Genetic Services Policy Project Final Report

Chapter 2: The Delivery of Genetic Services in the United States

A. What are genetic services?

The Genetic Services Policy Project defines genetic services as genetic testing, diagnosis of genetic conditions, genetic counseling, and treatments for individuals with genetic disorders. Genetic services occur across the lifecycle, and they affect behavior, disease monitoring, and treatment for those individuals being tested and their families.

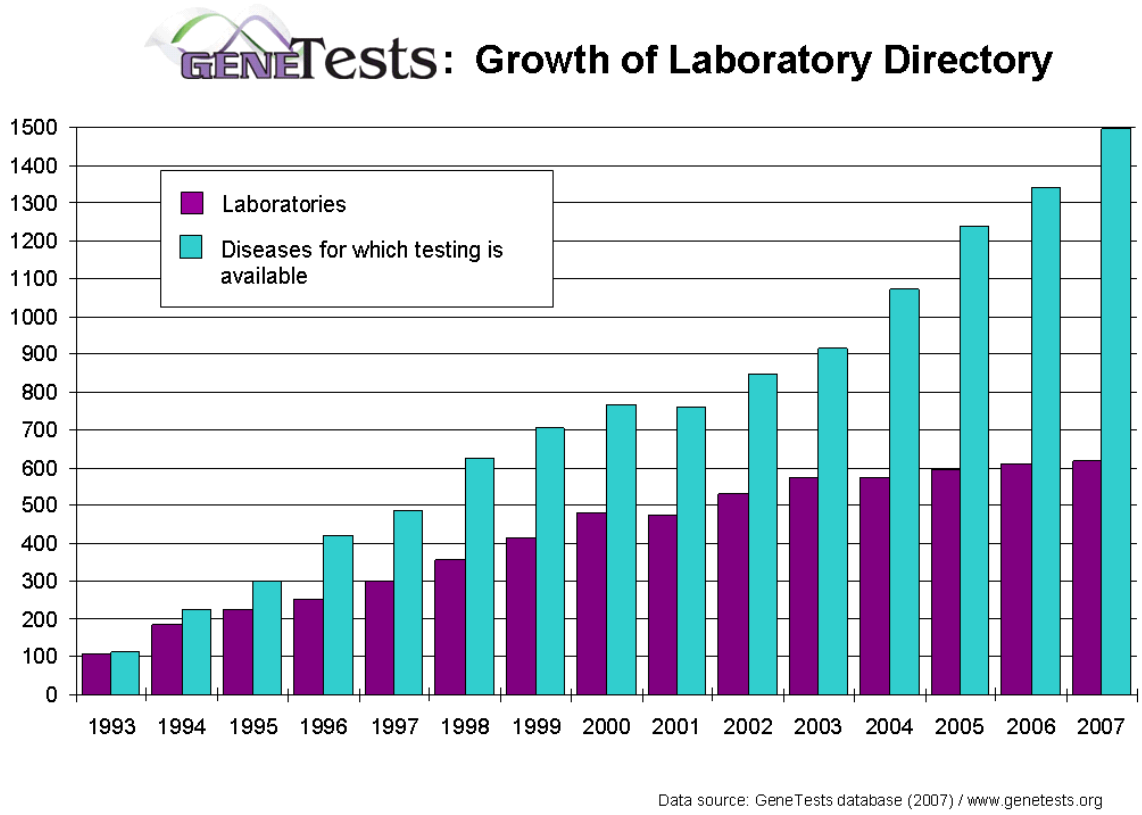
Genetic testing is defined as the laboratory analysis for DNA, RNA, chromosomes, or gene products (GeneTests: Educational: Fact Sheet). The most common types of genetic tests include:

- *Diagnostic testing,* to confirm or rule out known or suspected genetic disorders in symptomatic individuals (Jansson, 2007);
- *Screening* of a population, to identify an increased risk of illness or abnormality;
- *Predictive testing,* to identify future health risks in asymptomatic individuals with a family history of a genetic disorder (Jansson, 2007);
- *Pharmacogenetic testing,* to predict an individual's response to one or more drugs;
- *Carrier testing,* to determine whether an individual carries a mutation associated with a recessive disease;
- *Prenatal testing,* during pregnancy to provide information about genetic conditions or birth defects, using methods such as ultrasound, chorionic villus sampling, amniocentesis, and DNA studies of the fetus; and
- *Preimplantation genetic diagnosis,* to test embryos for known or suspected genetic conditions prior to implantation in the uterus.

Diagnosis and treatment of genetic disorders involves:

- *Genetics examination*: either a complete physical examination primarily to identify dysmorphology or a directed genetic examination to find evidence of a specific genetic condition;
- *Genetic counseling,* to help people understand the medical, personal, and family implications of genetic contributions to the disease. The process includes:
    - collection and interpretation of family and medical histories to assess the chance of disease occurrence or recurrence;
    - education about inheritance, testing, management, prevention, resources and research; and,
    - counseling to promote informed choices and adaptations to the risk or condition (Resta et al., 2006).
- *Genetic therapies* or other clinical interventions arising from genetic research and investigation, including pharmaceuticals, diets, enzyme replacement, gene therapy; and,
- *Pharmacogenomics,* the use of genomic concepts in the development and clinical application of pharmaceuticals to improve drug treatment by decreasing adverse drug reactions and increasing drug effectiveness.

Genetics services are expanding rapidly. Since 1993, the number of diseases for which genetic testing is available has grown from slightly more than 100 to over 1500 as of March 2008 (GeneTests: Growth).



