

Tests for over 900 genetic conditions are available to assist health care providers with patient management decisions. Genetic tests are used to diagnose disease, predict risk of future disease, inform reproductive decision-making, and manage patient care. In the past most genetic testing was for rare, so-called “single gene,” genetic disorders. They were ordered by geneticists familiar with the nuances of genetic testing and test interpretation and performed in academic labs with which the geneticist had an established relationship. But genetics is increasingly being practiced in other medical specialties as the genetic contributions to numerous disorders become known. And as knowledge about the genetic underpinnings of common, complex diseases – diabetes, cancer, heart disease, and the like – expands, so will the use of genetic information in routine health care. Thus, the practice of genetic medicine is transitioning from the academic-based genetics clinic to other medical specialties and, increasingly, to the community-based primary care provider’s office.

It is the responsibility of the health care provider to correctly use and interpret genetic tests. The clinician must know when it is appropriate to test, the correct test to order, what information the test can provide, the limitations of the test, how to interpret positive and negative results in light of the patient’s medical or family history, and the medical management options available. However, studies continue to show that many health care providers are ill prepared to use genetic tests in clinical practice [1].

Health care provider organizations can play an important role in developing clinical practice guidelines to aid clinical decision-making and in disseminating new information and technology. Clinical practice guidelines are “systematically developed statements to assist practitioner and patient decisions about appropriate health care for specific clinical circumstances” [2]. Practice guidelines include an assessment of the quality of the available evidence for a clinical scenario and provide recommendations for the clinician and patient based on that evidence.

According to the Conference on Guidelines Standards [4], the best guidelines:

- 1) provide an overview of the guideline and the date developed;
- 2) describe the focus,
- 3) the goal,
- 4) the user and setting,
- 5) and the target patient population;
- 6) identifies the developer and potential conflicts of interest;
- 7) names the source of funding;
- 8) describes the methods used to search the literature,
- 9) the criteria used to rate the quality of the evidence, and
- 10) the method for synthesizing evidence;
- 11) identifies how the proposed guideline was reviewed before being released, and
- 12) the plan for updating;
- 13) provides definitions of unfamiliar terms;
- 14) indicates the quality of the evidence and the strength of the recommendations;
- 15) reviews harms or benefits of implementing the guideline;
- 16) outlines the role of patient preferences;
- 17) provides a graphical description of the stages and decisions; and
- 18) describes implementation barriers.

Guidelines vary tremendously, however, in both quality and usefulness [3]. Guidelines may be evidence-based, in which rigorous methods are used to rate the quality of the evidence available, or consensus-based, that is, developed as the result of agreement amongst experts in the field. The Conference on Guidelines Standards provides 18 elements to include in a guideline (see Box).

Even the best evidence-based guidelines, however, developed with the most rigorous methodologies and incorporating all of the recommended elements, will be of little value if not utilized by the clinicians for whom they are intended. Getting guidelines to the point of care and changing clinical behavior are not trivial matters and studies have described numerous barriers to implementing guidelines [5,6,7]. However, when effectively disseminated and utilized, guidelines have been shown to improve clinical outcomes [8,9,10].

In the era of genetic testing, practice guidelines can assist professional judgments about when genetic testing is appropriate, how the information from a test may be used in clinical decision-making, how to frame discussions with patients, when referral for genetic counseling is appropriate, and how to interpret both positive and negative results. As an example, in 2001 The American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) published guidelines in response to a National Institutes of Health Consensus Develop Panel report Genetic Testing for Cystic Fibrosis [11]. The ACOG/ACMG guidelines recommended *offering* cystic fibrosis (CF) carrier testing to individuals with a family history of CF, reproductive partners of individuals who have CF, and couples for whom one or both partners are Caucasian and planning a pregnancy or seeking prenatal care. Testing should be *made available* to individuals at lower risk, i.e., couples in other racial and ethnic groups, for whom the test may be less sensitive [12]. The guideline developed was a collaborative effort in which each organization contributed its area of expertise – ACMG, the laboratory expertise in recommending which of the hundreds of CF mutations should be included in the screening panel, and ACOG, the clinical expertise of how to provide carrier screening in the obstetrical and gynecology practice. When disseminated, the guidelines were supplemented with physician and patient educational materials. Two years after implementation of the guidelines, a survey of obstetricians and gynecologists revealed that 86.6% provided information about CF and CF screening to their pregnant patients and 65.8% offered CF carrier screening - although not exactly according to the guidelines. Fewer ObGyns offered preconception testing as recommended and there was difficulty in practice of making the distinction between *offering* CF carrier testing versus *making available*. As a result, *all* pregnant couples tended to be offered testing as opposed the more defined set of patients based on ethnic background or family history as recommended by the guidelines. Thus, the survey confirmed the role of genetic testing guidelines in positively impacting clinical practice, while highlighting implementation and practice issues important for future genetic testing guidelines [13,14].

