Chapter 10

THE ROLE OF GENETIC COUNSELING

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THE SCOPE AND PROCESS OF GENETIC COUNSELING

Genetic counseling is a communication process, which aims to help individuals, couples, and families understand and adapt to the medical, psychological, familial, and reproductive implications of the genetic contribution to specific health conditions. This process integrates:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence,
- Education about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources, and research,
- Counseling to promote informed choices in view of risk assessment, family goals, and ethical values, and
- Support to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (National Society of Genetic Counselors [NSGC], 2006).

Genetic test options and results can seem quite complex and emotionally charged for the average person; therefore, this type of information must always be presented in a supportive and nonjudgmental manner and be relevant and responsive to the needs of the family/individual. The primary goal of genetic counseling is to facilitate the ability to use genetic information in a personally meaningful way that minimizes psychological distress and increases personal control (Biesecker & Peters, 2001). A genetic diagnosis as a result of newborn screening has significant implications for the couple's baby, their future pregnancies, and for the risk status of close relatives. The process of genetic counseling for a single positive newborn dried bloodspot screening (NDBS) result may involve multiple phases, including preclinic contact with individual/families, clinical consultation, additional testing, follow-up, and review. It may extend to consultation with other “at risk” relatives or coordination of tests in future pregnancies. It also entails confidential record keeping and may require extensive literature and database searches and/or liaison with laboratories or genetics units in other states or countries.

Genetic counseling, as a health care profession, is a relatively young discipline and formal registration has not yet been achieved in many states or countries. However, best practice guidelines and codes of ethics, which are shared by many national Human Genetic Societies (Table 10-1), aim to encourage and
Table 10-1. Human Genetics Societies

<table>
<thead>
<tr>
<th>Country/Region</th>
<th>Professional Body</th>
<th>Acronym</th>
<th>Year Est.</th>
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<tr>
<td>Canada</td>
<td>Canadian Association of Genetic Counsellors (Association Canadienne des Conseillers en Génétique) Canadian College of Medical Geneticists (Collège Canadien de Généticiens Médicaux)</td>
<td>CAGC</td>
<td>1987</td>
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<tr>
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<tr>
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Maintain consistency of practice standards among genetic health professionals in any particular country. Apart from ensuring high-quality health service delivery, adherence to uniform practice guidelines also ensures that relatives of the same family who are seen in different genetic units for counseling and/or cascade genetic testing within a country will receive comparable service, access to testing and support. Many health professionals may be involved in the provision of genetic counseling, particularly for complex genetic conditions that require a multidisciplinary focus; however, the core provision of genetic counseling should be conducted by appropriately trained professionals in order to maintain these high standards and provide optimal family support. In Australasia, Canada, the United Kingdom, and the United States, fully qualified genetic counselors have completed a professional, board approved, postgraduate degree program followed by a certification process. In many other countries, approved pathways to become a recognized, fully qualified genetic counselor are currently under development. In most regions, genetic counselors practice in a team setting with clinical geneticists.
or other medical specialists. However, in the United States, which has the widest range of employment opportunities for genetic counselors, they may work independently in private practice or in less traditional settings.

It should be noted that as genetic services may be administered by either public or private health facilities and funding for genetic tests may depend on government support, institutional policy, or individual insurance policies, access to clinical genetics services will vary widely between states and countries. In some cases, this may limit the availability of genetic counseling services for family support and access to cascade testing. Public awareness of genetic counseling services in some regions also may be limited; thus, individuals might not proactively seek such support if it is not offered routinely after a NDBS positive result. In some regions, the NDBS program employs a genetic counselor designated to provide follow-up education and support for all positive screen results. Other regions have a system of referral for genetic counseling follow-up as deemed necessary. Newborn hearing screening (NIHS) programs are more likely to utilize the latter system, as not all congenital hearing loss is due to genetic cause. It is most appropriate to refer an individual/family for genetic counseling following a positive NDBS result or laboratory diagnosis of a genetic cause of congenital hearing loss. Thus, these scenarios are usually included in the listed indications for referral for most genetics services.

ELEMENTS OF THE GENETIC COUNSELING PROCESS (ADAPTED FROM HUMAN GENETICS SOCIETY OF AUSTRALASIA [HGSA], 2008)

Referral

This usually will involve a referral letter or phone call from the referring health professional or the individual seeking counseling. Written referrals are preferred, as this facilitates continuity of care following the consultation, although verbal and self-referrals may be acceptable to some genetic services. Referrals are assessed regarding their urgency and appropriateness, and appointments arranged, as required.

Preconsultation Contact

The genetic counselor will often contact the referred individual by telephone prior to the scheduled clinic appointment. The purpose of this initial contact is to confirm appointment details and identify who is planning to attend (i.e., individual and/or family members). The genetic counselor will outline the process and aims of the consultation, addressing any concerns that the individual may have regarding the nature of the consultation, ascertain the needs and expectations of the individual or family (their agenda), as well as identify any special requirements (e.g., wheelchair access or interpreter services) or any social and/or cultural issues which may impact on the consultation. Research on counseling after NDBS positive result for cystic fibrosis demonstrates that exploring the individual/family agenda and matching counseling to those expectations will reduce parental stress, whereas ignoring parent preferences will increase anxiety about their infant’s well-being (Tluczek et al., 2006). At this time, the genetic counselor also may gather standardized pedigree information (Bennett, 1999; Bennett et al., 2008; Bennett, Steinhouse, & Ulrich, 1995) and determine additional requisite documentation, such as hard-copy confirmation of pathology results and whether signed consent forms will be required to facilitate release of the information. The genetic counselor also will clarify any unrealistic expectations of the clinical consultation.