In addition, technological advances are allowing increasingly larger numbers of conditions to be tested rapidly via a single test, e.g., microarray-based comparative genomic hybridization (CGH) analysis (Bejjani et al., 2005). Ancillary genetic services, such as parentage testing and DNA banking for future testing, are also growing service areas.

B. Who receives services?

Traditionally genetic services have focused on individuals and families with, or at risk for, rare single gene disorders or chromosomal abnormalities. This focus is now shifting as genetic testing and other services are being incorporated into diverse areas of medical practice. For example, genetic testing for the Her2 gene is now recommended in the clinical care of all patients with invasive breast cancer to determine the appropriate course of therapy. Positive Her2 status, which is present in approximately 20 percent of invasive breast cancer cases, has been shown to predict beneficial response and improve survival with the drug trastuzumab (Herceptin) (Carlson et al., 2006). In addition to new applications for specific diseases, efforts are underway to incorporate genetic information into the provision of routine clinical care, including preventive care. The Department of Veterans Affairs is implementing a VA Genomic Medicine Program that will link the individual genetic information of volunteers in the system to their electronic health records. These data will "eventually enable VA healthcare providers to

consider patients' genetic profiles when prescribing treatments or recommending preventive measures" (U.S. Department of Veterans Affairs, 2006). Duke University has launched an innovative "Prospective Medicine" initiative that includes plans to integrate biomarkers and genomic information, as they become available, with other clinical information into individualized risk assessment, health planning and coaching (Snyderman and Williams, 2003).

To date, the provision of genetic services has been closely associated with the life cycle and occurs across five broad stages: preconception, prenatal, newborn, pediatric, and adult. The following table shows which genetic services are provided during different life stages and the questions they address.

Preconception: *What is "our" risk of having an affected child? Should I get pregnant?*	**Prenatal:** *How will I manage my pregnancy?*	**Newborn, Pediatric, and Adult:** *How might my genetics affect my health?*
• Genetic counseling • Carrier test • Predisposition / susceptibility test • Diagnostic test	• Genetic counseling • Carrier test • Predisposition / susceptibility test • Diagnostic test • Preimplantation genetic diagnosis • Prenatal testing	• Genetic counseling • Carrier test • Predisposition / susceptibility test • Diagnostic test • Genetic evaluation • Pharmacogenetics • Gene therapy

The following list includes a number of common reasons why individuals and families are referred to, or seek, genetic services (GeneTests: Educational: About Genetic Services).

- *Preconception/prenatal*:
  - Mother will be 35 years or older at delivery. Note: Maternal age of 35 years has historically been used as a decision point for referral to genetic services, particularly evaluation for Down syndrome. New guidelines suggest all pregnant women regardless of age should be screened and/or offered diagnostic testing for Down syndrome. (American College of Obstetrics and Gynecology, 2007)
  - Abnormal results from a multiple marker maternal serum screen or fetal ultrasound
  - Personal or family history of a known or suspected genetic disorder, birth defect, or chromosomal abnormality
  - Exposure to a known or suspected teratogen
  - Mother has a medical condition known or suspected to affect fetal development
  - Two or more pregnancy losses
  - Close biological relationship of parents
  - Increased risk of certain genetic disorders associated with ethnicity.
- *Pediatric*:
  - Abnormal newborn screening results
  - One or more major malformations in any organ system

    - Abnormalities in growth
    - Mental retardation or developmental delay
    - Blindness or deafness
    - Presence of a known or suspected genetic disorder or chromosomal abnormality
    - Family history of a known or suspected genetic disorder, birth defect, or chromosomal abnormality
- *Adult*:
    - Mental retardation
    - Personal or family history of hereditary cancers
    - Personal or family history of a known suspected genetic condition or chromosomal abnormality
    - Development of a degenerative disease including blindness or deafness
    - Risk assessment for pregnancy planning
    - Infertility including multiple pregnancy losses

Unfortunately, data about who actually utilizes genetic services are extremely limited. In other clinical areas, billing information can provide valuable data about utilization. Prior to 2007, genetic evaluation and/or genetic counseling services were billed using evaluation and management codes (e.g., "consultation" or "office visit"), the same codes used by all sub-specialists. As a result, data on their utilization are incomplete and unreliable. As of January 1, 2007, medical genetic services and genetic counseling are specifically included in the Current Procedural Terminology (CPT) codes that describe health services for billing and reimbursement purposes (McDermott, 2007). It is hoped that the new CPT code, 96040, will improve data collection specifically about these services and the individuals who utilize them.

In the meantime, by recognizing the distribution of genetic services across the life cycle, it is possible to make inferences about the people using them. For example, nearly all of the 4.1 million babies born in the United States every year are screened for several genetic and metabolic disorders (National Newborn Screening and Genetics Resource Center website). Fourteen percent of women giving birth are 35 years and older (U.S. Centers for Disease Control and Prevention: Births, 2006), the age at which they would be offered amniocentesis. Increasingly, younger pregnant women are undergoing screening. Therefore, it is likely that at least 600,000 U.S. women are offered some type of genetic screening during pregnancy in each year (Rabin, 2007). Recent guidelines from the American College of Obstetrics and Gynecology (2007) recommend that all pregnant women be offered multiple marker screening for neural tube defects and chromosomal abnormalities. The results of these screening tests could lead to invasive diagnostic testing such as chorionic villus sampling or amniocentesis (American College of Obstetrics and Gynecology, 2007). The impact of this change on genetic service utilization is not yet known. In addition, ACOG recommends that information about cystic fibrosis carrier screening be provided to all couples and, at a minimum, screening should be offered to all couples where both partners are Caucasian, European or Ashkenazi Jewish prior to conception or in early pregnancy (American College of Obstetrics and Gynecology, 2005).

