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PROFESSIONAL ISSUES

Introduction to the Special Issue on Developmental Disabilities

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Abstract This special issue of the Journal of Genetic Counseling focuses on developmental disabilities and includes commentaries, original research, personal accounts, and practice guidelines.

Keywords Developmental disabilities · Etiology · Social services · Special education

> Developmental disabilities are a diverse group of severe chronic conditions that are due to mental and/or physical impairments. People with developmental disabilities have problems with major life activities such as language, mobility, learning, self-help, and independent living. Developmental disabilities begin anytime during development up to 22 years of age and usually last throughout a person's lifetime.
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> Centers for Disease Control and Prevention (2012)

Genetic factors have long been implicated in the causation of developmental disabilities (DD). There is also a large environmental component to these conditions, and the relative contributions of nature versus nurture have been debated for decades. Current theories on causation focus on genetic factors, and over 1,000 known genetic conditions include DD as a symptom (OMIM 2012). This number does not yet reflect the growing body of research on copy number variants and other findings of uncertain significance that may eventually prove to confer predisposition to neurodevelopmental disorders such as autism.

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The conceptualization of DD as the direct result of genetic disruption oversimplifies a complex and heterogeneous group of chronic conditions. Current special education and social service systems emerged from a backlash against a hopeless eugenics model of DD that marginalized environmental influences on outcome (Finucane 2010). For much of the 20th century, "two cultures" of professionals interested in DD evolved in parallel, professionally and philosophically isolated from each other. Professionals in the nonmedical DD services culture embraced a set of philosophies and interests that rarely included the etiology-based approaches of the medical genetics culture (Hodapp and Dykens 1994). Just as the field of medical genetics has undergone profound changes over the past two decades related to diagnostic technology, changes involving disability rights and social support have transformed the nonmedical DD services world. As of 2012, there has been some encouraging cross-fertilization between the two groups, particularly at the academic research level with regard to behavioral and cognitive phenotypes. In the hands-on work of schools and service agencies, however, the practical implications of genetic diagnoses have yet to be realized, and much work still needs to be done to bring the two cultures together.

Caught in the middle of this divide are families struggling to navigate separate diagnostic systems and conflicting opinions on the value of genetic testing. This has a direct impact on the utilization of genetic services for the evaluation of DD. Wydeven et al.'s study in this issue of the Journal found that parents of children with autism spectrum disorders lack awareness about genetic services and/or experience obstacles in accessing such services within the current healthcare system. This is an important finding that highlights the continuing disconnect between genetic diagnostic advances and the everyday experiences of families living with DD.

For genetic counselors, the dichotomy between postnatal counseling about genetic disabilities is inextricably linked to its flip side, the discussion of choice related to prenatally-identified genetic diagnoses. The genetic counseling profession is unique in its ability to navigate both sides of this issue, although by its nature it is an uneasy conflict that causes us to regularly examine our relationship with the disability community (Madeo et al. 2011). Research by Farrelly et al. cuts to the heart of the dilemma by asking the question, "Where is the discussion about disability in prenatal diagnosis?". Such questions remind us as individuals and as a profession to continually examine genetic counseling assumptions and practices, particularly in the face of new prenatal diagnostic technologies as well as postnatal interventions for genetic conditions. Melissa Lenihan's firsthand account of how her work as a prenatal genetic counselor changed after the birth of her son with cerebral palsy offers a poignant window into her altered viewpoint from the other side of disability experience. Also speaking from a patient perspective, Anne Powell's startling narrative about pre- and postnatal genetic testing for Lesch-Nyhan syndrome in her child and brother reminds us that patients also struggle to walk the blurred line between disability advocacy and reproductive choice.

In addition to highlighting general aspects of diagnosis and genetic counseling of DD, this special issue includes articles on fragile X and 22q11.2 deletion syndromes, specific conditions that have particularly relevant implications for genetic counselors. When fragile X syndrome (FXS) was first identified many decades ago as a cause of DD in males, no one could have predicted that it would go on to be recognized as a complex disorder with rippling family implications across several areas of clinical practice. Among the most important aspects of this condition are the genetic counseling considerations, with an inheritance pattern that gives pause to even the most seasoned practitioners. Visootsak et al. describe a qualitative assessment of the experience of African American families both before and after a diagnosis of FXS, identifying potential challenges for genetic counselors working with this population. The National Society of Genetic Counselors' practice guidelines on counseling and testing for FMR1 mutations provide an updated reference on current best practices. From technological advances that improve our ability to interpret the implications of intermediate alleles, to targeted pharmaceutical treatments that will soon be a game-changer for pediatric and prenatal discussions of FXS, the evolution of knowledge about FMR1 mutations is arguably the most important development in DD research in the last half century.