The Consultation

The physical setting for the genetic counseling consultation is important. The consultation room should ensure total privacy for the individual or family, as the matters discussed are confidential. There should be a limit on the number of professionals present during the consultation. The development of rapport with the individual or family is paramount for building the trust relationship that will encourage full disclosure and open discussion (Weil, 2000, pp. 96–97) and should not be compromised by the educational
needs of professionals and trainees. Consent should be obtained from the individual or family for other health professionals, such as students and trainees, to be present at the consultation. The following elements of the counseling process may be addressed over more than one consultation. Discussion must use language and explanations that can be easily understood by the individual and interpreters must be utilized if there is any concern about the individual’s ability to cope with in-depth discussion in the oral language of the genetic counselor.

Confirmation of the Individual/Family and Counselor Agendas. The agenda for the consultation should be set early, taking into consideration the needs and expectations of the individual or family, as well as the essential responsibilities of the professional. It is important for the counselor not to proceed with her or his own agenda without establishing the individual’s, couple’s, or family’s pre-existing knowledge and perceptions.

Personal Information Management. The individual or family should be informed about the possible uses of their information including their right to access it and that the information may be given to another medical practitioner in the course of their management (if relevant). Although local health service requirements for record keeping must be observed, clinical genetics units usually maintain individual or family files that are separate from records of the institution to which they are attached. This is due to the highly confidential nature of genetic information, the need to maintain the record for future generations, the aim to have a complete, comprehensive file for each family, and for potential access to the records (under appropriate consent conditions) by health professionals caring for extended family members in other regions or countries. In the context of newborn dried bloodspot screening and genetic testing, stored bloodspot samples (Guthrie cards) also can be considered part of the individual’s personal health information and will be stored for varying lengths of time according to local law. All health records are confidential and subject to relevant professional standards or legislative requirements related to privacy and release of information. Federal and local government privacy laws/codes may vary between regions and disclosure of confidential information is permitted only as consented to by the individual and/or required for medical management or as permitted/required by law.

Gathering Specific Health Information. Information may be gathered in several ways. The construction of the standard minimum three generation family pedigree (Bennett et al., 1995; Bennett et al., 2008) is an important part of the genetic counseling process, and as family information can be highly sensitive, great care needs to be taken to maintain confidentiality. Legally, health professionals are entitled to record a family medical history if it is necessary for the care of the individual or family. The genetic counselor is always mindful of the private nature of the family pedigree and the possibility that not all related family members have the same knowledge of genetic relationships within the extended family. For this reason, the genetic counselor usually will record the pedigree from each related person who is subsequently referred for counseling or cascade testing. Ethnic ancestry often is important in the determination of additional tests to be considered. Details such as undisclosed adoption, half-siblings, consanguinity, assisted conception using donor gametes, and misattributed paternity are vital for accurate genetic risk assessment but also may create a confidentiality dilemma for the genetic counselor in consultation with various relatives. Information about the individual or family medical history may also be accessed via the referring doctor and hospital/medical records. Results of previous tests and other investigations also may be accessed via the referring doctor, hospital/medical records, or directly from the laboratory.

Verification of the Diagnosis. This may be achieved via information obtained before or during the consultation. Information gathering that requires access to pathology/postmortem reports or genetic test results of relatives will need consent from the appropriate individuals, or from the next-of-kin or legally responsible person/agency.

Education About the Condition. Education includes discussion about the cause of the condition, pattern of inheritance, natural history, potential complica-
tions, and treatment options. All health professionals have a responsibility to provide the most up-to-date information. Information about genetic conditions should be given in a balanced manner, which not only lists the medical challenges but also raises the possible range of prognosis, the potential for good quality of life, and positive capabilities of an individual with the condition. The individual’s understanding should be assessed throughout the consultation, particularly before moving on to increasingly complex concepts. It also is important to provide support in the form of linguistically and culturally appropriate educational resources that will aid consolidation of understanding over time.

Discussion of the Medical, Emotional, and Social Implications. Genetic information often has a profound impact on an individual and his or her family. This should be acknowledged in the counseling process and individuals must be given a safe environment in which to express emotional or psychological responses. Some will experience guilt because they have passed on a genetic mutation or may inappropriately direct anger or blame towards family members. Others may repress their reactions, project emotions onto other family members, or focus only on intellectual aspects as a way of avoiding their own psychological response (Weil, 2000, pp. 8–12). As well as dealing with genetic information, the genetic counselor will be empathic in response to any distress, will normalize the individual’s reactions and work to increase his or her sense of personal control. This comprehensive support can assist individuals/families to work through these reactions as they go through the initial period of adjustment toward acceptance of the revealed genetic status and will encourage coping in an empowered manner.

Education About Recurrence Risk/Carrier Status. Once a genetic diagnosis is made in a family, the individual risk for carrier status and recurrence risk for offspring of the same couple can be estimated using empirical data, inheritance patterns, pedigree information, clinical expertise, and test results. As mentioned previously, confirmation of the veracity of genetic relationships within the family is paramount. When the genetic counselor has developed a good rapport with the individual it is more likely that this trust relationship will facilitate disclosure of accurate private information. The counselor should also ensure that the individual or family understands the risk figures, which can be presented in a variety of formats, feels empowered to make their own meaningful interpretation of high versus low risk and understands the limitations of such risk calculations.

Discussion of Implications for Genetic Relatives. A genetic diagnosis or positive newborn screening (NBS) test result may have direct risk implications for other family members. Genetic counseling should be made available to those relatives deemed to be “at risk” of being a carrier of a specific genetic mutation, as appropriate. Some individuals will require support and assistance in conveying genetic information to “at risk” relatives, either because of their lack of confidence about explaining genetic information or due to poor family dynamics and/or communication patterns. The genetic counselor can provide an explanatory letter for distribution or offer their genetic service contact details for dissemination throughout the family.

