

# CRS Report for Congress

## Genetic Exceptionalism: Genetic Information and Public Policy

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# Genetic Exceptionalism: Genetic Information and Public Policy

## Summary

The Human Genome Project, from inception through completion, has generated a great deal of debate over the appropriate uses, as well as potential misuses, of genetic information. Legislation that is specific to genetics is predicated on the concept of genetic exceptionalism, or the premise that genetic information is unique and, therefore, merits both special and different, or exceptional, treatment. As applied to public policy, this translates into genetics-specific legislative approaches to various health policy issues such as privacy and discrimination in health insurance. Amidst great concern over the potential for the misuse of genetic information, most states passed genetics legislation during the past decade and a half in areas such as genetic privacy, genetic discrimination in health insurance, and genetic discrimination in employment. A genetic exceptionalist approach is now being pursued at the federal level, as the 110<sup>th</sup> Congress debates the Genetic Information Nondiscrimination Act of 2007 (H.R. 493/S. 358).

This report provides an overview of the nature of genetic information and its implications for individuals, family, and society. Individuals utilize genetic information to guide health care and other decisions, when possible, and may experience anxiety as a result of genetic test results. Genetic test results for an individual may often be informative for other close family members and thus influence their care decisions. Society must grapple with the effect genetic information may have on our conception of disease, as well as its impact on issues like privacy and equity. The report ends by summarizing the main policy issues involved with a genetic exceptionalist approach to public policy, including defining genetic information; physically separating genetic information from other medical information; unintended disparities between “genetic” and “nongenetic” disease; and the effect of legislation on participation in genetic research, on uptake of genetic technology and on the delivery of high quality health care.

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# Genetic Exceptionalism: Genetic Information and Public Policy

## Introduction

Advances in human genetics, and especially the completion of the Human Genome Project, have focused attention on the potentially numerous ways that genetic information may be used to improve and benefit health and health care. Despite excitement about the anticipated benefits that the use of such information may generate, lawmakers have raised concern about the potential for misuse of this information as well. Genetic information can be defined in many ways, but for the purposes of this report, it means information about genes, gene products, or inherited characteristics generated through either genetic testing or analysis of family history.<sup>1</sup>

The term “genetic exceptionalism” refers to the concept that genetic information is inherently unique, should receive special consideration, and should be treated differently in law. This idea is based on the supposition that genetic information itself embodies several characteristics that may make it special and differentiate it from other medical or even personal information. According to the perspective of genetic exceptionalism, the characteristics of genetic information that make it different include the following: it can be predictive of future disease; it is a unique identifier; it can reveal information about family members; it is vertically transmitted (passed from parent to child); it can impact communities; it can be used to discriminate and stigmatize; and it can cause serious psychological harm. In addition, although not specific to genetic information, the following characteristics also describe genetic information: it can be obtained from small samples (possibly without consent); it may be used for purposes other than those for which it was collected; it is of interest to third parties (such as employers and insurers); it may be important for determining susceptibility to disease and effectiveness of treatment; and it can be recovered from stored specimens after many years.<sup>2</sup> These characteristics are not unique to genetic information; other medical information may also have some of these characteristics in common with genetic information. However, it may be argued that there is little medical or personal information that shares *all* of these characteristics with genetic information.

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<sup>1</sup> For a more detailed discussion of the definition of genetic information, see CRS Report RL33903, *Genetic Discrimination: Overview of the Issue and Proposed Legislation*, by Amanda K. Sarata and Erin D. Williams.

<sup>2</sup> Nuffield Council on Bioethics. *Pharmacogenetics: ethical issues*. (2003), at [[http://www.nuffieldbioethics.org/fileLibrary/pdf/pharmacogenetics\\_report.pdf](http://www.nuffieldbioethics.org/fileLibrary/pdf/pharmacogenetics_report.pdf)].