Yet despite their utility in aiding clinical decision-making, relatively few guidelines for genetic testing have been developed by health care provider organizations. A review of statements related to genetic tests published in the literature, posted on societies' websites, or on the National Guideline Clearinghouse website [15], reveals that only a limited number of such statements exist. [See Table 1] Some of them cover the same test, although from different perspectives. For example, the technical aspects of Fragile X testing are addressed by the American College of Medical Genetics, the care of children with Fragile X by American Academy of Pediatrics, prenatal diagnosis of Fragile X by the American College of Obstetricians and Gynecologists, and genetic counseling of Fragile X families by the National Society of Genetic Counselors. Statements also address counseling or ethical issues such as informed consent, direct to consumer testing, or genetic testing in adoption. The statements are variably classified by the organization as a "clinical report", "policy statement", "committee opinion", "position statement", or simply, a "statement". Sometimes the nomenclature reflects a well-defined internal process (such as when the level of evidence supports ACOG's use of the term "practice bulletin" versus an "opinion statement") but all too often the variability reflects inconsistent methodology, scope, and focus. Few of the published statements would meet the 18 recommended components of a clinical practice guideline [4].

This modest menu of statements reflects the history of genetic testing - organizations whose practices were impacted first by genetic tests have more numerous and more robust guidelines. The majority of guidelines were developed by medical geneticists and pediatricians, who diagnose and care for children with genetic disorders, and obstetricians who care for women at-risk to have a child with a genetic condition. More recently, the identification of mutations in genes associated with hereditary cancers has prompted the development of practice guidelines for genetic testing in oncology.

Few guidelines exist beyond those clinical areas where genetics historically has been practiced. Yet it is inevitable that genetic testing will continue to move into the medical mainstream. Already genetic variations have been identified that increase an individual's risk for thrombosis, cardiomyopathy, diabetes, and the like. Genetic variations are known that will predict a person's response to a treatment modality [16] or that will indicate the person is at higher risk for an adverse reaction to a drug [17]. Direct-to-consumer marketing of tests will put added pressure on clinicians to stay abreast of what tests are available and to understand the risks, benefits, and limitations of testing to appropriately advise their patients, or interpret results if a patient has ordered a test directly over the internet [18]. These are issues that will cut across medical specialties.

However, developing practice guidelines is not a trivial undertaking for professional health care provider organizations. The Genetics and Public Policy Center interviewed 20 health care provider organizations that develop practice guidelines about their policies and practices. They describe a development process that is time consuming and expensive. Most organizations rely on busy professionals to volunteer their time to conduct the review and develop the guidelines, although some of the larger

organizations have support staff to help move the process along. With the increasing emphasis placed on guidelines that are evidence-based, many organizations have developed detailed methods for guidelines development and extensive training materials for their expert volunteers. Some have commissioned research organizations to do the brunt of the evidence review, assuming those organizations have a certain level of competency already in this area. This process has met with variable success; ultimately the expert members must review the work (sometimes duplicating what has been done) and make the recommendations, thus not saving much time. Extensive internal and external vetting and approval processes within the organization also add to the development time. Indeed, it is not unusual for guidelines to take two years from the time of conception to dissemination to the membership. Once published, most organizations do not have the resources to monitor compliance or assess how well the guideline works in clinical practice. Despite the difficulty and expense, however, all of the organizations interviewed felt that guidelines are an important service they provide their members and when surveyed, members view practice guidelines as one of the top benefits of membership and one of the most visited and downloaded sites on the association website.