Presentation of Options. Options may include offer of additional genetic testing and discussion of future reproductive options. Additional genetic tests following a positive NDBS result might include additional DNA analysis to identify a rare mutation that was not included as part of the newborn screening panel, or mutation testing for the parents. Whether or not the individual elects to have any genetic test will depend on multiple factors. In general, the types of genetic tests that may be available include carrier, screening, diagnostic, predictive, presymptomatic, and donation of samples for research purposes. However, only some of these will be appropriate following particular NDBS positive results (refer to Appendix 10-A for further explanation of the various classes of genetic tests). After discussion of NDBS results and implications for the family, the individual’s/couple’s values, goals, and needs also should be explored. The individual should be informed of the reason that the test might be indicated, the potential information to be gained by taking the test, the nature of the sample required, the limitations of the test result interpretation, and the possible implications of the result. As DNA analysis is not a routine blood test, it is important to
give a realistic estimate of how long it will take to receive results, as well as an accurate estimate of any associated financial cost to the individual. At this stage, it also is important to discuss any ethical issues that arise (see Discussion of Ethical Challenges section later).

**Discussion About the Potential Interests of Third Parties.** Genetic information, if given to other family members or outside agencies such as insurers or employers, has the potential to result in discrimination and stigmatization so this issue is discussed during the consultation. Third parties outside the family who may have an interest in genetic test results/diagnoses may include life insurance companies, health insurance companies, and employers. Where insurance companies may have an interest, it is important to explain to the individual or family (in advance of the consent process) the common requirement to disclose to these companies, at the time of application, the genetic diagnosis, or genetic test result. Health professionals must comply with current federal and/or local government privacy laws at all times and may disclose genetic information only as documented by the individual during the informed consent process (signed and witnessed) and/or required for medical management or as permitted or required by law. The genetic counselor will provide support for determining and conveying the appropriate amount of genetic information to “at risk” relatives.

**Support for Informed Decision-Making.** It is essential that informed decision-making occurs in the context of a nonjudgmental and noncoercive approach. It is most important to confirm the individual’s comprehension throughout the consultation. In order to make an informed decision, there must be good comprehension of the concrete information, all available options must have been presented without bias, the advantages and disadvantages of each option should be discussed not only with the genetic counselor but also with significant others, which may include the individual’s partner, close friend, extended family or primary care provider, and the individual’s perception and acceptability of potential consequences of each option should be explored. The individual or family’s well-deliberated, informed decision should be respected.

**Coordination of Genetic Testing, Delivery, and Explanation of Results.** At the time of consent to the test, the genetic counselor and individual or couple should agree on a contract for delivery of the results, that is, which health care professional will be responsible, when, where, and how they will be delivered, and who else may receive the result. The appropriate request forms should be completed and a system of follow-up or tracking of outstanding results must be set in place for efficient reporting and action. Information about test results should be clearly explained to the individual/couple using language and explaining concepts in a way that they are able to comprehend. The counselor must confirm the individual’s comprehension of the test result.

**Contact Details for Community-Based Support.** Thanks to the Internet, it is now possible to find and access support groups for quite rare genetic conditions. However, this increased availability also raises the danger of accessing inaccurate, outdated information on natural history and single case accounts that do not realistically represent the specific condition. It is important that the individual or family understands the inherent bias toward worst and best case scenario reporting and receives some guidance about reputable support groups and Web sites. Some newborn screening systems have a Web site that includes direct links to reputable support and information services. Email and online discussion boards allow families to communicate with others who are in a similar situation while protecting their own privacy. Genetic counselors can arrange contact with community-based support groups or appropriate individuals or families according to the individual or family’s requirements.

**Postconsultation**

The genetic counseling service does not necessarily conclude at the end of a single consultation. Many of the following tasks are considered standard practice.

**Summary Letter.** It is customary for many genetic services to send the individual/couple a letter that summarizes the most important information discussed in the consultation. This practice has developed in response to research that has shown that the average person finds genetic concepts and tech-
nology too difficult to remember; even if they feel that they understood them during the consultation. The summary letter provides an easy reference which serves as a plain language explanation that can be shared with the extended family.

Ongoing Support. It often is not possible to cover all of the necessary information and thoroughly explore psychosocial responses in a single session. Thus, an offer of follow-up, either in another consultation or by telephone might be important to adequately address agendas of both counselor and referred individual. Individual or family follow-up is an essential component of the genetic counseling process. With follow-up, the counselor can ascertain that the individual or family has understood the information, create an opportunity to address any new questions, as well as provide additional psychosocial support to the individual or family. The individual and extended family members should also feel welcome to E-mail or telephone with new concerns as they arise.

Liaison with Relevant Health Care Professionals. If a formal referral has been received, the genetic counselor will send the referring health professional a letter that briefly notes the consultation details as well as any decisions made about additional testing. A copy of the genetic counseling summary letter sent to the referred individual may also be included for his or her medical record. The genetic counselor may facilitate referral, personally or via the referring medical practitioner, of an individual or family for treatment or ongoing clinical management and/or emotional and psychological support. With individual/family consent, copies of relevant correspondence and reports should be sent to the individual’s other current health care providers.

ROLE OF THE GENETIC COUNSELOR IN NEWBORN SCREENING

In many cases an infant diagnosed with a genetic condition as a result of a newborn screening system will be referred to a specialist clinic for ongoing management by a multidisciplinary team. Some teams will include a genetic counselor, whereas others will adhere to a protocol that recognizes the need to refer parents for genetic counseling support around the time of diagnosis as well as refer the affected individual to a genetic counselor as they approach reproductive age or express interest in understanding the genetic cause of the condition and implications for future offspring (Comeau et al., 2004; Comeau et al., 2007; Robinson & Rowell, 2008; Southern et al., 2007). However, coordinated approaches are not universal, and in some settings, a variety of health professionals assume the role of counseling parents. In some regions, responsibility for follow-up and coordinating care for the diagnosed infant may fall to the primary care physician who may not feel adequately prepared for this task (Kaye et al., 2007; Kemper, Uren, Moseley, & Clark, 2006). Programs might rely on the use of fact sheets; however, documentation of provision of written information alone certainly does not provide evidence of adequate understanding in the recipient (Clayton, 2005) and research has identified the importance of addressing parental psychosocial needs alongside genetic education (Clayton, 2005; Skirton, 2001; Tluczek et al., 2006). In general, there has been very little attempt to conduct quality assurance auditing of the counseling provided in association with newborn screening systems, which means that, in some areas, families might be less than adequately educated and supported after a positive finding on newborn screening (Ciske, Haavisto, Laxova, Rock, & Farrell, 2001; Farrell, Certain, & Farrell, 2001; Farrell, Deuster, Donovan, & Christopher, 2008; La Pean & Farrell, 2005; Lafayette, Abuelo, Passero, & Tantravahi, 1999; Wilford & Gollust, 2005).