Genetic information is viewed as powerful, perhaps uniquely so, by society and has been regarded with some degree of awe and respect not afforded other types of health or personal information.<sup>3</sup> This may be due to the sense that genetic information is immutable and, therefore, that its use merits more caution than might the use of other information. This heightened caution may be warranted simply because of the perception that genetic information cannot be modified or altered, and that it is not chosen by the individual who bears it. The differential regard for genetic information may also be related to the idea of genetic determinism, or the concept that our genes exclusively predict or foretell our fate, including health, behavior, and physical characteristics. Although efforts have been made by responsible scientists to disabuse the public of this notion by stressing the role of environmental factors in disease, it is still fairly persistent, as seen in overly glowing portrayals of genetic discoveries and technologies by the media. It is difficult to specifically determine what gave rise to the concept of genetic exceptionalism; however, it does appear that, broadly, the public regards genetic information as particularly sensitive and perhaps even as unique.

The influence of the concept of genetic exceptionalism is reflected in genetics-specific public policy at the state level and proposed legislation at the federal level. As of 2007, the National Conference of State Legislatures reports that 48 states and the District of Columbia have passed genetic nondiscrimination legislation that places various restrictions on health insurers' use, collection, or acquisition of genetic information to restrict or deny health insurance coverage. Similarly, a majority of states have passed legislation to protect the privacy of genetic information or to prohibit employers from using such information to discriminate against employees.<sup>4</sup> At the federal level, Congress has debated genetic nondiscrimination legislation in every Congress since the 104<sup>th</sup>. In the 109<sup>th</sup> Congress, the Senate passed the Genetic Information Nondiscrimination Act of 2005 (S. 306). The House did not bring its version to a vote (H.R. 1227). In the 110<sup>th</sup> Congress, the House passed the Genetic Information Nondiscrimination Act of 2007 (H.R. 493). However, the Senate has not yet voted on its version (S. 358).<sup>5</sup> Because the idea of genetic exceptionalism has had a significant influence on public policy to date, Congress may want to consider how to treat genetic information in future legislative efforts.

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<sup>3</sup> Roche PA and GJ Annas, "Protecting genetic privacy," *Nature Reviews Genetics*, 2:392-396, 2001.

<sup>4</sup> NCSL. Genetic Technologies Project, at [<http://www.ncsl.org/programs/health/genetics/charts.htm>].

<sup>5</sup> For more detail on S. 358 and H.R. 493, see CRS Report RL33988, *Genetic Nondiscrimination in Health Insurance: A Side-by-Side Comparison of the Title I Provisions in S. 358 and H.R. 493*, by C. Stephen Redhead, and CRS Report RL33987, *Genetic Nondiscrimination in Employment: A Comparison of Title II Provisions in S. 358 and H.R. 493, 110<sup>th</sup> Congress*, by Nancy Lee Jones. Genetic discrimination in health insurance (and employment) are discussed in more detail in CRS Report RL33903, *Genetic Discrimination: Overview of the Issue and Proposed Legislation*, by Amanda K. Sarata and Erin D. Williams, and CRS Report RL30006, *Genetic Information: Legal Issues Relating to Discrimination and Privacy*, by Nancy Lee Jones and Amanda K. Sarata.

This report provides an overview of the implications of genetic information for individuals, family, and society; examines public opinion about genetic information; and summarizes the main policy arguments for and against incorporating genetic exceptionalism into public policy.

## **Genetic Information vs. Other Medical Information**

The differences between genetic information and other types of medical information have important implications for the individual, for families, and for society as a whole.<sup>6</sup> The discussion below provides a broad overview of these implications, but recognizes that there may be specific examples of nongenetic tests that have the same implications as genetic tests. However, the discussion below is meant to provide a general comparison across the two categories of tests — genetic versus nongenetic — and the information they generate.