A 2003 survey of medical geneticists describes children (birth through adolescence) as the major client group for services, accounting for 76 percent of all new patients, with reproductive genetics patients accounting for 11 percent of those who use services, and "other adults" totaling

13 percent (Cooksey, 2005). A 2006 National Society of Genetic Counselors (NSGC) study found that, from 2000-2002, respondents shifted specialty areas away from prenatal (59 to 56 percent) and toward adult (26 to 28 percent) and cancer genetics (34 to 42 percent).

The GSPP analyzed utilization trends at nine regional genetics clinics in Washington State from 1995-2004 and found that the number of visits grew by an average of 8 percent a year, a rate surpassing that of other health care providers (Wang and Watts, 2007). The analysis did not include visits for prenatal services. The analysis revealed changes in the patient mix. The share of female and urban clients increased, as did the share of adults 35 years and older. The study found that in 2000, the number of adults visiting the clinics surpassed the number of children, and the trend has continued since then. Adults 20 years and older comprised 40 percent of all visits in 2004, compared with 29 percent in 1995.

Distribution of genetic services, and therefore access to these services, is dependent on broad issues of resources and public awareness, which are strongly influenced by the activities of the advocacy groups organized around specific genetic conditions or diseases. These groups play important roles in mobilizing public support for research and application of new technologies, shaping public policies, and providing clinical services directly. The number of these groups has grown significantly with the work of the Human Genome Project. More than 3,000 support groups, networks, and informational web sites are listed under Google's directory of genetics-associated diseases and conditions, including 148 groups associated with cystic fibrosis, 111 for Down Syndrome, 30 for Huntington's Disease, and 19 for sickle cell disease (Google Directory).

Most of the advocacy groups were established by parents of children with genetic-related disabilities and the organizations that work with them. The Washington, D.C.-based Genetic Alliance, formed in 1985, identifies itself as a coalition of "millions of individuals" and more than 600 organizations, from the Aasrskog Syndrome Parents Support Group to the Y-ME National Breast Cancer Organization (Genetic Alliance website).

In common with most of the largest genetics-related advocacy groups, the Genetic Alliance raises both public and private resources—the alliance is partly supported by funding from the federal Health Resources and Services Administration (HRSA) and the Centers for Disease Control and Prevention (CDC). Many of the groups advocate for specific public policies. The March of Dimes, for example, has become strongly supportive of legislative efforts to mandate a broad, consistent panel of conditions for newborn screening by the states (March of Dimes website). The reach and influence of the advocacy groups is not proportionate with the prevalence or public health impact of the specific conditions that are their focus. For example, the Cystic Fibrosis Foundation annually raises more than 30 times the resources raised by the Sickle Cell Disease Association of America, despite the fact that sickle cell disease's national prevalence is more than twice that of cystic fibrosis (80,000 v. 30,000). The inequity, which reflects the ethnicity and socio-demographics of the individuals and families affected, has led to significant gaps in federally sponsored research and in application of gains in clinical care (Smith et al., 2006). However, recent data indicate that the National Institutes of Health (NIH) funding for cystic fibrosis research has been reduced from $117 million in 2003 to $85 million in 2006, which is less than 2006 funding for sickle cell disease at $91 million (National Institutes of Health, 2007).

C. Who provides genetic services?

The genetic services workforce consists primarily of health professionals providing diagnosis, counseling, testing, and test interpretation. Nearly all licenses and credentials to perform genetics services are conferred by private organizations and trade associations.

Providers fall into two general categories: those who are specifically trained to provide genetic services and are certified by professional organizations, and those who perform genetic services but are licensed in another discipline and have received little or no formal training or certification *specifically in genetics*.

In the first category—those with formal training—are four types of providers that are essential to genetic services delivery. These professionals provide direct patient care, as well as work to educate the public and their colleagues, participate in research, and administer public and private programs.

These four provider types are:

- **Medical geneticists**, all of whom have medical or doctorate degrees, who provide counseling about the risk for genetic disorders as well as diagnosis, management, and treatment. The American Board of Medical Genetics (ABMG) certifies five categories of practice: PhD medical genetics (for which the certification exam has been dropped in 2007), clinical genetics, clinical cytogenetics, clinical biochemical genetics, and clinical molecular genetics. Certification requires at least 48 months of full-time training in addition to MD, DO, or doctoral degrees. Patients often see medical geneticists when a screening test comes back positive and they are seeking confirmation or interpretation of test results and a diagnosis. Most medical geneticists work in academic medical centers (see page 8), and may also direct laboratories, research, and genetics-related organizations. Approximately 2,300 individuals have received certification in one or more of the ABMG categories of practice since 1982, with 166 individuals becoming certified in 2007 (American Board of Medical Genetics website).

- **Genetic counselors,** who must complete master's level training at an American Board of Genetic Counseling (ABGC)-certified program. Instruction includes principles of human genetics, applicability of related sciences to the practice of clinical and medical genetics, psychosocial issues, and the ethical, legal, and social issues associated with genetic services delivery. According to the NSGC, these providers "identify families at risk, investigate the problem present in the family, interpret information about the disorder, analyze inheritance patterns and risks of recurrence and review options with the family. [They] also provide supportive counseling to families, serve as patient advocates and refer individuals and families to community or state support services." (National Society of Genetic Counselors: FAQs) Since 1993, 2,035 genetic counselors have been certified by the ABGC, with about 400 new counselors certified nationwide every two years (American Board of Genetic Counseling website). Primary sub-specialties are prenatal, pediatric, adult, cancer, specialty disease, and molecular/cytogenetic testing (NSGC Professional Status Survey, 2006).

