Pediatric Psychology Training and Genetics: What Will Twenty-First-Century Pediatric Psychologists Need to Know?

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Objective To demonstrate the importance of genetic knowledge in coming decades and to outline necessary areas of genetic education. Method This article reviews research involving genetic testing of children for cancer syndromes, development disabilities, psychiatric problems, and other conditions. Results The developmental, clinical, research, and consultation skills of well-trained pediatric psychologists will make them valuable collaborators with genetics professionals. Pediatric psychologists study the genetic etiology of psychiatric conditions and outcomes of genetic testing for physical disease. Conclusions Pediatric psychologists will need training in the concepts and methods of the New Genetics. They should understand the implications of risk notification and genetic test disclosure and should be aware of related ethical concerns.

Key words genetic testing; hereditary cancer syndromes; psychiatric genetics.

This article is directed at those responsible for the training of twenty-first-century pediatric psychologists. It briefly reviews current genetic knowledge as it relates to physical and psychiatric diseases of childhood and describes the roles of pediatric psychologists in the clinical care of patients with genetic concerns. It outlines some of the research questions regarding inherited disease predisposition and related ethical issues and provides an overview of the training in genetic concepts, methodologies, and outcomes that pediatric psychologists will need to meet clinical and research challenges.

Genetic Testing of Children
Clinical Roles on Genetics Teams Assessing Genetic Predisposition for Physical Illness

Geneticists, genetic counselors, nurses, specialty physicians (e.g., developmental pediatricians, oncologists, neurologists), psychologists, and other mental health professionals all make important contributions in helping children and families cope with genetic disease. There is often some overlap in roles between members of different professions represented on a genetics team, with geneticists, genetic counselors, and nurses offering information about the inheritance patterns, risk estimates, and options for prevention and several team members providing psychological support. However, some roles are typically assumed by clinical psychologists. Psychologists working in pediatric settings may provide the team with developmental assessment of the child’s cognitive understanding of the role genetics plays in the etiology of the condition that runs in his or her family. Psychologists typically assess psychological readiness for genetic testing and can judge the emotional value of genetic information to parents and children. Pediatric psychologists assist genetics professionals in the preparation of age-appropriate materials for use with children of different ages to describe relevant inheritance issues and options. Psychologists may help children and parents cope with the results of genetic testing and may aid in family dissemination of genetic information and related emotional concerns. They may help children reevaluate the meaning of their genetic risk when the findings acquire particular saliency because of developmental changes. Psychologists may also advise the genetics team on ways to improve their approach to patients or their follow-up procedures or may make recommendations.
Concerning issues relevant to subgroups of patients, such as patients with developmental delays.

**Cancer as a Model**

Cancer is the first major disease for which genetic testing is available for the detection of inherited illness predisposition among members of some high-risk groups or families. Most genetic cancer testing has been conducted in adults for malignancies typically diagnosed in adulthood. The most common types of genetic cancer testing currently performed are for BRCA1 or BRCA2 genes that predispose to breast or ovarian cancer or testing for the several genes that predispose carriers to colon cancer. Professional medical organizations have issued guidelines suggesting that children not undergo genetic testing for adult-onset disorders unless treatment in childhood would substantially affect the outcome of the serious adult disorder (American Medical Association, 1995; American Society of Human Genetics/American College of Medical Genetics, Statement on Children, 1995), and most professionals concur. However, several pediatric conditions involve disposition to pediatric cancers for which genetic testing is available and is, in some cases, considered standard patient care. Discussion of these conditions provides a template for raising general questions about the genetic testing of children.

