Published in final edited form as:

J Commun Disord. 2008 ; 41(5): 444–457. doi:10.1016/j.jcomdis.2008.03.001.

Ethical and social implications of genetic testing for communication disorders

Kathleen S. Arnos*

Department of Biology, Gallaudet University, 800 Florida Avenue, NE, Washington, DC 20002, USA

Abstract

Advances in genetics and genomics have quickly led to clinical applications to human health which have far-reaching consequences at the individual and societal levels. These new technologies have allowed a better understanding of the genetic factors involved in a wide range of disorders. During the past decade, incredible progress has been made in the identification of genes involved in the normal process of hearing. The resulting clinical applications have presented consumers with new information and choices. Many of the same gene identification techniques are increasingly being applied to the investigation of complex disorders of speech and language. In parallel with gene identification, studies of the legal, ethical and psychosocial impacts of the clinical application of these advances and their influence on specific behaviors of individuals with communication disorders are paramount, but often lag behind. These studies will help to ensure that new technologies are introduced into clinical practice in a responsible manner.

Learning outcomes—As a result of this activity, the participant will be able to (1) explain the differences between Mendelian and complex forms of inheritance and why these differences complicate the ethical impact of genetic testing, (2) explain how publicly funded genome research through the Human Genome Project, the International HapMap Project and others have examined the ethical, legal and social implications of genome research, (3) list some of the ethical complexities of prenatal, newborn and predictive testing for various genetic disorders and (4) discuss the importance of evidence-based practice to the development of public policy for the introduction and clinical use of genetic tests.

1. Introduction

New technologies in genetics have led to exciting developments that have improved the ability of scientists to identify and characterize genes for many human traits. The application of these technologies to human health and disease is one of the major goals of the efforts which followed the completion of the Human Genome Project in 2003, through publicly and privately funded research efforts such as the HapMap Project, the Genes and Environment Initiative and the Human Diploid Project. As applications to human health arise, particularly in regard to more common, complex traits such as heart disease, cancer, diabetes, and dementia, more genetic tests will become available and more “personalized” testing and

© 2008 Elsevier Inc. All rights reserved.

* Tel.: +1 202 651 5258; fax: +1 202 651 5179. kathleen.arnos@gallaudet.edu..

Publisher's Disclaimer: This article appeared in a journal published by Elsevier. The attached copy is furnished to the author for internal non-commercial research and education use, including for instruction at the authors institution and sharing with colleagues. Other uses, including reproduction and distribution, or selling or licensing copies, or posting to personal, institutional or third party websites are prohibited. In most cases authors are permitted to post their version of the article (e.g. in Word or Tex form) to their personal website or institutional repository. Authors requiring further information regarding Elsevier's archiving and manuscript policies are encouraged to visit: <http://www.elsevier.com/copyright>

treatments will be made possible, leaving consumers and their health care providers faced with decisions about undertaking these tests. Applications to the improvement of human health are exciting and show great potential to improve quality of life and enable low-cost preventative intervention for many conditions however, concerns regarding the ethical, legal and social implications of these new technologies continue to be expressed. Such issues as discrimination, privacy and confidentiality, informed consent, access to genetic information by employers and insurance companies and appropriate use of testing for prenatal or pre-symptomatic diagnosis are basic issues that are the continuing topics of discourse by bioethicists (Arnos, 2003). Evidence-based guidelines from translational research to enable the clinical application of genetic discoveries to health care in an ethically responsible manner to maximize benefits and minimize harm are often lacking (Khoury et al., 2007). The introduction of genetic tests without consumer and professional education, cost/benefit analysis, data collection to determine reliability and quality control, and public evaluation can result in financial and social health care disparities which may lead to genetic elitism, worsening of health outcomes and the creation of a sub-class of the “worried well” (McGuire, Cho, McGuire, & Caulfield, 2007). The use of evidence-based practice to address clinical decision making for applications of new technologies or diagnostic techniques is recognized as important and is occurring more frequently in the fields of genetics (Burke, Zimmern, & Kroese, 2007; Khoury et al., 2007) and speech and hearing science (Dollaghan, 2004; Robey, 2004).

Incredible progress in the last decade has allowed the discovery of dozens of genes involved in the normal process of hearing and a handful associated with speech and language (Kochar, Hildebrand, & Smith, 2007; Morton & Nance, 2006). Many of the same clinical and ethical issues that apply to genetic testing for more common conditions also apply to genetic testing for different forms of communication disorders. The additional linguistic and cultural differences of some individuals with communication disorders, specifically those with hearing loss, add an interesting and challenging component to these already complex issues.