- **Genetic nurses,** who are trained to assess and discuss genetic disease risk with patients and families. The Genetic Nursing Credentialing Commission oversees two types of credentials for genetic nursing: Advanced Practice Nurse in Genetics (APNG), which requires a master's in nursing, and Clinical Genetic Nurse (CGN), for baccalaureate degree nurses. Credentialing, which has been available only since 2001 for APNG and 2002 for CGN also requires a practice portfolio indicating a 50 percent or greater genetic practice component. As yet, no states require licensure and certification for genetic nurses, beyond standard nursing requirements (Greco and Mahon, 2003; Lea et al., 2006).

- **Genetic technologists,** who work in laboratories and generally do not interact with patients. The National Credentialing Agency for Laboratory Personnel offers certification for cytogenetic technologists and molecular technologists, who must have bachelor's degrees in a scientific discipline or medical technology and at least 6 months to one year of laboratory experience in their discipline (National Credentialing Agency for Laboratory Personnel website). They must also meet competencies in areas such as specimen collection, handling, processing, and the use of general laboratory skills.

The second category of genetic services providers—those without formal training or certification specifically in genetics—consists of:

- **Physicians,** particularly those in primary care and in specialties in which they see high numbers of patients who are undergoing some type of genetic testing. These specialties include, but are not limited to, oncology, neurology, obstetrics-gynecology, psychology, and endocrinology (Washington State Department of Health, 2006). All physicians receive some basic genetics training in medical school with the specific curriculum dependent on the particular school. Residency programs may also offer genetics rotations to non-geneticist trainees. Physicians in practice qualify for continuing medical education credits for courses related to genetics and also may receive genetics training from their professional associations.

- **Other clinicians,** including nurse practitioners, midwives, physician assistants, and social workers. Genetics-related competencies have been developed in some of these areas (e.g., social work), but they are unevenly implemented across the country.

The genetic services workforce is growing. Genetic counselors serve more than 1.2 million clients each year, and the number of clinical patients they see has been increasing by 5 percent each year since 2000 (NSGC Professional Status Survey, 2004). But in some areas, the workforce is not growing at a rate consistent with expansion of knowledge and clinical applications. Medical geneticists, in particular, do not appear to be entering the field in numbers that would keep pace with population needs, and they are distributed unevenly across the country (Cooksey et al., 2005). For example, Georgia has been providing genetic services via telemedicine for several years. Several other states are beginning to explore this venue as well, including three western states, Hawaii, Oregon, and Washington, who have piloted programs in "telegenetics," which provide opportunities for genetic consultations in real-time, interactive video (Western States Genetic Services Collaborative website). Such programs can help overcome problems of access to genetic services caused by poor representation of providers in

rural areas and geographic barriers, and have received funding from the federally supported Region 7 Genetics Collaborative. In the Midwest, the Region 4 Genetics Collaborative is also exploring telemedicine as a vehicle for genetic services delivery as part of an overall goal to facilitate access to genetics expertise for underserved populations (Region 4 Genetics Collaborative website). Within the region, in Wisconsin, the Marshfield Clinic began offering telemedicine for genetics in November 2006.

Provider type	**Number**
Medical geneticists	2,342 (American Board of Medical Genetics: Numbers)
Genetic counselors	2,035 (American Board of Genetic Counseling: Fact Sheet)
Genetic nurses	27 with Advanced Practice Nurse in Genetics credential; 9 with the Clinical Genetic Nurse credential (Genetic Nursing Credentialing Commission)
Genetic technologists	1,200* (Association of Genetic Technologists)

*Note: There are 1,200 members of the Association of Genetic Technologists; the absolute number of genetic technologists (including those who are not members) could not be found.

State governments play several important roles in delivering genetic services for life stages from prenatal to adult.

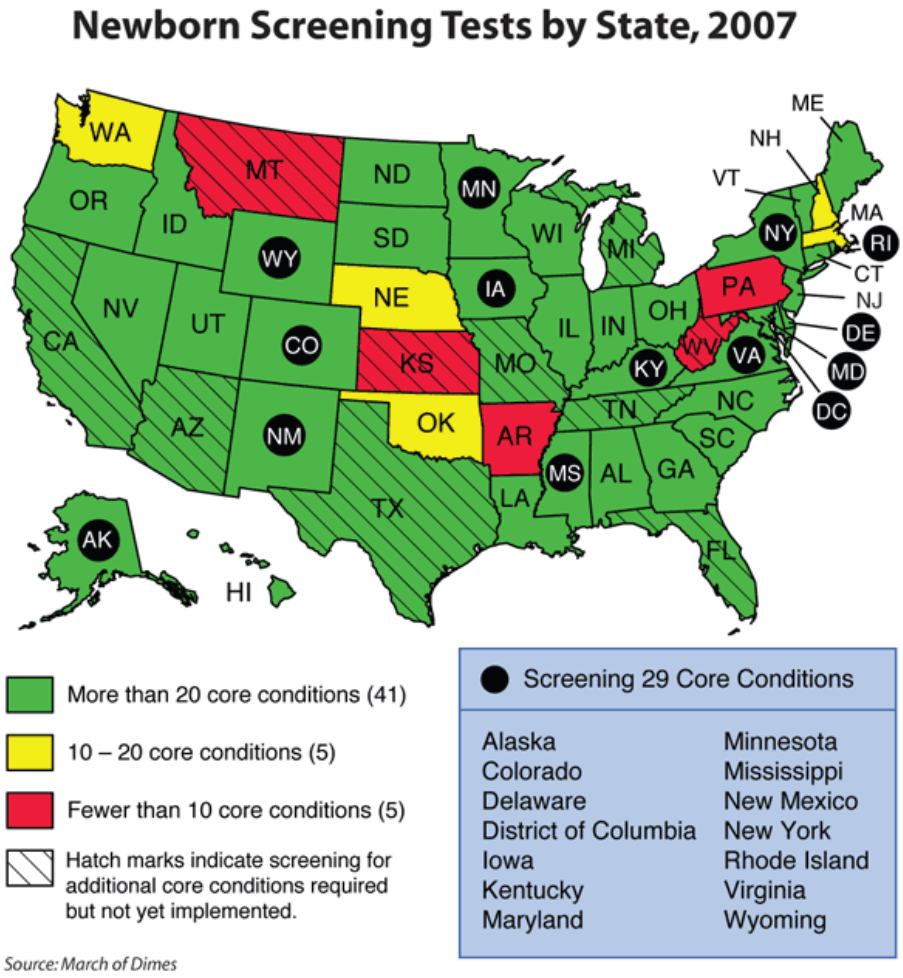
In California, the Genetic Disease Branch of the state health department administers the Expanded AFP (XAFP) Screening Program to assure that all pregnant women are offered prenatal screening and follow-up for certain birth defects and conditions, including open neural tube defects, Down syndrome and Trisomy 18. Participation in the program is voluntary and fee-based. California insurance providers are required to cover this service (California Genetic Disease Branch website).

