

Genetic Testing and Screening in the Age of Genomic Medicine

Executive Summary

Genes and Chromosomes

- Genes are the blueprints of heredity. Genes are made of hundreds to thousands of DNA bases.
- Each gene directs cells to produce one or more specific proteins, including enzymes and structural proteins.
- The human genome is the complete set of genes that every person inherits from his or her parents. It is present in virtually every cell of the body.
- The human genome consists of tens of thousands of pairs of genes. Each person inherits one copy of each gene from each parent.
- Genes are organized along string-like structures called chromosomes. Each individual inherits two sets of twenty-three chromosomes, one from each parent: two sets of twenty-two autosomes and one set of sex chromosomes (X, X or X, Y).

Genetic Variations, Mutations, and Human Disease

- The DNA base sequence of human genes is about 99.9 percent identical among individuals. About 1 of every 1,000 DNA bases varies among individuals, accounting for inherited differences in traits and disease susceptibility.
- Changes in a DNA base sequence, called mutations, account for inherited gene variations. Mutations may be harmful if they prevent a gene from making a normal copy of its specific protein. These mutations can cause, or increase susceptibility to, specific diseases.
- Single-gene diseases are relatively rare diseases that result when a person inherits one gene with a harmful mutation or a pair of genes in which each has a harmful mutation. Inheritance of these mutated genes generally results in a 100 percent chance of developing a specific disease. Single-gene diseases include autosomal dominant diseases (e.g., Huntington disease), autosomal recessive diseases (e.g., sickle cell disease), and X-linked diseases (e.g., Duchenne muscular dystrophy).
- Most diseases result from a complex set of both genetic and environmental causes. Inheritance of some harmful gene mutations increases the chance, although it does not ensure, that a person will develop a specific disease. These mutations are called inherited susceptibility mutations.

The Human Genome Project and Health Care

- The Human Genome Project, an international research project, has now deciphered the more than three billion DNA letters of the human genome. Follow-up research is expected to discover the structure and function of thousands of new genes.
- Genetics research will lead to the development of new predictive and diagnostic genetic tests. It also will lead to the development of new preventive and treatment interventions. Generally, the development of interventions lags years, even decades, behind gene discovery and genetic test development.

Genetic Testing

- Genetic testing for inherited genetic variants is performed for several purposes: diagnosis of individuals with symptoms, determination of future disease risks in asymptomatic individuals, determination of genetic risks for progeny, guidance of medical treatment, research, and individual identification.
- Genetic testing for inherited genetic disease risks is an analysis of DNA, chromosomes, or gene products to provide specific information about variations in the number or form of genes or chromosomes in an individual or his or her progeny.
- Genetic information is information about specific variations in genes or chromosomes learned by genetic testing or by other means.
- DNA-based testing directly analyzes the DNA base sequence of a gene.
- Phenotypic testing identifies specific inherited gene variations indirectly, by detecting specific variations in the structure of a protein encoded by a gene or variations in a protein's enzyme activity.
- Karyotype analysis and fluorescent in situ hybridization analysis detect variation in form or number of chromosomes.
- New testing technologies that will promote genetic testing in health care include DNA chip technology and tandem mass spectrometry.

Assessing the Accuracy and Usefulness of Genetic Tests

- Analytical validity of a genetic laboratory test is a measure of how well the test detects what it is designed to detect. It encompasses analytical sensitivity (the probability that the test will detect a gene variant it is designed to detect when present in a sample) and analytical specificity (the probability that the test will be negative when a specific variant tested for is not present in a sample).
- Clinical validity measures the extent to which an analytically valid test result can diagnose a disease or predict future disease. For predictive genetic tests, it includes positive predictive value (the ability to predict that an individual will develop a disease) and negative predictive value (the ability to predict that an individual will not develop a disease).
- For DNA-based testing, clinical validity is limited by genetic heterogeneity and incomplete penetrance. Genetic heterogeneity means that different mutations in a specific gene, or mutations in different genes, are associated with the same disease. Incomplete penetrance means that within a population, not everyone who tests positive for a specific gene mutation will develop the associated disorder.
- Utility of a test is a measure of how useful test results are to the person tested. Clinical utility is a measure of how a test may guide clinical decisions. In some circumstances, predictive genetic testing may not provide medical preventive or treatment options but may help reduce anxiety and/or aid planning for the future.

Predictive Genetic Testing to Assess Reproductive Risks

- Reproductive genetic tests detect heritable genetic variations that are associated with disease. This type of testing includes carrier testing, prenatal testing of fetal cells, and pre-implantation testing of embryos formed by in vitro fertilization.
- Reproductive genetic tests generally are offered to individuals and couples who are at increased genetic risk for a specific disorder based on family history or membership in a racial or ethnic group that has identified genetic variants that increase risk for a specific disease.

- Carrier testing generally is performed to determine the risk of a healthy individual or couple of having a child with a recessive disorder. It may be performed before or after conception.
- Prenatal testing of fetal cells includes amniocentesis and chorionic villus sampling.
- Pre-implantation testing of embryos formed by in vitro fertilization is performed using single cells removed from individual embryos to detect specific gene mutations or chromosomal anomalies.

Predictive Genetic Testing to Assess Future Disease Risks in Healthy Adults

- Presymptomatic genetic testing is predictive testing of apparently healthy adults to determine whether they are at risk for a single-gene disorder. These disorders occur with virtually 100 percent incidence in persons who have inherited a specific gene mutation.
- Susceptibility (predispositional) testing is predictive genetic testing of apparently healthy adults to determine whether they are at increased risk, relative to the general population, for a specific future disease. A positive test result (finding a mutation) does not necessarily mean that a person will develop a future disease.
- For susceptibility testing, establishing a test's clinical predictive value may require years of research.
- Pharmacogenetic testing is genetic testing of individuals to guide their pharmaceutical or other medical treatment. Pharmacogenetic testing seeks to promote a favorable response and to prevent an adverse response to a drug or other treatment based on genetic predisposition.

Misunderstandings of and Misperceptions about Genetics

- Throughout human history, people have understood that physical and behavioral traits have a genetic component. Proponents of a centuries-long debate, referred to as the "nature-nurture" debate, have disagreed about the relative contribution of genetic and environmental factors to human behavioral and cognitive (intelligence) traits.
- Scientific evidence, including evidence from molecular genetics research, shows that genes may influence complex behavioral and cognitive traits and mental illness. Generally, however, behavioral and cognitive traits and mental illnesses result from a complex and cumulative interplay of many genetic and environmental factors.
- Many commentators express concern that the general public and the popular press overemphasize the role of genetic inheritance for health as well as for the development of behavioral and cognitive characteristics, taking an overly deterministic view of genetics. These views may lead people to overestimate the meaning of genetic testing and to misconstrue genetic test results.
- Throughout the past century, some countries, including the United States, have promoted and/or endorsed eugenic policies that aim to promote the births of certain types of individuals and to discourage births of other types of individuals.
- In the first half of the twentieth century, eugenic attitudes contributed to the passage of federal legislation to limit immigration of people into the United States based on country of origin. Eugenic attitudes also were a cause of federal and state court decisions and state legislation that prevented or discouraged marriages of people from different racial groups and promoted involuntary sterilization of individuals who were deemed "unfit." Included among the "unfit" were the mentally ill, the "feebleminded," and habitual criminals.
- "Genetic exceptionalism" is the belief that medical genetics is sufficiently different from other areas of medicine to warrant special protections. Commentators disagree on whether genetic testing should be treated differently from other forms of medical testing.