2. Mendelian and multifactorial inheritance—progress in identifying genes associated with communication disorders

Genetic conditions, including various forms of communication disorders, can be caused by a single gene or several genes in combination with environmental risk factors. Conditions which are inherited in a Mendelian fashion are caused by mutations in a single gene. Examples of Mendelian forms of communication disorders include non-syndromic forms of hearing loss caused by mutations in the *GJB2* (connexin 26) gene or syndromic forms of hearing loss such as Usher syndrome (a group of disorders caused by mutations in more than 11 separate genes), Waardenburg syndrome type 1 (caused by mutations in the *PAX3* gene) and Pendred syndrome (caused by mutations in the *SLC26A4* gene) (Kochar et al., 2007; Morton & Nance, 2006). Mendelian inheritance is characterized by three specific patterns called autosomal dominant, autosomal recessive and X-linked recessive. A detailed explanation of inheritance patterns associated with hereditary hearing loss is provided by Welch (2006).

Over the past decade, the rate at which genes for hearing loss have been identified and characterized has increased dramatically. More than 100 genes for syndromic and nonsyndromic deafness have been mapped and several dozen have been fully characterized so that the protein product is known and genetic testing to identify specific mutations can be performed. In several cases, complex interactions between separate genes (e.g. digenic inheritance) or between genes and environmental factors (e.g. genetic susceptibility to hearing loss caused by aminoglycosides) have been documented, emphasizing the need for

the guidance of geneticists in the interpretation of genetic test results and determining the impact on individuals and families. This knowledge has led to a greatly increased understanding of the physiologic and molecular mechanisms of hearing. Tests for common forms of hereditary deafness, such as *GJB2* (connexin 26), are now considered standard of care as part of the follow up done for infants in whom hearing loss has been confirmed and in the future, may become part of routine newborn genetic screening at birth (Morton & Nance, 2006).

Most human traits, including many common diseases are caused by a combination of several genes and environmental factors. This is known as complex or multifactorial inheritance. Each of the different genes and environmental factors usually has a small, additive effect on causing the trait. These factors are often difficult to identify and any single gene or variant will typically contribute only a small risk to the onset and/or severity of the condition. It can be very difficult to elucidate the environmental factors which contribute to any particular condition. Examples of multifactorial conditions associated with hearing loss include the genetic susceptibility to hearing loss caused by aminoglycoside antibiotics (Pandya, 2007) as well as presbycusis and noise induced hearing loss (DeStefano, Gates, Heard-Costa, Myers, & Baldwin, 2003; Gates, Couropmitree, & Myers, 1999; Zhu et al., 2003). Much of the recent research elucidating the causative factors involved in speech and language disorders suggests that disorders such as autism, dyslexia, stuttering, aphasia, specific language impairment and other disorders of articulation, vocabulary skill and verbal memory exhibit complex or multifactorial inheritance (Bishop, 2002; Felsenfeld, 2002; Gibson & Gruen, 2008; Hayiou-Thomas, 2008). Like hearing loss, progress has been made in the discovery of genes involved in speech and language disorders, although at a much slower pace. The lack of specific diagnostic criteria to allow grouping of the extremely heterogeneous disorders of speech and language is but one of the many challenges faced by researchers who work in gene identification. Excellent reviews of the evidence for a significant genetic contribution to disorders of speech and language, the challenges of molecular genetic studies in identifying the specific factors involved in these complex disorders, and potential research strategies to overcome these challenges are presented by Felsenfeld (2002), Bishop (2002), and Gibson and Gruen (2008).

3. The role of genome science and ethics research

Over that past 10 years, several technological developments in molecular biology have greatly enhanced the progress in identifying genetic factors associated with both Mendelian disorders and multifactorial conditions. High-throughput DNA sequencing methods allow the rapid identification of the specific order of the chemical components of a particular piece of DNA, gene, or genome. Rapid sequencing of DNA, together with powerful computational methods, has allowed the determination of the majority of the DNA sequence in the human genome, as well as variants in the genome that occur from person to person. These variants occur in many forms, one known as single nucleotide polymorphisms or SNPs. SNPs are differences in single letters of the DNA sequence that are a normal genetic variation. Identification of the more than 10 million SNPs is useful since these variants can serve as markers to locate genes or DNA sequences associated with particular physical characteristics or diseases. Techniques such as microarray-based comparative genomic hybridization (targeted and whole-genome arrays) and genome wide association studies have been essential in the search for genes which contribute to complex phenotypes and the applications of these tests to clinical care (Manning & Hudgins, 2007; McGuire et al., 2007). Many of the new technologies have provided the means to examine the genetic components of complex diseases and traits for which other, older techniques such as genetic linkage studies or candidate gene approaches have not been successful. As these techniques have been increasingly introduced to clinical care for diagnosis of complex traits, patient

misunderstandings of the predictive power of these tests, and overhyped conclusions regarding the causality of genetic variants have been problematic (Tabor & Cho, 2007).

