Managed Genetic Care in the Largest HMO: The Challenge of Providing Genetic Services To 2.7 Million Members

By Ronald P. Bachman, MD
Edgar J. Schoen, MD

Comprehensive clinical genetic services are offered to 2.7 million members in Northern California by the Kaiser Permanente Medical Care Program (KP), a not-for-profit HMO. Four genetics centers are staffed by clinical geneticists, genetic counselors, nurses, and laboratory technologists, who together provide patient and physician education, genetic screening, and prenatal, infant, and adult evaluation. These centers provide genetic care for 19 KP medical centers and 16 satellite clinics. Besides offering lectures and teleconferences, the geneticists publish a newsletter for up to 1200 pediatricians, obstetricians, and other relevant specialists on how to use genetic services. Clinical services offered to members include individual, group, and telephone consultations, and easy access to geneticists is provided for primary care providers who have questions about genetic care. In-house laboratory services include blood and tissue cytogenetic analysis, DNA testing, and testing of prenatal blood and amniotic fluid; specialized testing for inborn errors of metabolism is centralized at KP Northern California Divisional Laboratory. An Interdivisional genetics data system is being established to link Northern and Southern California and the Northwest (Portland and Hawaii). Under the newly established national KP Care Management Institute (CMI), we propose to offer selective genetic services to KP facilities across the country using computer linkages and new technologies such as telemedicine consultation.

"Geneticists are rapidly acquiring new abilities because of technologic advances which have largely resulted from the Human Genome Project."

"The goal is to offer appropriate, up-to-date, comprehensive, high-quality genetic services which are also cost-effective."

Introduction

Genetics is a unique specialty: provision of genetic services encompasses all medical fields and age groups. Geneticists are rapidly acquiring new abilities because of technologic advances which have largely resulted from the Human Genome Project.

The Kaiser Permanente Medical Care Program (KP), a not-for-profit HMO, offers comprehensive clinical genetic services to 2.7 million health plan members in Northern California; Southern California has a similar membership. These services are coordinated by a group of clinical geneticists who provide current genetic care and plan for future services. The goal is to offer appropriate, up-to-date, comprehensive, high-quality genetic services which are also cost-effective.

In Northern California, KP is concentrated in the San Francisco Bay area but extends over 200 miles, including 19 hospitals with outpatient departments as well as 16 freestanding outpatient facilities. Genetic services are provided subregionally at 4 centers—San Francisco, Oakland, San Jose, and Sacramento. Each genetics center is staffed by clinical geneticists and genetic counselors and may also have nurses and laboratory technologists.

Some genetic centers are responsible for doing specific tasks for the entire area. For instance, only 2 cytogenetic laboratories and 1 molecular genetics laboratory exist. All genetic perinatal screening programs are administered from a single location (Oakland). The 4 genetics departments are staffed by 9 clinical geneticists (MDs), 35 genetic counselors (MSs), 4 laboratory directors (PhDs in cytogenetics and molecular genetics), 20 laboratory technologists (MSs or equivalent), and 10 genetic nurses or metabolic nutritionists (MSs).

Services are coordinated through meetings with geneticists who provide similar services in other areas in which KP operates (the Northwest and Southern California). In the 3 areas, KP serves about 5.8 million members, a population similar to that of several small European countries (eg, Norway, Denmark).

Divisional genetic policy is established through periodic meetings of the geneticists in charge of the 4 genetic centers with representation from the genetic counselors and laboratory personnel. A single divisional genetics budget is shared by the 4 centers. Budgetary decisions are arrived at by consensus based on current and future genetic advances relative to estimated costs.

Comprehensive services include patient education, provider education, genetic screening, prenatal, neonatal, child, and adult evaluation, multispecialty clinic services, laboratory services, resident education, and research.

Patient and Provider Education

Providers, physicians, and other professionals must know what services are available and how to use the system best. The genetics program functions most efficiently with appropriate referrals and requests for genetic laboratory tests.

