Introduction to the Special Issue: Psychological Aspects of Genomics and Child Health

Kenneth P. Tercyak, PhD
Lombardi Comprehensive Cancer Center, Georgetown University Medical Center

The much-anticipated completion of the sequencing of the human genome (Lander et al., 2001; Venter et al., 2001) is expected to significantly alter our understanding of the genetic basis of several common chronic diseases and related health conditions, including diabetes, heart disease, high cholesterol, high blood pressure, osteoporosis, and some forms of cancer (Pang, Baum, & Lam, 2000). As a result of this breakthrough, the ability to predict the onset of disease will be enhanced, and more advanced and effective disease prevention, control, and treatment strategies will likely result (Gottesman & Collins, 1994). In light of this, both the practice of medicine and the ways in which health care will be delivered are expected to change considerably (Taylor, Edwards, & Ku, 2006). Notable changes will include the offer of newer and greater numbers of diagnostic and predictive genetic tests to individuals (Chung, 2007), and the incorporation of genetic testing into routine, preventive medical care (Burke, 2004; Knottnerus, 2003; Qureshi, Modell, & Modell, 2004). At a population level, these changes may ultimately result in a more healthful nation (Collins, 1999).

Predictive genetic tests are genetic tests typically offered to healthy persons so as to assist in the determination of their risks of developing given diseases later in life (Chung, 2007). Some of the more well-known predictive genetic tests are those for Huntington’s disease (Tibben, 2007), breast cancer (Bradbury & Olopade, 2007), and hemochromatosis (Burke, Reyes, & Imperatore, 2002). When partnered with information such as family history, environmental exposures, and lifestyle behaviors, risk information learned through genetic testing can be powerful and guide personal and medical decision making—including informing decisions about prevention and treatment options that are efficacious for a given individual’s risk profile (Beery & Williams, 2007; Reid & Emery, 2006).

Though there are many reasons to be hopeful and optimistic about the benefits of genetic testing for individuals, potentially at-risk family members, and the general population, knowledge generated from predictive genetic tests is not without hazard or debate. This includes a host of ethical, legal, social, and psychological issues that are raised by genetic forecasting of a future health state, including risks of social stigmatization, loss of privacy over protected health information, educational, employment, and insurance discrimination, uncertainty, psychological distress, and regret, and fatalistic thinking about the genetic determinism of disease (Burke, Pinsky, & Press, 2001). These issues are present not only for the individual who participates in genetic testing, but also for his or her family members who may share similar biological and environmentally moderated disease predispositions (Gaff et al., 2007; Jacobs & Dearthick, 1999).

Nowhere are the above-mentioned issues more pronounced than when genetic testing is raised in the context of children’s health and health care (American Society of Human Genetics & American College of Medical Genetics, 1995; Dinc & Terzioglu, 2006). There are many diagnostic and predictive genetic tests that are beneficially utilized with children (Twomey, 2006), but predictive genetic testing in children is often limited to only those disease circumstances where known prevention and intervention options already exist (e.g., familial adenomatous polyposis, multiple endocrine neoplasia type 2a). Presently, this represents a fraction of diseases—though it is expected to grow rapidly over time (Chung, 2007). Moreover, many of the newer genetic tests will likely be offered to the young and healthy, especially children, given the potential for these individuals to engage in health-promoting decision making and take additional steps toward primary disease prevention at early stages of life (Kenner, Gallo, & Bryant, 2005; Moore, Khoury, & Bradley, 2005).
As genetic testing proliferates, greater numbers of adults will also be posed with decisions about participating in such testing and learning about their risks for disease. These adults are, oftentimes, parents of minor-age children who question whether or not their children could have inherited parental disease risks (Tercyak et al., 2007). Whether children directly participate in predictive genetic testing or not, they likely become aware of their family’s history of disease, their parents’ genetic test results, and outcomes related to these circumstances—generating many questions and uncertainties about their own future health status (van Oostrom et al., 2006; Tercyak, Streisand, Peshkin, & Lerman, 2000). These and other issues are propelling a reexamination of what is known (and still unknown) about the risks and benefits of predictive and other forms of genetic testing in children, including testing for adult-onset diseases (Kopelman, 2007; Pelias, 2006; Rhodes, 2006).