The more rapid development of many, if not all of these technologies was made possible by work done as part of the Human Genome Project (HGP). The HGP was initiated in 1990; a major goal of this project was to determine the exact DNA sequence of the human genome as well as the genomes of several other species. Other goals included an examination of the ethical, legal and social issues of genomic research, the development of publicly available databases to store genomic information and the development of new computational tools for data analysis (Human Genome Project Information, 2005). Early versions of the human genome sequence were first published by HGP researchers and a privately funded group in 2001 (Lander et al., 2001; Venter et al., 2001). The National Human Genome Research Institute led the HGP (Human Genome Sequencing Consortium), an international effort which culminated in the completion of the full human genome sequence in April 2003 (International Human Genome Sequencing Consortium, 2004). The private and federally funded teams had different technical approaches to sequencing of the human genome using DNA from different human donors; their combined accomplishments set the groundwork for additional public and privately funded genome studies. The expanded mission of the National Human Genome Research Institute as it currently exists is to perform studies which permit “understanding the structure and function of the human genome and its role in health and disease” (National Human Genome Research Institute [NHGRI]). These expanded studies are designed to understand gene expression by identifying the full complement of human proteins (Service, 2003), the relationship between variations in the genetic sequence and phenotype, and the interaction of proteins with each other. As part of an important priority, the National Human Genome Research Institute continues to devote a significant proportion of its annual budget to a program known as Ethical, Legal and Social Implications (ELSI) which supports research into the ethical, legal and social issues such as intellectual property, translation of genetic information to improved human health, use of genetic information in non-health care situations, and the impact of genomics on the concepts of race, ethnicity and group identity. The ELSI program also supports public and professional education on a wide variety of issues (NHGRI, 2007).

The International HapMap Project was conducted between 2003 and 2005 and was designed to catalog information regarding common DNA variants that occur in various populations around the world (International HapMap Project, a). The HapMap database is intended to provide a resource for researchers who can utilize the information to link genetic variants to specific diseases. The HapMap is unique in that it characterizes how SNPs and other DNA variants are organized within the genome; variants that are near to each other are inherited as part of a haplotype. There are a few haplotypes in humans in which linked DNA variants are found which have proven to be quite useful to researchers seeking to identify disease-causing genes. The scientists associated with the International HapMap Consortium maintained close collaborations with bioethicists through an ELSI group to address issues related to privacy, cultural and religious implications, and “community engagement” from country to country (International HapMap Project, b). For example, due to its emphasis on working with specific populations, HapMap research may have identified genetic variants more common in some populations that may be associated with a higher-than-average risk of a disease. ELSI researchers studied mechanisms to avoid group stigmatization and discrimination that may occur to members of those populations due to identification of these variants. There was also a risk that HapMap research findings may have undermined established cultural or religious traditions or political or legal status in some groups by challenging existing social and cultural methods of determining group membership. Geneticists and bioethicists worked together in the communities participating in HapMap to provide community education and community engagement through meetings, focus group

discussions, public surveys and individual interviews. This process helped researchers to understand and respond to attitudes, beliefs and concerns of the community. As described below, similar efforts have been undertaken in recent years by researchers studying hearing loss to understand the attitudes towards and beliefs regarding genetic technologies for hearing loss in parents of children with hearing loss as well as deaf and hard of hearing adults.

The recently published Diploid Genome Sequence (called HuRef) is the complete sequence of only one person, which includes assembly of the sequences obtained from each parent (Levy et al., 2007). While the original Human Genome sequencing required more than \$500 million of public and private funds, the HuRef sequence, a privately funded project, was completed at a fraction of the cost. The advantage of having the DNA sequence from only one person is that the problem of underestimation of the variability in the human genome could be overcome. HuRef demonstrated that there was a five times greater level of variability in the DNA sequences inherited from each of the parental chromosomes than expected.

The Genes and Environment Initiative is a collaborative effort between geneticists and environmental scientists which received federal funding in late 2007 (Genes, Environment and Health Initiative, 2006). This initiative has two main components: (1) the analysis of SNPs in groups of patients with common chronic diseases such as diabetes, childhood asthma, obesity and autism and (2) the creation and validation of methods to monitor environmental exposures (toxins, dietary intake and physical activity) which interact with genetic variations to produce human disease. The first component of the project is administered by the National Human Genome Research Institute, while the second is led by The National Institute of Environmental Health Sciences, both at the National Institutes of Health. This is an exciting new project with great potential to uncover elusive environmental factors which contribute to multifactorial conditions; discoveries made regarding these factors will surely have important implications for understanding other multifactorial conditions, including those involving disorders of speech, language and hearing.

4. Ethical, legal and social implications of genetic testing

Research in genetics and genomics is rapidly leading towards clinical applications and has already resulted in new genetic tests becoming available for many conditions. Some genetic tests are offered on a routine basis through commercial laboratories; some are available only through carefully controlled research or clinical protocols; an increasing number are being made available directly to consumers (called direct to consumer genetic testing). Types of genetic testing include prenatal, newborn screening, carrier screening, and diagnostic testing, as well as predictive or presymptomatic testing to detect gene mutations which lead to conditions that occur later in life (such as Huntington disease or polycystic kidney disease) or to detect genes which may predispose to disorders such as heart disease, cancer or Alzheimer disease.