It is just as important for the parent(s) and relatives of babies determined to be unaffected carriers of an autosomal recessive mutation to receive comprehensive genetic counseling. It has been estimated that in the United States, there are about 12,000 false positive results for each 1,000 infants diagnosed with a metabolic disorder by newborn screening systems (Waisbren, 2006). Research has identified a risk of increased parental stress and impaired infant-parent relationship associated with false positive newborn screening results, but these untoward effects can be ameliorated by improved communication at the time of detection (Gurian, Kinnamon, Henry, & Waisbren, 2006). Thorough education and counseling about the implications of carrier status will reduce inappropriate anxiety
about the health impact of carrying a single nonfunctioning autosomal recessive gene and should ease the guilt that some parents experience over passing on their nonfunctioning gene. Because the infant’s health and well-being has been questioned, naturally this issue is the parents’ primary focus. Once it becomes clear that there is no immediate health risk to their child, parents of carrier infants sometimes fail to recognize or remember the potential importance of cascade testing to clarify their risk in future pregnancies and/or the risk status of their close relatives (Kladny, Gettig, & Krishnamurti, 2005). It appears that the majority of individuals/families directly offered an opportunity for specialized genetic counseling after NBS detection do accept this option (Comeau et al., 2004; Kladny et al., 2005). On the matter of advising relatives about potential carrier status, many individuals report concern about family reactions or are reluctant to initiate conversations about issues that they consider to be emotionally charged and complex. However, there is no evidence that communicating information about carrier risk to other family members impairs relationships and it could be argued that nondisclosure has more potential to cause family discord and harm than disclosure of risk status.

Individuals with a sound understanding of the risk of recurrence and implications for other family members are more likely to disseminate this information proactively, which increases awareness of cascade testing and elective prenatal testing or preimplantation genetic diagnosis (PGD) in future pregnancies of those couples identified to be “at risk” for an affected child. It has been noted that communication coaching and psychosocial support around the issue of disclosure to relatives can make the task less burdensome for the proband family and facilitate wider dissemination (Gaff et al., 2007). A decline in birth rate of infants affected by cystic fibrosis has been observed since the introduction of genetic testing and NDBS (Dankert-Roelse & Mérelle, 2005; Dupuis, Hamilton, Cole, & Corey, 2005).

It is often claimed that other health care professionals are capable of taking responsibility for the various tasks that comprise the role of the genetic counselor. This is not disputed; however, it is rare for a single professional to routinely undertake all of these tasks or complete them all with the same level of expertise. Individual or family support after delivery of newborn screening results can be a time consuming and challenging task and it is often devolved to a number of different health care professionals, often without reference to a standardized protocol, which increases the chance that support might be suboptimal for some families. It also has been noted that some health care professionals may not have had sufficient counseling training to adequately assess comprehension (Farrell & Kuruvilla, 2008) or recognize the importance of the psychosocial impact of familial genetic information (Freyer et al., 1999). Accurate risk assessment for members of the extended family is vital but many health professionals lack the expertise or confidence to proactively investigate important aspects of a genetic history and family pedigree, such as ethnicity, consanguinity (Bishop, Metcalfe, & Gaff, 2008; Maradicgue & Edwards, 2006), and conception with donor gametes. Advanced genetic technology, such as simultaneous multigene mutation detection by microarray in infants with sensorineural hearing loss (Gardner et al., 2006), or expanded DNA cystic fibrosis mutation panel analysis by multiplex polymerase chain reaction (PCR), are not easily understood or well explained by many health care professionals. Though the technology tends to become less complicated for the scientist and less costly for health care providers and consumers over time, it can yield increasingly complex results, which sometimes are equivocal or have variable diagnostic power and/or significant limits to interpretation, so it is imperative that these results are delivered in accurate yet simple terms to the family. Individuals and families are best served by receiving explanation of these results in the context of full genetic counseling support.

Genetic counselors are allied health professionals who are specially trained to support and empower individuals and families during the difficult journey of learning and adaptation that must occur after a positive result on NDBS. Not only do they provide thorough education of the relevant scientific and medical information and test options in plain language but they also attend to the psychosocial impact that inevitably accompanies such issues. Over the last few decades, the concept of perceived personal control (PPC) has been recognized as an important aspect of adaptation in response to health
challenges. PPC has been defined as, "the belief that one has at one’s disposal a response that can influence the aversiveness of an event" (Thompson, 1981). Research has demonstrated that developing a positive PPC is an important coping strategy, which enables enhanced self-management of health threats and improved mental and physical health outcomes (Berkenstadt, Shiloh, Barkai, Kattelson, & Goldman, 1999). It is evident that the psychosocial issues surrounding genetic diagnosis and heritability of causative gene mutations are associated with a sense of loss of control. The three dimensions of PPC are central to the goals of genetic counseling, that is, the consultation process aims to build the individual’s ability to mentally process and cope with the perceived threat of a genetic condition (cognitive control); it aims to present opportunities to alter the physical characteristics of the stressful event, thereby altering its actual threat in the future (behavioral control); and it aims to foster the individual’s confidence to choose among options in a manner that reduces the likelihood of decisional regret (decision control) (Wang, Gonzalez, & Merajuer, 2004).