The aim of this Special Issue of the Journal of Pediatric Psychology on Psychological Aspects of Genomics and Child Health is to summarize and present some examples of contemporary behavioral science research at the interface of child health psychology and medical genetics/genomics research. As there is growing interest in understanding how genes interact with each other and with the environment to affect health throughout the lifespan (Cutfield, Hofman, Mitchell, & Morison, 2007), articles were sought addressing a spectrum of research—from newborn screening paradigms that detect serious or life-threatening conditions before symptoms begin, to predictive testing scenarios in childhood for late-onset medical disorders presenting during adulthood. The resulting collection of papers highlights some of the challenges and complexities confronting scientists in these areas, including the selection of appropriate research designs and methods to answer questions of interest, characterizing genotypes and phenotypes and interpreting their correspondence, and anticipating the needs and interests of parents, children, and health care providers as genetic knowledge emerges. As will be seen, the field of pediatric psychology has much to offer in beginning to address these challenges, to help blaze the trail for future research endeavors, and to promote health and well-being in the genomic era of medicine.

This issue is in keeping with the Journal’s renewed interest in prevention-focused research under the Editorship of Dr Dennis Drotar, including representation of works addressing a spectrum of risk, from healthy to medically affected populations (Tercyak, 2008). In doing so, we recognize that the activities of many scientist-practitioners in the field take place around advanced disease stages and in tertiary and acute care settings, where children have already been diagnosed and are receiving care for physical illnesses. However, as a field, we also recognize the value of primary prevention to help avert disease, its course, and impact. We seek to help change the outcome with respect to disease management, quality of life, and receipt of health care services, but also to broaden the populations, settings, and outcomes under investigation. This includes primary prevention of disease among healthy and at-risk populations, reaching children in their homes, schools, and communities, and examining the interplay among biological, psychological, and social influences that may promote and compromise children’s health and wellness. Thus, the Special Issue is consistent with the Journal’s focus on public and population health, as charged under the Editorship of Dr Ronald Brown (Brown, 2007), and as called upon during the editorial tenure of Dr Anne Kazak (Kazak, 2002) and other leaders in the field (e.g., Black, 2002; Roberts, 1986).

Special Issue

All six of the articles contained within this Special Issue share a common, primary focus on how the emergence of genomics in medicine (and genomics in pediatrics in particular) has the potential to influence the health and well-being of children and their family members. For some, this influence is direct and may ultimately affect the ways in which both scientists and practitioners approach increasingly complex questions surrounding the etiology and management of inherited disease risks, or risks arising from the interactions of genes and the environment (e.g., genomics). For others, this influence is distal—indirectly affecting the manner in which we consider the developing child as part of a large system that is shaped by emerging knowledge and technologies of a genomic nature.

The first article, by Kral and Faith (2008), provides a behavioral genetics perspective on children’s eating patterns, the development of weight, and their relationship to obesity. Behavioral genetics is a rapidly growing field, dedicated to the study of genetic underpinnings of behavioral characteristics such as eating, substance abuse, violence, personality, and cognitive traits and abilities. The authors offer an expert review of the state of the science for familial associations between parents’ and children’s weight statuses, correspondence in their eating behaviors, and intergenerational food preference
symmetries. As is often the case with research addressing highly complex and multiply determined health outcomes, they adopt a biopsychosocial model of obesity that examines key linkages among biology, child development, child rearing practices, and the environment to clarify how genetic and nongenetic factors play a role in hereditary obesity risk. Central to their review is the concept of ‘eating in the absence of hunger,’ or disinhibited eating tendencies that are related to children’s weight gain. An important point is raised about how children’s responses to interventions for diet and weight management may be influenced by genetic factors. This is exciting and relatively unexplored research territory, and could serve as fertile ground for additional child health psychology studies in preventing and controlling obesity throughout the lifespan.

The next two articles focus on problem behaviors that are typically manifest during adolescence—substance use and tobacco use. In their review of the literature, Rende and Slomkowski (2008) argue that a behavioral genetic approach to the study of substance use etiology has the potential to offer deep insights into the timing of gene-related effects on alcohol, tobacco, and cannabis use, the identification of substance use behaviors that are particularly sensitive to gene influence, and environmental controls that may accelerate or decelerate the substance use behavior continuum from initiation through regular use to cessation. They lay the necessary groundwork of a conceptual blueprint that could inform the development of pediatric psychology studies on gene, environment, and gene × environment interaction paradigms of substance use risk and resilience in childhood.