- Genetic testing shares characteristics with other forms of medical testing. However, some forms of predictive genetic testing, notably DNA-based testing of inherited genetic variants, differ from other medical testing in important ways. For example, predictive DNA-based genetic testing has exceptionally long-range predictive powers; it can predict disease, or increased risk for disease, in the absence of clinical signs or symptoms; it reveals the sharing of genetic variants within families at precise and calculable rates; and, at least theoretically, it has the potential to generate a unique identifier profile for individuals.

Genetic Screening for Adult Health and Reproductive Risks

- Genetic screening differs from genetic testing in that it targets populations rather than at-risk individuals. Genetic screening generally is performed to detect future disease risks in individuals or their progeny for which established preventive interventions exist. Examples of genetic screening include newborn screening for phenylketonuria, carrier screening for sickle cell disease, and prenatal screening of fetal cells to detect chromosomal or other congenital abnormalities.
- Although predictive genetic screening to detect future disease risks in adults has not yet been offered, commentators predict that predictive genetic screening for hemochromatosis and other adult-onset diseases will be offered in the next decade.
- Some commentators express concern that if screening tests become routine practice, individuals may be pressured to undergo testing that they would not choose to undergo in a different context. Commentators also express concern about possible discrimination and/or stigmatization against individuals and groups who are the subjects of genetic screening because of their racial, ethnic, or geographic origin.
- Commentators maintain that a number of factors should be evaluated in determining whether a particular screening test should be implemented, including the purpose of the screening test, the test's analytical validity, clinical validity, and clinical utility, and the cost of the screening test.
- Multiplex genetic testing is genetic testing for two or more completely different conditions in a single testing session. Some commentators oppose multiplex genetic testing if it is performed only because it is technologically possible. Other commentators maintain that multiplex genetic testing is generally inappropriate unless the tests provide clear and useful options to the persons being tested. A key issue for both reproductive and late-onset multiplex testing is how to bundle tests together to allow for appropriate pretest education, counseling, and consent.

Conclusions and Recommendations of the Task Force

Purpose of Predictive Genetic Screening

- The purpose of predictive genetic screening should be to benefit the individual or couple tested. Screening tests offered to healthy individuals who do not perceive themselves or their offspring to be at increased risk for disease based on family and/or personal history should provide clear medical benefits or expanded reproductive options.

Predictive Value of Screening Tests

- Predictive genetic screening tests should have a sufficient level of confirmed predictive value in healthy populations to justify their use for individuals who are not known to be at increased disease risk.

How to Offer Predictive Genetic Screening Test

- Predictive genetic screening tests should be voluntary and should be offered only when accompanied by adequate education, counseling, informed consent, test follow-up, and efforts to ensure confidentiality.

Special Concerns about Offering Genetic Screening to Determine Risks for Late-Onset Disorders

- Genetic screening tests to determine future risk for late-onset disorders should have confirmed clinical utility, and screening should be offered on an age-appropriate basis to ensure maximum medical benefit and minimal risks.

Special Concerns about Offering Genetic Screening to Determine Reproductive Risks

- Genetic screening tests to predict reproductive risks should provide individuals and couples with useful options. Providers should make clear that despite the routine offering of tests, some individuals may wish to decline if they think that the test will not be useful to them. Providers should offer screening tests in a timely manner to maximize the reproductive options of tested individuals.

Federal and State Governments Should Not Require Genetic Screening by Law

- It is generally inappropriate for federal or state governments to mandate population genetic screening. New York State should repeal legislation that mandates sickle cell carrier screening for some couples seeking a marriage license.

Role of Study Panels and Professional Guidelines

- Study panels that include national experts, community representatives, and others, as well as professional medical societies such as the American College of Obstetricians and Gynecologists and the American College of Medical Genetics, should determine the appropriateness of offering specific genetic screening tests based on the test's validity and utility. For reproductive screening tests, for which follow-up options may include decisions about pregnancy termination, professional guidelines should consider the seriousness of the disorder tested for, its penetrance, its age of onset, and the variability of disease symptoms.

Genetic Screening of Minors

- Generally, minors should not be offered genetic screening tests to determine future health or reproductive risks, unless screening provides a clear and timely medical benefit and has minimal psychosocial risks.

Multiplex Genetic Testing Panels

- Genetic tests that provide information about future risks for unrelated disorders should be included in multiplex testing panels only when they meet all criteria for genetic screening tests. Tests should be grouped based on similar issues and implications to allow for adequate counseling and consent. For tests to determine risks for late-onset diseases, tests placed in multiplex panels should provide a demonstrated, significant medical benefit and should be offered on an age-appropriate basis. For reproductive carrier testing, tests placed on a

multiplex panel should be for diseases of similar seriousness.

Newborn Screening

- Newborn screening is the most widely performed type of genetic testing in the United States today. Newborn screening programs exist in all fifty states, the District of Columbia, Puerto Rico, and the Virgin Islands. The goal of newborn screening is to detect infants affected by conditions for which prompt application of confirmed interventions can prevent or reduce disease, disability, and/or death.
- As a result of the Human Genome Project, the discovery of the genetic variations that underlie inherited disorders and the technology to detect them are expanding rapidly. These developments will present state screening programs with new testing methods and expanded lists of disorders for which testing is possible.
- Most states, including New York, have added tests to their newborn screening panels without formal criteria or processes to guide them. Many commentators recommend that newborn screening programs form advisory committees composed of medical and laboratory professionals and community participants to establish criteria for screening tests and to review screening test panels and program outcomes.
- Most states, including New York, mandate newborn screening and do not require parental consent. New York and other states exempt from newborn screening children whose parents have religious objections to it. Commentators disagree over whether parental consent to newborn screening should be required.
- Some benefits of newborn screening are reduced morbidity and mortality of children and cost savings to society through early prevention and treatment of childhood disease. Some of the risks of newborn screening include parental anxiety about false positive results; harm that can be caused to the parent-child relationship by parental misperceptions about the meaning of a child's carrier status; and the possibility that children will be subjected to needless, and potentially risky, medical interventions or monitoring.
- Most newborn screening programs, including New York's program, store residual newborn blood samples (bloodspots) and use them for research. Some commentators maintain that it is appropriate to use residual screening samples for research if the samples are anonymized. Others contend that ethical concerns about the use of residual newborn blood samples may be greater than for other tissue samples obtained in the clinical context because the collection of newborn screening samples is mandated by law. Commentators also have discussed the appropriate research uses of identified and coded newborn samples and whether parental consent for and/or notification about the research use of residual newborn screening samples should be required.