Since Berkenstadt et al. (1999) proposed the value of using PPC questionnaires to evaluate client satisfaction with genetic counseling consultation, several researchers have determined that professional genetic counseling is correlated with a significant increase in PPC (Berkenstadt et al., 1999; Lipinski, Lipinski, Biesecker, & Biesecker, 2006; Payne et al., 2007; Smets, Pieterse, Aalfs, Ausems, & van Dulmen, 2006). The genetic counselor's focus on development of rapport with, and empowering of, the individual establishes a trust relationship that fosters an increased capacity and willingness to communicate sensitive information that often is relevant to accurate risk assessment (Davey, Rostant, Harrop, Goldblatt, & O’Leary, 2005; McConkie-Rosell & Sullivan, 1999).

When a genetic counselor takes responsibility for educational and psychosocial support post-newborn screening, follow-up will be conducted in a coordinated manner, which provides the individual and their extended family with a single reference point and this facilitates information consistency, comprehensive understanding, enhanced family communication, and equitable access to high standards of supportive health care. Genetic counselors can also enhance communication and sharing of knowledge between multidisciplinary health care providers, many of whom were trained before genetics had any significant impact on clinical care. This clearly is of increasing importance as genetic science further translates into primary care and specialist medicine (Drury, Bethea, Guilbert, & Qureshi, 2007; Gaff, Williams, & McInerney, 2008; Puryear, Watson, Mann, Strickland, & van Dyck 2006; Thompson, Ahrens, LeRoy, Brown, & Berry, 2005).

Optimal outcomes for newborn screening systems are achieved by protocols that ensure appropriate family communication and adequate genetic counseling alongside best practice methods of analysis and follow-up medical care provided by specialized multidisciplinary teams (Therrell, Lloyd-Puryear, & Mann, 2005).

**EXPANDED NDBS FOR INHERITED DISORDERS: ETHICAL CHALLENGES**

There have been some ethical concerns raised about expanded NDBS. These are based on concerns about the potential to cause more harm than good in some specific situations, the storage and secondary uses of samples, and the process of informed consent.

The clinical and ethical justification of newborn screening systems rest on the principles that:

- there exists a suitable, accurate, and reliable test for the condition,
- there is benefit to the baby, family, and society from early diagnosis,
- the benefit is reasonably balanced against financial and other costs, and
- there is a satisfactory system in operation to deal with diagnostic testing, counseling, treatment, and follow-up of patients identified by the test (Holland, Stewart, & Masseoia, 2006; HGSA, 2004).

Newborn screening systems are designed to identify neonates with a range of medical conditions before the onset of adverse clinical symptoms, so that they can benefit from protective strategies or health surveillance to detect early symptoms and initiate treatment. There is little doubt that the
families of children affected by rare, severe, but treatable, genetic disorders benefit from early detection by newborn screening. Not only can parents expect an improved prognosis for the child, but they also can make reproductive decisions in a timely manner; whereas, in the past, a second affected child may have been born before the elder sibling’s diagnosis. Early genetic diagnosis reduces the number of specialist referrals and investigations that the infant (or child) must endure, and reduces the sometimes protracted period between observation of clinical symptoms, diagnosis, and subsequent implementation of treatment, a time which entails significant anxiety for parents. Additionally, universal state or nationally funded screening systems also reduce the diagnostic delays that so often are related to a child’s ethnicity, family poverty, and limited access to health care services (Farrell & Farrell, 2005).

While cystic fibrosis is considered one of the more common genetic disorders, most of the conditions included in NDBS are rare; that is, most have a birth incidence of less than 1 in 50,000. Although there is reasonable certainty that individuals who carry two recognized disease-causing mutations for conditions such as phenylketonuria (PKU) or cystic fibrosis in fact will develop a degree of clinical disease, the prognosis for some other metabolic genetic disorders is much less certain. One of the ethical problems raised by an expanded newborn screening protocol is that it creates a capacity to diagnose some infants, who therefore will have a recognized but unquantifiable risk for conditions that also may have a variable age of onset. In the first large published series of results from the use of tandem mass spectrometry in NDBS, it was observed that additional cases (above the usual clinical rate of detection of these rare disorders) have been genetically diagnosed (Wilcken, Wiley, Hammond, & Carpenter, 2003). What is not clear is whether these infants actually will develop clinical disease or not, given that in one instance the gene positive 5-year-old sibling of one NBDS diagnosed infant had no clinical symptoms despite an expected onset of disease in infancy for that particular condition (Holtzman, 2003). As has been noted, phenotypes are invariably broadened when defined by laboratory criteria rather than clinical evaluation. Therefore, one should be cautious about treating all genetically diagnosed individuals with a therapy that was developed for the clinically defined cohort. The test-defined cohort may contain normal individuals who do not require the therapy and may even be harmed by it (McCabe & McCabe, 2008).

Studies confirm that there is potential for anxiety and grief reactions in association with genetic diagnosis of an infant who is well, and it has been suggested that this might place families “at risk for impaired parent-child bonding, disrupted relationships, personality problems, and the development of psychogenic symptoms or some variant of the vulnerable child syndrome” (Farrell & Farrell, 2003, p. 710). The imposed suspicion of health threat can impact parental perceptions of the child’s health, and parent-child interactions, and negatively influence the associated health monitoring behaviors of the family. Parents also have noted concern that the knowledge of the genetic diagnosis might impact their patterns of discipline and interactions with the diagnosed child as compared to his or her siblings (Campbell & Ross, 2003). Although these are possibilities, only research at various time frames beyond implementation will confirm whether these indeed are significant risks to be weighed against the benefits of expanded NDBS.

