

Expanding Genetics Knowledge Drives Demand for Care Services

Debra Lochner Doyle

Sequencing the human genome is opening the floodgates for the identification of thousands of disease-causing genes and ultimately for better risk assessment and diagnosis for genetic disorders. New strategies for prevention, management, and treatment of genetic diseases will benefit those with these disorders and their families.

Information evolving from the Human Genome Project is already significantly influencing medical practices. While some view this revolution in genetics with great promise, others harbor serious concerns. At this relatively early stage, many issues related to genetic testing and counseling have sparked widespread professional and public policy discussions. As the demand for genetic testing grows, what are the implications for Washington's system of Regional Genetics Clinics? What is public health's role in this revolution?

Growing Pains as a Discipline Evolves

Clinicians were able to visualize chromosomes as early as 1957, and most states implemented newborn screening programs in the 1960s. Amniocentesis and ultrasound technologies catapulted prenatal genetic services into the mainstream of medical care in the 1970s. Laboratory techniques such as molecular genetic testing (DNA testing) and fluorescent insitu hybridization (rapid identification of specific DNA sequences located on chromosomes) initiated in the 1980s and 1990s further confirmed that such tests can be cost-effective, invaluable medical tools.

Yet, the American Board of Medical Subspecialties did not formally designate medical genetics as a subspecialty until 1991. Prior to 1997, few genetic diagnostic tests had Current Procedural Terminology (CPT®)* billing codes. Such codes are still lacking for the labor-intensive genetic counseling services that precede and follow most laboratory procedures. This systems shortfall makes it extraordinarily difficult for clinics to bill for their services.

Only recently has medical genetics become a mandatory topic for study in medical schools. Many practitioners have little or no training in this subject. Yet, a recent March of Dimes survey reported that over 90% of those surveyed would seek answers to their genetics questions from their primary care provider. A key concern is the readiness of health professionals to recognize when genetic testing and counseling services may be appropriate and to provide such services or referrals.

Initiatives in Washington

The Washington State Department of Health (DOH), with funding from the federal Department of Health and Human Services, worked with genetic service providers and others to develop a statewide system for genetic health care services in the 1970s. The goal was a system accessible to all residents that would prevent premature death or disability related to genetic risk factors.

At Regional Genetics Clinics, Washington residents can receive a genetic consultation from a health care provider certified by either the American Board of Genetic Counseling or the American Board of Medical Genetics. Western Washington has 12 regional clinics and Eastern Washington has four (Figure 1). In addition, 11 clinical laboratories and several specialty clinics provide comprehensive services for complex conditions (e.g., sickle cell anemia [Figure 2], PKU, and hemophilia).

These regional clinics and laboratories are located in and managed by academic, private, and public institutions. DOH oversees and coordinates some clinic functions and funds visits by medical geneticists to clinics in Tacoma, Walla Walla, Yakima, Wenatchee, and Spokane. Each regional clinic compiles data on prenatal services (focused on the fetus), clinical services (for children and adults), and laboratory services. The clinics gather basic demographic data (e.g., county of residence, financial status, and self-reported ethnicity of the family), reasons for referral,

and the testing or screening services rendered.

The most common reasons for referral for prenatal services include advanced maternal age, an abnormal result on a screening test (e.g., ultrasound), and a family history of a genetic disorder or birth defect. Pediatric referrals continue to dominate the clinical visits, with the most common referrals being multiple congenital anomalies, skeletal abnormalities, and mental retardation. An important trend, however, is the increasing number of persons seeking genetic services for adult-

number of families seen. Staffing limitations prevent several clinics from expanding service provision. The number of molecular genetic tests performed (DNA tests) also has risen sharply in recent years.

These data reflect only those services and tests provided through the Regional Genetics Clinics. Health care providers such as obstetricians or oncologists also offer genetic services, but DOH does not compile data on services obtained through individual providers. As scientists continue to identify disease-causing genes and better understand how they affect our health, the demand for genetic services will continue to increase.