### **Implications for Individuals**

Genetic tests are similar to many other medical tests in that they are often used to diagnose a disease or condition. Most of these diagnostic tests, both genetic and nongenetic, provide information about treatable maladies. Unlike most other medical tests, however, genetic tests may be used to determine that a currently healthy individual is susceptible to — at increased risk of developing — a particular illness or condition (susceptibility genetic testing). Genetic tests may also be used to predict that a currently healthy person will develop an illness or condition in the future with certainty (predictive genetic testing). It is often the case that many of the illnesses and conditions tested for using predictive or susceptibility testing cannot be cured or prevented by any currently available medical intervention. For example, a genetic test is available for Huntington's disease, a disease for which no prevention or cure (or even effective treatment) currently exists.

Because of this general lack of curative measures, sometimes the only options for reducing one's risk of future disease rely on modification of personal health related behaviors; for example, undergoing frequent screening, eating healthy foods, and exercising regularly. It is unclear how knowledge of genetic risk factors will influence the modification of such behaviors. While some individuals may modify their behavior because of knowledge of their genetic risk factors, some may have the perception that because they cannot change their genetic makeup, modifying their behavior will have little effect on decreasing their risk for disease. It is possible that these individuals may even increase unhealthy behaviors out of a sense of hopelessness. If no preventive or curative options exist, predictive genetic test results may increase a patient's anxiety level if they are positive or provide a false sense of reassurance if they are negative. Susceptibility genetic test results may also increase anxiety if they indicate an increased risk of disease while offering no options for mitigating this risk. For example, a certain allele, the APOE4 allele, has been found to increase an individual's risk of developing Alzheimer's disease. However, testing

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<sup>6</sup> For more information about genetic testing, see CRS Report RL33832, *Genetic Testing: Scientific Background for Policymakers*, by Amanda K. Sarata.

for this particular allele has been discouraged by health professionals because Alzheimer's disease cannot be prevented or cured, and, therefore, knowing one's risk status may simply increase anxiety. Furthermore, the actual increase in risk is not absolute in any sense; one may carry two copies of the APOE4 allele and still not develop Alzheimer's disease, while one may carry no copies of the APOE4 allele and still develop the disease.

Unlike most other medical testing, genetic testing may unintentionally reveal information about an individual or may reveal information with unknown significance given current scientific knowledge. For example, genetic testing may reveal unanticipated instances of non-paternity (where it is revealed that the purported father is in fact not the father). The development of microarray technology provides an increasingly cost-effective mechanism for testing hundreds or even thousands of genetic health indicators at one time.<sup>7</sup> Genetic testing on this scale could likely lead not only to the revelation of an enormous amount of health information about individuals, but also to a large amount of unexpected health information. This raises concerns about informed consent, as obtaining truly voluntary informed consent for each individual test might be prohibitively time consuming. Banking and storage of this vast amount of information, along with personal identifiers, provides unique challenges to existing privacy protections. Consumer advocates caution that although economic incentives favor testing for a large number of genetic characteristics at one time, testing should be limited to allow for thorough informed consent before and genetic counseling both prior to and after testing.

Genetic information can be used not only in the clinical setting, but also by forensic experts and the criminal justice system to identify individuals with a high degree of certainty. Genetic information has been used to help convict and exonerate individuals accused of committing crimes. It has been used to help identify the remains of war and accident victims. Because of these potential applications, tissue samples and certain types of genetic information collected for health care purposes may later be used in law enforcement, and vice versa. The fact that genetic information collected for health care may have forensic applications may increase lawmakers' concern with the increasingly widespread availability and use of genetic information. Concern about use of genetic information in nonhealth settings may also decrease uptake of new genetic technologies in health care and decrease participation in genetic research.