Conclusions and Recommendations of the Task Force

Basic Requirements for Newborn Screening Tests

- New York's Newborn Screening Program panel should be restricted to tests that detect congenital disorders characterized by serious and irreparable harm that can be avoided or minimized only by prompt application of confirmed medical interventions. The analytical and clinical validity of the screening tests also must be confirmed.

Statutory Authorization for New York's Newborn Screening Program

- New York Public Health Law § 2500-a should be amended to delete the names of individual disorders screened for by the Newborn Screening Program. The law should designate the Commissioner of Health to specify in regulations those congenital disorders for which screening should be performed.

Informing Parents about Newborn Screening

- The Commissioner of Health should promulgate regulations to require the Newborn Screening Program to provide educational materials about screening to prenatal care providers, as well as to hospitals and institutions of birth. Prenatal care providers should be required to provide and be available to discuss these materials during the course of prenatal visits. Program materials should be multilingual and at appropriate reading levels for a general audience. They should explain the purpose of screening and provide a description of the disorders screened for, their population incidence, and the follow-up process for infants with a positive screen test result.

Mandatory Newborn Screening

- New York's Newborn Screening Program should be mandatory for all infants born within the state, provided that several conditions are met: (1) all screening tests must meet the criteria described above in the recommendation concerning the basic requirements for newborn screening tests; (2) parents must be informed and receive educational materials about the program, its goals, and the screening process; and (3) the state must ensure that newborns identified as positive in screening tests are promptly diagnosed and that identified newborns and their families have access to follow-up medical care and counseling related to the disorder, regardless of their ability to pay. New York Public Health Law § 2500-a should be amended to remove the right of parents to assert religious objections to screening.

Follow-up Evaluation and Diagnosis of Screen-Positive Newborns

- The Newborn Screening Program should ensure that follow-up testing and diagnostic evaluation of newborns who test positive on a screening test is rapid and readily accessible, to maximize treatment benefits for affected newborns and to minimize potential anxiety associated with an initial false positive test result.

Follow-up Medical Care for Newborns of Confirmed Positive Newborns

- New York State should ensure that newborns detected to have a congenital condition by newborn screening receive necessary long-term medical and preventive care, into and through adulthood, regardless of ability to pay. The Newborn Screening Program should facilitate efforts to ensure that affected newborns identified by the program obtain necessary and appropriate medical care. The program should assist treatment centers in locating and treating children who are lost to follow-up.

Establishment of a Newborn Screening Advisory Committee

- New York's public health regulations should establish a newborn screening advisory committee to act in an advisory capacity to the Commissioner of Health and the Newborn Screening Program. The committee should include outside professional and community representatives and should be independent from the screening program. It should meet at least annually to consider new screening tests, solicit community input, and evaluate program infrastructure, policies, and outcomes.

Review and Implementation of Newborn Screening Tests

- A newborn screening advisory committee, and ad hoc specialty subcommittees established by it, should review all tests currently on or under review for New York's screening panel, as well as potentially valuable new tests, and make recommendations to the Commissioner. For tests for which a confirmed medical benefit has not been sufficiently demonstrated, tests should be viewed as human subject research and should require parental informed consent. These tests should be subject to review by an institutional review board to determine the information that should be provided as part of obtaining parental informed consent. All new screening tests should be subject to periodic follow-up evaluation to determine test accuracy and effectiveness of medical interventions.

Universal Performance of Newborn Screening

- Newborn screening tests should be performed for all newborns, rather than targeted to specific minority populations perceived to be at higher-than-average risk for a particular disorder.

Financing of the Newborn Screening Program

- A permanent, stable funding source is needed to enable the program to consider additional tests, implement new tests as needed, consider changes in testing technologies, improve processes and follow-up evaluation, and support the activities of the advisory committee.

Research Use of Anonymized Newborn Bloodspots

- The Newborn Screening Program, consistent with the recommendations in Chapter 7 concerning research use of samples obtained in the clinical context, should permit the use of anonymized samples for research. The program should inform parents that residual bloodspots may be anonymized and used for quality assurance activities or research. Parents should be informed of the potential research value of the samples and of the impossibility of linking research results to any individual newborn.

Research Use of Identified Newborn Bloodspots

- Research use of identified newborn bloodspots should be permitted in accord with recommendations in Chapter 7 concerning the research use of identified samples obtained in the clinical context. In addition, investigators who seek to use identified newborn blood samples for research should demonstrate why unidentified samples or alternate sample sources would not suffice. The use of identified samples should require recontact by the New York State Department of Health and informed consent of parents for each research use. The New York State Department of Health should not release samples that retain identifying data to researchers outside the department except for rare circumstances in which the research is directly relevant to the health of a specific newborn.

Research Use of Coded Newborn Bloodspots

- Research use of coded newborn bloodspots should be permitted in accord with recommendations in Chapter 7 concerning the research use of coded samples obtained in the clinical context. The use of coded samples should require recontact by the New York State Department of Health to obtain the consent of parents for the future research use of the samples.

Policies for Storage of Newborn Bloodspots

- The Newborn Screening Program should establish a formal policy for the storage of residual identified and anonymized bloodspots. The policy should specify potential uses for stored bloodspots and a maximum period of time for which samples may be maintained with personal identifiers.

Notification of Parents of Newborn Carrier Status

- When carrier status for a recessive genetic disease is determined as an incidental finding of a newborn screening test, New York's Newborn Screening Program should report that finding to the authorized physician. Ideally, parents of carrier newborns should be informed of that result and offered appropriate education, counseling, and testing by appropriately trained and credentialed professionals.

Informed Consent

- Informed consent to a medical procedure is an agreement to allow a medical procedure to go forward after having been advised of relevant facts necessary to make that agreement an intelligent one. Relevant facts include the patient's diagnosis, the nature and purpose of the proposed procedure, and the risks and benefits of, and the alternatives to, the procedure.
- Obtaining a patient's informed consent to medical procedures is both a legal necessity and a basic requirement of medical ethics, and most commentators maintain that the requirement of informed consent applies to decisions about predictive genetic testing. Some of the issues that commentators recommend that health care providers should discuss with their patients as part of obtaining informed consent to predictive genetic testing include: (1) the purpose of the test; (2) a description of the testing process; (3) the accuracy of the genetic test and the meaning of its results; (4) the risks and benefits of, and alternatives to, genetic testing; and (5) confidentiality issues.
- It is unclear whether New York's general law on informed consent to medical procedures covers predictive genetic testing. However, in 1996 and 1997, New York enacted laws that require persons performing predictive genetic tests to obtain the individual's written informed consent prior to testing. The laws require the consent form to contain some, but not all, of the information commentators have recommended for informed consent to predictive genetic testing.
- Multiplex genetic testing is predictive genetic testing for more than one condition in a single testing session. Some commentators argue that health care providers should obtain full informed consent from patients for each test in a multiplex testing panel. Others contend that a patient's generic consent to all of the tests in the panel would be sufficient if the consent process highlights broad concepts and common-denominator issues for all of the tests.
- Commentators disagree about the proper method for obtaining informed consent to predictive genetic tests for gene variants that have been identified as having multiple, seemingly unrelated health effects (pleiotropic genetic tests). One contends that health care providers have an obligation to disclose to patients the risks associated with learning information about all of the conditions detected by the tests and must provide counseling and other support services as required by testing protocols for each individual condition. Another maintains that outside of the reproductive genetic testing context and situations where there are "special concerns" about the psychological state of a patient to be tested, health care providers need only inform their patients about the different clinical uses of the test and need not provide any special counseling or support services.