**Li-Fraumeni Syndrome and p53 Genetic Testing.** Li-Fraumeni syndrome (LFS) is a rare cancer syndrome transmissible to disorders in a single gene, p53. The p53 gene is autosomal dominant; thus, each child has a 50% chance of inheriting the deleterious mutation. LFS family members who are mutation carriers have a 40% chance of developing cancer by age 20 (Williams & Strong, 1985), far above the population risk of malignancy in childhood. In addition, the cancers that develop in LFS tend to develop earlier. Secondary primary malignancies are common, and third and even fourth malignancies in the same person have been reported (Hisada, Garber, Fung, Fraumeni, & Li, 1998). Families are often devastated by cancer in multiple generations, and multiple cases of rare pediatric cancers can occur in the same nuclear family. Members of LFS cancer families often report anxiety originating in childhood about the patterns of cancer in their family. Unfortunately, there are no appropriate screening mechanisms for most of the target cancers (sarcomas, breast cancer, brain tumors, bone cancers, adrenocortical carcinomas). Hence, the direct benefits of genetic testing of adults or children are limited to (1) allowing for the recognition of the 50% of family members who are not carriers and, thus, not at increased risk for cancer; (2) alerting parents, patients, and physicians to the high likelihood of cancer in even quite young individuals who are mutation carriers and encouraging early investigation of symptoms; and (3) providing clarity for patients and parents about whether children are mutation carriers. Because of the limits of direct medical benefit, there has so far been little genetic testing of unaffected children in LFS families. However, in cases where children are diagnosed with target cancers and the family history involves or is suspicious for LFS, testing is likely.

**Multiple Endocrine Neoplasia 2: Cancer Curable With the Help of Genetic Testing.** Multiple endocrine neoplasia 2 (MEN2) is a cancer syndrome marked by early development of medullary thyroid cancer, as well as several other conditions. Genetic testing is the standard of care currently in children whose parent has MEN2 due to a mutation in the RET gene (Johnston et al., 2000). MEN2 is the rare case where identification of a genetic mutation is usually quickly followed by a targeted treatment. Surgical removal of the thyroid gland is recommended before age 5 (and even earlier by some experts) in mutation carriers to prevent metastasis and to reduce potentially fatal outcomes (van Heurn et al., 1999). Testing allows the 50% of children found not to be RET mutation carriers to be spared the recurrent biochemical testing recommended for all children from families with hereditary MEN2 (Gagel et al., 1995).

Several studies of the reactions of children, adolescents, and adults to MEN2 genetic testing reveal generally normal levels of postdisclosure distress (Grosfeld, Beemer, Lips, Hendriks, & ten Kroode, 2000; Michie, Bobrow, & Marteau, 2001), although, in one Dutch study, the 15- to 20-year-old patients had scores above population levels. Parents of 27% of the children testing negative in that study wished to continue calcitonin testing (Grosfeld, Beemer, et al., 2000).

**Familial Adenomatous Polyposis.** Familial adenomatous polyposis (FAP) is also a condition for which genetic testing prevents unnecessary medical screening for children who are not mutation carriers. Children in FAP families usually have colonoscopies beginning around age 10 to detect the presence of precancerous polyps, which occur in great abundance in individuals with FAP (Neugut, Jacobson, & De Vivo, 1993). If not removed, these polyps can lead to colon cancer. Surgery occurs as early as the mid-teens in many families. Genetic testing for mutations in the APC gene that conveys predisposition to FAP are likely to be negative in half of the cases. Thus, 50% of children in families at risk for FAP who might have had a childhood marked by recurrent, unpleasant medical tests, overlaid with repeated parental anxiety about the test results, may be spared these potentially traumatic experiences if they do
not carry the familial APC mutation. This represents a significant psychological and economic consideration. Such testing comes, however, as part of a gamble with fate, which includes the possibility of finding that the child is a mutation carrier. Carriers can be monitored carefully and have timely surgery.

A 3-month follow-up study of 41 children tested for APC showed that, regardless of their genetic status, the children overall remained within normal limits of psychological distress (Codori, Petersen, Boyd, Brandt, & Giaraldiello, 1996). However, subclinical increases in distress were noted, depending on the child’s genetic status and the health status of the same-sex parent. Mutation-positive children with mothers who also had FAP had significantly higher depression scores, and both mutation-positive and mutation-negative children had higher anxiety scores if their mother was affected (vs. having an affected father). Unaffected parents of both mutation-negative and -positive children had increased depression scores at 3 months.