NDBS systems also have the potential for incidental detection of carriers for some of the target genetic conditions. For ethical reasons, the testing of minors (individuals under the age of legal adulthood) for carrier status generally is considered inappropriate in other genetic testing settings, but it is accepted as an unintended consequence of the NDBS. As the very young minor cannot give consent for this type of test, there is a concern about his or her loss of autonomous choice as an adult to know or not know this information, and concern that it might have an impact on self-esteem (Borry, Nys, & Dierickx, 2006). Knowledge of carrier status has no clinical utility for the infant but may lead to cascade testing of parents and other relatives in order to clarify their reproductive risks. These concerns have been raised as a result of studies which have investigated the emotional consequences of knowledge of CF carrier status in adults and found that carriers expressed more negative feelings than noncarriers, and that they believed there was a degree of social stigma associated with CF carrier
status (Gordon et al., 2003). There also have been older studies that confirm the association of elevated anxiety in response to knowledge of carrier status, even several years beyond the time of confirmation of carrier status (Marieau, 1992). As long as there is a supportive protocol that ensures provision of appropriate genetic counseling to clarify the significance of carrier status, then it could be expected that the likelihood that harm will result from this revelation will be reduced. It also may transpire that with greater knowledge of carrier status for increasing numbers of individuals, it becomes more commonplace, and thus less personally threatening.

For childhood onset metabolic conditions, for which there is currently no reliable evidence of effective treatment, rationalization for early detection rests on the justification of the elucidation of each condition's clinical natural history to detect earliest symptoms and to develop or improve treatments so that they are more efficacious. Early identification of affected offspring is also considered useful because this will allow parents to modify parental employment situations, plan access to support and financial resources, and prepare family members for adjustment, as well as enable informed reproductive decisions in future pregnancies (Campbell & Ross, 2003). It has been postulated that for an untreatable disorder of childhood, in the absence of both benefit and harm to the diagnosed infant, a significant benefit to the family is considered to satisfy the principles of newborn screening (Wicken, 2003).

The number of conditions currently included in standard NDBS varies from state to state and from country to country, as does the length of time the blood sample cards are stored. The Guthrie card, which is used to collect blood samples from neonates between 24 to 72 hours after birth, is usually considered to be a health record, and as such is stored and treated with the same level of confidentiality and security as other medical records. However, because the card contains the residual dried bloodspot samples, including DNA which can be extracted and analyzed, in many regions its legal status is unclear.

Many U.S. states have mandated NDBS (Baily & Murray, 2008), but this is not the case in other countries (HGSA, 2004). Not all NDBS programs include a requirement for fully informed consent and this raises some specific ethical concerns (Baily & Murray, 2008). In the absence of pretest education, most parents are likely to assume that the sample is simply used for the panel of NDBS tests and then discarded, but this is rarely true. In addition to their primary use, in Australia at least, the Guthrie sample cards also might be utilized for:

- confirmation of laboratory normal ranges (using anonymized samples),
- modification of existing screening tests (using either anonymized or identified samples),
- development of new screening tests (using either anonymized or identified samples),
- epidemiologic or public health research (using anonymized samples),
- testing of deceased members of a family if a specific disorder is suspected or known (using identified samples), and
- as evidence in legal and forensic investigations (using identified samples) (HGSA, 1999).

In the past, there have been several examples of these retained pathology samples being released by legal order of law enforcement, without the consent of the individuals, for investigative purposes other than those for which they were collected (Skene & Bankier, 2004). An American study determined that several screening facilities would provide insurance companies, employers or law enforcement agencies with the samples without seeking consent from the relevant individuals or family members (Elkin & Jones, 2000). However, by most ethical interpretations, this would be a clear breach of confidentiality and lack of respect for the individual's rights (O'Neill, 2001).

In fact, unless legislation specifically allows otherwise, all potential secondary uses of the Guthrie samples are ethically unacceptable unless the parent has been informed of these possibilities before consenting to sampling of their offspring. So, it has been noted that newborn screening programs have an obligation to prohibit the release or use of identified samples after completion of the agreed screening tests, unless written parental or
individual consent is secured (Wilcken, 2003). A study of parental attitudes towards newborn screening confirmed that some individuals were even concerned about unauthorized use of the samples for research and felt that they should be advised of potential additional uses before their consent was sought (Campbell & Ross, 2003).

The requirement for fully informed consent specifically assists in the protection of the recognized human rights of autonomy, personal freedom, and self-determination (Neale, 2000). Fully informed consent usually assumes full disclosure of the relevant information, sufficient time for the individual to discuss, question, and consider the personal implications of the information, and a signed and witnessed document (Schwartz, Preece, & Hendry, 2002, pp. 52-55). Clearly, newborn screening is performed on minors who cannot give informed consent for either the initial biochemical screen or any additional genetic tests. As for other pediatric medical situations that call for decision-making and consent, the onus falls on the parent(s) or legal guardian to make choices for their offspring. It is assumed that such decision-making will be based on the principle of beneficence and that the best interests of the child are considered paramount (Elkin & Jones, 2000).

The legal requirement for informed consent for newborn screening has been considered problematic by some institutions due to the practical limitation of health resources. Unfortunately, the process of gaining fully informed consent necessarily is labor intensive. The complete process, which would not only inform parents of the facts about the screening process (including its limitations), the possible psychological impact of a positive result, and give the family control over the potential use of derived information and future use of the retained sample, would add significant costs to any newborn screening program. Not surprisingly, inclusion of this time-consuming activity reduces the economic viability of adding several rare or complex disorders to the list of targeted conditions (Dhanda & Reilly, 2003).

Some NDBS programs use printed fact sheets to inform parents about screening in order to facilitate the consent process. However, it has been noted that, "It is doubtful that, in a busy maternity ward, and with short-stay patients, informed consent processes that would stand the test of law" are achieved by this approach (Skene & Bankier, 2004, p. 70). Information brochures, with contact numbers for those who wish to learn more about the newborn screening program, are disseminated during prenatal care in some institutions, but there is usually no verification process to ensure that parents have read or understood this information prior to giving consent for sampling. In fact, it has been noted that mothers tend to feel too preoccupied with their apparently healthy new baby to focus on complex discussions about possible rare genetic disorders (Campbell & Ross, 2003).