- Stored tissue samples, which today number at about 282 million in the United States, are used by medical researchers as their principal source of human biological materials. These tissues are most commonly collected during clinical medical procedures, and many of the patients from whom they are collected are not informed that their tissues will be stored and used for research.
- In some circumstances, federal regulations governing research involving human subjects require researchers to obtain a subject's informed consent before performing research on the subject's identified or coded tissue samples removed in the clinical context. These regulations do not require informed consent if the tissue samples have been anonymized. New York's statutes concerning research involving human subjects specifically exempt tissues removed in the clinical context from the statutes' coverage.
- Although most commentators agree that researchers should obtain a subject's informed consent before performing research on the subject's identified tissue sample, commentators disagree about whether, or what type of, consent is necessary before researchers may perform research on a subject's coded or anonymized tissue samples.

Conclusions and Recommendations of the Task Force

Necessity of Informed Consent for Predictive Genetic Testing in the Clinical Context

- Predictive genetic testing should not be performed without the informed consent of the subject of the test, except in the limited circumstances described below.

Power of the Commissioner of Health

- Those sections of New York's genetic testing statutes that list specific elements of informed consent should be replaced with an authorization for the Commissioner of Health to issue regulations on the process and content of informed consent to predictive genetic testing.

Content of Informed Consent to Predictive Genetic Testing in the Clinical Context

- Assuming that New York law is amended to authorize the Commissioner of Health to regulate informed consent to predictive genetic testing, the Commissioner should require the following information to be provided to the patient before obtaining the patient's consent (elements currently not required by New York law are italicized):
 1. The purpose of the test
 2. A general description of the testing process
 3. A description of the diseases or conditions tested for, *including their ranges of severity*
 4. *The risks and benefits of, and alternatives to, the predictive genetic test*
 5. Confidentiality issues, *including confidentiality protections, the circumstances under which results of tests may be disclosed without the patient's consent, and the names of persons and/or organizations to whom the patient has consented to disclose the results*
 6. *Protections against adverse uses of genetic information*
 7. *The chances of false positive and false negative results*
 8. The meaning of both positive *and negative* results
 9. *The ability, or lack thereof, of the test to predict a disease's severity and age of onset*
 10. *The possibility that no additional risk information will be obtained at the completion of the test*

11. *Available medical surveillance, treatment, and/or reproductive options following testing*
12. A statement that, prior to providing informed consent to genetic testing and after receiving the results, the individual may wish to obtain professional genetic counseling
13. *The risks of transmitting the relevant mutation to children and that the mutation may be present in other blood relatives*
14. A statement that no tests other than those authorized will be performed on the biological sample and that the sample will be destroyed at the end of the testing process, or not more than a specific period of time after the sample was taken, unless the subject consents to a longer period of storage
15. *That the test is voluntary*
16. *An offer to answer inquiries*
17. *The fees charged for the laboratory tests and pre- and posttest counseling*

Sufficiency of Signed Informed Consent Form as Evidence of Informed Consent

- Assuming that New York law is amended to authorize the Commissioner of Health to regulate informed consent to predictive genetic testing, the Commissioner should require that health care providers disclose the information described above in a manner that will enable the patient to make a knowledgeable evaluation. A signed informed consent form is not necessarily sufficient evidence that this goal has been achieved.

Use of Decision Aids in the Informed Consent Process

- Health care providers are encouraged to use decision aids, such as written materials, videos, group discussions, and CD-ROMs, as part of the informed consent process to predictive genetic testing. However, health care providers should not use decision aids as a substitute for discussing predictive genetic testing issues with their patients.

Persons Required to Obtain Informed Consent

- Assuming that New York law is amended to authorize the Commissioner of Health to regulate informed consent to predictive genetic testing, the Commissioner should require that the person who orders a predictive genetic test has the obligation to ensure that the subject's informed consent is obtained.

Responsibility of Testing Laboratories

- The New York State Department of Health should permit clinical laboratories to perform predictive genetic tests on biological samples only if the laboratories receive assurances that the subjects provided informed consent for the tests.

Professional Guidelines on the Process and Content of Informed Consent for Predictive Genetic Tests

- Professional organizations should issue guidelines on the process and content of informed consent for specific predictive genetic tests and should create model consent forms that are consistent with existing law and contain the information necessary for patients to make informed decisions about undergoing predictive genetic testing.

Health Care Providers Qualified to Order Predictive Genetic Tests

- Health care providers should order predictive genetic tests only when (1) they know the circumstances under which it is appropriate to order them and the meaning of their results, (2) they are capable of providing their patients with sufficient information to make informed decisions about undergoing the tests, and (3) they are able to provide their patients with comprehensive pre- and posttest counseling or can refer their patients to professionals who are able to do so.

Informed Consent to Multiplex Genetic Testing

- Ideally, a patient's full informed consent should be obtained to each test on a multiplex panel prior to testing. However, assuming that New York law is amended to authorize the Commissioner of Health to regulate informed consent to predictive genetic testing, generic consent to multiplex testing should be permitted if (1) the number of tests on the panel is so high or the information about the tests is so complicated that attempting to obtain full informed consent from the patient to each test would be confusing or otherwise burdensome to the patient; (2) the tests on the panel meet all of the criteria described in Chapter 5 for inclusion in a multiplex panel; (3) the patients are informed, prior to testing, that more detailed information about each test is available; and (4) the patients are given an opportunity to obtain that information prior to testing either from the health care provider offering the multiplex panel or from another health care professional.

Special Issues Related to Pleiotropic Information

- Before offering a predictive genetic test to a patient, providers should give the patient all information necessary for the patient to provide informed consent to the intended use of the test, that is, information relevant to any condition about which the patient intends to receive test results. If the test also may reveal confirmed, clinically valid information about conditions for which the patient has not sought testing, the provider should inform the patient of this fact, specifying (1) the condition(s) about which the test may reveal information; (2) the consequences of having this additional information in his or her medical record; and (3) opportunities, including genetic counseling, for the patient to obtain further information about aspects of the test unrelated to its intended use. If the patient expresses an interest in learning how his or her test results relate to conditions for which testing was not originally sought, the provider should ensure that the patient provides informed consent to obtaining this additional information. Providers should respect patients' right not to learn pleiotropic information revealed by genetic tests.