Prevention. Genetics is likely to enhance our understanding of what enables some people to avoid health risks and what drives others toward risky behaviors. Research is ongoing on genetic factors related to the age at which individuals start smoking and their later success in giving it up (Lerman et al., 2001). Such research has broad implications for the development of targeted interventions to teens at highest risk of smoking and lowest levels of ability to control their smoking behavior, once initiated. If some young people have a relatively low genetic risk of developing cancer due to smoking, they may be encouraged to take up smoking, with other negative health consequences. Psychologists have much to offer in the framing of genetic health risk information to teenagers.

Summary. Many questions arise about the impact of early genetic testing on the identity development and self-esteem of children in high-risk cancer families. Research in this field is needed to understand how children react to early knowledge of hereditary risk for a variety of conditions. Long-term follow-up of larger samples of children tested for p53, APC, and RET and their family members is warranted. Defining the emotional reactions to genetic identification of at-risk children in families with hereditary cancer syndromes is a complex process. I have discussed relatively rare cancer syndromes attributable to alterations in single genes. In the future, hereditary contributions to more common pediatric cancers, like leukemia, probably involving multiple genes, will be found. Parents may be confused about the etiology of their child’s disease, may misunderstand requests to have their child’s blood drawn for genetic studies, or be confused about the nature of testing to which they offered their consent. Almost a third of parents of children tested for the RET gene for MEN2 believed that the test would determine if their child had the disease rather than assessing genetic predisposition (Grosfeld, Lips, et al., 2000). Parental anxiety about the etiology of a child’s disease could also lead to requests for unnecessary and uninformative genetic testing. About half of mothers of pediatric oncology patients questioned said they would have their child undergo genetic testing if a test were available, even if there were no direct benefit to the child (Patenaude et al., 1996). Pediatric psychologists in oncology settings will need to keep abreast of rapidly changing knowledge in pediatric cancer genetics. Their research will aid genetics professionals in clearly informing parents about the nature, risk, and benefits of genetic testing.

Nonmalignant Genetic Diseases of Childhood

Of course, many other genes for pediatric-onset conditions, such as those for cystic fibrosis (CF), diabetes, and asthma, have been, or will be, found in the coming decade. The discovery of the cystic fibrosis gene in 1989 led to current consideration of population screening for the 1 in 25 individuals who are CF carriers (Grody & Desnick, 2001). However, identification through genetic testing of an individual carrier of a serious illness may itself have psychological effects (Fanos, 1997; Zeesman, Clow, Cartier, & Scriver, 1984). A follow-up study of adults who were CF carriers showed that, 3 years after testing, the carriers described themselves as significantly less healthy than non-carriers, despite the lack of any actual physical effects on CF carriers (Axworthy, Brock, Bobrow, & Marteau, 1996). These reports and others (Michie & Marteau, 1996) suggest that children, adolescents, and adults attach different meanings to knowledge of genetic risk.

Developmental Disabilities and the New Genetics

Advances in genetics enable more accurate and earlier molecular diagnosis of children with symptoms or family histories suggestive of developmental syndromes. New genetic technology led to surprising findings of non-Mendelian mechanisms underlying Prader-Willi syndrome (PWS) and Angelman syndrome. About 30% of PWS cases reveal imprinting, an unusual instance where the expression of a genetic condition differs depending on the sex of the parent from whom the altered chromosome is inherited (Muir, 2000). Subtle phenotypic differences have been found in birthweight, facial features, coloring, and age of onset depending on the mode of inheritance. Like PWS, some cases of Angelman syndrome are also due to uniparental disomy. Further study of phenotype-genotype correlations and molecular findings in these two disor-
Major Research Questions: Genetics of Physical Illness

