Commentary: Trailblazing a Research Agenda at the Interface of Pediatrics and Genomic Discovery—a Commentary on the Psychological Aspects of Genomics and Child Health

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Unprecedented advances in human genome science are underway with potential to benefit public health. For example, it is estimated that within a decade, geneticists and epidemiologists will complete a catalog of the majority of genes associated with common chronic diseases. Such rapid advances create possibilities, if not the mandate, for translational research in how best to apply these and other anticipated discoveries for both individual and population health benefit. Driving these discoveries are rapid advances in infrastructure (e.g., the International HapMap Project to catalog human genetic variation; <http://www.hapmap.org>), analytical methods, and technology. This expansion in capabilities quickly has taken us from a *genetics* paradigm—where the influence of individual genes on health outcomes is paramount, to a *genomics* paradigm—where the complex influence of individual genes is considered in concert with each other and with environmental exposures on health outcomes. We discuss these and similar groundbreaking discoveries with an eye toward understanding their importance to child health and human development, and the role of behavioral science research conducted at the interface of pediatrics and genomic discovery.

Many diseases of childhood, both rare (e.g., cystic fibrosis) and common (e.g., asthma), involve genetic factors in both their etiology and in individual response to the disease and to our therapies. There also is a strong evidence-base documenting that those health behaviors and lifestyle habits established in childhood (e.g., cigarette smoking), that are the leading contributors to chronic illnesses of adulthood, also have substantial heritability (Lesso, Swan, Ring, Khroyan, & Lerman, 2004). Evidence to support the role of environment and social influence in chronic disease is, of course, also strong (Institute of Medicine, 2006). We expect that genomic science will enable disaggregation of the conjoint, interwoven influences of genes and environment that influence health outcomes.

Setting and embarking on a research agenda to apply genomic knowledge raises numerous issues, some of which can be anticipated. For example, advances in genomic knowledge combined with existing risk assessments could identify individuals at increased risk for common health conditions or new foci for intervention approaches that could improve primary prevention efforts.

Such interventions would have the greatest benefit if targeted to early childhood, a time of critical importance for primary prevention of adult-onset common conditions (Tercyak & Tyc, 2006).

The articles in this special issue of the *Journal of Pediatric Psychology* on psychological aspects of genomics and child health come from scientists whose research is blazing a trail at the interface of genomics discovery and pediatric health promotion. Each has imagined the potential trajectory of genomic discovery and conducted research to replace speculative rhetoric with a scientific evidence base (McBride et al., 2008). The early and elemental questions addressed here span a spectrum of emerging issues where research is needed. Findings of this research and future iterations will be required to inform best practices in applying genomic information to improve child health and related policy debates.

For example, Wilfond and Ross (in press) consider the issues that will be raised by identification of an ever broadening array of genetic variants associated with disease susceptibility. Debates about expansion of newborn screening

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and direct-to-consumer marketing of genome scans put new pressures on parents relating to if, and under what circumstances, children should be tested. Accordingly, it will be important to consider carefully (and reconsider as knowledge evolves) parental and family preferences and needs related to genomic testing of newborns and minor children. This raises the need for an evidence base in best practices to support parental decision making, acquisition of comprehensible test-related information, and for providing ongoing assistance in the implications of test results for children's health and appropriate use of available health delivery systems (Bailey, Armstrong, Kemper, Skinner, & Warren, in press; Wilfond & Ross, in press).

Considerable ongoing research is identifying behavioral phenotypes that are heavily influenced by genetics (e.g., eating in the absence of hunger and drug metabolism). These behaviors are evident in childhood and are associated with increased likelihood of negative health outcomes in adulthood (Kral & Faith, in press; Rende & Slomkowski, in press). New knowledge emerging from this research will necessitate creation of a translation process such as that suggested by Tugwell and colleagues (2007) in which such knowledge is synthesized, adapted to context (such as stage of child development), and any barriers to use are evaluated: all this is done to pave the way for new knowledge to be used to inform new primary prevention interventions.

This new thinking and developing controversy about genetic testing of minor children also raises the need for psychometrically sound measures to evaluate parents' attitudes and behaviors (Peshkin et al., in press). These measures could enable rigorous evaluation of the effects of parents' deliberations about and uptake of genetic testing for themselves and their children on downstream health outcomes, such as family adherence to national health recommendations, children's weight trajectories, and use of pediatric and adult health services. Moreover, such measures of parental attitudes and behaviors improve the internal validity of the research as they enable more complex consideration of theory-based moderators and mediators of the above specified outcomes.

O'Neill's and colleagues' report (this issue) suggests the importance of an informed health care delivery system. Pediatric health providers will be important partners in the delivery and interpretation of new knowledge to parents. Those results suggest that clinicians will be heavily influenced by parental preferences, and they may be most comfortable with genetic susceptibility testing for behavioral risk factors in high risk contexts (e.g., adolescent is already smoking, or has a preexisting condition that would be

aggravated by smoking) than in primary preventive contexts. Realizing the primary prevention potential and optimizing parent-clinician communication and decision making around genetic testing for children will require a good deal of continued research.

Taken together, these six articles raise more research questions than they answer. Moreover, the articles underscore that families contribute more than genes to their children's health outcomes and that research and translation must integrate these other contributions, not just genome sequences. The charge now is to continue to identify and tackle the questions that will be essential to closing the gap between genomic discovery and applications to improve health, and children's health in particular. What makes this an especially exciting time for child health researchers and practitioners and those from other collaborating disciplines is that they can play an active role in shaping the research agenda by anticipating the ways genomic discovery might be used to improve child health. In so doing, those in the field cannot only participate in the research agenda, but also proactively set the agenda.

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