The Challenge for Service Delivery

In the next few years the genetics and public health communities need to collaborate to facilitate access for all Washington residents to high-quality, comprehensive genetic health care services. In September 1999, the State Genetics Advisory Committee, composed of geneticists, consumers, teachers, and public health staff, identified the following priority issues to be addressed in the coming years:

- **Education** — Educational endeavors are needed for diverse audiences, including policy makers, teachers, adoption workers, the media, health care providers, and consumers.
- **Quality assurance** — Mechanisms are needed to allow monitoring of both clinical and laboratory genetic services to improve their quality. Approaches might include proficiency testing for laboratories providing genetic testing or continuing medical education (CME) credits to health care providers who receive additional training in genetics.
- **Economic stability of the Regional Genetics Clinic system** — If health care payers treated medical genetics like all other mainstream medical services and reimbursed genetic services accordingly, then genetics clinics would be less dependent on state support. In turn, these public health resources that now ensure access to genetic services could be redirected to studies of genetic epi-demiology or to policy development.
- **Confidentiality/privacy regulation** — Policies that ensure the privacy and the confidentiality of genetic information have been introduced at both the state and federal level,

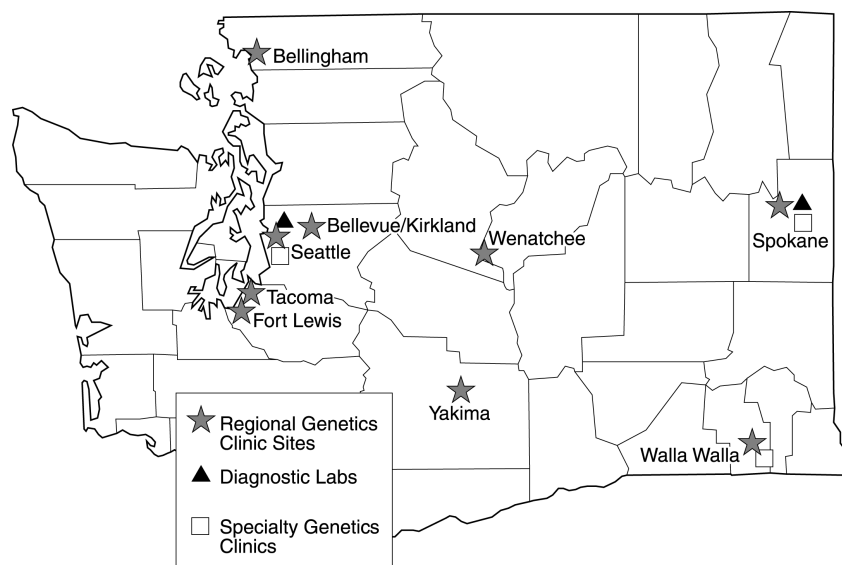


Figure 1: The Regional Genetics Clinic system.

onset disorders. For example, the number of families seeking services related to a family history of cancer increased 20-fold from eight in 1994 to 163 in 1998. This increase is likely a direct result of the discoveries of genes for both breast and colon cancer realized through the Human Genome Project's mapping and sequencing efforts.

Another indicator of expanding need for genetic consultation services is the nearly four-fold increase in outreach clinics funded by DOH and conducted by a medical geneticist from Children's Hospital and Regional Medical Center. From 16 outreach clinics in 1991 in Spokane, Tacoma, Yakima, and Walla Walla, the number expanded to 62 in 1999. Nevertheless, some Regional Genetics Clinics have waiting lists of up to two months for services. Complete data for 1999 are still being compiled, but staff at some clinics have already reported a 20% increase over 1998 in the

but none have passed. Policy makers will need to balance consumer concerns with public health needs and legitimate research interests. They must consider both a person's right to privacy and the societal benefits that can be derived from genetic epidemiology research.

Educational Concerns

The lack of genetics knowledge among consumers and professionals poses several hazards. Especially significant are missed health promotion opportunities or missed preventive strategies if a patient or health care provider fails to recognize genetic risk factors. Incorrect genetic information may raise undue alarm and prompt ill-advised decisions. For example, in a 1997 study published in the *New England Journal of Medicine*, Francis Giardiello and his associates found that nearly 32% of physicians ordering genetic testing for familial adenomatous polyposis (a form of colon cancer) from a commercial laboratory misinterpreted the test results. While the study does not comment on the outcomes of these cases, clearly the potential exists in such circumstances for individuals to be informed that they have a gene for colon cancer when in fact they do not and perhaps to receive unnecessary treatment.