It is important to consider what particular information must necessarily be given if NDBS is expanded to include possible detection of serious debilitating conditions for which we have neither accurate understanding of natural history nor preventive or ameliorative therapy. Ideally, the parent should be provided with a simple fact sheet which briefly explains, in plain (language-appropriate) terms, the aims and limitations of expanded newborn screening, as well as stating that the program has potential to detect untreatable disorders or carriers for certain conditions. Contact details for the NDBS counselor should be included in case the parent feels the need for additional details, further discussion, or clarification. In this scenario, if the parent demonstrates unquestioning acquiescence or active authorization for the tests despite being offered the opportunity to be counseled or have access to more detailed information, this can often be understood to imply appropriate consent, albeit not necessarily fully informed consent. Under these circumstances, there is no strong requirement that the parent receives and completely understands all of the relevant information (Faden & Beauchamp, 1999; Forrester & Griffiths, 2001, pp. 135–139; Miesel & Kuczewski, 1996; Schwartz et al., 2002, pp. 51–55).

Of course, there remains scope for uncertainty and misunderstanding due to the paucity of actual communication between parent and health professional, which could lead to allegations of professional malpractice (Schwartz et al., 2002, pp. 51–55). And health professionals would have to be diligent and discerning in the identification of those parents who may not be able to read, or who have a low functional intelligence, and ensure that they personally are offered a verbal explanation. In reality,
the actual number of families that ultimately might feel aggrieved as a result of this process is likely to be very small, and thus it could be considered a risk worth taking in light of the increase in benefits associated with the expanded program to a greater number of families. A recent qualitative study of public attitudes towards consent issues noted that members of the public "recognize the necessity of a balancing act to resolve competing but legitimate interests (for example, individual versus community)" (Robling et al., 2004, pp. 108).

It is also important to maintain low operational costs for such programs in order to ensure that every newborn continues to have equitable access to this form of preventative medical care. The ethical challenge of expanded newborn screening is to minimize adverse events experienced by parents of identified carriers or infants with untreatable disorders, while gaining maximum benefit for the families of children affected by rare, severe, but treatable, genetic conditions. This can only be achieved by an integrated process that aims to attain appropriate consent, ensure relevant ongoing medical care, genetic counseling, and support for families of detected infants. It is also important to offer counseling support for parents of infants who are detected as carriers or determined to be at high risk for conditions for which there is no immediate medical management.

It should be remembered that in the main, the risks associated with expanded newborn screening are psychological and ethical rather than threats to physical function or life. In fact, for newborns, not being screened actually may constitute a risk to well-being and/or life, and their parents may unknowingly be at high risk for having more than one child affected with a potentially severe, debilitating genetic condition. It is important to realize that for those with an undiagnosed metabolic disorder that requires an environmental trigger, the first presentation with serious clinical symptoms can be fatal or cause irreversible impairment despite the best efforts of competent specialists. On the other hand, for diagnosed individuals who comply with medical management, early fatality is increasingly rare (Wilcken, 2003). The case for expanded newborn screening, inclusive of untreatable, severe, rare, genetic disorders with childhood onset, can be considered ethically justified as long as appropriate consent, based on printed material (that offers easy access to further discussion), is obtained.

In conclusion, we ought not to assume that harm will not ensue as a result of expansion of NDBS, and care must be taken to continue to monitor parental attitudes and family outcomes in order to ascertain the real social impact of this public health service.

GENETIC COUNSELING FOR INHERITED DEAFNESS: ETHICAL CHALLENGES

It is now known that genetic factors cause at least half of all cases of profound congenital deafness. In the past few years, more than 120 genes for deafness have been identified and current research is aimed at gaining insight into the pathophysiology of hearing deficit in order to design the most effective interventions, which will minimize functional hearing loss and maximize residual hearing. Genetic hearing loss can be associated with a spectrum of clinical symptoms affecting other body organs or systems (syndromic) or, as in around 70% of cases, is an isolated finding (nonsyndromic) (Nance, 2003). A statement on Universal Newborn Hearing Screening, produced by the American College of Medical Genetics (ACMG) in 2000 and reaffirmed in 2005, recommends that for deaf infants ascertained by these programs, the "provision of appropriate clinical, audiologic, and genetic follow-up, evaluation, and testing not only to confirm the presence of a hearing loss but to establish the etiology whenever possible" (ACMG, 2002).

One of the original guiding principles of newborn screening and genetic testing is that the disease must manifest in childhood with attendant "severe complications" or "high burden" (Ciarleglio, Bennett, Williamson, Mandell, & Marks, 2003). Genetic testing in childhood removes the possibility for that individual to make an autonomous decision about genetic testing as an adult; therefore, such tests are usually only considered appropriate for minors if there are clear and defined benefits to the child, usually in the form of effective intervention or surveillance aimed at prevention. Currently, in contrast
to testing for cystic fibrosis and the majority of metabolic conditions associated with NDBS, information derived from genetic tests for deafness offers very little advantage to the infant in terms of clinical utility, unless required for diagnosis or exclusion of a syndromic cause of congenital deafness. For this reason, a normative medical ethics stance would not support universal application of genetic tests for hearing loss detected via newborn hearing screening (Battey, 2003). On the other hand, parents often gain immense relief from understanding the precise cause of hearing loss and this can assist their acceptance of the diagnosis. On the negative side, identification of the inherited genetic mutations also may cause parents to experience varying degrees of guilt because they feel they are to blame for their child’s condition. Another potential utility of genetic diagnosis is that it can create additional reproductive choices for the parents in subsequent family planning.

As newborn dried bloodspot screening expands to include genetic testing for an increasing number of metabolic conditions that have unpredictable age of onset or degree of severity, and in particular for inherited deafness, it must be acknowledged that the subjective interpretation of the relative burdens associated with these conditions will vary significantly within health disciplines and from family to family. Failure to explore these issues early in the genetic counselling session may lead to a mismatch in perceptions and language between the counselor and the parent, which will impact negatively on the effectiveness of and satisfaction with the consultation (Tłuczek et al., 2006). Such exploration also will encourage parents to clarify their own ethical framework, within which they can effectively measure the available options.