1. How will children’s self-esteem, ambition, optimism, and general emotional well-being be affected by knowledge that they carry a disease-predisposing mutation?
2. How will families change because of testing of children and adult family members? Areas of interest include emotional impact of knowledge of carrier status, reproductive decision making, possible differential allocation of emotional and financial resources depending on genetic status, blame and guilt, and relationship to extended family members.
3. How do children of different ages and family histories understand genetic concepts?
4. What are the methods and timing for optimal communication with children and adolescents about genetic information?
5. What are the cultural influences on cognitive understanding and acceptance of genetic medicine among families of diverse ethnic origins?
6. How will knowledge of genetic predisposition affect adoption or avoidance of health behaviors? Are there interventions to aid children and adolescents at increased hereditary risk of disease in the acceptance of screening interventions that could be life-saving?

Genetics of Psychiatric Disease

A full review of current knowledge of psychiatric genetics with relevance to pediatrics is beyond the scope of this article. Readers are directed to the extensive review of the pediatric psychiatric-genetics literature by Rutter, Silberg, O’Connor, and Simonoff (1999b). I will, however, attempt to identify roles for pediatric psychologists associated with the growth of research in behavioral genetics.

Psychiatric genetic epidemiology has, through twin, adoptee, and family studies, led to many interesting and valuable observations about likely genetic factors in the etiology of affective disorders (Beardslee, Keller, Lavori, Stanley, & Sacks, 1993), schizophrenia (Moldin & Gottesman, 1997), autism (Bailey, Phillips, & Rutter, 1996), attention deficit/hyperactivity disorder (Biederman et al., 1992), substance abuse (Reed, Page, Viken, & Christian, 1996), antisocial behavior (Farrington, Barnes, & Lambert, 1996), and neurofibromatosis (MacCollin, Guttmann, Korf, & Finkelstein, 2001), which have changed our thinking about the role of nature and nurture in psychiatric illness. Thirty years ago, there was little recognition of genetic factors in schizophrenia, for example, but now it is clear that it and many other psychiatric conditions are multifactorial in etiology, requiring both genetic vulnerability and environmental triggers (Farrell, Tsuang, & Tsuang, 1999).

There were some false starts in psychiatric genetics with early findings of genetic links that were not replicated (Robertson, 1989). Attempts to locate candidate genes have been largely unsuccessful (Owen & Cardno, 1999). However, considerable excitement remains about the promise of genetic studies (Evans, Muir, Blackwood, & Porteous, 2001). Genetic findings should lead to breakthroughs in our understanding of the etiology of psychiatric disease and to new approaches to treatment.

The definition of the disease state in psychiatric disorders is based on observation of behaviors rather than on biochemical or other direct measures of physical status. Thus, the ability to accurately define and consistently recognize which participants meet the established phenotypic criteria is essential for rigorous, reliable research. Pediatric psychologists familiar with the manifestations of behavioral and psychiatric conditions can help define these criteria and maintain consistency of observations.

Pharmacogenetics. Pharmacogenetics refers to the growing body of knowledge indicating that individuals vary in sensitivity to adverse side effects of medications due to genetic differences (Wolf & Smith, 1999). This revolution in the way medicines are prescribed has tremendous economic, medical, and social ramifications. The hope is that adverse side effects of psychotropic medica-
tions could be reduced and positive effects optimized through individualization of prescribed doses, based on genetic testing. However, there also are concerns about whether the testing of customized, pharmacogenetic approaches may result in under- or overdosing of children (Kearns, 1995). Informed consent, the balance of risk and benefit, and confidentiality issues about the information generated by phenotype testing for pharmacogenetic purposes also concern ethicists and researchers (Issa, 2000). Pediatric psychologists will need to be aware of special issues concerning pharmacogenetic approaches to the treatment of pediatric behavior disorders and psychiatric illness.