In addition to the difficulties organizations face in developing any guideline, there are a number of challenges specific to genetic testing guidelines. One is limited genetics expertise to draw upon within the membership to develop the guidelines.

A second issue is the rapidity with which genetic tests move into the market and the minimal level of governmental oversight of the laboratories performing genetic testing. The clinical validity of a test is only subject to premarket review by the FDA if the test is to be marketed as an in vitro diagnostic device, or “test kit”. Of the 900 genetic conditions for which testing is clinically available, the FDA has approved only a handful as kits. The rest have been developed by laboratories as in-house tests or “home brews”. Although laboratories are subject to regulation under the Clinical Laboratories Improvement Amendments of 1988 (CLIA), no specialty area for genetic testing exist, leaving laboratories to develop their own methods to document accuracy and reliability. This minimalist regulatory landscape puts an added burden on the clinician ordering genetic tests and subsequently on guideline developers to incorporate laboratory testing issues into the guidelines.

The rapid pace of advances in genetics and ease with which tests move into the market means that clinicians need guidance about the appropriateness of testing, or not, before the efficacy of a test will have been studied in multiple randomized clinical trials (RCT) - considered the gold standard of evidence. The lack of this type of evidence may prevent health care provider organizations from developing guidelines, yet knowing the level of evidence available, the strength of that evidence and, importantly, the gaps in knowledge, can help clinicians and patients make informed judgments about the relative value of testing in a particular situation. A systematic review of the available evidence will also help prioritize future research agendas.

Proposal for a solution

A mechanism is needed to support health professional organizations to develop, disseminate, and monitor the implementation of monitor genetic testing guidelines. The piecemeal approach of the past will not be sustainable in the long run and testing will quickly outpace the efforts of individual organizations. Federal agencies whose mission is to translate research finding into clinical practice, as noted in the NIH Roadmap or HRSA mission statement, could provide this needed support.

A sustainable source of financial support to which organizations could apply would enhance the number and quality of genetic testing guidelines available and address many of the challenges to developing guidelines in this rapidly emerging area of medicine.

The availability of funding would raise the priority of genetic testing guidelines in organizations where genetic testing must compete with other clinical practice areas for organizational attention and resources. Organizations could compensate their expert members, support the work of external experts if needed, and devote resources to developing, testing, and disseminating supplemental educational materials for patients and providers.

Additionally, the funding agency could play an important role in improving the overall quality of genetic testing guidelines by setting criteria for applicants and by establishing standards for what should be included. Applicants could be required to document their expertise in a particular topic area and the methods by which they will review the evidence and make recommendations; detail plans for provider and patient education; outline how the guideline will be disseminated and monitored for its usefulness in practice, and revised or update as needed. Encouraging multidisciplinary applications would utilize resources more efficiently, reduce the redundancy of several organizations covering the same topic, and ensure a broader dissemination and implementation plan.

Summary

Genetic testing is moving beyond the practice of the clinical geneticist, pediatrician, or obstetrician and increasingly is being used in all areas of medicine. Yet guidelines to aid clinical decision-making are lagging behind. Many of the issues inherent in developing genetic testing guidelines cut across clinical practice areas. Regardless of their specialty area, health care providers need guidelines for genetic testing that are accessible – they need to be able to find them when they need them. The guidelines need to be clear and easy to use, and the strength of the evidence and how recommendations were developed transparent and trusted. Even knowing in what way the data are incomplete or insufficient can assist clinical decision-making. Consolidating efforts of several organizations would reduce the burden of developing guidelines by more efficiently using resources and sharing expertise. A sustainable funding source would support this effort and provide standards to ensure that translation of new genetic discoveries into the clinic occurs in a timely fashion, so that those who would benefit

from testing have access, yet also in a thoughtful and measured way to ensure that providers and patients understand the benefits and limitations of testing.