A natural extension of this line of research is how health care professionals may make use of these types of data to assist in substance use prevention and control efforts. Toward that end, O’Neill et al. (2008) conducted an analog study to determine the extent to which providers of adolescent medicine services might someday be ready and willing to incorporate the results of genetic susceptibility testing into the prevention of cigarette smoking and lung cancer. They surveyed attendees at a national adolescent health conference, asking providers about their willingness to recommend genetic susceptibility tests; the hypothetical tests were for different diseases (nicotine addiction and lung cancer), and stratified by adolescent patient lifestyle (nonsmoker and smoker), and other contextual factors (i.e., informed consent, provision of genetic counseling, insurance coverage, and comorbid chronic illness or substance abuse history). The results indicated that provider willingness may ultimately depend upon the extent to which parents and patients are adequately informed about the risks and benefits of such tests, as well as the family history of substance use. Differences were also identified with respect to genetic susceptibility testing for primary versus secondary prevention purposes (i.e., prior to the onset of disease, vs. among those already at risk), and disease state. From a genomic perspective, the timing and staging of adolescents’ smoking and lung cancer risks appear critical to providers. This information will become increasingly important as adolescent risk profiling takes into account a fuller complement of personal, familial, and social factors, including information derived from DNA-based testing.

The next two articles examine psychosocial and ethical aspects, respectively, of predictive genetic testing in children for adult-onset diseases. First, Peshkin et al. (2008) report on the development and validation of a new measure of parental attitudes about testing children for genetic susceptibility to inherited breast/ovarian cancer (the Pediatric BRCA1/2 Testing Attitudes Scale [P-TAS]). Though such testing is not recommended for children, parental inquiries about pediatric testing are not uncommon. In developing the P-TAS, the authors sought to characterize parental attitudes, beliefs, and motivations so as to facilitate research and clinical consultation in this matter. The measure was administered to a large clinical sample of mothers undergoing cancer genetic counseling and testing for the identification of familial mutations in major breast/ovarian cancer susceptibility genes (BRCA1 and BRCA2; BRCA1/2), as well as their spouses or partners. Factor analyses explored and confirmed the structure of the P-TAS and its validity. This new tool may prove important in assessing parents’ attitudes and interests in pediatric BRCA1/2 testing, and improve education and counseling efforts for parents and their children. It could also lead to adaptations of this measure, and extension studies of its potential applicability and validity for other adult-onset diseases.

Of course, any such efforts must co-address the bioethical implications of pediatric genetic testing. In their article, Wilfond and Ross (2008) take up this issue, exploring its risks and benefits, and advocate for an expanded role of parental decision-making in children’s genetic health care, particularly in light of emerging genomic technologies. Many of these technologies will likely offer a more comprehensive assessment of children’s health than previously available, and would need to be accompanied by empirical data on the social and
behavioral risks and benefits of testing. Pediatric psychologists are extremely well poised to contribute to this process, and help promote more informed decision-making by parents, their children, and their children’s health care providers.

In keeping with the theme of emerging genomic technology, expanded health information about children, and family adaptation, the sixth article in the Special Issue examines these concerns from the perspective of newborns and their parents. For years, most states in America have conducted public health screening programs to identify newborn children at risk for the development of diseases such as phenylketonuria and congenital hypothyroidism. The list of diseases for which newborn screening takes place is now growing rapidly, leaving many parents uninformed about, and sometimes unprepared to cope with, the feedback and uncertainties that screening raises. Bailey, Armstrong, Kemper, Skinner, & Warren (2008) propose a four-way support structure to promote familial adaptation to pre-symptomatic and untreatable conditions associated with newborn screening. These are as follows: (a) accurate and understandable information; (b) formal and informal support; (c) active surveillance; and (d) general and targeted interventions. Pediatric psychologists have important roles to play in these endeavors by assisting families’ understanding of genetic risk information, informing their decisions about screening, and facilitating coping with the iatrogenic psychosocial consequences of at-risk notification.