Court Orders for Predictive Genetic Testing

- New York law should be amended to permit courts to order predictive genetic testing without the subject's consent only when (1) absent the testing, there would be a clear and imminent danger to the public health; (2) such testing is authorized by federal and/or New York State statutes or regulations; or (3) in a civil or criminal litigation, the subject affirmatively places his or her physical or mental condition at issue and the genetic testing directly relates to that physical or mental condition.

Remedies for the Performance of Genetic Testing without Informed Consent

- New York law should be amended to expressly authorize private lawsuits by subjects of unconsented-to predictive genetic tests against persons who order and/or perform the tests.

- New York law should be amended to authorize the Attorney General to bring lawsuits on behalf of individuals who have undergone predictive genetic testing without informed consent.
- Persons and organizations licensed by New York State should be subject to professional discipline and/or other sanctions, including fines and license suspension and revocation, for performing or ordering predictive genetic testing without informed consent.

Research on Tissue Samples Obtained in the Clinical Setting

- New York's law on the protection of human research subjects should be amended to cover research on tissue samples obtained in the clinical context. The amendment should apply only to tissue obtained after the amendment's effective date.
- Research on identified tissue samples obtained in the clinical context should be permitted only after the subjects have provided full informed consent to the research and an institutional review board has reviewed and approved the research protocol.
- Research on anonymized tissue samples obtained in the clinical context should be permitted only after an institutional review board has reviewed and approved the research protocol. The institutional review board review should ensure that the samples are or will be truly anonymized and should determine whether the research is of such a sensitive nature that it is inappropriate to use anonymized samples without having obtained the subjects' specific consent to the research.
- Research on coded tissue samples obtained in the clinical context should be permitted only if (1) the patients have agreed to the storage and research use of their coded samples; (2) the patients have been told about the operation, tissue release policies, and confidentiality protections of the tissue repository; and (3) an institutional review board has reviewed and approved the protocols for the research. The institutional review board review should ensure that the samples are or will be truly coded and should determine whether the research is of such a sensitive nature that it is inappropriate to use coded samples without having obtained the subjects' specific consent to the research. The coding of the samples should be performed by a person who is not connected to the research and who will not learn the individual results of the testing.
- Patients should be informed that their decision about whether to consent to the research use of their coded and/or identified tissue samples is wholly voluntary and that their decision will not affect their access to, or quality of, care.
- Institutions should encourage clinicians to ask patients to consider authorizing the use of their tissue for research purposes, and clinicians should do so when they deem it appropriate.

Predictive Genetic Testing of Children

- When susceptibility to a genetic disorder is discovered within a family, parents may seek predictive genetic testing of their children to obtain a medical benefit for the child, to reduce the child's future disease risk, and/or to make life planning decisions. Adolescents also may initiate requests for predictive genetic testing to determine future disease or reproductive risks.
- Parents generally have the legal authority to control their children's medical care, and children may generally not obtain medical care without their parents' consent.
- Benefits of testing children for late-onset disorders can include parental recognition of the need for clinical surveillance and/or preventive measures available for their asymptomatic children and enhanced parental ability to make life planning decisions for their children. Possible risks include the use of unconfirmed clinical interventions on the children that may be unnecessary and/or harmful, discrimination against the children, and psychological harms to the children

and family.

- There are generally no benefits to genetic carrier testing of minors, except when adolescents are contemplating marriage or having children in the near future. Risks of such testing include stigmatization, discrimination, and parental misunderstanding of the meaning of the test results.
- Most commentators contend that the primary determinant of whether a child should undergo genetic testing is the best interests of the child. In the absence of a clear medical benefit to the child, these commentators opine that avoidance of potential testing-associated harms and the preservation of the minor's future autonomy should be the overriding considerations. Accordingly, these commentators maintain that children generally should not undergo genetic testing for late-onset disorders in the absence of a medical benefit and should not undergo genetic carrier testing for recessive disorders.
- Most commentators agree that health care providers play an important role in assessing the benefits and risks of testing a child to the child and family.

Adoption

- New York mandates that adoption agencies disclose to prospective adoptive parents the "available" medical histories of the prospective adoptee and the child's biological parents. These histories must include all available information about diseases or conditions believed to be hereditary. New York law does not require parties to an adoption to exercise reasonable efforts to collect this information if it is not already available.
- Commentators stress that the best interests of the prospective adoptees should be the guiding principle in determining whether they should undergo genetic testing. Some commentators contend that prospective adoptees should undergo genetic testing only in situations where it would be appropriate to test other children.

Conclusions and Recommendations of the Task Force

Predictive Genetic Testing to Determine Adult-Onset Disease and Reproductive Risks

- The best interests of the child, including respect for the child's future autonomy, should be the primary consideration in decisions about predictive genetic testing of children. Predictive genetic testing of children is clearly appropriate when test results will provide information relevant to current decisions about the child's care, such as decisions to institute prophylactic treatment. Where the benefits to the child are less clear, however, predictive genetic testing should be approached with caution, given that testing can also lead to significant harms.

Predictive Genetic Testing to Determine Risks for Pediatric-Onset Disease

- When a healthy child is at risk for a pediatric-onset disorder, predictive genetic testing to confirm or allay disease risks may be in the best interests of the child, even if preventive or therapeutic interventions are not available.

The Role of Health Care Providers in Guiding Predictive Genetic Testing Decisions

- Health care providers play a critical role in guiding decisions about predictive genetic testing of children. When faced with a parent's request for predictive genetic testing of a healthy child or with a request initiated by a healthy adolescent, providers should counsel the parents and the child, commensurate with the child's maturity, and help families balance potential benefits and

risks of testing. When the balance of potential risks and benefits is uncertain, providers should generally respect the decisions of parents.

Conflicts between Parents and Adolescents

- Ideally, predictive genetic testing of children will be performed with both the consent of the parents and either the assent or consent of the child, depending on the child's maturity. The Task Force members hold differing views about cases where parents and adolescents disagree about genetic testing decisions. Where the balance of benefits and risks is uncertain, some members believe that providers should generally defer to the wishes of the parent, even over the objection of a mature adolescent. Others would defer to the adolescent's decision in at least some cases, particularly when an adolescent opposes testing.

Disclosure of Test Results to Minors

- If a child or adolescent has provided assent or consent for predictive genetic testing, he or she also should be informed of test results and their meaning, commensurate with his or her maturity and with his or her desire to have this information.

Genetic Testing of Prospective Adoptees by Their Current Caregivers

- Caregivers of prospective adoptive children should ensure that the children undergo genetic testing when such testing is necessary for the children's current health care.