We found that a useful way to educate providers to properly use genetic services was through a periodic, short newsletter titled *The Screen*, which is directed to appropriate specialists, usually pediatricians and obstetricians. The mailing list of up to 1200 providers is tailored to the topic (eg, prenatal and neonatal hemoglobinopathy screening, triple-marker screening).

left RONALD P. BACHMAN, MD, is chief of the genetics department and has been practicing pediatrics and genetics with TPMG for over 28 years.
right EDGAR J. SCHOEN, MD, is senior consultant in pediatrics and director of regional perinatal screening in the genetics department for TPMG. He joined Permanente in 1954.

We have begun providing *The Screen* to other KP divisions and have uploaded it to the bulletin board section of our e-mail network, which has made it even more accessible. Other modes of communicating with our providers include lectures, minicourses, teleconferences, and personal interaction.

Health plan members are informed about genetic services through health education centers as well as through a quarterly newsletter, *Planning for Health*. The health education centers are located in each of the larger facilities and consist of a patient library with exhibits and cubicles for viewing videotapes; educational material is available for distribution, loan, or purchase.

We try to target genetics education to those patient groups for whom it is most relevant. For example, pregnant patients are informed of our services at prenatal classes.

Role of the Primary Care Physician

Primary care physicians (mainly obstetricians and pediatricians), as well as other providers, are kept informed of genetic services through a variety of interfacility communications such as in-service education at prenatal clinics and labor and delivery areas. Genetic counselors, nutritionists, and nurse coordinators provide outreach services at the smaller clinics through regularly scheduled meetings and in response to quality and utilization surveys and local requests. Geneticists and genetic counselors are available for informal telephone consultations. Patients as well as providers can call genetic counselors directly for answers to questions on such issues as teratogens, or family history of genetic disorders and can request a formal consultation.

In 1994, at the 4 genetics centers, 5464 phone consultations with patients and primary care providers and 8515 genetic care office visits were provided. All office consultations are followed by a medical report to the primary care provider and an explanatory letter to the patient.

At multidisciplinary clinics (metabolic, lipid, spina bifida, skeletal dysplasia, neurogenetics, and craniofacial) run by the Oakland genetics department, highly specialized care (eg, metabolic diets, plastic surgery) is coordinated in a periodic, centralized, one-stop way. At these multidisciplinary clinics, we are following up about 150 cases of inborn metabolic errors (eg, phenylketonuria, galactosemia), 160 cases of spina bifida, 250 cases (including some in adults) of craniofacial anomalies, and 70 cases of children from families with hereditary lipid disorders. The local primary care provider cares for the ongoing, daily medical needs of the patient close to home, and close communication occurs between primary and secondary providers.

At these clinics, psychosocial as well as medical and genetic counseling needs of patients are reviewed. These multidisciplinary specialty clinics have been well received by patients and primary care providers. National consultations using telemedicine could be provided to patients at distant sites by using our established team of experts.

Perinatal Screening and Genetic Services

The main mission of both HMOs and geneticists is disease prevention. Instituting preventive measures before conception is ideal, but often the family does not become concerned until conception has occurred.

The following genetics programs are offered during pregnancy: 1) genetic questionnaire; 2) hemoglobinopathy screening (for S, C, and E hemoglobins); 3) thalassemia screening (for α- and β-thalassemia); 4) maternal serum alphafetoprotein (MSAFP) screening (for neural tube defects, Down syndrome, and other abnormalities); 5) Tay-Sachs disease (in Ashkenazi Jewish and French Canadian persons); 6) fetal ultrasonography; 7) amniocentesis/chorionic villus sampling; and 8) "triple-marker" screening (an extension of MSAFP screening, mainly for Down syndrome, which has been offered through a state program since mid-1995).

In addition to prenatal testing, the genetics program manages mandated neonatal testing, including screening for phenylketonuria, galactosemia, hemoglobinopathy, and hypothyroidism. Tracking and follow-up of neonatal screening programs is implemented through a special contract with the California State Genetic Disease Branch.