Newborn screening is a major prevention-based, public health program for every state, usually performed to detect genetic and metabolic disorders. Maryland is the only state requiring parental consent to screen newborns, though most states have provisions for parents to opt out of screening for religious or other reasons (Therrell et al., 2006). Typically, blood samples are collected at birth and processed in state, regional, or contracted private laboratories. All states perform newborn screening for 5 to 50+ genetic and metabolic disorders, including disorders that cause mental retardation, disability, and premature death (National Newborn Screening and Genetics Resource Center website).

States differ considerably in the numbers of disorders on their screening panels, as well as how they count the number of tests they conduct; for example, Washington state counts hemoglobinopathy screening as one test while other states may count the same testing as 4 or more, including SS, SC, beta-thalassemia, and alpha thalassemia. In 2005, the American College of Medical Genetics recommended mandated screening for 29 disorders, including certain metabolic conditions and hearing loss (American College of Medical Genetics, 2006). But as of June 2006, only five states (Iowa, Maryland, Mississippi, New Jersey, and Virginia) and the District of Columbia required screening for all these conditions. At the same time, 31 states,

representing 64 percent of newborns, reported screening for more than 20 conditions. Most states have significantly expanded their newborn screening panels in recent years (March of Dimes website).

The following map shows how the state newborn screening panels compare as of 2007 (McDowell, 2007):



The decision regarding which conditions to include in the panels involves complex social, ethical, and political issues. Especially since the late 1990s, with the availability of technology that allows laboratories to test for many disorders with a single blood sample, the technical capabilities of newborn screening have outpaced public health policy in this area. Advocacy groups for children's health and those organized around genetic conditions, as well as many professional groups and researchers, have encouraged expansion of the newborn screening panels in recent years. One example is the Save Babies Through Screening Foundation. During 2006, the U.S. Senate considered legislation that would have established a standard slate of conditions subject to newborn screening and required states to contribute to a national database on screening results (S.3743). The companion bill was not introduced in the House.

At the same time, some health policy makers in the genetics field have proposed that the issue be addressed more consistently in the context of evidence-based public health, which requires

consideration of the balance of benefits, risks, and costs—including opportunity costs—of using public resources to screen newborns, particularly for rare conditions (Grosse et al., 2006). In this "new paradigm," the panels would also address potential harm to children and families, including the parental stress and expense caused by ambiguous test results, false positives, false negatives, labeling of children with mild or benign conditions, and unnecessary or otherwise misapplied therapies.

Another issue associated with expansion of the newborn screening panels is the availability of follow-up services for infants receiving abnormal test results. Families who, under these circumstances, are advised to seek genetic counseling may not possess the stable health insurance coverage needed to facilitate access to services. An analysis of 2001 data collected for the National Survey of Children with Special Health Care Needs (CSHCN) showed that 19 percent of families of CSHCN who needed genetic counseling—123,117 CSHCN families—did not receive it. CSHCN with interrupted insurance coverage and those without insurance were significantly less likely to receive needed genetic counseling services (Wang and Watts, 2007).

Few states employ genetic services providers. As of 2002, 21.6 percent (11) of the state genetics coordinators were certified by either ABMG or ABGC, but the majority did not have formal training in genetics (Coalition of State Genetics Coordinators, 2002).

Eighteen states have developed genetics plans or needs assessments, some with the support of federal funds from HRSA (National Newborn Screening and Genetics Resource Center: State Genetics Plans). These plans identify broad areas of genetics activities: newborn metabolic and hearing screening, clinical genetic services including testing and counseling across the lifespan; programs providing treatments and support to individuals and families with genetic disorders; educational activities; birth defects or other genetics related surveillance programs; policy development (e.g., genetic legislation, privacy and discrimination); and public health genetics research/evaluation activities. Some plans also aim to create Integrated Child Health Information Systems, an effort to improve health outcomes for children by increasing data sharing efficiencies and program coordination. Most state activities are directed by state genetics units, newborn screening programs, or federally supported CSHCN units.