Genetic Testing of Prospective Adoptees at the Request of Prospective Adoptive Parents

- Genetic testing should be performed on a prospective adoptee at the request of prospective adoptive parents only when (1) the testing is medically indicated and can reveal that a child is highly likely to develop extraordinary health care needs during childhood, (2) the testing will help ensure that the child is placed with a family who is capable of dealing with those needs, and (3) the prospective parents are otherwise committed to adopting the child.

Collection and Disclosure of Prospective Adoptees' Medical Histories

- New York law should be amended to require that parties placing a child for adoption make reasonable efforts to collect a complete medical and genetic history of the child and provide it to the prospective adoptive parents. New York law also should be amended to require the parties to make reasonable efforts to collect the medical and genetic histories of the birth parents and close blood relatives of the prospective adoptee and disclose them to the prospective adoptive parents. The parties should collect and disclose this information in a manner that respects the privacy of the persons from whom it is obtained and the subjects of the information. For example, the medical and genetic histories of the prospective adoptees' relatives should be disclosed to prospective adoptive parents with all identifying information removed.

Confidentiality

- Numerous persons and organizations, including insurance companies and the government, have access to individuals' health and genetic information.
- Although legal protections for health and genetic information confidentiality exist on both the federal and state levels, they are often limited in scope and do not provide adequate

safeguards.

- Some commentators maintain that genetic information is more sensitive than other health information and should receive special confidentiality protections because it has been used in the past to discriminate and perpetrate terrible horrors against those deemed to be genetically unfit and because it reveals not only personal health information but also information that has implications for one's family. Other commentators contend that genetic information and nongenetic health information should receive the same levels of confidentiality protections because nongenetic health information can be just as sensitive as genetic information and it is impractical to provide varying levels of protection to different categories of health information.
- New York passed laws in 1996 and 1997 that provide greater confidentiality protections for predictive genetic information than for other health information. However, the laws do not protect the confidentiality of all genetic information, and they do not protect as confidential the fact that an individual has used or inquired about genetic services. The laws also do not provide individuals with legal remedies against those who violate the laws' provisions, do not appear to prohibit waiver of its confidentiality protections, and do not make clear whether anonymous genetic testing is permissible.
- Commentators disagree about whether health care providers ever have the obligation to disclose to a patient's relatives, over the patient's objection, the medical ramifications to the relatives of the patient's genetic information. Some commentators maintain that health care providers should not make such disclosures over the patient's objection and that health care providers should fulfill any obligations they might have vis-à-vis the patient's family by informing the patient of these ramifications and, when appropriate, advising the patient to disclose the information to the patient's family. Other commentators contend that health care providers should disclose the information directly to the patient's family, over the patient's objection, if the patient refuses to do so and if doing so would avert serious harm that is highly likely to occur absent such a disclosure.
- Some commentators have recommended that, to encourage individuals to take genetic tests and to prevent unconsented-to acquisition of genetic information by insurers, employers, and others, patients should be permitted to take certain types of genetic tests anonymously. Others believe that anonymous genetic testing is generally inappropriate because it interferes with proper pretest and posttest genetic counseling.

Conclusions and Recommendations of the Task Force

Confidentiality Protections for Genetic Information and Other Medical Information

- All personal medical information, including genetic information, should receive a uniform, high level of confidentiality protection. Absent new, comprehensive federal legislation or regulation that provides such protection, New York should enact comprehensive medical confidentiality legislation that does so.
- Assuming that comprehensive medical confidentiality protections are not adopted, New York's genetic confidentiality statutes should be amended to protect the confidentiality of all genetic information.

Confidentiality Protections for the Use of Genetic Services

- Assuming that comprehensive medical confidentiality protections are not adopted, New York's genetic confidentiality statutes should be amended to protect the confidentiality of the fact that an individual has obtained and/or inquired about genetic testing and/or counseling. The

statutes also should be amended to protect the confidentiality of the content of the inquiries and/or counseling.

Scope of Consented-to Disclosure of Genetic Information by Persons Other Than the Subject of the Information

- Assuming that comprehensive medical confidentiality protections are not adopted, New York's genetic confidentiality statutes should be amended to limit the disclosure of genetic information by persons other than the subject of the information to the amount necessary in light of the reason for the disclosure. The statutes also should be amended to limit such disclosures to those persons who have a need for the information in light of the reason for the disclosures.

Permissible Third-Party Disclosures of Genetic Information without the Subject's Consent

- Assuming that comprehensive medical confidentiality protections are not adopted, the legislature should review and, if appropriate, amend the genetic confidentiality statutes in light of the recommendations of the Special Committee on Medical Information Confidentiality of the New York State Public Health Council about legitimate disclosures of medical information without patient consent.

Waivers of Genetic Confidentiality Protections

- Assuming that comprehensive medical confidentiality protections are not adopted, New York's genetic confidentiality statutes should be amended to render nonwaivable all of the confidentiality rights they provide.

Disclosure of Genetic Information to Relatives

- Health care providers should discuss with their patients the medical ramifications of the patient's genetic information for the patient's relatives. Health care providers should encourage their patients to disclose genetic information to relatives when the disclosure is likely to help the relatives avert or treat disease or to make reproductive decisions. Health care providers should not disclose their patient's genetic information to the patient's relatives without the patient's consent or a court order. Courts should be authorized to permit health care providers to make such disclosures only when (1) the patient refuses to disclose the information to an identified relative despite attempts by the health care provider to convince him or her to do so; (2) without disclosure, serious harm to the relative is highly likely to occur; (3) with disclosure, the harm can be averted or its chances of occurring significantly minimized; and (4) the harm that may result from failure to disclose outweighs the harm that may result from the disclosure.

Court Orders for Disclosure of Genetic Information

- Other than court orders for the disclosure of genetic information to a patient's relatives, New York law should be amended to permit court orders for the disclosure of genetic information to third parties without the subject's consent only when (1) absent the disclosure, there is or would be a clear and imminent danger to the public health; (2) the third party is entitled to the disclosure under federal and/or New York statutes or regulations; or (3) in a civil or criminal litigation, the subject of the information affirmatively places his or her physical or mental condition at issue and the genetic information to be disclosed directly relates to that physical or mental condition.

Remedies for Unlawful Disclosure or Solicitation of Genetic Information

- Assuming that comprehensive medical confidentiality protections are not adopted, New York law should be amended to (1) expressly authorize private lawsuits by victims of unlawful disclosures or solicitations of genetic information against persons who make such disclosures or solicitations and (2) authorize the Attorney General to bring lawsuits on behalf of individuals whose genetic information has been or will be unlawfully disclosed or solicited. In addition, persons and organizations licensed by New York State should be subject to professional discipline for unlawfully disclosing or soliciting genetic information.
- Private and public institutions that deal with genetic information should create their own internal sanctions against persons who unlawfully disclose or solicit genetic information.