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Table 1: Genetic Testing Guidelines

Organization	Date	Type	Condition	Title
American Academy of Allergy, Asthma and Immunology	2005	Guidelines	Immunodeficiency	Practice Parameter for the Diagnosis and Management of Primary Immunodeficiency
American Academy of Neurology	1996	Practice Parameter	Genetic Testing	Genetic Testing
American Academy of Neurology, Child Neurology Society	2003	Guidelines	Developmental Delay/MR	Practice Parameter: Evaluation of the Child with Global Developmental Delay
American Academy of Pediatrics	2005	Clinical Report	Achondroplasia	Health Supervision for Children with Achondroplasia
American Academy of Pediatrics	2004	Clinical Report	Prenatal Dx	Prenatal Screening and Diagnosis for Pediatricians
American Academy of Pediatrics	2003	Clinical Report	Turner Syndrome	Health Supervision for Children with Turner Syndrome
American Academy of Pediatrics	2002	Clinical Report	Hemoglobinopathies	Health Supervision for Children with Sickle Cell Disease
American Academy of Pediatrics	2001	Policy Statement	Down Syndrome	Health Supervision for Children with Down Syndrome
American Academy of Pediatrics	2001	Policy Statement	Genetic Testing	Ethical Issues with Genetic Testing in Pediatrics
American Academy of Pediatrics	2001	Policy Statement	Phenylketonuria	Maternal Phenylketonuria
American Academy of Pediatrics	2001	Policy Statement	William Syndrome	Health Care Supervision for Children with Williams Syndrome
American Academy of Pediatrics	2000	Policy Statement	Ambiguous Genitalia	Evaluation of the Newborn with Developmental Anomalies of the External Genitalia
American Academy of Pediatrics	2000	Technical Report	Congenital Adrenal Hyperplasia	Congenital Adrenal Hyperplasia
American Academy of Pediatrics	1996	Policy Statement	Marfan	Health Supervision for Children with Marfan Syndrome
American Academy of Pediatrics	1996	Policy Statement	Newborn Screening	Newborn Screening Fact Sheets
American Academy of Pediatrics	1996	Policy Statements	Fragile X	Health Supervision for Children with Fragile X Syndrome
American Academy of Pediatrics	1995	Policy Statements	Neurofibromatosis	Health Supervision for Children with Neurofibromatosis
American Academy of Pediatrics	1993	Policy Statements	Congenital Hypothyroidism	Newborn Screening for Congenital Hypothyroidism: Recommended Guidelines
American Academy of Physician Assistants	2001	Policy Brief	Genetic Testing	Genetic Testing in Clinical Practice
American Association for the Study of Liver Disease	2003	Practice Guideline	Wilson Disease	A Practice Guideline on Wilson Disease
American Association for the Study of Liver Disease	2001	Practice Guideline	Hemochromatosis	Diagnosis and Management of Hemochromatosis
American College of Medical Genetics	2006	Technical Standards and Guidelines	Genetic Testing	Technical Standards and Guidelines for Clinical Genetics Laboratories

Note: List obtained from review of the published literature, societies' websites, and the National Guideline Clearinghouse as of February 1, 2006 and not meant to be all inclusive