States do coordinate initial screening and confirmation testing of infants identified through the newborn screening programs (both mandated and non-mandated) as well as follow-up activities when infants are diagnosed with a condition. Some states only do so through the primary care provider being notified to refer the infant for further testing and others oversee care coordination. In some states (e.g., Washington, Idaho, Alaska), state-sponsored genetics outreach clinics offer general testing, evaluation, and counseling services to individuals and families in geographic areas that would not normally have access to genetic services providers. Some states also provide treatment and direct care programs for individuals diagnosed with genetic conditions. In particular, many states provide and coordinate direct care for children and adults with genetic disorders such as sickle cell disease and hemophilia.

States also conduct a wide variety of activities that focus on surveillance for genetics related conditions. Forty-two states either have or are developing birth defects surveillance programs, financed primarily by federal grants, service-related fees and private sources (Wang et al., 2005).

In addition to these direct effects, states also have indirect influence on genetic services through their purchasing power. States are purchasers of genetic services for their employees, and they help shape public coverage of services through their work with their legislatures, insurers in their state, and state Medicaid programs.

D. Where are services provided?

Genetic services are provided across both clinical and laboratory settings. Clinical settings include private and group practices, hospitals, community health centers, specialty programs, state programs, and hospitals. Laboratory providers work in public (including state) laboratories and commercial labs. The following table shows different provider types and the settings in which they most often work.

Provider type	**Practice settings**
Medical geneticists	Academic medical centers (62%), hospitals (9%), commercial labs (9%), medical practices (9%), other settings (10%) (Cooksey et al., 2005)
Genetic counselors	Academic medical centers (41%), private hospitals or medical facilities (21%), public hospitals or medical facilities (11%), diagnostic laboratories (7%), HMOs (4%), physician's private practice (4%), and other (4%) (NSGC Professional Status Survey, 2004)
Genetic nurses	Specialty genetics clinics, primary health settings, cancer centers, prenatal and reproductive centers, research centers, industrial health, school health, biotech and insurance industries
Genetic technologists	State public health labs, commercial laboratories, biotech industry

The GeneTests Laboratory Directory website, a voluntary listing of molecular genetic testing offered by laboratories, indicates that, as of March 2008, 608 U.S. laboratories offer genetic testing and 1,144 clinics offer one or more of the following services:

- Adult genetics
- Pediatric genetics
- Prenatal diagnosis
- Cancer genetic counseling and risk assessment
- Telemedicine
- Preimplantation genetic diagnosis

In general, genetic services and consultations are more readily available in urban areas (such as those with academic medical centers) as opposed to rural areas (Acheson et al., 2005).

Advances in genomics as well as technological and cultural shifts toward the use of Internet-based services have contributed to the booming direct-to-consumer commerce in genetic tests. These retail tests bypass the traditional role of the health care provider to recommend genetic testing and interpret results. More than three dozen companies already offer such services online, ranging in cost from $200 to $3,456 (Google.com, 4/19/07). The new e-commerce is raising many consumer protection issues. While at least one online company offers genetic

counseling over the phone, most do not. Consumers who lack genetics knowledge and are unable to gauge tests' analytic or clinical validity may be vulnerable to inflated promises from advertisements. They may lack the knowledge to interpret test results and thus risk making ill-advised, even harmful health care decisions (Gollust et al., 2002). Direct-to-consumer tests have provoked a range of regulatory questions, as the federal government does not currently regulate the safety of most genetic tests (Genetics & Public Policy Center: FDA Regulation, 2006). In July 2006, the Federal Trade Commission issued an alert warning consumers to be skeptical about "at-home genetic tests" and to consult with their doctors or health care practitioners prior to purchasing or undertaking any tests and to discuss test results with a doctor or a genetic counselor (U.S. Federal Trade Commission, 2006). On the other hand, there are several potential benefits to online testing services, including increased uptake by the public of valuable services if people perceive greater privacy, and decreased concerns about insurance discrimination, especially if services are paid out-of-pocket.

E. Who pays for services?

As new genetic tests and services continually become available, consumers, providers, and payers must develop mechanisms to pay for them. Several factors present barriers to the integration of genetic services in the U.S. health care system and its current methods of financing, particularly reimbursement by third-party payers (Secretary's Advisory Committee on Genetics, Health and Society, 2006). These factors include the use of multiple providers (medical geneticists, genetic counselors, laboratory technicians, and others), uncertainty concerning the role of genetic factors in the actual onset of disease, complex issues of ethics, privacy and confidentiality, historical experience, and continuing issues of insurance coverage and reimbursement. In addition, many payers require providers to be licensed, and only six states require licensing of genetic counselors (National Conference of State Legislatures: Genetic Counselor Licensing).

Medicare reimbursement for genetic services is complicated by a longstanding U.S. Centers for Medicare and Medicaid Services (CMS) policy to exclude coverage of predictive and pre-symptomatic tests and services (Secretary's Advisory Committee on Genetics, Health and Society, 2006). Private health plans often follow CMS policies. Not all genetic services providers may bill Medicare directly; those who are statutorily eligible to bill directly for their services are physicians, nurse practitioners, physician assistants, certified nurse specialists, certified nurse midwives, clinical psychologists, and clinical social workers. Other non-physician providers must bill "incident to a physician," often using CPT codes that do not cover the full time required to provide care (Secretary's Advisory Committee on Genetics, Health and Society, 2006). Experience with the new CPT code for medical genetic services and genetic counseling, described earlier in this paper, is too limited at this time to determine the impact on billing and reimbursement; however, CMS Medicare has already decided it will not cover this code (McDermott, 2007).