Anonymous Genetic Testing

- Although anonymous genetic testing has significant drawbacks, it should be an option available to those who desire it. New York law should be amended to eliminate potential barriers to anonymous genetic testing.

Insurance and Employment

Insurance

- Currently, insurers do not require applicants to take predictive genetic tests because the tests are very expensive and reveal only a limited number of serious genetic abnormalities. Commentators disagree about whether insurers use genetic information to make adverse insurance decisions. Insurers maintain that they may wish to use genetic information for insurance underwriting in the future.
- Some commentators argue that insurers should be prohibited from using genetic information for insurance underwriting because otherwise individuals, who could potentially benefit from genetic testing, may refrain from undergoing it and thereby endanger their health. Insurers argue that prohibiting the use of genetic information in underwriting could lead to adverse selection and would unfairly favor people who have genetic conditions or identifiable predispositions.
- Some commentators contend that although most Americans appear to consider access to health care a basic right, public attitudes concerning access to life, disability, and long-term care insurance are less clear. These forms of insurance can be seen as means to protect assets rather than as providing access to an important social good, such as medical care. Therefore, according to these commentators, the justifications for prohibiting insurance companies from underwriting using genetic information for these forms of insurance may be less compelling.
- Federal law and the New York Insurance Law prohibit group health insurance plans from making adverse insurance decisions against individuals based on their health status, medical history, and genetic information or from treating genetic information as a pre-existing condition. New York's community rating, open enrollment, and other insurance laws prohibit individual and small group health insurers from making adverse insurance decisions based on genetic information or from treating genetic information as a pre-existing condition.
- Federal law and the New York Insurance Law do not prohibit life, disability, and long-term care insurers from making adverse insurance decisions based on genetic information. However, New York law requires the insurers' decision to be actuarially justified and requires insurers to notify individuals in writing if it charges a higher-than-standard premium or denies an individual

insurance based on genetic test results. New York law also prohibits insurers from placing an individual's genetic test results into a nonconsenting relative's records and from drawing, using, or communicating an adverse inference about the relative's genetic status based on these results.

Employment

- Commentators disagree over whether employers make adverse employment decisions based on genetic predispositions to disease. Most commentators agree, however, that it is generally inappropriate for employers to fire, refuse to hire, or otherwise discriminate against qualified individuals in the terms and conditions of employment because they have a genetic predisposition to disease.
- Although federal protections against adverse employment decisions based on genetic predispositions to disease are limited, New York law generally prohibits such decisions.

The Impact of the Americans with Disabilities Act

- Because it is unlikely that genetic predispositions to disease are "disabilities" within the meaning of the Americans with Disabilities Act, the Act's protections against adverse employment and insurance decisions based on such predispositions are limited.

Conclusions and Recommendations of the Task Force

Health Insurance and Individual Medical Underwriting

- Access to health care is a necessity for all Americans, and for most Americans, health insurance provides such access. By limiting individual medical underwriting in health insurance, New York's community rating and open enrollment laws appropriately seek to make access to medical care more equitable.

Current Protections against Adverse Insurance Decisions by Health Insurers Based on Genetic Information

- The combination of New York and federal laws currently protects New Yorkers from adverse insurance decisions by health insurers based on genetic information.

Use of Genetic Test Results by Life, Disability, and Long-Term Care Insurers

- New York insurance law should be amended to require a moratorium on requests for genetic test results and the use of genetic test results in underwriting, by life, disability, and long-term care insurers. Insurers should be permitted to use these results for underwriting only when (1) the subjects of the tests voluntarily provide the results to the insurer and (2) the insurers will use the results for the subjects' benefit.

Use of Genetic Information by Employers

- New York Law provides significant protections against adverse use of genetic information by employers. As a result of these protections, it is not necessary to consider further legislation in New York prohibiting the use of genetic information by employers.

Public Health Role in Genetic Services

- As opposed to clinical medical practice, which focuses on the health of individual patients, public health focuses on disease prevention for whole populations. Public health's core functions include assessment (the systematic collection, assembly, analysis, and dissemination of information about the health of a community), policy development, and assurance of the safety and reliability of, and access to, health services.
- Public health assessment activities in the genetic context include population surveillance and molecular genetic epidemiology research. Policy development activities include the translation of scientific and medical discoveries about genetics into guidelines, regulations, and legislation to promote the public's health. Assurance activities include oversight by the federal government and New York State of genetic testing laboratories.
- New York State has the most far-reaching requirements for genetic test approval and laboratory oversight in the United States. New York State mandates genetic testing laboratories to engage in quality assurance and to employ personnel that meet certain standards. New York also reviews clinical genetic tests for their analytical and clinical validity and requires all laboratories that are located in New York State or test specimens from New York State to obtain a permit for the testing from the New York State Department of Health.
- Commentators have expressed concern about the oversight of predictive genetic testing. They note the current limited degree of federal oversight, the rapid expansion of genetic technologies and clinical genetic tests, and the complexity of genetic test performance and interpretation. Concerns about New York's program of oversight of genetic testing laboratories include disincentives to out-of-state laboratories to participate, timeliness of the program's responses to genetic test approval requests, difficulty locating information about laboratories with New York State permits, and the program's lack of clear criteria for assessing a predictive test's clinical validity.
- The federal and New York State governments, as well as some other states and nongovernmental organizations, promote genetics education for the public and have established and are continuing to establish genetics education programs. The federal and New York State governments also have developed programs to help ensure access to genetic services.

Conclusions and Recommendations of the Task Force

Oversight of Genetic Testing by Federal and State Government Agencies

- Federal government agencies should strengthen their oversight of clinical laboratory genetic tests, including tests provided as services, to ensure that tests have adequate analytical and clinical validity. New York State should continue its oversight of clinical genetic testing laboratories and should re-examine its criteria and processes for test approval and laboratory oversight.

Approval Process for Genetic Tests

- The New York State Department of Health's Laboratory Reference System, and its Clinical Testing Review Panel, should review proposed genetic tests expeditiously, within a specified time period. Approval decisions for individual genetic tests should be made on a case-by-case basis, based on analytical validity and clinical validity data for the test's intended use. The program also should require testing laboratories to provide educational materials to providers ordering the test.

Establishment of Criteria for Genetic Test Approval

- The New York State Department of Health should develop clear guidelines to delineate the assessment criteria the Laboratory Reference System will use for approval of genetic tests. The guidelines document should be open for public comment by interested parties, including genetic testing laboratories and clinical geneticists practicing in New York State, prior to adoption by the department.

State Oversight of Laboratory Quality Assurance

- The New York Laboratory Reference System should continue to require that permitted genetic testing laboratories meet specified certification, performance, and personnel standards and participate in quality assurance programs.

