children in four different families. For two of the families, the story was: "I went to my obstetrician and asked him about being tested for TSD. He stated that I should not worry as I already have two healthy children."
3. The very religious segment of the Ashkenazi community (approximately 5 percent) is not at all interested in the artificial interruption of a pregnancy because of TSD and for the most part will not participate in screening. When the disease has occurred in such families, some will want to know the genotype of their children and their children's possible marriage partners. We know of instances in which marriages in such families were not consummated because both partners were carriers of the TSD gene.
4. We have learned that TSD also occurs among non-Ashkenazi Jews but as yet do not know its frequency. Cases have also been seen in offspring of couples of mixed Jewish ethnic origin and in one couple in which only one parent was Jewish.

Mainly because of the marked decline in proper referrals, some members of our TSD screening program have begun to question the cost benefits of the program. This matter certainly needs to be carefully evaluated, in view of our serious budgetary restrictions. It is now obvious that if any genetic screening program is going to be worthwhile and have longevity, its staff must make a concerted effort to keep both the medical community and the lay community properly informed.

Prenatal Diagnosis

A few years ago, the Israeli Government enacted a law that provides for free amniocentesis for all women 37 years old and above. This law has been most useful in preventing the birth of fetuses that are malformed and usually mentally retarded because of mainly chromosomal disorders. As I said earlier, it is not my aim to praise this program, though praise is deserved, but rather to call attention to some of the issues that have arisen during its implementation.

1. Our female cutoff age of 37 for a high-risk pregnancy was selected on the basis of budgetary and laboratory considerations. This is unfortunate. In the United States, 35 was once regarded as the age marking the beginning of maternal high risk for chromosomal disorders—especially Down's syndrome. Now, more recent data would move the age to 30 (*5*). Such information makes it mandatory for us to know how many infants who are malformed because of a chromosomal disorder are being born to mothers and fathers below the age of 37.
2. Not only maternal age, but also paternal age, is a factor in the etiology of certain chromosomal disorders, and this has not been adequately considered or emphasized in formulating the indications for amniocentesis in Israel.
3. In the past, it was not our policy at the Chaim Sheba Medical Center to tell parents the sex of the fetus after amniocentesis unless the procedure was done for the purpose of sexing (X-linked disorders). Objection to this policy was almost nil from those parents who were traditional or observant in their religious views, while less than half of those in the secular community wanted to know the sex of their fetus. Today, when parents request the sex of the fetus the information is given. You may be interested in knowing some of our past reasons for not telling the sex: (*a*) mistakes in sexing would be noted in the testicular feminization syndrome, and so on; (*b*) sex preference might result in the interruption of a seemingly normal pregnancy; (*c*) when the sex of a fetus is known, there is closer natural identification, and in miscarriages under these conditions, greater psychological trauma has been noted.
4. It is not uncommon for obstetricians to refer patients to our clinic for amniocentesis and not understand the diagnostic limitations of the procedure. Frequently, they also give false hope to their patients by telling them that we will be able to know if their baby is perfectly healthy.
5. At times we have been faced with difficult moral and ethical problems, such as (*a*) the request to interrupt an apparently normal pregnancy because the fetus is not the sex of the parents' choice and (*b*) requests from young unmarried women who are pregnant by choice, either

naturally or by artificial insemination, and who want as much assurance as possible that all is well with the pregnancy.

In general, the matter of prenatal diagnosis, as I have briefly viewed it from the Israeli scene, is not overwhelmingly plagued by problems. Although the present period may be relatively tranquil, the new technology using recombinant DNA studies, gene identification, and new genetic linkage information is going to expand beyond all expectations our ability to make prenatal diagnoses of genetic disorders. Then we shall be faced with tremendous moral dilemmas: Where shall we draw the line? When shall we interrupt or not interrupt a pregnancy? Who shall be the decisionmaker? Will the parents have the final word? These extremely difficult issues are just around the corner, and it would be prudent for all who work in this area to begin considering an approach to these matters.

Genetics and the Mentally Retarded Child

To the credit of the health services of our country, we have established regional centers for assessment of mentally retarded children. The largest of these centers is located at the hospital in which I work, and I serve as its consultant in medical genetics. Over the years, I have come to appreciate the center's difficult task but also have become critical of its diagnostic evaluation. However, the comments I am about to make are not to be construed as meant solely for this particular center but also reflect my impressions of other centers that I have had contact with, both in Israel and abroad.