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Organization	Date	Type	Condition	Title
American College of Medical Genetics	2005	Technical Standards and Guidelines	Neural Tube Defects	Prenatal Screening for Neural Tube Defects
American College of Medical Genetics	2004	Technical Standards and Guidelines	Down Syndrome	Prenatal Screening for Down Syndrome
American College of Medical Genetics	2004	Policy Statement	Cystic Fibrosis	Cystic Fibrosis Population Carrier Screening: 2004 Revision of the American Board of Medical Genetics Mutation Panel
American College of Medical Genetics	2004	Policy Statement	Neural Tube Defects	Second Trimester Maternal Serum Screening for Fetal Open Neural Tube Defects
American College of Medical Genetics	2004	Practice Guideline	Developmental Delay/MR	Cytogenetic Evaluation of the Individual with Developmental Delay or Mental Retardation
American College of Medical Genetics	2004	Practice Guideline	Fragile X	Fragile X Diagnostic and Carrier Testing
American College of Medical Genetics	2004	Statement	Direct to Consumer	Direct to Consumer Testing
American College of Medical Genetics	2004	Technical Standards and Guidelines	Factor V Leiden	Technical Standards and Guidelines: Venous Thromboembolism (Factor V Leiden and Prothrombin 20210G>A Testing)
American College of Medical Genetics	2004	Technical Standards and Guidelines	Genetic Testing	Molecular Testing for Ultra Rare Disorders
American College of Medical Genetics	2003	Policy Statement	Huntington	Technical Guidelines and Standards for Huntington Disease Testing
American College of Medical Genetics	2002	Statement	Hearing Loss	Genetics Evaluation Guidelines for the Etiologic Diagnosis of Hearing Loss
American College of Medical Genetics	2002	Technical Standards and Guidelines	Cystic Fibrosis	Technical Standards and Guidelines for CFTR Mutation Testing
American College of Medical Genetics	2001	Consensus Statement	Factor V Leiden	Consensus Statement on Factor V Leiden Mutation Testing
American College of Medical Genetics	2001	Guidelines	Cystic Fibrosis	Laboratory Standards and Guidelines for Population-based Cystic Fibrosis Carrier Screening
American College of Medical Genetics	2001	Policy Statement	Managed Care	Genetics and Managed Care
American College of Medical Genetics	2001	Position Statement	Genetic Discrimination	Points to Consider in Preventing Unfair Discrimination Based on Genetic Disease Risk
American College of Medical Genetics	2001	Statement	Fragile X	Technical Standards and Guidelines for Fragile X
American College of Medical Genetics	2001	Statement	Uniparental Disomy	Diagnostic Testing for Uniparental Disomy

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American College of Medical Genetics	2000	Position Statement	FISH	Technical and Clinical Assessment of Fluorescence In Situ Hybridization: Technical Considerations
American College of Medical Genetics	2000	Recommendations	Sequence Variation	Recommendations for Standards for Interpretation of Sequence Variations
American College of Medical Genetics	2000	Statement	Colon Cancer	Genetic Testing for Colon Cancer: A Joint Statement of the American College of Medical Genetics and the American Society of Human Genetics
American College of Medical Genetics	2000	Statement	Newborn Screening	Universal Newborn Screening
American College of Medical Genetics	2000	Statement	Newborn Screening	Tandem Mass Spectrometry in Newborn Screening
American College of Medical Genetics	1999	Clinical Guideline	Multiple Congenital Anomalies	Evaluation of the Newborn with Single or Multiple Congenital Anomalies: A Clinical Guideline
American College of Medical Genetics	1999	Guidelines	Breast Cancer	Genetic Susceptibility to Breast and Ovarian Cancer: Assessment, Counseling and Testing Guidelines
American College of Medical Genetics	1999	Policy Statement	Recontacting	Duty to Recontact
American College of Medical Genetics	1999	Position Statement	Gene Patents	Position Statement on Gene Patents and Accessibility of Gene Testing
American College of Medical Genetics	1998	Position Statement	Canavan	Position Statement on Carrier Testing for Canavan Disease
American College of Medical Genetics	1997	Report	Screening	Principles of Screening: Report of The Subcommittee on Screening of the American College of Medical Genetics Clinical Practice Committee
American College of Medical Genetics	1996	Consensus Statement	Breast Cancer	Statement on Population Screening for BRCA-1 Mutation in Ashkenazi Jewish Women
American College of Medical Genetics	1996	Statement	Paternal Age	Statement on Guidance for Genetic Counseling in Advanced Paternal Age
American College of Medical Genetics	1995	Statement	Storage of DNA	Statement on Storage and Use of Genetic Materials
American College of Medical Genetics (with American Society of Human Genetics)	2000	Statement	Adoption	Genetic Testing In Adoption
American College of Medical Genetics (with American Society of Human Genetics)	2000	Statement	Genetic Testing in Children	Genetic Testing in Children and Adolescents, Points to Consider: Ethical Legal and Psychosocial Implications of
American College of Medical Genetics (with American Society of Human Genetics)	1998	Statement	Homocystinuria	Measurement and Use of Total Plasma Homocysteine
American College of Medical Genetics (with American Society of Human Genetics)	1996	Report	Prader Willi/Angelman	Diagnostic Testing for Prader-Willi and Angelman Syndromes