Clinical Roles for Pediatric Psychologists in Psychiatric Genetics. As knowledge about genetic factors grows, more people with family histories of psychiatric disorders will seek genetic counseling and associated psychological services to understand possible hereditary risks to themselves and their children. However, genetic testing or specific, genetically based interventions will not be available for some years (Welch & Burke, 1998). This may be difficult for many desperate parents or patients to understand.

Research into the molecular basis for many psychiatric and behavioral conditions promises to lead to genotypic diagnostic groupings that will allow for objective and definitive diagnosis. This specificity will, in turn, enhance the development of targeted treatments and interventions. The finding of genetic effects, however, is not consonant with understanding the cause of the condition. As Rutter (1999a) notes, “The road from gene localization (or even identification) to an understanding of causal mechanisms is likely to prove a long and arduous one, especially in the case of multi-factorial disorders” (p. 18). With both genetic and environmental risks it is essential to move beyond the quantification of each to the study of which genes and which environmental factors provide the risk and how they operate” (p. 12).

Careful observation and clinical ability, however, will continue to be central factors in the success of any psychological treatment. Flexible thinking, as well as knowledge of patterns of genetic comorbidity, will be useful in determining which questions to ask in taking a family history to ascertain hereditary links. When a genetic vulnerability is established, pediatric psychologists can help children and adults find a balance between the helplessness of genetic determinism and the self-blame caused by belief in purely environmental causes for children's psychiatric illness. In psychotherapy, confusion and anger about the unfairness of inherited vulnerabilities to psychiatric disease may be addressed. Psychologists may help parents and children understand why genetic investigation of sensitivity to psychotropic medication may be helpful in optimizing and personalizing prescription recommendations.

Major Research Goals: Psychiatric Illness
Researchers should address the following goals:

1. Isolation of the molecular genetic features that predispose to the major psychiatric conditions with strong family patterns of transmission;
2. Improvement in our understanding of mechanisms underlying genetic heterogeneity;
3. Improvement in our understanding of whether intermediate phenotypes (like schizo-affective disorder) represent a continuum of phenotypic manifestation of similar underlying genetic mechanisms;
4. Improved understanding of the ways in which environmental factors trigger (or fail to trigger) genetic predisposition to mental illness and behavioral conditions;
5. Communication of complex genetic information about risk for psychiatric disease to affected families and research on the outcomes of risk notification; and
6. Acceptability of pharmacogenetic targeting of treatment for psychiatric illness.

Impact of Parental Testing on Children and Family Communication
Although much has been written concerning the potential effects of genetic testing of family members on children (Clarke, 1998; Wertz, Fanos, & Reilly, 1994), little empiric research to date addresses children's involvement in or reaction to genetic testing of their parent or other relative. Researchers have been reluctant to directly approach children of tested adults, both because of confidentiality and because of the research complexities in assessing and comparing how children of varying ages understand genetic testing. However, studies are beginning to emerge in this area. Geller, Tambor, Bernhardt, Wissow, and Fraser (2000) interviewed 10- to 17-year-old daughters of mothers with breast cancer from high-risk breast cancer families about whether the daughters would want to be genetically tested themselves. The daughters initially could not see any reasons not to be tested, but with time and additional information, they understood more about the risks and benefits of testing, suggesting a strong need for counseling of teenagers deciding about testing. Hamman
et al. (2000) conducted telephone interviews of 104 women and men with children under age 18 who had been tested for mutations in their BRCA1 gene regarding their beliefs about whether minor children should be tested for BRCA1. Seventeen percent reported that they would want to test their own children, whereas 83% did not want their children tested as minors. Tercyak et al. (2001) asked 133 parents who had been tested for BRCA1/2 mutations and who had children under age 18 about their decisions to tell or not tell their child(ren) their test result 1 month after disclosure. Mothers were nearly evenly split, but 71% of the fathers did not disclose their result. Mothers and parents who had high baseline levels of distress were more likely to tell their children their result. Prior cancer history, carriers’ status, and age of the child did not predict disclosure patterns. In a related study, investigators found that disclosure to a minor child of a parent’s BRCA1/2 test result was inversely associated with parental education level, suggesting that greater knowledge about the ethical dilemmas and social stigma pertaining to genetic predisposition is associated with more hesitation about sharing results with children (Lerman et al., 1998). Data on the role parental anxiety and other factors play in decisions about sharing genetic information will be critically important as more disease genes are cloned and genetic testing programs instituted.