While there can be many health benefits of genetic information in the identification and treatment of disease, the risks of genetic tests are usually not physical, but are psychological, financial and social. Ethical concerns occur at the individual, family and societal levels. Specific concerns of the use of genetic information and testing center around privacy and confidentiality, the provision of informed consent which adequately provides information about risks and benefits of testing (McGuire & Gibbs, 2006), the appropriate uses of prenatal diagnosis, use of testing to predict the future onset of diseases for which there may be no prevention or treatment, and discrimination in employment and insurance (Arnos, 2003). Testing for genes which predispose to complex or multifactorial conditions such as

communication disorders carries with it additional concerns. The genetic complexity of most diseases tends to lessen the predictive power of genetic tests for predisposing genes, particularly regarding aspects which seem to be the most important to patients, such as predicting the severity and time of onset of the disease as well its treatability (Juengst, 2004). Early identification of these genetic variants in people who may or may not go on to actually develop the disorder in question may lead to social and medical stigmatization and discrimination related to employment and health insurance. In response to concerns regarding employment and insurance discrimination based on the results of genetic testing, many states and the federal government have now enacted legislation that restricts the use of genetic information by insurance companies and bans the use of genetic screening for employment decisions (NHGRI, 2007).

An overriding issue is the provision of genetic counseling as part of the genetic testing process, so that accurate information is conveyed pre- as well as post-test, especially concerning the predictive power of tests for complex disorders. Families and clinicians are very motivated to understand the cause of neurodevelopmental, speech, hearing and language, and other disorders which have no clear familial pattern. Priorities are to have tests with clear diagnostic and predictive value that may lead to better treatment and allow prenatal detection of disorders. As genetic tests become more common and are increasingly ordered by primary care providers for their patients, problems arise with the lack of pre- and post-test genetic counseling by qualified professionals. The value of competent genetic counseling cannot be overestimated, particularly when testing for genes which predispose to multifactorial disorders and which may confer only a small risk of disease or for which the precise risk of disease may not even be clearly known. An extreme example of the potential danger of the disconnect between genetic testing and pre- and post-counseling is direct to consumer genetic testing (Lewis, 2007). Commercial genetic testing laboratories are now marketing genetic tests for breast cancer (*BRCA1* & *BRCA2*), hemochromatosis, and cystic fibrosis as well as non-clinical tests for nutrition, aging and behavior directly to consumers (NHGRI, 2004). Direct to consumer marketing has been shown to lead to inappropriate test utilization, misinterpretation of test results, and lack of necessary follow-up. The limitations of genetic testing are rarely accurately conveyed via print and television advertisements. An increasing number of companies are now using the Internet to market genetic tests to consumers and a variety of "home testing" kits are now available. The rapid increase in direct to consumer genetic testing in recent years has prompted the American College of Medical Genetics to issue a position statement on this practice regarding the potential harm of genetic tests for which no appropriate pre- and post-test genetic counseling is provided (ACMG Board of Directors, 2004).

Problems with misunderstandings about the clinical power of genetic tests may be exacerbated by the language and hype in the professional scientific literature and in the media which is used to describe research about the identification of genetic factors in complex disorders. An example is research on autism, an extremely heterogeneous disorder that occasionally (about 10% of cases) has a single gene or chromosomal etiology (e.g. fragile X or Rett syndrome, tuberous sclerosis, chromosome 15q11–q13 duplication). Autism is largely a multifactorial disorder with a strong genetic component, caused by susceptibility genes and environmental factors that remain largely unknown (Herman et al., 2007). A recent review of the ethical concerns related to the identification of genetic variants that may contribute to or predispose to disorders with complex inheritance examined recent literature describing research in autism (Tabor & Cho, 2007). The research examined a recently developed technique known as array comparative genomic hybridization to examine copy number variants that may be associated with autism. Tabor and Cho identified substantial inconsistency for evaluation of causal criteria for genetic variants, which often led to overstatement of the causal nature of variants that were thought to be associated with

autism. The use of more cautious language was advised when describing causation, to avoid misinterpretation of the predictive value of these variants by clinicians, families and the media. Ethical concerns were also raised about specific research criteria related to study design, including selection of the population and sample size, which have an important impact on determining causality and the generalizability of results.

Prominent bioethicists have contended that the enduring ethical concerns of genetic information go beyond the issues mentioned above. Four fundamental and enduring moral concerns in genetics are identified by Juengst (2004); genetic information can disclose essential secrets about individuals which can affect their identities in terms of familial role, ancestral origin, community memberships, and ethnic affiliations. Some argue that ethical problems in genetics are time limited and will resolve as genome medicine catches up with genome science, however, “the basic social challenges of genetic information are not the clues it can give us about future health risks. . . as long as people use familial role, ancestry, community membership or ethnic identity as indicators of social standing, genetic information will continue to be socially potent” (Juengst). As mentioned above, the recognition of these risks prompted federally funded projects such as the International HapMap Project and the Human Genome Project to devote a significant proportion of their budgets to community education and discussion with community members regarding the role of genetic testing in defining or challenging community membership, ethnic identity or familial roles.