Increased awareness about genetic conditions and genetics issues may also decrease social stigmatization sometimes reported by adults and parents of children with genetic disorders. They frequently describe a lack of sensitivity toward affected family members in health care settings, in media coverage of genetics issues, and in general societal attitudes.

In 1997, the Department of Health convened a group of professionals active in genetics education to help develop a plan for addressing the broad educational issues. This group identified 14 target audiences along with recommendations and information needs for each. Target audiences ranged from adoption workers, lawmakers, and attorneys, to health care professionals, teachers, and the general public. Given the number of audiences and the varied educational goals set for each, the educational needs are vast. Full implementation of this plan will require the cooperation of the public health community and numerous community partners.

Quality Assurance

Laboratories providing genetic testing should be able to demonstrate that they have the capacity to perform the tests requested and report accurate results. The Clinical Laboratory Improvement Act (CLIA) does require extensive documentation of quality assurance and quality improvement plans and outcomes, but proficiency testing is not part of this federal regulation. The College of American Pathology offers proficiency testing for molecular genetic tests (DNA tests), but laboratory participation at present is entirely voluntary.

Genetic service providers should also demonstrate a commitment to quality care. For example, Regional Genetics Clinics in Washington have written mission statements and policies and procedures concerning the privacy of their patients and the confidentiality of patient information. Most public and private clinics have developed programs to monitor the quality of their services; these quality improvement efforts should be encouraged and supported by public health entities and others. In addition, clinics should routinely solicit consumer input (e.g., regarding patient satisfaction) to aid planning and evaluation services, especially given the rapid advances in medical genetics.

Economic Stability

The Regional Genetics Clinic system is witnessing a rising demand for genetic services. Yet, several factors threaten the continued viability of clinics with this specialized expertise. Many regional clinics heavily depend on state support for their basic infrastructure because health care billing and reimbursement practices have failed to keep pace with advances in genetics. Mandated benefits for prenatal genetic services have allowed pre-natal specialty clinics to be fairly self-reliant. Genetics clinics offering both pediatric and adult services must rely on institutional, state, and local support because it is virtually impossible for them to be self-sustaining based on billable services.

Besides the lack of CPT[®] codes mentioned previously, many third-party payers continue to deny coverage for genetic services by claiming that they are "experimental" or because they deem "counseling" services nonessential. Patients frequently must cover the costs,

although none are turned away due to inability to pay. When services are reimbursed, the amount paid is typically too low to cover the clinic's actual costs. Therefore, the dependence of the regional clinics on public health funding limits the ability to conduct other core public health functions such as assessment, policy development, and quality assurance, including health education.

Confidentiality and Privacy Policy Issues

Every person has genes that carry the risk or certainty of eventual disease or disability. Many people are concerned that misuse of genetic information could exacerbate discriminatory practices in insurance and in employment. Over the past four years, both state and federal legislative proposals have attempted to address this issue of "genetic discrimination." However, to date, no federal or state protections specifically bar the misuse of genetic information. In public hearings conducted in 1998, residents from around Washington State reported that without comprehensive protections covering both employment and health care, they would be fearful of possible discriminatory practices and would be reluctant to participate in clinical genetics studies. This potential decrease in patient participation would slow medical and scientific progress.

For genetics advances to benefit the general population, access to, and quality of, genetic health care services must be ensured. Furthermore, health and social service providers and the general public should be educated about genetics issues. The public health community is in the unique position to help move the discoveries gained through sequencing the human genome into appropriate medical and public health practice. Public health professionals have a long track record of working with partners in the health care system, community groups, higher education, local government, schools, and business to improve people's health. It is exactly these types of partnerships and opportunities for dialogue that will help to resolve some of the complex issues already confronting our health care delivery system as a result of the Human Genome Project.

Recommended Reading

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Author

Debra Lochner Doyle, M.S., C.G.C., is the state coordinator for genetic services with the Washington State Department of Health. She is board certified by the American Board of Medical Genetics and the American Board of Genetic Counseling and is a past president of the National Society of Genetic Counselors.

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 Box 354809, Seattle, Washington 98195-4809
 (206) 685-2617, Fax (206) 543-9345