Cultural differences also are likely to influence family communication about genetics. There are, as yet, no studies of differences by ethnicity or culture of how genetic information is shared with children. However, Hughes (1997) found that African American women revealed their breast cancer genetic testing result to their spouse and parents 27% of the time, whereas Caucasian women informed their spouse 66% of the time and their parents 40% of the time.

Ethical and Social Issues

Much concern about the testing of children revolves around finding a balance between utilizing genetic technologies to reduce morbidity and mortality of disease for children and protecting them from harm. Ethical issues about the genetic testing of children include autonomy, consent, confidentiality, and avoidance of discrimination.

Autonomy

Testing in childhood precludes the option not to know one’s genetic status, a choice made by a significant number of adults offered genetic testing for cancer genes and Huntington’s disease. Some experts, however, believe that teenagers are capable of making such decisions and that respecting the autonomy of their decisions about genetic testing avoids paternalism and is in their best interest (Elger & Harding, 2000). Others believe that when medical benefit is not imminent, it is preferable for the individual to defer decision making about genetic testing until adulthood (Clarke & Flinter, 1996; Collins, 1996). Views of what constitutes “medical benefit” in childhood are, of course, variable.

Informed Consent

Parents must provide consent for the genetic testing of their children. There is much need for research on the motivations for surrogate decision making regarding genetic testing of children for physical and psychiatric illness. Some parents of children with cancer have indicated greater willingness to test their minor, healthy children for inherited risk of cancer than to be tested themselves (Patenaude et al., 1996). Whose task is it to inform children about their hereditary risk? What “duty to warn” considerations apply for the provision of genetic risk information to previously tested, but uninformed, children when they reach legal maturity (Clayton, 1998)?

Confidentiality

Confidentiality about genetic matters is a complex issue for individuals of all ages. There is substantial variation between states in the kinds of legal protection offered. The rights of third-party payers, health providers, employers, and patients are difficult to balance, and it is not clear what path best protects the patient’s health and who needs to have access to a child’s genetic information. For example, should information about a child’s genetic risk for disease be routinely shared with pediatricians, schools, camps, or teachers?

Difficult ethical questions arise about the use of genetic testing in adoption. Concern has been raised about whether genetically testing a potentially adoptable child objectifies the child (Freundlich, 1998), while also precluding the right to make later decisions about testing. In some cases, genetic testing could show that a child is not a carrier of a deleterious mutation known to exist in his or her biological family, which might increase the child’s chance for adoption. Alternatively, testing could make clear that the child is at increased risk for the condition, possibly foreclosing many options for adoption. There is no clear policy yet about the genetic testing of preadoptive children and about the conditions under which genetic testing should be undertaken to satisfy potential adoptive parents’ questions.
Discrimination
Given the eventual needs of many mutation carriers for expensive medical care, it is worrisome that children might be genetically tested for adult-onset conditions when they do not have health insurance that can be carried into adulthood (Wertz et al., 1994). Identification of children's genetic status could limit their options to acquire reasonably priced health insurance as adults and could also result in employment-related discrimination.

What Pediatric Psychologists Need to Know About Genetics
To function optimally in clinical or research roles, especially (but not only) when working as part of genetics teams, psychologists need to refresh their knowledge of basic genetic terms and concepts and to receive updated teaching about the impact of modern genetic technology on their particular areas of inquiry or practice (Jenkins et al., 2001). Such training may occur in continued education courses; in multidisciplinary, disease-specific training workshops; and, ultimately, in graduate school curricula.