5. Clinical use of genetic tests and the development of public policy

The availability of genetic tests does not necessarily mean that their routine clinical use for diagnostic testing or screening is appropriate. However, rather than a rational analysis of data concerning accuracy, reliability, quality control, acceptable costs and provisions for consumer and professional education, as well as public participation in evaluation issues related to the testing, development of policy for the clinical use of such tests is often driven by consumer demand, legal forces and professional practice (Wilfond & Nolan, 1993). Researchers and clinicians in both genetics and speech and hearing science increasingly recognize the importance of the use of evidence-based guidelines and translation research to address clinical decision making for applications of new technologies or diagnostic techniques that maximize benefits and minimize harm (Burke et al., 2007; Dollaghan, 2004; Khoury et al., 2007; Rosenbek, McCullough, & Wertz, 2004; Yoshinaga-Itano, 2004). Evaluation of genetic tests can be difficult given the variability of such tests in predictive value, the range of technologies used, and the wide range of clinical applications—prenatal diagnosis, screening based on family history or clinical symptoms, population screening based on ethnicity, etc. (Burke et al., 2007).

One example of translation research to evaluate the potential clinical, patient and economic outcomes associated with the use of a genetic test for a communication disorder is a very recent outcomes-based study of a gene which confers susceptibility to aminoglycoside-induced hearing loss (Veenstra et al., 2007). The A1555G mutation and other variants in the mitochondrial 12S ribosomal RNA gene, *MTRNR1* are associated with predisposition to aminoglycoside ototoxicity and/or late-onset sensorineural hearing loss (Pandya, 2007). Hearing loss associated with *MTRNR1* associated aminoglycoside ototoxicity is bilateral and severe to profound, occurring within a few days to weeks after administration of any amount (even a single dose) of an aminoglycoside antibiotic. Penetrance of hearing loss in those who carry the mutation and are exposed to aminoglycosides is reported to be 100% however this estimate is based on limited research data. The test for the A1555G mutation has been clinically available for several years and is directly marketed to clinicians. Some researchers have proposed the addition of a screening test for the A1555G mutation to the

newborn screening panel performed on all babies at the time of birth, along with three other genetic tests for common forms of hearing loss (Morton & Nance, 2006). Among the advantages of this would be the early identification of late-onset or preventable forms of hearing loss.

Veenstra et al. (2007) estimated the incremental cost-effectiveness of screening for the A1555G variant compared with the current standard of care of not screening in a hypothetical population of patients with cystic fibrosis, a group that is commonly exposed to aminoglycosides to treat respiratory infections. This model predicted small improvements in patient outcomes, counteracted by a lack of cost-effectiveness and the possibility of worse patient outcomes due to the avoidance of first-line antibiotic therapy. Based on a few studies, it is known that the prevalence of this mutation is quite low in most ethnic groups. Studies of the association of this mutation with hearing loss in individuals exposed or not exposed to aminoglycosides are lacking. For these reasons, the authors advised caution in both the clinical interpretation of these results and the possible decision to avoid aminoglycoside therapy based on a positive test result. The need for additional research on the prevalence of the mutation, its association with hearing loss and the clinical outcomes of testing for mutations of *MTRNR1* in patients likely to be exposed to aminoglycosides was emphasized.

6. Recent studies of individual and community concerns regarding genetic testing for hearing loss

A number of recent studies provide an opportunity to understand issues that are important to consumers with hearing loss and also serve as a starting point for the process of public education where community concerns related to ethical and legal issues can be addressed. Several studies have documented the interest of hearing and deaf parents in using genetic information for planning purposes, not just for reproductive decision-making (Brunger et al., 2000; Middleton, Hewison, & Mueller, 2001; Steinberg, Kaimal, Bain, Krantz, & Li, 2007; Stern et al., 2002). While parents are often highly motivated to undergo genetic testing once the deafness in their child is identified, it is important that they have appropriate expectations of genetic evaluation and testing (Burton, Withrow, Arnos, Kalfoglou, & Pandya, 2006; Li et al., 2007; Parker, Fortnum, Young, & Davis, 2000); parental frustration and disappointment have been expressed when genetic evaluation and testing leads to lack of a clear diagnosis or cause. Several early studies documented predominantly negative attitudes towards genetic testing by members of the Deaf community, including the belief that genetic testing devalues deaf people (Middleton, Hewison, & Mueller, 1998; Stern et al., 2002). While there has been a very recent trend for increased interest and support for genetic evaluation and testing from the Deaf community, deaf adults have continued to express fear that these advances may be used to eliminate or 'cure' deafness (Burton et al., 2006). Other studies have shown that deaf adults are generally supportive of genetic testing for deafness and support individual choice, including prenatal diagnosis, but only when full information about all relevant aspects of genetic testing are given to potential consumers of those services (Burton et al.; Guillemin & Gillam, 2006). At least one study has examined parental attitudes towards the inclusion of genetic tests for common forms of hearing loss as part of newborn screening done for all babies at the time of birth (Burton et al.). While some parents who already have deaf children support including genetic testing for genes for deafness because it has the potential to identify at-risk children missed by audiologic screening, they also express concerns about cost effectiveness and the appropriate use of limited medical resources. The attitudes and behavior of the deaf with respect to "genotypic mate selection," the selection of a mate on the basis of genotype in order to prevent or ensure the birth of a deaf child, is an emerging issue that may be unique to this group (Burton et al.). All of these studies document the need of health care providers to be aware