## Implications for the Family

Genetic tests for *acquired* markers — genetic changes that an individual does not receive from his or her parents but rather acquires sometime post-conception —

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<sup>7</sup> Microarray technology is defined as “a new way of studying how large numbers of genes interact with each other and how a cell's regulatory networks control vast batteries of genes simultaneously.” Accessed at [<http://www.genome.gov/glossary.cfm?key=microarray%20technology>].

have direct medical implications only for the individuals taking the tests.<sup>8</sup> Most other medical tests also have direct medical implications only for the individual taking the test. However, unlike most other medical tests, genetic tests for *inherited* markers — markers that are transmitted from parents to their offspring — have implications for biological relatives of the individual. Parents, siblings, and children have a predictable risk of sharing certain inherited genetic markers. Furthermore, the generally unchanging nature of inherited DNA means that genetic testing for inherited markers can be performed at any stage of human development on most tissue types. This offers the possibility of testing before implantation or prenatally, and may possibly allow for fetal intervention.

Some genetic tests cannot be interpreted for an individual unless other members of her family are also tested. For example, breast cancer genetic testing on the BRCA1 and BRCA2 genes relies on sequencing the entire gene in an affected individual to pinpoint the specific familial mutation. Then other family members may be tested for that specific mutation to help accurately determine if they are at increased risk of developing breast and/or ovarian cancer and, if so, the magnitude of that risk. In those cases, all family members may or may not want to know their test results. Sometimes testing family members can also provide unwelcome information about sensitive issues such as paternity.

## Implications for Society

Unlike most other medical information, genetic information challenges traditional medical perceptions of health and disease, what is normal or abnormal, and raises questions about what constitutes manifested disease. For example, lactose intolerance was long thought to be an abnormal state. Research in different populations elucidated that, in fact, it is the normal state, and the ability to digest lactose was the result of mutations that were under strong selective pressure in areas of the world where domestication of animals led to increased consumption of dairy products. These concepts are becoming increasingly blurred as genetic advances uncover the true range of “normal” and “abnormal” in many human traits.

Many experts predict that genetics may soon provide the opportunity to test large groups of people for disease susceptibilities. However, fear caused by the possibility that a job may be lost or insurance may be cancelled — whether real or perceived — affects individuals’ decisions regarding seeking potentially beneficial genetic testing in clinical or research settings. If people decline to participate in genetic research in large numbers, the ability of scientists to gather the data that are necessary to determine the utility or ultimately the value of genetic testing may be compromised. This may delay realization of the potential benefits of genetic advances for the larger population.

Genetic information may be used in order to link a person to a distinct group or heritage. This type of information may have consequences, particularly for

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<sup>8</sup> For example, the *oncotype Dx* test offered by Genomic Health profiles a tumor’s gene expression patterns to determine whether the tumor is likely to spread or respond to different types of treatment. Expression patterns in tumors are largely due to acquired mutations.



individuals who have rights based upon heritage, such as Native Americans. Advocates of these groups are wary of the potential effect that genetic testing and information could have on their current rights and protections. Others argue that definitions of race and heredity are irrelevant or that genetic information will not supplant current nongenetic determinants of ancestry. Finally, because the prevalence of disease-causing genes often varies by ethnicity, there is potential for discrimination against groups of people that share similar genetic information.

The meaning of genetic information evolves with scientific progress, which has special implications both for privacy and for equity. In the privacy arena, a person could grant others access to genetic data that is benign by today's standards. However, with scientific advancements these same data may in time reveal more sensitive information. In the equity arena, those with health conditions for which genetic tests currently exist may suffer discrimination in the near future, while those with similarly debilitating health conditions for which there is no genetic test may escape the same discrimination simply by virtue of the current state of science. Certain initiatives in genetic research, such as large-scale banking of genetic information, could raise both privacy and equity concerns, even though they may also have widespread public health benefit — such as facilitating characterization of genetic markers associated with rare adverse drug events.

## Public Opinion About Genetic Information

Although there has been an ongoing academic debate about the concept of genetic exceptionalism, it is perhaps more relevant in terms of public policy to examine how the public views genetic information. Several studies have been undertaken in an attempt to elucidate whether the public views genetic information as inherently different from other information, whether it deserves special protections, and who should have access to it.