Conceptual Knowledge
While the extent and specific nature of the genetics training needed by pediatric psychologists will vary depending on their area of specialization, pediatric psychologists should be trained in three basic aspects of genetic knowledge: (1) an updating of their conceptual understanding of genetics, including some knowledge of methods for advancing genetic knowledge; (2) education about the advantages and limitations of different approaches to evaluating the role of heredity and environment in human behavior and illness; and (3) awareness of potential individual, familial, and societal ramifications of genetic testing.

The genetics of Mendel familiar to most educated individuals emphasized single-gene conditions with strict phenotype-genotype concordance. Discussion of the “gene for blue eyes” suggests a 1:1 relationship between a single gene and a physical characteristic. But much of the New Genetics concerns more complex situations where multiple genes (gene-gene interactions) and gene-environment interactions together produce the phenotypic characteristic or behavior. This vision enlarges the scope of genetic studies enormously, but complicates it to a similar degree. At least some awareness of techniques for studying genetic effects and localizing genes within chromosomes, like fluorescent in situ hybridization techniques or “knockout genes,” is needed to understand how genetic knowledge grows. Genetic fluency will include familiarity with the concepts of multifactorial interactions, understanding the challenge of finding appropriate ways to measure differential genetic and environmental effects, and recognition of terms like “expanding trinucleotide repeats,” “imprinting,” and “penetrance” as genetic characteristics influencing disease presentation.

Advantages/Limitations of Methods of Evaluating Genetic and Environmental Effects
Understanding terminology and genetic methodology is a necessary tool for in-depth evaluation of research studies illustrating genetic or environmental effects. This is an extremely complex area. Rutter et al. (1999a) review the intrinsic biases in twin and adoptee studies of psychiatric disorders, as well as the benefits, limitations, and biases of affected relative linkage designs and association strategies, new approaches in genetic research on psychiatric illness. Rutter et al. (1999a) also point out that until better techniques are developed, negative results still leave the presence of an association open to question.

Individual, Familial, and Social Implications of Genetic Risk Notification, Counseling, and Testing
Finally, genetic education must include awareness of how the availability of genetic risk information changes individuals’ views of themselves, their families, and society. Knowledge of research findings on short- and long-term outcomes of risk notification and genetic testing will be essential. Emerging studies suggest complexity of response and a need to evaluate individual outcomes in a family context (Smith, West, Croyle, & Botkin, 1999). Data on the impact of genetic risk notification on children's health, anxiety, somatization, and goal setting will be helpful in designing future interventions. Regular revisiting of the ethical literature will be required to stay abreast of changing local and federal statutes on genetic privacy and professional recommendations about confidentiality of personal genetic information of relevance to other family members.

Genetic Resources
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Genetic Resources
The training of pediatric psychologists should involve guidance about sources for the necessary updating of genetic information and for locating appropriate referral sources for patients and research participants with genetic concerns.

Finding networks of genetics professionals trained in the delivery of specialized genetic services (e.g., cancer or psychiatric genetics) and developing liaisons with local members of that network can help pediatric psychologists make appropriate referrals. Pediatric psychologists also will find genetics professionals to be helpful colleagues.
who are knowledgeable about the implications of identification of new genes and important mutations, and who can provide information about the relevance of genetic findings to specific patients. In turn, because most genetic counselors have little direct experience working with children, pediatric psychologists can provide useful advice on approaches to teaching children and parents about genetics and on ways to optimize family communication about genetic issues.