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American College of Obstetricians and Gynecologists	2005	Committee Opinion	ART	Perinatal Risks Associated With Assisted Reproductive Technology
American College of Obstetricians and Gynecologists	2005	Committee Opinion	Cystic Fibrosis	Update on Carrier Screening for Cystic Fibrosis
American College of Obstetricians and Gynecologists	2005	Committee Opinion	Tay-Sachs	Screening for Tay-Sachs Disease
American College of Obstetricians and Gynecologists	2005	Practice Bulletin	Hemoglobinopathies	Hemoglobinopathies in Pregnancy
American College of Obstetricians and Gynecologists	2004	Committee Opinion	Cord Blood	Routine Storage of Umbilical Cord Blood Storage for Potential Future Transplantation
American College of Obstetricians and Gynecologists	2004	Committee Opinion	1st Tri Screening	First-Trimester Screening for Fetal Aneuploidy
American College of Obstetricians and Gynecologists	2004	Committee Opinion	Canavan	Screening for Canavan Disease
American College of Obstetricians and Gynecologists	2004	Committee Opinion	Jewish Screening	Prenatal and Preconception Carrier Screening for Genetic Disease in Individuals of Eastern European Jewish Descent
American College of Obstetricians and Gynecologists	2004	Committee Opinion	Phenylketonuria	Maternal Phenylketonuria
American College of Obstetricians and Gynecologists	2003	Committee Opinion	Newborn Screening	Newborn Screening
American College of Obstetricians and Gynecologists	2003	Committee Opinion	Paternal Age	Advanced Paternal Age: Risk to the fetus
American College of Obstetricians and Gynecologists	2003	Practice Bulletin	Neural Tube Defects	Neural Tube Defects
American College of Obstetricians and Gynecologists	2003	Practice Bulletin	Breast Cancer	Breast Cancer Screening
American College of Obstetricians and Gynecologists	2002	Technology Assessment	Genetic Testing	Genetics and Molecular Diagnostic Testing
American College of Obstetricians and Gynecologists	2001	Committee Opinion	Stillbirths	Genetic Evaluation of Stillbirths and Neonatal Deaths
American College of Obstetricians and Gynecologists	2001	Practice Bulletin	Prenatal Dx	Prenatal Diagnosis of Fetal Chromosomal Abnormalities
American College of Obstetricians and Gynecologists	2000	Committee Opinion	Fragile X	Fragile X Syndrome

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American College of Obstetricians and Gynecologists	1997	Educational Bulletin	Teratology	Teratology
American College of Physicians	2005	Ethics Manual	Genetic Testing	Genetic Testing
American College of Physicians	2005	Practice Guideline	Hemochromatosis	Screening for Hereditary Hemochromatosis
American Gastroenterological Association	2001	Guidelines	Colon Cancer	Hereditary Colorectal Cancer and Genetic Testing
American Gastroenterological Association	2001	Technical Review	Colon Cancer	Hereditary Colorectal Cancer and Genetic Testing
American Geriatrics Society	2000	Position Statement	Alzheimer's Disease	Genetic Testing for Late-Onset Alzheimer's Disease
American Psychiatric Association	2004	Practice Guideline	Alzheimer's Disease	Practice Guidelines for the Treatment of Patients with Alzheimer's Disease and Other Dementias of Late Life
American Society for Reproductive Medicine	2002	Educational Bulletins	Infertility	Information on Commonly Asked Questions about Genetic Evaluation and Counseling for Infertile Couples
American Society for Reproductive Medicine	2002	Guidelines	Gamete Donors	Minimal Genetic Screening for Gamete Donors
American Society of Clinical Oncology	2003	Policy Statement	Cancer	Genetic Testing for Cancer Susceptibility
American Society of Colon and Rectal Surgeons	2003	Practice Guideline	Colon Cancer	Practice Parameters for the Treatment of Patients with Dominantly Inherited Colorectal Cancer (Familial Adenomatous Polyposis and Hereditary Nonpolyposis Colorectal Cancer)
American Society of Human Genetics	1998	Policy Statement	Confidentiality	Professional Disclosure of Familial Genetic Information
American Society of Human Genetics	1995	Policy Statement	Alzheimer's Disease	Statement on Use of apolipoprotein E Testing for Alzheimer Disease
American Society of Human Genetics	1995	Policy Statement	Genetic Testing	Genetic Testing and Insurance
American Society of Human Genetics	1994	Policy Statement	Breast Cancer	Genetic Testing for Breast and Ovarian Cancer Predisposition
American Society of Human Genetics	1992	Policy Statement	Cystic Fibrosis	Cystic Fibrosis Carrier Screening
American Urological Association (w/ASRM)	2001	Best Practice Policy	Infertility	Report on Optimal Evaluation of the Infertile Male
International Society of Nurses in Genetics	2005	Position Statement	Confidentiality	Privacy and Confidentiality of Genetic Information: the Role of the Nurse
International Society of Nurses in Genetics	2005	Position Statement	Informed Consent	Informed Decision-Making and Consent: The Role of Nursing
International Society of Nurses in Genetics	2003	Position Statement	Access	Access to Genomic Healthcare: The Role of the Nurse
International Society of Nurses in Genetics	2002	Position Statement	Vulnerable Populations	Genetic Counseling for Vulnerable Populations: The Role of Nursing