Medicaid coverage is subject to state variation, but most state programs reimburse for "medically necessary" genetic services, including tests such as amniocentesis, maternal-serum screening for neural tube defects and Down syndrome, and chromosomal analysis from amniotic fluid (Secretary's Advisory Committee on Genetics, Health and Society, 2006).

Private health insurance plans are financing a significant share of genetic services—in general, covered services are those related directly to an enrollee's health, not those suggested by a disease or condition of a family member. Once again, the inconsistent methods of billing and reimbursement make it difficult to know which payers are financing which services. A 2004 survey of genetic counselors revealed that 57 percent report billing under their supervising physician's name, 9 percent bill under their own name and the supervising physician, and 14 percent do not bill for services at all (NSGC Professional Status Survey, 2004).

Established services, such as amniocentesis for pregnant women older than 35, are the most likely to be covered. Generally, private insurance plans cover genetic testing for chromosomal abnormalities, prenatal and neonatal diagnosis, and some pre-implantation genetic diagnosis services. Coverage decisions are often based on what competing plans are doing and which services are moving from experimental to standard practice. Many specifically exclude certain types of genetic testing, such as tests for Alzheimer's disease, which are considered experimental. This exclusion creates a particular problem with rare diseases, whose low prevalence presents difficulty in establishing clinical validity. Other considerations in coverage decisions include whether a technology has received FDA approval, if clinical trials demonstrate medical effectiveness, and whether practice guidelines exist to justify its use, as well as costs and projected cost savings (Secretary's Advisory Committee on Genetics, Health and Society, 2006). The 1996 Health Insurance Portability and Accountability Act (HIPAA) prohibits group health insurers from denying coverage based on genetic information.

Federal genetics-related expenditures—primarily for research through agencies within the U.S. Department of Health and Human Services—amount to about 15 percent of HRSA spending and 10 percent of Centers for Disease Control and Prevention (CDC) spending projected for 2007. The National Institutes of Health will spend more than $1.06 billion on the Human Genome Project in 2007 and nearly $4.8 billion on other genetics-related activities, including $355 million on gene therapy, $32 million on gene therapy clinical trials, $417 million on genetic testing, and $103 million on "conditions affecting unborn children" (National Institutes of Health, 2007).

States finance their genetic services activities through an array of public and private funding sources, including newborn screening fees (ranging from $0 to $139 with a mean of $54) (National Newborn Screening and Genetics Resource Center: Summation, 2008), state general funds, federal Maternal and Child Health Block Grant resources, CDC grants, and private grants. States receive reimbursement for the direct services they provide through private insurers, Medicaid, State Children's Health Insurance Plans (SCHIP), CSHCN programs, and consumers' out-of-pocket payments. States, for the most part, have published little expenditure data about the costs and reimbursement for the genetic services they provide (Wang, 2006).

F. How are genetic services regulated?

Regulation of genetic services includes government oversight of genetic testing, licensing of laboratories and their personnel, and quality assurance and control. This regulatory environment is changing rapidly and incorporates federal agencies, state initiatives, and professional organizations.

At the **federal level**, the Clinical Laboratory Improvement Act (CLIA), the Food, Drug, and Cosmetic Act, and the Federal Policy for the Protection of Human Subjects regulate both the development and application of genetic tests (Genetics & Public Policy Center: Who Regulates, 2006). But CLIA, which has general regulatory authority for all laboratory testing, has not yet implemented genetic-specific regulations, except for the cytogenetics specialty. As a result, genetic tests are not subject to uniform quality control and proficiency testing requirements—no single government agency has these responsibilities. It is often unclear to health care providers which laboratories are qualified to perform genetic testing. CMS is responsible for implementing CLIA and its standards, and the agency has come under growing pressure in recent years from health care provider and consumer groups advocating that it implement a genetic testing specialty (Javitt and Hudson, 2006). In 2006, the Government Accountability Office exposed the uneven oversight of genetic tests sold over the Internet and highlighted potential harms to consumers who may be misled by the information received from this testing, such as recommendations for nutritional supplements that are not only expensive but unnecessary or even contraindicated (U.S. Government Accountability Office, 2006).

The Food and Drug Administration (FDA) regulates genetic tests that are sold to laboratories. These "test kits" are considered diagnostic devices and are subject to agency approval for their safety and efficiency. But the test kits are available for only a small share of genetic tests compared with the far greater number of "homebrew" tests that are developed in-house and are marketed as clinical laboratory services. The agency has implemented regulations for some of the active ingredients in the homebrew tests (analyte specific reagents), and in recent years, it has suggested that it will reconsider additional controls. But it has resisted more stringent regulation so as not to discourage innovation, a situation that puts developers of the test kits at a competitive disadvantage to homebrew makers in the rapidly expanding market.

CDC collaborates with other public agencies and private-sector groups to develop both regulatory and voluntary laboratory standards and to promote integration of validated genetic tests into clinical and public health practice. The Office of Genomics and Disease Prevention at CDC (now the National Office of Public Health Genomics) has initiated two projects: the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) and an evaluation of the analytic validity, clinical validity, clinical utility, and associated ethical, legal and social implications (ACCE) of genomic test data. These projects support the first phases of a coordinated process for evaluating genetic tests that are in transition from research to clinical and public health practice (U.S. Centers for Disease Control: Genetic Testing). The CDC National Center for Environmental Health offers, in partnership with the Association of Public Health Laboratories, a Newborn Screening Quality Assurance Program to help state agencies and laboratories improve the quality of test results.