Establishment of a Genetic Testing Advisory Committee

- The New York State Department of Health should create a genetic testing advisory committee, composed of departmental members and representatives of New York's clinical and diagnostic laboratory genetics community, to meet at least annually to review New York's Laboratory Reference System's genetic test approvals, the approval process, and outcomes. The committee also should serve as a sounding board for the clinical genetics and genetic laboratory communities and aid the department in its efforts to disseminate genetic testing information among health care providers.

Categorization of Approved Genetic Tests

- For genetic test approval, the New York Laboratory Reference System should move from its current categories of test approval, "generally accepted" and "investigational," to a single category of approved tests in which test-specific limitations or restrictions that are important to patients, providers, and/or payers are noted. For example, approval should specify, when relevant, the need for ongoing data collection to establish a test's clinical validity for its intended application.

Provider Access to State Oversight Information

- The New York State Department of Health should ensure that an up-to-date database listing of New York State-approved genetic tests and the laboratories authorized to perform them is readily accessible to health care providers in New York State.

Exemption from State Regulations for Laboratory Licensure

- New York's Public Health Law, which requires state licensure for all laboratories performing tests on specimens obtained in New York, should be amended to permit the New York State Department of Health to grant exemptions on a case-by-case basis.

Ongoing Collection, Evaluation, and Dissemination of Clinical Data

- Federal and state health agencies should work with laboratories, providers, and other partners to promote the ongoing collection, evaluation, and dissemination of clinical validity and utility data for predictive genetic tests.

Role of Institutional Review Boards

- Organizations seeking New York State approval for genetic tests that require ongoing collection of clinical data should be required to submit evidence that they have obtained approval of an institutional review board.

Assessment of Population Needs

- Federal and state health agencies play an important role in assessing the population's genetic and environmental risk factors. The New York State Department of Health should continue its activities in statewide assessment of the population's genetic health and genetic epidemiology research.

Public and Provider Education

- Federal and state health agencies play an important role in educating the public about genetics generally and about particular genetics services that are available to the public. They also should support production and dissemination of genetics educational materials to health care providers.

Coordination of State Agency Genetics Activities

- The New York State Department of Health should assure coordination of activities of departmental personnel and programs that promote genetics health and research activities throughout the department. The department also should promote coordination of its efforts with those of other partners outside the department.

Integrating Genetics Services into Clinical Care

- Clinical genetics services encompass the application of genetics technology in a wide array of clinical contexts, including treatment and management of genetic disorders and genetic testing for diagnostic and predictive purposes. Genetics services providers include physicians and nurses with special training in genetics, genetic counselors, and others.
- Studies indicate that primary care physicians, who will be utilized for genetic services more frequently as the demands for such services grow, have limited training in genetics and often do not have the knowledge to integrate genetics into primary care services. Commentators have recommended a number of ways to increase genetics knowledge among physicians, including a greater emphasis on genetics in medical and postgraduate medical training programs, continuing medical education in genetics, and the creation of clinical guidelines for genetic medicine.
- Genetic counselors provide patients with counseling services regarding the occurrence or risk of occurrence of genetic conditions or birth defects. Although no state currently requires genetic counselors to be licensed or certified in order to practice or use the title genetic counselor, genetic counselors can receive board certification from the American Board of Genetic Counselors. In the mid-1990s, about 66 to 75 percent of New York's approximately 150 genetics counselors were board certified.
- Most genetic counselors work in institutional settings as part of a genetics services delivery team and under the supervision of a physician. Under New York law, genetic counselors may not independently order genetic tests for patients. Because there are no specific medical billing codes (CPT codes) for genetic counseling, genetic counselors cannot directly bill third-party payers for the counseling services they provide. Third-party payers do not consider genetic counselors as reimbursable providers in part because the states do not license or certify genetic

counselors.

- Increasingly, insurers are covering the costs of predictive genetic testing and counseling for individuals who are at risk for adult-onset disorders or disorders in future offspring. In general, insurers will cover counseling by medical geneticists with M.D. or D.O. degrees, although some insurers will cover counseling by Ph.D. geneticists or nongeneticist physicians.

Conclusions and Recommendations of the Task Force

State Licensure or Certification of Genetic Counselors

- To ensure an adequate level of competency of genetic counselors and to support the viability of the profession of genetic counseling, New York State should create a process for state certification of genetic counselors who are certified by the American Board of Genetic Counseling or American Board of Medical Genetics.

Scope of Genetic Counseling Practice

- Ideally, all genetic counselors should work within a team of health care providers, which may include medical geneticists, Ph.D. geneticists, primary care physicians, and physician specialists, such as oncologists, obstetrician-gynecologists, and neonatologists, to provide genetic counseling as an integrated component of the patient's health care. If genetic counselors practice independently, they should maintain the same level of professional standards as genetic counselors who work within institutional settings and should strive to achieve the benefits of working in an integrated health care team by consulting with other genetics and nongenetics professionals.

Authorization to Order Genetic Tests

- Under New York law, genetic counselors can order genetic testing for their patients only through licensed physicians or other persons who are authorized by law to do so, such as dentists, podiatrists, and nurse practitioners. The Task Force does not recommend any changes to the current law.

Direct Billing by Genetic Counselors

- The Task Force encourages the American Medical Association to adopt changes to the CPT codes that would allow nonphysician genetic counselors to bill directly for genetic counseling services.

Training Genetic Counselors about Legal and Ethical Issues

- All genetic counselors should receive training in and be knowledgeable about legal and ethical issues relevant to genetic counseling, such as confidentiality and medical privacy. Professional societies of genetic counselors should develop standards and guidelines for educating and training genetic counselors about legal and ethical issues.

Genetics Training in Medical School and Postgraduate Education

- Medical schools should incorporate genetics education into their core curriculum. Medical schools and postgraduate training programs should integrate genetics into clinical practice training to teach the necessary skills and attitudes for recognition and assessment of the

genetic component of disease.

Physician Licensure Examinations with Genetics Requirements

- Physician licensing examinations should assess knowledge of basic genetics issues.

Genetics Education through Clinical Guidelines

- Professional medical associations should promote development of comprehensive and up-to-date clinical guidelines to help physicians recognize appropriate genetic testing opportunities, provide a source of continuing genetics education, and ensure that patients receive adequate counseling and appropriate specialty referrals. National and state health agencies and private partners should support the development, updating, and dissemination of professional guidelines.

Specialty Board Certifications with Genetics Requirements

- The American Board of Medical Specialties and the individual specialty boards should ensure that specialty board certification and recertification examinations adequately assess genetics competencies.

Medical Organization-Based Requirements for Genetics Education

- Managed care and other medical practice organizations should promote genetics education of their member practitioners for the appropriate integration of genetic testing and counseling services and for specialty referrals.

Genetics Education and Training for Nursing and Allied Health Professionals

- Nursing and allied health professionals, working with the genetics community, should continue their efforts to incorporate genetics into all levels of nursing practice and allied health services and to promote research to assess and monitor the integration of genetics into all nursing and allied health practices.

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