It is my contention that many physicians, mainly pediatricians, working in such centers often lack the diagnostic capabilities needed for the proper evaluation of the mentally retarded child with or without congenital malformations. When correct diagnoses are not made, then other areas involving care, prevention, and counseling not uncommonly are neglected or are not given their proper attention. I hasten to add that I do not feel it is right to totally condemn these physicians for their inadequate diagnostic skills, for in most instances they were never trained to be diagnosticians in the area of childhood mental retardation. Furthermore, the recent advances in knowledge about genetic causes (genetic syndromes with associated mental retardation) have become so numerous (*6*) that even experts in the field have a difficult time keeping abreast. Before offering a possible solution to what should be considered a rather common problem, I want to stress again that, as in all areas of clinical medicine, mentally retarded patients and their families can suffer enormously by being given an incorrect diagnosis.

' . . . the new technology using recombinant DNA studies, gene identification, and new genetic linkage information is going to expand beyond all expectations our ability to make prenatal diagnoses of genetic disorders. Then we shall be faced with tremendous moral dilemmas . . . '

I think the time has arrived to begin training pediatricians for a new subspecialty that I have termed "pediatric developmentology." This new area of training would center its attention on the acquiring of skills required to diagnose, treat, and counsel congenitally malformed and mentally retarded children in a family setting. Without such a training program, I feel certain that we shall continue along the lines of mediocrity.

Furthermore, I would urge that studies be done to evaluate the effectiveness of our existing centers for assessing mental retardation. If they are found to be inadequate, then consideration should be given to the establishment of regional birth defect centers. Under such a new roof, with a qualified staff, it is conceivable that more congenitally malformed children, with or without mental retardation, could be better evaluated and treated at a lower financial expenditure.

Genetic Counseling

Of the four segments of medical genetic services in Israel being discussed, it is the area of genetic counseling that I view as the most problematic—for here we know very little about the results of our efforts. Although genetic counseling is done in eight other centers in Israel, the Chaim Sheba Medical Center is the largest and can be considered as representative of the Israeli scene. If I could speak for my colleagues, I think the majority would agree that despite our good intentions we are frustrated by (*a*) poor followup studies, (*b*) inadequate storage and exchange of information, (*c*) lack of periodic assessment, with discussion of ways to improve genetic counseling, and (*d*) a shortage of personnel and funds to rectify the situation.

Without delving into the many philosophical questions related to genetic counseling, I do want to touch upon some of the practical issues that confront us.

1. For a small country, we are extremely heterogeneous in all spheres of life, and, as such, we need to better

understand the thoughts and motivations of the people who come to us for counseling.
2. Our referrals to genetic counseling are not covering a broad enough expanse of medicine, and this is due in part to our failure to inform the medical profession of their responsibility to patients who have genetic disorders. On the other side of the coin is nonrecognition of the genetic etiology of disorders being seen and treated by practicing physicians.
3. Weekly, we are counseling couples who are consanguineous (usually first cousins) and are considering marriage. We must do followup studies on such couples, because we need to know the influence of genetic counseling on their decision to marry or not to marry. If they do marry, we later need to know the health status of their offspring.
4. Most patients and families referred to genetic counseling need to be followed, and better facilities for this should be available. Patients with genetic diseases are periodically in need of consultation for numerous reasons, and usually it is the physician or genetic counselor who must aid them.
5. There is a tendency on the part of some parents to reject and even abandon the child who is born congenitally malformed, mentally retarded, or both. We need to better understand this situation so that we can improve our ability to help those involved.

Suggestions for improving genetic counseling in Israel include the following:

1. Establishment of a National Registry for Genetic Diseases. Such a registry would not only tremendously aid in the counseling and care being given but would advance our research efforts pertaining to the recognition of new genetic diseases and those specific disorders involving certain of our ethnic communities.
2. Creation of a computer center for the storage, retrieval, and analysis of all information regarding genetic counseling in Israel.
3. Initiation of national periodic review conferences to discuss ways of improving all aspects of genetic services, including that of counseling.
4. Publication of a quarterly bulletin or newsletter to keep physicians abreast of the rapid progress in genetics and the applicability of genetics in medicine.
5. Physical improvement of outpatient facilities for genetic patients.
6. Involvement of psychiatrists and psychologists in genetic counseling to help us better understand our patients.
7. An increase in the number of university training programs for those who wish to become genetic counselors.

Conclusion

On a practical note, it must be mentioned that the present financial burdens of our government and universities are of such a nature that it would not be logical to expect a great deal of support for much of what I am advocating. Thus, it will be necessary to seek funding from private sources or develop binational programs with other countries where there is such a need and funds are available. Nevertheless, not all of these recommendations require large funding, and some could be implemented with relative ease.

I hope that this presentation will serve as a focal point in the exchange of ideas pertaining to (*a*) cost benefits of genetic screening, (*b*) the best means of evaluating and caring for the congenitally malformed and retarded child, (*c*) approaches to prenatal diagnosis based on the "new genetic technology," and (*d*) ways to improve genetic counseling. These four issues are of extreme importance to health care planners in countries where family size has been markedly reduced by choice or governmental direction.

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