The 22q11.2 deletion syndrome (22qDS), which encompasses outdated diagnostic entities such as velocardiofacial and DiGeorge syndromes, is not only common but highly variable. Its associated features include a wide range of physical, intellectual, and psychiatric effects, including a 25 % risk for schizophrenia. Despite this, Martin et al. found that genetic counselors do not consistently discuss the psychiatric aspects of 22qDS when the diagnosis is made. The authors surmise a number of possible reasons for this, including a general discomfort among genetic counselors related to the discussion of mental illness. Given the reticence of professionals to discuss certain aspects of the psychiatric phenotype, it is not surprising that parents of children with 22qDS also find it difficult to talk with their affected children about the diagnosis. Faux et al. surveyed parents to learn about their experiences discussing 22qDS with their children and found that many would welcome guidance in this area. The authors conclude that genetic counselors should play a role in preparing caregivers for conversations with their children about their genetic diagnosis.

Recognition of the impact on families of raising a child with special needs remains an important aspect of the psychosocial training of genetic counselors. Several articles in this issue bring together familiar themes of emotional reaction and family adaptation to DD. The personal stories by Iannuzzi, Schuler, and Nieder highlight experiences at different points in the parental journey. Recognizing that societal acceptance of children with DD directly affects family coping, Hurst et al. describe a school-based disability awareness program. Mathiesen et al. explore the value of parent-to-parent support networks for families of children with structural birth defects, reinforcing the important role of healthcare providers in fostering these connections. Navon points out that genetic counselors are intimately involved with advocacy organizations which by their nature offer syndrome-specific support and resources for parents. By contrast, families of children without a causative diagnosis may feel isolated, particularly as perceptions of DD move away from sweeping concepts such as "intellectual disability" toward more fine-tuned etiological characterizations. Lewis et al. document the development of an evidence-based information booklet to support parents of children without a known etiological diagnosis.

This special issue of the Journal includes commentaries from expert contributors outside the genetics field who view genetic counselors as an important bridge between etiological diagnoses and the social and educational service systems that support individuals with DD. Daniel Navon highlights the crucial role of genetic counselors in facilitating the formation of syndrome support organizations and expanding the knowledge base about genetic disorders. As a sociologist taking a fresh look at family support, Navon sees genetic counselors as activists in an era of "genotype-first" conditions waiting to be clinically described. He proposes that genetic counselors are particularly well-placed to "steer the new wave of genomic diagnoses towards the mobilization

of resources, research and community organization" because our unique training puts us at the crossroads of many different specialties.

Related to the advocacy role of genetic counselors is an understanding of the implications of genetic syndromes on child and family outcomes. Robert Hodapp and Elisabeth Dykens propose expanding the definition of syndromic phenotypes to include aspects of family functioning. Like Navon, they also see a role for genetic counselors in helping families and professionals to understand syndrome-specific effects of behavioral and non-behavioral symptoms and how such characteristics influence real-life outcomes. Elliott Simon's article on social service systems for people with intellectual disabilities acknowledges the growing importance of genetic information for enabling an integrated team approach to support affected individuals through the lifespan. Given our specialized training, genetic counselors have much to contribute, yet we remain on the periphery of the extensive DD social service system. Simon proposes a model that would foster a closer working relationship between genetic counselors and community-based supports and services for people with disabilities.

Taken together, the commentaries by Navon, Hodapp and Dykens, and Simon represent a call to action for genetic counselors to use our unique skill set toward more fully integrating genetic diagnoses into the care of families with DD. They challenge us to take on an expanded role beyond hospital walls and throughout the lifespan of people with genetic diagnoses. It is particularly compelling that this call comes from leaders in the DD world, given its past rejection of genetics practices. For our profession, this is a hopeful sign of new opportunities for genetic counselors to more fully support families with genetically-based DD outside of the traditional medical model.

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