Congress is increasingly addressing legislation to regulate the protection and use of genetic information. Since 1995, healthy individuals with a genetic predisposition to certain disabling diseases fall within the scope of the Americans with Disabilities Act's nondiscrimination rules (Americans with Disabilities Act Compliance Manual, 1995). As mentioned previously, the 1996 HIPAA legislation (HIPAA Title I) prevents insurers from discriminating against individuals in enrollment and eligibility for benefits in group health plans, based on health status

including genetic information (McAndrew, 2007). In addition, group insurers must not increase premiums or use genetic information in the absence of a diagnosis as the basis for a "pre-existing condition." For a number of federal legislative sessions, Congress has debated various bills focused on discriminatory practices against people without signs or symptoms of illness but who possess a known genetic mutation for illness. In 2005, the Genetic Information Nondiscrimination Act of 2003 passed the Senate, but was stalled in the House. A new bill, the Genetic Information Nondiscrimination Act of 2007 was introduced in January 2007. The bill passed the House on April 25, 2007 and passed the Senate unopposed on April 24, 2008. It was signed into law on May 21, 2008 and became Public Law No: 110-233 (S.358, 2007; H.R.493, 2007).

In 2006, Sen. Barack Obama (D-IL) introduced the Genomics and Personalized Medicine Act, which would promote research and application of new genetic technologies by improving access and utilization of "valid, reliable and accurate molecular genetic tests by all populations, thus helping to secure the promise of personalized medicine for all Americans" (S.3822, 2006). The measure would expand research programs in genetics and personalized medicine ("new methods of molecular analysis to better manage a patient's disease or predisposition towards a disease") (Personalized Medicine Coalition), promote translation of research findings into clinical and public health applications, and require more extensive federal review of the safety and efficacy of genetic tests. It would also convene stakeholders in a "Personalized Medicine Interagency Working Group" including the National Institutes of Health, CDC, FDA, CMS, HRSA, and the Department of Energy. Sen. Obama reintroduced the bill in 2007 (S.976, 2007).

An important section of the Obama bill—one that was also addressed by Sen. Edward M. Kennedy (D-MA)—was clarification of the FDA's authority to regulate homebrew tests. The Obama measure would have required developers of homebrew tests to submit to the FDA evidence to support their tests' analytical and clinical validity for inclusion in a public database. Although new homebrew tests could be marketed without FDA clearance, labeling would indicate whether the tests were FDA-cleared or -approved.

States conduct various levels of genetic services regulation (National Conference of State Legislatures: Genetic Technologies Project), including:

- *Mandated screening of all newborns*. State legislatures appropriate funds or authorize fees for newborn screening programs. State statutes may address newborn screening panels, privacy and confidentiality issues, and laboratory standards.

- *Licensing of genetic counselors*. Only six states (California, Illinois, Massachusetts, Oklahoma, Tennessee and Utah) require licenses and define minimum qualifications for licensure, as of January 2008 (National Conference of State Legislatures: Genetic Counselor Licensing).

- *Use of genetic information in health insurance*. Most states (44) have enacted legislation prohibiting insurance companies from using genetic information for decisions on eligibility for policies; 41 states prohibit use of genetic information for risk selection or risk classification; and 27 states prohibit insurance companies from disclosing information without informed consent.

- *Protection of genetic privacy*. Most states have imposed rules requiring more rigorous protection of genetic information than other types of health information. Twenty-seven states require consent to disclose genetic information, and 17 have laws requiring informed consent for a third party to perform a genetic test or obtain genetic information. Washington State alone treats genetic information the same as other health information under its state health privacy protections.

- *Laboratory practice standards*. New York is the only state that has implemented specific laboratory practice standards for several genetic specialties. New York and Washington State conduct their own laboratory inspections because they have more stringent standards than CLIA.

- *Regulation of embryonic and fetal research*. State laws may promote or prohibit stem cell research, and they may restrict funding for this purpose. About half of all states have implemented laws that, in various ways, restrict research on aborted fetuses and embryos.

- *Genetic nondiscrimination in employment*. Thirty-four states have implemented laws prohibiting discrimination in employment decisions of hiring, promotion and/or termination based on the results of genetic tests. Most states also restrict employer access to genetic information.

In the **private sector**, professional societies and other organizations are involved in developing guidelines for genetic tests. These organizations include the Association of Public Health Laboratories, the American College of Medical Genetics, the College of American Pathologists, the National Committee on Clinical Laboratory Standards, and the Commission on Office Laboratory Accreditation.

In addition, the federal Agency for Healthcare Research and Quality (AHRQ) supports the development and use of evidence-based quality measures in health care practice, including genetics related measures (Agency for Healthcare Research and Quality website). Professional organizations, such as the American Medical Association Physician Consortium for Performance Improvement, are engaged in performance improvement initiatives that include the use of quality measures (American Medical Association). Though limited in number, examples of genetics related measures include percentage of prenatal patients aged 35 years and older at the time of expected delivery who are offered amniocentesis or chorionic villus sampling and percentage of prenatal patients less than 35 years who are offered multiple marker testing for congenital anomalies (National Quality Measures Clearinghouse).

G. The Future of Genetic Services

The science behind genetic services is expanding rapidly. New services, particularly new genetic tests, follow closely on the heels of scientific discovery. Thus, public policy must address not only issues arising from the delivery of current services but also translational issues such as when genetic tests are ready for clinical application. What level of evidence, and what kind of evidence, is sufficient before a test reaches the marketplace? Who should provide the

evidence? Who should pay for its development? When the test is marketed, what standards of practice should apply to its use? Who should pay for the test? Who should provide it? These questions are among the translational policy issues we address in the remainder of this report.

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