Because of our ability to track and monitor prenatal and neonatal patients with computer programs, we have added infectious disease monitoring to our genetics program, including monitoring of prenatal and neonatal patients for syphilis, hepatitis B, and human immunodeficiency virus (HIV). We are currently developing computer linkage between Northern and Southern California to track all prenatal hepatitis B and syphilis cases from the 60,000 KP newborn infants born annually in the state. The potential exists for national expansion of such programs. We are also evaluating how to determine the best way to prevent group B streptococcus infection in newborn infants and are developing a system for tracking mammography results as part of a breast cancer management program.

Clinical Genetic Services

Aside from the screening programs, referred neonates, infants, and older children (as well as some

"In 1994, at the 4 genetics centers, 5464 phone consultations with patients and primary care providers and 8515 genetic care office visits were provided."

"At these clinics, psychosocial as well as medical and genetic counseling needs of patients are reviewed."

adults) are evaluated for birth defects or genetic syndromes by teams of genetic counselors and clinical geneticists (MDs). Pre-evaluation information (records and tests) is accumulated by the genetic counselor, who establishes a relationship with the family and constructs a genetic pedigree.

This information is reviewed by the clinical geneticist before seeing the patient and family. After the patient is seen by the geneticist and counselor, the genetic counselor is responsible for follow-up evaluation and for helping the patient to receive the recommended testing and treatment. The genetic counselor also sends a written summary to the family of the affected person.

Presymptomatic or predictive testing, an outgrowth of new technologic advances, is also available through the genetic departments. Huntington's disease, which can be diagnosed before onset of symptoms, is an example of a disorder for which presymptomatic counseling and testing are available for patients at risk.

If a patient is seen at >1 facility or has laboratory work done at different centers, all medical data are available through a computer medical record stored in a genetics data base.

"Our large population of members, including about 30,000 deliveries annually, has permitted us to develop our own genetics laboratories."

Genetic Laboratories

Our large population of members, including about 30,000 deliveries annually, has permitted us to develop our own genetics laboratories. We believe that internal provision of services allows for improved quality as well as cost control.

In 1994, our genetic laboratories analyzed 3770 amniocentesis and 148 chorionic villus samples as well as 17 percutaneous umbilical vein blood specimens. Cytogenetic testing was done on 1082 blood and 265 cancer samples. Our DNA laboratory began operation in mid-1994 and is currently analyzing about 25 specimens weekly; work includes testing for Fragile X syndrome, Huntington's disease, Prader-Willi syndrome, cystic fibrosis, and sickling disorders as well as doing Y-probes. We plan to improve cost-effectiveness by offering to do molecular studies for other KP divisions.

"Currently we have an active program for breast cancer risk counseling as well as an intramural innovation research grant for tracking breast cancer patients."

Most cytogenetic studies use amniocentesis specimens, but peripheral blood, bone marrow, chorionic villus sampling, and fluorescent in situ hybridization (FISH) comprise about 25% of the studies. In addition, we are doing an increasing number of bone marrow studies for oncologists.

Currently we have an active program for breast cancer risk counseling as well as an intramural innovation research grant for tracking breast cancer patients.

To avoid duplication and to improve efficiency, highly technical functions have been divided. In Southern California, a laboratory was developed which specializes in metabolic testing for genetic disorders (eg, amino and organic acid disorders); practitioners in KP Northern California send the KP Southern California laboratory specimens for metabolic testing and use the consulting services of their metabolic specialists. Similar cooperative use of the KP Northern California molecular laboratory is planned.

"Although providing clinical service is the primary role of an HMO, a unique opportunity exists to engage in relevant clinical research such as outcome analysis and studies of the cost-effectiveness of genetic services."

Clinical Research

Although providing clinical service is the primary role of an HMO, a unique opportunity exists to engage in relevant clinical research such as outcome analysis and studies of the cost-effectiveness of genetic services.