The diagnosis of congenital deafness can provoke a sense of panic, regret, loss, or shame in the parents and family of an affected infant or alternatively for some Deaf parents, happiness and relief (Harris, 1995, pp. 15–17, 37). Around 90% of congenitally deaf children have hearing parents (Lané, 1993, p. 5; Levy, 2002). However, one survey estimated that 33% of deaf adults could be classified as “culturally Deaf,” suggesting that a significant number of profoundly deaf individuals born to hearing families ultimately choose a Deaf identity (Middleton, Hewison, & Mueller, 2001). The genetic coun-


er must explore the individual’s personal beliefs and values around the state of deafness in order to be respectful of their worldview, because when it comes to genetic testing, the individual’s right to make an informed autonomous decision, which harmonizes with his or her own beliefs and values, is an ethical consideration of paramount importance.

Recurrence risks for nonsyndromic deafness in the future offspring of the same parental couple commonly range from 25% to 50%, which is considered to be in the “high risk” range by most hearing parents. The option of prenatal diagnosis is certainly raised for couples if both parents are known carriers of noncomplementary autosomal recessive gene mutations (i.e., mutations in the same gene) or one parent carries a single autosomal dominant gene mutation that is associated with the inherited deafness. Prenatal testing potentially raises the emotionally distressing choice of electing to continue or terminate a pregnancy if the fetus is diagnosed with genetic deafness (Nance, Liu, & Pandaya, 2000; Sutton, 1988). Some “at risk” couples will find the option of prenatal testing morally unacceptable so many choose to take their chances in future pregnancies, consider adoption, or decide not to have any more children together. For others, the capacity to choose prenatal testing in future pregnancies theoretically, could be utilized to prepare for the birth of another deaf offspring or to create the potential option of termination of an otherwise healthy deaf fetus, or alternatively, in the case of Deaf parents who desire Deaf offspring, lead to potential termination of a healthy hearing fetus. Of course, effective realization of any of these options will be limited by ethical, legal, and economic policies of institutions and countries or economic capability of the couple to seek out and pay for these services through a private provider. In fact, some health authorities have an explicitly stated ethical stance on the issue (National Ethics Committee on Assisted Human Reproduction [BCART], 2005; Human Genetics Commission [HGC], 2006; National Health and Medical Research Council [NHMRC], 2007). The manner in which individuals and couples seek to use such genetic technology, if it is available, varies widely depending on individual worldview, personal or familial exposure to Deaf persons and their innate understanding of deafness, personal religious beliefs, or other factors associated
with current prevailing medical approaches and management options (Brunger et al., 2000; Middleton, Hewison, & Mueller, 1998; Rehm, 2003).

There has been some interesting published research and opinion on the application of genetic tests for deafness. Several studies have found that Deaf or hard-of-hearing individuals and their hearing parents generally have a positive attitude to genetic testing when it is used to determine cause, etiology, and prognosis of hearing deficit. Most also maintain that they do not have a preference for children of a particular hearing status and are of the opinion that prenatal diagnosis would only be useful for preparation and that termination of pregnancy on the basis of hearing status is generally unacceptable. However, these early studies also confirm that attitudes vary significantly, depending on whether or not the participant identifies with Deaf culture. It appears that around 6 to 16% of deaf, hard-of-hearing, and hearing respondents would consider termination if genetic deafness was diagnosed in the fetus and a smaller percentage, about 2%, of culturally Deaf individuals surveyed indicated that they have a strong preference for Deaf offspring and would consider termination if early prenatal diagnosis determined that the fetus had not inherited their familial deafness genes (Brunger et al., 2000; Martinez, Linden, Schummeni, & Palmer, 2003; Middleton et al., 1998, 2001). Of course, these data relate to expressed intentions and should in no way be interpreted to be a definite indication of expected actual behavior, as it has been well documented, in the case of prenatal diagnosis for other genetic conditions, that decisions often change when in the clinical setting, couples are faced with the tangible reality of their choices (Green & Statham, 1996). Whether or not a woman or couple elects to engage in prenatal diagnostic testing for other genetic conditions has been the subject of numerous research studies. These investigations have identified the principal influential factors as: attitudes to abortion, subjective perception of risk (rather than objective probability), perceived burden of disability, attitudes to the risks associated with invasive procedures, manner in which tests are presented, response to perceived pressure to use available technology, and anticipated decision regret. Certainly, many of these factors can be positively or negatively influenced by the attitudes or demeanor of the health care professionals who provide prenatal genetic counseling (Green, Statham, & Snowdon, 1992; Marteau, 1991; Marteau, Plenican, & Kidd, 1993; Tymstra, 1989).

The genetic counselor has appropriate training to explore the potential ethical challenges associated with the decision-making demands that may arise from results of prenatal diagnostic tests. Without sufficient information and opportunity to discuss personal concerns, individuals and couples are rarely able to feel confident that they have accurately compared the risks and benefits and made the most meaningful decision for their personal situation. The current status of genetic knowledge has created numerous challenging ethical scenarios because valuable information about the causes of human variation, disease, and disability is available but their corresponding remedial treatments or cures are not yet a reality. Genetic counselors and clinical geneticists are uniquely qualified and experienced in dealing with the ethical aspects of genetic medicine in a responsible manner.

New technologies and health care services that enable choices based on genetic information inevitably will continue to provide new ethical challenges as they widen the scope of reproductive possibilities and reveal implications for other family members. For the average individual, a combination of knowledge, personal history, personality, family culture, and desired goals create unique meaning that underpins personal choice. Essentially, those who counsel at the cutting edge of genetic technology should promote freedom, within legal boundaries, for the individual to make a personally meaningful choice as he or she must live with the consequences of that decision.

FURTHER READING

For an in-depth review of the process, aims, and challenges of genetic counseling refer to the following texts:


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