

Genetic Services Policy Project Case Studies Overview

The field of genetic services is emerging as quite complex, with many ethical, legal, social, financial, and policy implications. Because the nuances of the issues vary across different types of genetic services, we have chosen to use a case study approach to investigate service delivery. The case study as a research strategy applies a host of data collection techniques to study a “contemporary phenomenon within its real life context.”¹ In other words, the case study allows us to explore the current issues surrounding genetic services as the issues develop and surface in the broader health care system.

Specific case study topics were chosen based on the systematic application of selection criteria. Topics were to encompass the six major types of genetic services:

1. screening (not diagnostic)
2. predictive/susceptibility testing
3. pharmacogenetic testing
4. testing that leads to personalized, non-drug therapies
5. gene therapy
6. genetic counseling

In addition, we sought variation across the following dimensions:

- Cultural effects/issues
- Delivery system(s) involved
- Financing mechanism
- Target population
- Old and new technology effects/issues
- Evidence of effectiveness
- Association to major health problem
- Stakeholder interests and involvement

Based on these criteria, we chose seven case study topics. Five are clinical studies, addressing these conditions: aminoglycoside induced hearing loss; cystic fibrosis; hereditary breast and ovarian cancer; multiple congenital anomalies; and sickle cell anemia and beta-thalassemia. The remaining two studies address infrastructure issues.

Clinical Studies:

Variations in the selection criteria across the case studies foster comparative analyses.

	Population (Race/Ethnicity Prevalence)	Age of onset	Disease Severity	Purpose of Genetic Testing	Public Programs	Retail Test for Genetic Mutation(s)
Amino-glycoside Induced Hearing Loss	Higher prevalence in some Mediterranean populations		Moderate, Varies	Prediction	No	Yes

Cystic Fibrosis	Higher prevalence in European-American populations	Childhood, adult (rare)	Severe	Diagnosis, Carrier detection, Prenatal diagnosis	Yes	Yes, carrier screening
Hereditary Breast and Ovarian Cancer	Higher prevalence in Ashkenazi and Icelandic populations	Adult	Severe	Diagnosis, Prediction	No	Yes
Multiple Congenital Anomalies	Varies by specific condition; non-standardized definition	Birth	Varies	Diagnosis, Prenatal diagnosis	Yes, under the larger category of programs for children with special health needs	No
Sickle Cell Disease and Beta-thalassemias	Higher prevalence in African-American, some Mediterranean, and some Asian populations	Childhood	Varies	Diagnosis, Carrier detection, Prenatal diagnosis	Yes	No

Aminoglycoside induced hearing loss is an example of the growing field of pharmacogenomics (PGx), the study of drug-gene interactions. PGx is of interest to multiple stakeholders, particularly payers, because of the prospects for increasing safety and reducing therapy costs by better targeting drug response. Testing for the gene variant associated with aminoglycoside induced hearing loss is commercially available and marketed to the provider community. However, the clinical utility of this test remains unclear. To provide additional data for providers, payers, and the public, this case study involves a cost utility analysis of the genetic test in relation to clinical outcome.

Cystic fibrosis addresses several different types of genetic services. Historically, the disorder was diagnosed through testing after an individual began to experience symptoms. Newer technologies have introduced screening of asymptomatic individuals, including newborns, at relatively low cost. Testing and screening also affect genetic counseling for patients with cystic fibrosis as well as carriers of the gene variants.

In addition to involving a variety of genetic services, the unique care systems for cystic fibrosis warrant exploration. The presence of specialty centers throughout the United States allows for inquiry into access and payment issues. The development of improved therapies has increased life expectancy, which affects public programs and financing systems. Finally, the impact of an active advocacy group and the involvement of specific ethnic communities can also be explored.

Hereditary breast and ovarian cancers are complex diseases for which screening and genetic testing exist. Because genetic testing may, to a degree, quantify susceptibility, the role of inherited risk impacts genetic counseling to patients and families. Research and development by

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drug manufacturers have also resulted in the marketing of testing related to more personalized treatment of cancers.

The elucidation of genetic variability around breast and ovarian cancers introduces challenges around the definition of the appropriate target population for genetic testing. In turn, evolving testing guidelines may have downstream effects on the financing of testing and may affect health outcomes. In addition, this case study provides the opportunity to explore the role of direct-to-consumer marketing of genetic tests in populations that may or may not benefit from testing.

Multiple congenital anomalies are complex conditions and may be defined as the presence of two or more major abnormalities. Although the causes of multiple congenital anomalies are not well defined, the multi-factorial etiology involves genetic as well as environmental factors.

Uncertainty surrounding etiology and disorder severity affects the role of screening, testing, and counseling for all families. Introduction and dissemination of technologies have the potential to affect public and private payment of screening services.

Sickle cell disease and beta-thalassemias are recessive conditions of varying severity. Although testing and screening began as targeted efforts toward specific ethnicities, the disorders are now included in state newborn screening programs. Prenatal screening and carrier testing are available both in public and private delivery systems. Specialty centers and public programs have developed to treat and assist patients in the management of disease. Because detection and care for these disorders have improved, patient life expectancy continues to increase.

This case study enables us to explore how services have developed and are currently delivered. We will raise some of the controversies surrounding ethnicity, culture, and the history of sickle cell testing. Access to services and financing of services will be considered in the context of continuing disparity in health outcomes.

Infrastructure Studies:

We chose two non-clinical topics because progress in genetic services, and the delivery systems for those services, will also be substantially affected by trends in both health care delivery and technology. The two topics are: the use of enabling technologies (specifically the electronic medical record/personal health record (EMR/PHR) and gene chip technology) and direct-to-consumer retail sale of genetic testing.

Enabling technologies. Individuals can already view their complete medical records electronically. In the near future, individuals will also be able to obtain their full genome on a device or on a personal computer at relatively modest expense. Our case study examines the impact of these technologies on consumers, patients, providers and other institutional players in the health care arena.

We will examine:

- the nature of the technology;
- security and access controls;

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- how soon, with what level of maturity, and at what price points the technology is expected to be available;
- what initial marketing plans show about expected market share by type of vendor;
- total market penetration estimates;
- whether any modeling of the impact of the technology in health care has been undertaken; and,
- whether any synergistic technology exists that would fundamentally change projections about social, economic and health care impacts.

Direct to consumer (retail) genetic services. The convergence of accelerating discovery in genetics, rapidly advancing applications and technology, and policy initiatives enabling greater consumerism in health care (e.g., health savings accounts) creates favorable conditions for the emergence of a direct to consumer genetic services market, principally in testing, risk analysis, and educational tools and content. Some retail genetic services providers already exist. We will track how this sector grows, specifically the types of provider entities that enter this market, the scope of services they offer, the extent of their adherence to explicit and implicit professional standards, and their reception among both consumers and medical professionals. We will also track the activities and success of existing providers, including monitoring how their business model changes over time.

References

1. Yin RK, Campbell DT. *Case Study Research: Design and Methods*. 3rd ed: Sage Publications, Inc.; 2003.