To address the question of whether genetic information merits special protections, a 2007 study examined opinions about genetic information through a series of focus groups with members of a health maintenance organization (n=46). Focus groups conducted with these members found that, broadly, participants did not feel that genetic information is special or inherently different from other medical information. In addition, the participants generally felt that all sensitive medical information, including but not limited to genetic information, should be protected. Sensitive medical information was agreed to be that information that has the potential to be used to discriminate against or stigmatize an individual.<sup>9</sup>

A second study compared the experiences, attitudes, and beliefs of persons with genetic conditions (cystic fibrosis and sickle cell disease) to those with other serious medical conditions (diabetes, HIV, breast cancer, and colon cancer) and to persons at risk for developing a disease (breast or colon cancer) due to strong family history.

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<sup>9</sup> Diergaard B. et al., "Genetic Information: Special or Not? Responses From Focus Groups With Members of a Health Maintenance Organization," *American Journal of Genetics Part A*, 143A: 564-569, 2007.

The authors found that in most instances, patients felt strongly that their health information needed to be protected regardless of whether it was genetic. In fact, respondents indicated that information about non-genetic stigmatizing conditions — such as abortion history, mental health history, drug and alcohol history, HIV status, and sexually transmitted disease — needed special protection. Based on their findings, the authors concluded that separate privacy policies for genetic and non-genetic health information would be unwarranted.<sup>10</sup>

Other studies and public opinion polls suggest that patients and members of the community desire and may benefit from additional protections for their genetic information. A 2003 study of 470 people with a family history of colorectal cancer showed that 39% rated their level of concern about genetic discrimination in health insurance as the most significant reason to forego testing. Those individuals with high levels of concern indicated that they would be significantly less likely to consider meeting with a health care professional to discuss genetic testing, or to undergo testing.<sup>11</sup> A 2007 survey by the Center for Genetics and Public Policy found that 93% of survey respondents thought employers should not have access to their genetic test results, and 93% opposed letting insurance companies have access to results.<sup>12</sup> A 2006 survey conducted by Cogent Research found that 72% of survey respondents believe that laws are needed to protect the privacy of genetic information, and 85% believe that if protections are not established, employers would use genetic information discriminate.<sup>13</sup>

Findings from these selected studies suggest that people view genetic information as particularly sensitive, but not more so than other sensitive medical information such as HIV status. These studies also found that people believe that genetic information needs to be kept private from employers or insurers, and that special protections are required to achieve this goal.

## Policy Issues

When considering public policy to legislate the treatment of genetic information, Congress may want to address several key issues, such as defining genetic information; physically separating genetic information from other medical information; unintended disparities between “genetic” and “nongenetic” disease; and the effect of legislation on participation in genetic research, on uptake of genetic

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<sup>10</sup> Plantinga L. et al., “Disclosure, Confidentiality, and Families: Experiences and Attitudes of Those with Genetic Versus Nongenetic Medical Conditions,” *American Journal of Medicine*, General Part C, vol. 119C (2003), pp. 51-59.

<sup>11</sup> Hadley D.W. et al., “Genetic counseling and testing in families with hereditary nonpolyposis colorectal cancer,” *Arch Intern Med*, 163(5): 573-82, 2003.

<sup>12</sup> Williams S., “The Impact of Genetic Discrimination,” *Genetics and Public Policy Center Issue Brief*, Updated April 2007.