Because genetic knowledge is changing rapidly, reeducation about genetics will require periodic updating to remain current. Hence, knowledge of useful resources is crucial, both for the pediatric psychologist and for helping affected families find updated information sources. Resources for both basic and specific genetic information are increasing exponentially. Some useful current resources include Internet access to a glossary of genetic terms offered by the National Human Genome Research Institute at www.nhgri.nih.gov/DIR/VIP/Glossary/pub_glossary.cgi. Specific disease information is also available through government agencies such as, the Centers for Disease Control and Prevention (online at www.cdc.gov/genomics/default.htm) and disease organizations (such as, Genetic Alliance online at www.geneticlelliance.org). The National Cancer Institute maintains an updated summary of cancer genetic research findings, including related psychosocial research at www.cancernet.org. GeneClinics (www.geneclinics.org) offer an online, peer-reviewed genetics textbook and information about genetic counseling. The American Medical Association Web site offers a great deal of genetic information, much of it pertaining to children (www.ama-assn.org/ama/pub/category/1799.html). There is also a growing reference library of books that offer information on genetic issues relevant to the pediatric psychologist (see Clarke & Flinter, 1996; Farone et al., 1999; Offit, 1998; Marteau & Richards, 1996).

Discussion

Well-trained pediatric psychologists will offer essential clinical services to children and parents with concerns about genetic disease predisposition and will conduct research in this new field. The impact of genetic testing on children's self-esteem, identity, health behaviors, emotions, and outlook needs to be explored. Research that can separate the effects of parental illness, cognitive level, coping style, gender of child and parent, parental distress, social support, cultural factors, and the impact of test results of other relatives will be needed. These data can be used to set policy about the genetic testing of children. Pediatric psychologists can help us understand what children want to know about inherited disease and how best to convey that information. Children's phobias or separation fears about their own or other relatives' future health may increase. Parental advice not to widely discuss family genetic information may be misinterpreted by children and arouse feelings of isolation or deficit. Pediatric psychologists will need a basic understanding of the genetic issues within families to help parents and children facing these issues.

In some cases, it may be as important to help children and families understand that a medical or psychiatric condition is not likely to be due to genetic factors. Because of widespread news coverage about genetic advances in medicine, many parents will voice concern about whether their child's condition is due to inherited factors and if future generations will be affected. Children are also learning a good deal about genetics at earlier ages, due to increased educational efforts related to the Human Genome Project. When a disease occurs in a family, children wonder about their own hereditary risks. Because early genetic advances have tended to involve single-gene disorders, this is the model many will be using. When that is the relevant model, children may need information about particular risks involved, age of onset, and preventive or screening measures. When that is not the appropriate model, children and adults may need help in understanding how genes work together or with environmental factors in leading to disease. Such discussion may be helpful in reducing fatalistic thinking about illness.

It will also be important to help children and adults understand the changing nature of genetic information. Even in relatively well-studied genetics fields, much remains unknown, and even when knowledge is fairly advanced about the genetic contribution to a disease state, the impact on treatment or prevention efforts often is minimal for a long time. Psychologists in pediatric subspeciality areas who know when there is reputable genetic evidence to suggest specific diagnostic or prognostic information or treatment direction and when there is not will help families make informed decisions about the use of genetic technologies. Keeping up with the literature about genetic contributions to the conditions that affect the children and families in one's practice or research area will be an essential part of being a well-informed professional in the twenty-first century. Possessing such knowledge will allow one to be a resource for patients and families, for professional colleagues, and for those seeking to make policy concerning the use of genetic technology. Knowledge of the psychological vulnerability of children of different ages or in different situations will be important to share with those who actually make the laws that govern genetic technology.
Psychological factors are central in understanding how at-risk individuals perceive and utilize genetic information. The research skills of pediatric psychologists will be essential in designing clinical and research paradigms and interventions that capture the critical elements of emotional and health–behavioral impact of genetic information on family systems.

The development of good clinical and research skills will be, as always, the keystone in the arch of unique professional contributions pediatric psychologists have to offer. We need to complement the development of these skills with training in genetics to help pediatric psychologists understand the importance of this new frontier in medicine and the intrinsic research challenges it offers. The answers to the many dilemmas in genetics are not simple, but the questions themselves will remain fascinating for many decades.

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References