of cultural differences and maintain a flexible approach in recommendations for genetic evaluation and testing that is respectful of differing attitudes and motivations.

7. Summary and conclusions

Advances in genetics and genomics will surely result in more widespread development and use of technologies such as prenatal diagnosis and gene therapy for hearing loss as well as other genetically based communication disorders. These applications are not without controversy, at both the societal and individual/family level. Respect for patient autonomy and the right to individual choice is highly valued in the United States and other developed countries, and there appears to be a trend towards a model of genetics focused on individual needs and desires. Technologies which empower individuals with information and choices can also be threatening and have unintended consequences for them and their family members. Adequate understanding of the needs and desires of parents and family members as well as adults who are themselves affected with various communication disorders is essential in order to guide the application of new genetic technologies to clinical practice.

New studies of the ethical and social impact of genetic testing and other technologies for communication disorders are underway. These studies are examining the psychosocial implications and the influence of genetic testing on specific behaviors of individuals with communication disorders and their family members. Other studies of the impact of more widespread use of genetics technologies for population screening for carriers of genes or for those at risk for communication disorders are also warranted. Finally, it is clear that evidence-based guidelines and outcomes-based research are important in determining accuracy, reliability, quality control, and cost-effectiveness of testing. Along with provisions for consumer and professional education, as well as public participation in evaluation of testing, attention to these issues will allow the development of policy for the clinical use of such tests which will maximize benefits and minimize potential harm. The implications of the developments in genetics for the diagnosis and treatment of communication disorders will continue to be wide-ranging and controversial. Integration of genetic tests for communication disorders into clinical care in a responsible manner will require multidisciplinary collaboration between speech and hearing professionals, geneticists and bioethicists. Careful consideration of the ethical, legal and social implications of these tests will be essential.

Appendix A. Continuing education

1. Examples of publicly funded genome projects which included an ELSI component are
 - a. The Human Genome Project and the Diploid Genome Project (HuRef).
 - b. The International HapMap Project and the Diploid Genome Project (HuRef).
 - c. The Genes and Environment Initiative and the Human Genome Project.
 - d. The International HapMap Project and the Human Genome Project.
 - e. None of the publicly funded genome projects included an ethics component.
2. Ethical and legal concerns related to genetic testing which occur at an individual level are
 - a. Privacy and confidentiality of genetic information.

- b. Discrimination in employment and health insurance based on genetic information.
 - c. The potential uses of prenatal diagnosis.
 - d. Use of testing to predict the future onset of diseases for which there may be no prevention or treatment.
 - e. All of the above.
- 3. An example of evidence-based (outcomes-based) translation research for genetic testing of a communication disorder is
 - a. The evaluation research studies of genetic testing for genes which contribute to autism.
 - b. The evaluation of screening for the A1555G variant of the *MTRNR1* gene in a hypothetical population of patients with cystic fibrosis.
 - c. A study of the use of *GJB2* (connexin 26) testing for children with hearing loss.
 - d. A study of genetic testing for specific language disorder.
 - e. Outcomes-based research has yet to be performed for genetic testing in a communication disorder.
- 4. Decisions to introduce genetic tests into clinical practice are usually made based on
 - a. Public participation in evaluation issues related to testing.
 - b. A thoughtful evaluation of accuracy and reliability of the test.
 - c. Provisions being in place for consumer and professional education.
 - d. Consumer demand and legal forces.
 - e. None of the above.
- 5. Studies of individual attitudes and community concerns regarding genetic testing for hearing loss
 - a. Have not yet been undertaken in the United States.
 - b. Have been performed with parents of deaf children, but not with deaf adults.
 - c. Have documented concerns on the part of the deaf community about the development of a 'cure' for deafness.
 - d. Have indicated that parents of deaf children are highly supportive of molecular genetic testing for deafness as part of newborn screening done for all babies at the time of birth.
 - e. Both c and d are true.