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Organization	Date	Type	Condition	Title
National Society of Genetic Counselors	2005	Position Statement	Prenatal Dx	Preconception/Prenatal Genetic Screening
National Society of Genetic Counselors	2005	Practice Guideline	Cystic Fibrosis	Cystic Fibrosis Prenatal Screening in Genetic Counseling Practice
National Society of Genetic Counselors	2005	Practice Guideline	Fragile X	Genetic Counseling for Fragile X syndrome
National Society of Genetic Counselors	2005	Practice Guideline	Multiple SABs	Genetic Counseling and Evaluation of Couples with Recurrent Miscarriage
National Society of Genetic Counselors	2004	Practice Guideline	Cancer	Genetic Cancer risk Assessment and Counseling
National Society of Genetic Counselors	2003	Position Statement	Cystic Fibrosis	Cystic Fibrosis
National Society of Genetic Counselors	2002	Position Statement	Adoption	Genetic Testing and Adoption Position
National Society of Genetic Counselors	2002	Position Statement	Confidentiality	Confidentiality of Test Results
National Society of Genetic Counselors	2002	Position Statement	Genetic Testing	DNA Sequencing Position
National Society of Genetic Counselors	2002	Practice Guideline	Consanguinity	Genetic Counseling and Screening of Consanguineous Couples and their Offspring
National Society of Genetic Counselors	2002	Practice Guideline	Fabry	Fabry Disease in Genetic Counseling Practice
National Society of Genetic Counselors	1997	Position Statement	Adult onset	Genetic Testing for Adult-onset Disorders
National Society of Genetic Counselors	1995	Position Statement	Genetic Testing in Children	Prenatal and Childhood Testing for Adult-Onset Disorders
National Society of Genetic Counselors	1995	Practice Guideline	Pedigree Nomenclature	Recommendations for Standardized Human Pedigree Nomenclature
National Society of Genetic Counselors	1994	Position Statement	Screening	Genetic Screening
North American Society for Pediatric Gastroenterology, Hepatology and Nutrition	2005	Clinical Guideline	Celiac Disease	Guideline for the Diagnosis and Treatment of Celiac Disease in Children: Recommendations of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition
Society for Inherited Metabolic Diseases	2004	Policy Statement	Newborn Screening	Statement on Newborn Screening and Treatment of Individuals with Inborn Errors of Metabolism Detected by Newborn Screening
U.S. Preventative Services Task Force	1996 (under review)	Guidelines	Down Syndrome	Screening for Down Syndrome
U.S. Preventative Services Task Force	1996 (archived)	Guidelines	Neural Tube Defects	Screening for Neural Tube Defects
U.S. Preventative Services Task Force	2005	Guidelines	Breast Cancer	Genetic Risk Assessment and BRCA mutation Testing for Breast and Ovarian Cancer Susceptibility

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