Finally, McBride and Guttmacher (in press) provide commentary on the future of genomic research as it relates to pediatric populations and our nation’s health. From their vantage point at the National Human Genome Research Institute of the National Institutes of Health (where Dr McBride is currently the Chief of its Social and Behavioral Research Branch and Head of its Public Health Genomics Section, and Dr Guttmacher is the Institute’s Acting Director), they foresee an increasingly important role of children and families in research on health, beyond that of just genes. They ask ‘How will we translate what we know about the genomic underpinnings of health and disease into meaningful preventive interventions for children?’ Asking questions about how we translate knowledge into action is not unique to child health psychology, as it permeates all areas of evidence-based medicine. What is unique, however, is that preventive medicine (to which child health psychologists are important contributors) will soon be able to take advantage of a far wider array of knowledge about genes, the environment, and the social contexts in which we all live, work, and play to design, implement, and evaluate intervention efforts that seek to effect positive health and lifestyle change. Many will need to be called upon to fulfill this important mission. As demonstrated herein, child health psychologists have valuable roles to play in such efforts, and in shaping the course of the research agenda that lies ahead.

Comments and Conclusions

As scientific knowledge emerges about the relationships among genes, behaviors, and the environment, the practice of medicine and the delivery of health care will be altered. Though we cannot know exactly how soon these changes will come about (or what form they will take in pediatrics), I assert that we should not become complacent by awaiting their arrival. Rather, we should heed the call to become more fully engaged research partners, and look ahead to take on the important challenges awaiting us. This is true for psychology in general (Patenaude, Guttmacher, & Collins, 2002), and child health psychology in particular (Patenaude, 2003).

Is medicine already changing? To answer this, one need only look at the proliferation of online direct-to-consumer marketing of genomic services that seek to put the promise of disease prediction into almost everyone’s hands. For better or worse, such companies are already offering personal genomic profiling services to the general public, and do with little regulatory oversight (Geransar & Einsiedel, 2008). For a relatively modest financial cost and a sample of DNA, such tests will yield information about your risks for a wide range of diseases and traits, including information about predisposing genetic risks for major illnesses like cancer and diabetes. Physicians and other health care providers are usually not involved in such testing, and it remains to be seen if or how this model of genomic information service delivery could or would be sustainable, and/or become integrated into mainstream health care.

Further evidence of change comes from the growing emergence of ‘systems biology’ as a paradigm for understanding complex interactions within and between the genome (i.e., the world inside an organism) and the environment (i.e., the world outside an organism) to transform the understanding of human health and disease (Collins, Green, Guttmacher, & Guyer, 2003). A systems biology perspective seeks to advance health care by developing better predictive models of disease based on genomic and other quantitative data, and it pairs these predictions with preventive interventions that stop disease before it
starts (Assmus, Herwig, Cho, & Wolkenhauer, 2006). The aim is to personalize the health care that an individual receives by tailoring it to one’s genetic makeup—largely discarding a one-size-fits-all model of medical intervention, and replacing it with a model of individualized prevention.

Systems biology-type approaches require voluminous amounts of data, including data about the natural history of the earliest origins of disease onset and progression, and disease risk and protective factors. For children, we are already seeing the building blocks of these activities put into place through the National Children’s Study. The National Children’s Study is a longitudinal epidemiologic cohort study designed to follow 100,000 American children from birth to young adulthood to uncover preventable risks for chronic disease (Landrigan et al., 2006). Among the sources of data that this study will collect are maternal and child DNA samples so that genomic effects can be explored, as well as assessments of environmental exposures, psychosocial functioning, growth, and development. Their goal is to develop a “blueprint for disease prevention in children” (p. 2173).

As child health psychologists, we are concerned with reciprocal influences of health on children’s behavior and of children’s behavior on their health. The works contained herein highlight the value of our becoming more conversant in genomics to inform our research and practice. In doing so, I hope we can ensure that adequate provisions are in place to appropriately educate and counsel children and their parents about the risks and benefits of genomic information. There is also promise that more efficacious behavioral interventions will result when enacted with genomics in mind. Furthermore, perhaps for most of all, I hope that this work promotes the translation of genomic knowledge into improved health and health care.

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