References

- American College of Medical Genetics Board of Directors. ACMG statement on direct-to-consumer genetic testing. *Genetics in Medicine*. 2004; 6(1):60. [PubMed: 14726812]
- Arnos KS. The implications of genetic testing for deafness. *Ear & Hearing*. 2003; 24:324–331. [PubMed: 12923423]

- Bishop DVM. The role of genes in the etiology of specific language impairment. *Journal of Communication Disorders*. 2002; 35:311–328. [PubMed: 12160351]
- Brunger JW, Murray GS, O'Riordan M, Matthews AL, Smith RJ, Robin NH. Parental attitudes toward genetic testing for pediatric deafness. *American Journal of Human Genetics*. 2000; 67(6):1621–1625. [PubMed: 11062052]
- Burke W, Zimmern RL, Kroese M. Defining purpose: A key step in genetic test evaluation. *Genetics in Medicine*. 2007; 9(10):675–681. [PubMed: 18073580]
- Burton SK, Withrow K, Arnos KS, Kalfoglou AL, Pandya A. A focus group study of consumer attitudes toward genetic testing and newborn screening for deafness. *Genetics in Medicine*. 2006; 8(12):779–783. [PubMed: 17172941]
- DeStefano AL, Gates GA, Heard-Costa N, Myers RH, Baldwin CT. Genomewide linkage analysis to presbycusis in the Framingham heart study. *Archives of Otolaryngology Head and Neck Surgery*. 2003; 129:285–289. [PubMed: 12622536]
- Dollaghan CA. Evidence-based practice in communication disorders: What do we know, and when do we know it? *Journal of Communication Disorders*. 2004; 37:391–400. [PubMed: 15231419]
- Felsenfeld S. Finding susceptibility genes for developmental disorders of speech: The long and winding road. *Journal of Communication Disorders*. 2002; 35:329–345. [PubMed: 12160352]
- Gates GA, Couropmitree NN, Myers RH. Genetic associations in age-related hearing thresholds. *Archives of Otolaryngology Head and Neck Surgery*. 1999; 125:654–659. [PubMed: 10367922]
- Genes, Environment and Health Initiative. National Institutes of Health.. Determining Genetic and Environmental Roots of Common Diseases. 2006. Available from <http://www.gei.nih.gov/genetics/index.asp>
- Gibson CJ, Gruen JR. The human lexinome: Genes of language and reading. *Journal of Communication Disorders*. 2008; 41:409–420. [PubMed: 18466916]
- Guillemin M, Gillam L. Attitudes to genetic testing for deafness: The importance of informed choice. *Journal of Genetic Counseling*. 2006; 15:51–59. [PubMed: 16525898]
- Hayiou-Thomas ME. Genetic and environmental influences on early speech, language and literacy development. *Journal of Communication Disorders*. 2008; 41:397–408. [PubMed: 18538338]
- Herman GE, Henninger N, Ratliff-Schaub K, Pastore M, Fitzgerald S, McBride KL. Genetic testing in autism: How much is enough? *Genetics in Medicine*. 2007; 9(5):268–274. [PubMed: 17505203]
- Human Genome Project Information. About the Human Genome Project. 2005. Available from http://www.ornl.gov/sci/techresources/Human_Genome/project/about.shtml
- International HapMap Project(a). Available from <http://www.hapmap.org>
- International HapMap Project(b). How Are Ethical Issues Being Addressed?. Available from <http://www.hapmap.org/ethicalconcerns.html.en>
- International Human Genome Sequencing Consortium. Finishing the euchromatic sequence of the human genome. *Nature*. 2004; 431:931–945. [PubMed: 15496913]
- Juengst ET. FACE facts: Why human genetics will always provoke bioethics. *The Journal of Law, Medicine & Ethics*. 2004; 32(2):267–275.
- Khoury MJ, Gwinn M, Yoon PW, Dowling N, Moore CA, Bradley L. The continuum of translation research in genomic medicine: How can we accelerate the appropriate integration of human genome discoveries into health care and disease prevention? *Genetics in Medicine*. 2007; 9(10):665–674. [PubMed: 18073579]
- Kocher A, Hildebrand MS, Smith RJH. Clinical aspects of hereditary hearing loss. *Genetics in Medicine*. 2007; 9(7):393–408. [PubMed: 17666886]
- Lander ES, Linton LM, Birren B, Nusbaum C, Zody MC, Baldwin J, et al. Initial sequencing and analysis of the human genome. *Nature*. 2001; 409:860–921. [PubMed: 11237011]
- Levy S, Sutton G, Ng PC, Feuk L, Halpern AL, Walenz BP, et al. The diploid genome sequence of an individual human. *PLoS Biology*. 2007; 5(10):1–32.
- Lewis, R. *Human genetics: Concepts and applications*. 8th ed.. McGraw-Hill; New York: 2007.
- Li Y, Steinberg AG, Bain L, Yaeger D, Bieler A, Ewing R. Assessing parental attitudes toward genetic testing for childhood hearing loss: Before and after genetic consultation.. *American Journal of Medical Genetics. Part A*. 2007; 143(A):1546–1553.