An interregional genetic data base has been established and has stored enough data for selected clinical research. The data base is housed and supported by the Center for Health Research, the research arm of the KP Northwest Division in Portland, Oregon.

The size and organization of our genetic and perinatal screening services has permitted interested clinicians to engage in clinical research, and scientific presentations and publications have resulted.[1-6]

Outcome Measures

The perinatal screening and interdivisional genetic data bases provide information which allows the outcome of selected genetic and metabolic disorders to be measured. As an example, we studied outcomes for more than 160 patients with congenital hypothyroidism whose cases were followed up since 1979, as well as for a group of children with the D/G heterozygotic form of galactosemia. We have completed an evaluation of prenatal congenital toxoplasmosis screening[6] and are beginning a cost-effectiveness analysis of prenatal human immunodeficiency virus (HIV) testing. Periodic patient surveys indicate a high level of satisfaction with the genetic services offered to members.

Care Management Institute (CMI)

The CMI was recently established jointly by the Permanente Federation and the Kaiser Foundation Health Plan (KFHP) to improve the quality and effectiveness of care delivered to KP members nationally and to discover and share new knowledge with the health care community. The aims of the KP Northern California Genetics Program to collaborate with other divisions in a number of pilot projects are relevant to these goals. Current programs which could serve as models for transportability under CMI include 1) prevention of perinatal hepatitis B and syphilis transmission with tracking of cases; 2) voluntary prenatal HIV testing; 3) consultation for newborn

screening tests; 4) special genetic laboratory studies; 5) metabolic nutrition services and consultation for inborn metabolic errors; 6) a system for tracking breast cancer; and 7) genetic and multispecialty clinic consultations through telemedicine.

Centralized management could implement these programs through computer linkage, on-site consultation, and staff training to provide standardized, cost-effective in-house KP genetic services.

Summary and conclusion

Comprehensive, effective, and highly technical genetic services can be developed in a managed care system. Providing preventive care—the primary mission of an HMO—coincides with providing genetic services. Coordination and divisional-interdivisional cooperation are the keys to comprehensive, cost-effective genetic care.

The extensive national KP membership (currently 8.8 million members) permits us to develop a program to export certain genetic services. Although smaller divisions might not be able to develop these highly specialized programs, they could use computer linkage and new technologies such as telemedicine to work collaboratively with the larger areas such as KP Northern California to provide current, cost-effective genetic services to these members. ❖

Acknowledgment: The Medical Editing Department, Kaiser Foundation Research Institute, provided editorial assistance.

This article was extracted with permission from related publication: Bachman RP, Schoen EJ: Managed genetic care in a large HMO. HMO Pract 1996 Jun;10(2):54-8.

References

1. Bachman RP, Schoen EJ, Backstrom MV, et al. Phenylalanine embryopathy in three siblings. Am J Dis Child 1993;147:22-23.
2. Bachman RP, Schoen EJ, Stembridge A, et al. Compliance with childhood cholesterol screening. Am J Dis Child 1993;147:382-385.
3. Witt DR, Blumberg B, Schaefer C, et al. Cystic fibrosis carrier screening in a prenatal population [abstract]. Am J Hum Genet 1992;51(4 Suppl):A16.
4. Schoen EJ, Marks SM, Clemons MM, Bachman RP. Comparing prenatal and neonatal diagnosis for hemoglobinopathies. Pediatrics 1993;92:354-357.
5. Schoen EJ, Weber PM. Proposed classification of congenital primary hypothyroidism. Clin Pediatr (Phila) 1990;29:731-732.
6. Schoen EJ, Black S, Cohen D. Screening for neonatal toxoplasmosis [letter]. N Engl J Med 1994;331:1458.

"Comprehensive, effective, and highly technical genetic services can be developed in a managed care system."

A Dollar Song

"They're only puttin' in a nickel, but they want a dollar song."

Song Title