<sup>13</sup> Presentation by Ms. Christy White at the Ninth Meeting of the Secretary’s Advisory Committee on Genetics, Health, and Society. March 28, 2006. Accessed at [<http://www4.od.nih.gov/oba/SACGHS/meetings/March2006/White.pdf>].

technology, and on the delivery of high quality health care. Policy makers may take an exceptionalist approach, meaning they may chose to treat genetic information differently from other medical information. For example, H.R. 493/S. 358 (110<sup>th</sup>), the Genetic Information Nondiscrimination Act of 2007, takes this approach. Conversely, policy makers may wish to take a non-exceptionalist approach, meaning they may wish to treat genetic information the same way as they treat other medical information. For example, the HIPAA Privacy Rule treats genetic information in a non-exceptional way. Some of the policy issues Congress may want to consider are outlined below.

If a genetic exceptionalist approach to public policy at the federal level were to be employed, several policy considerations may be raised. Experts highlight the challenge of effectively defining genetic versus nongenetic information for public policy purposes.<sup>14</sup> All disease is now thought to have a genetic component; hence, the term genetic disease is slowly growing more obsolete. Nearly all medical information might arguably be defined as being in part genetic in nature, creating challenges for making such a distinction.

In addition, physically separating genetic from non-genetic information in medical records may pose a real practical challenge. Requirements regarding the physical separation of different types of medical information may be so complicated and costly as to actually interfere with the delivery of effective and high-quality care. These requirements may further frustrate attempts by physicians to provide optimal care to their patients, and may impede the sharing of relevant medical information. Importantly, adoption of electronic medical records may facilitate such a separation (both for purposes of maintaining the information and releasing the information).<sup>15</sup>

Several experts have voiced concern that genetic exceptionalist laws may result in unintended disparities.<sup>16</sup> For example, they argue that such laws may create a situation where individuals affected by a disease or condition with a *known* genetic basis (i.e., breast cancer in the presence of a BRCA1 mutation) would be protected from discrimination while those with breast cancer with no *known* genetic basis would not be protected.

Experts often assert that without genetic-specific legislation in place, individuals would not utilize and, therefore, may lose out on the clinical and other benefits of new genetic technologies out of concern about misuse of their genetic information. In addition, these experts maintain that without protections in place, genetic research will be stifled.<sup>17</sup> It is possible, however, that genetic exceptionalist laws might reinforce perceptions among the public that genetic information is special, unique,

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<sup>14</sup> Rothstein, M.A., "Genetic Exceptionalism and Legislative Pragmatism," *The Journal of Law, Medicine, and Ethics*, 35(2):59-65, 2007.

<sup>15</sup> *Ibid.*

<sup>16</sup> Diergaarde B. et al., "Genetic Information: Special or Not? Responses From Focus Groups With Members of a Health Maintenance Organization," *Am J Genet Part A*, 143A: 564-569, 2007.

<sup>17</sup> Slaughter L.M., "Your Genes and Privacy," *Science*, 316(5826): 797, 2007.

or deterministic. Although many argue that protections are needed to encourage participation in genetic research and the uptake of genetic medical technologies and tests, it is possible that reinforcing the notion that genetic information is uniquely powerful (i.e., has such high potential for misuse that it requires its own special protections) might have the unintended consequence of decreasing participation in genetic research or decreasing utilization of important genetic tests that could guide treatment. This is especially a concern if genetic exceptionalist legislation were to be passed that is perceived by the public to be weak or lacking thoroughness in the protection it offers.

In an environment where special protections are not extended to genetic information, patients may be hesitant to share relevant genetic information because of concerns about possible misuse, and health care providers may have difficulty providing the best care to their patients.<sup>18</sup> With a non-exceptionalist policy approach, it may be challenging to adequately safeguard genetic information. A broader law may be more unwieldy and, therefore, may provide challenges to providing the appropriate protections for medical information with varying degrees of sensitivity. As there is some evidence that the public views genetic information as deserving of special protections, if not of being special itself, treating genetic information the same as other medical information may not resonate with the public (see “Public Opinion about Genetic Information” above).

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<sup>18</sup> Hadley D.W. et al., “Genetic counseling and testing in families with hereditary nonpolyposis colorectal cancer,” *Arch Intern Med*, 163(5): 573-82, 2003.