- McGuire AL, Cho MK, McGuire SE, Caulfield T. The future of personal genomics. *Science*. 2007; 317:1687. [PubMed: 17885116]
- McGuire AL, Gibbs RA. Currents in contemporary ethics: Meeting the growing demands of genetic research. *The Journal of Law, Medicine & Ethics*. 2006; 34(4):809–812.
- Manning M, Hudgins L. Use of array-based technology in the practice of medical genetics. *Genetics in Medicine*. 2007; 9(9):650–653. [PubMed: 17873654]
- Middleton A, Hewison J, Mueller RF. Attitudes of deaf adults toward genetic testing for hereditary deafness. *American Journal of Human Genetics*. 1998; 63:1175–1180. [PubMed: 9758618]
- Middleton A, Hewison J, Mueller RF. Prenatal diagnosis for inherited deafness—What is the potential demand? *Journal of Genetic Counseling*. 2001; 10(2):121–131. [PubMed: 11767801]
- Morton CC, Nance WE. Newborn hearing screening—A silent revolution. *New England Journal of Medicine*. 2006; 354(20):2151–2164. [PubMed: 16707752]
- National Human Genome Research Institute. National Institutes of Health. Available from <http://www.genome.gov>
- National Human Genome Research Institute. National Institutes of Health.. Direct to Consumer Marketing of Genetic Tests. 2004. Available from <http://www.genome.gov/12010659>
- National Human Genome Research Institute. National Institutes of Health.. Policy & Ethics: Critical issues and legislation surrounding genetic research. 2007. Available from <http://www.genome.gov/PolicyEthics/>
- Pandya, A. [November 11, 2007] Nonsyndromic hearing loss and deafness, mitochondrial. 2007. from GeneTests, GeneReviews Web site: <http://www.genetests.org>
- Parker MJ, Fortnum HM, Young ID, Davis AC. Genetics and deafness: What do families want? *Journal of Medical Genetics*. 2000; 37(E26)
- Robey RR. A five-phase model for clinical-outcome research. *Journal of Communication Disorders*. 2004; 37:401–411. [PubMed: 15231420]
- Rosenbek JC, McCullough GH, Wertz RT. Is the information about a test important? Applying the methods of evidence-based medicine to the clinical examination of swallowing. *Journal of Communication Disorders*. 2004; 37:437–450. [PubMed: 15231424]
- Service RF. Public project gears up to chart the protein landscape. *Science*. 2003; 302:1316–1318. [PubMed: 14631011]
- Steinberg AG, Kaimal G, Bain L, Krantz I, Li Y. Parental narratives on genetic testing for children with hearing loss: A qualitative inquiry. *American Journal of Medical Genetics. Part A*. 2007; 143(14):1533–1545. [PubMed: 17542005]
- Stern SJ, Arnos KS, Murrelle L, Welch KO, Nance WE, Pandya A. Attitudes of deaf and hard of hearing subjects towards genetic testing and prenatal diagnosis of hearing loss. *Journal of Medical Genetics*. 2002; 39(6):449–453. [PubMed: 12070258]
- Tabor HK, Cho MK. Ethical implications of array comparative genomic hybridization in complex phenotypes: Points to consider in research. *Genetics in Medicine*. 2007; 9(9):626–631. [PubMed: 17873651]
- Veenstra DL, Harris J, Gibson RL, Rosenfeld M, Burke W, Watts C. Pharmacogenomic testing to prevent aminoglycoside-induced hearing loss in cystic fibrosis patients: Potential impact on clinical, patient, and economic outcomes. *Genetics in Medicine*. 2007; 9(10):695–704. [PubMed: 18073583]
- Venter JC, Adams MD, Myers EW, Li PW, Mural RJ, Sutton GG, et al. The sequence of the human genome. *Science*. 2001; 291:1304–1351. [PubMed: 11181995]
- Welch KO. Fundamentals of human genetics. *Seminars in Hearing*. 2006; 27:127–135.
- Wilfond BS, Nolan K. National policy development for the clinical application of genetic diagnostic technologies. Lessons from cystic fibrosis. *Journal of the American Medical Association*. 1993; 270(24):2948–2954. [PubMed: 8254856]
- Yoshinaga-Itano C. Levels of evidence: Universal newborn hearing screening (UNHS) and early hearing detection and intervention systems (EHDI). *Journal of Communication Disorders*. 2004; 37:451–465. [PubMed: 15231425]

Zhu M, Yang T, Wei S, DeWan AT, Morell RJ, Elfenbein JL. Mutations in the γ -actin gene (ACTG1) are associated with dominant progressive deafness (DFNA20/26). *American Journal of Human Genetics*. 2003; 73:1082–1091. [PubMed: 13680526]

\$watermark-text

\$watermark-text

\$watermark-text