General Principles in the Care of Children and Adolescents With Genetic Disorders and Other Chronic Health Conditions

ABSTRACT. The intent of this statement is to describe the breadth of issues that have special pertinence to pediatricians who care for children and families affected by genetic disorders and other chronic health conditions. The Committee on Children With Disabilities believes that because these children are leading healthier and longer lives, pediatricians are the more highly qualified to serve them, by virtue of their training and experience, and to provide them a “medical home.” This statement is designed to assist pediatricians in the treatment of these patients by describing their potential roles in relationship to their patients’ changing needs, as they work with various members of the health care team and as they respond to the requirements of government agencies and various third-party payers.

During the last decade we have witnessed dramatic changes in the diagnosis and treatment of genetic disorders in children that have fundamentally altered the way pediatricians view such conditions. Children with genetic disorders now live longer, most into adulthood. Using the latest scientific information, pediatricians are in the unique position of assisting children who have genetic conditions to reach their full potential. This goal is best achieved by facilitating the integration of the child and family into the community while minimizing the effects of the genetic condition on the child’s overall growth and development.

Individuals with genetic and other chronic health conditions and their families confront a seemingly endless series of stressors in their daily lives, many of which reflect having a chronic condition rather than being specifically related to the underlying disorder. These stresses may burden families emotionally, socially, and financially and may involve them with complex and often bureaucratic health, habilitation, education, and health insurance requirements. Pediatricians have a critical role in diagnosis, interdisciplinary planning, acute care delivery, and long-term treatment of children and adolescents with genetic disorders and other chronic health conditions.

Families play a central role in coordinating care and making decisions for the children. As the children mature, they may assume greater roles in the decision-making process. This process requires that pediatricians work with families to define and improve coping skills and to build a partnership that educates the parents, defines the care desired, and coordinates and assigns responsibility for the provision of that care. The role of each care provider should be clearly stated to the parent. Pediatricians have a key role in the provision of family-oriented, community-based services that recognize the following issues.

THE MEDICAL HOME

Lifelong chronic conditions are often characterized by periods of unexpected medical crises that may be life threatening, interspersed with periods of relative quiescence. Pediatricians should remain accessible through all these crises, providing a “medical home” for all such patients. They often advocate for the family when dealing with third-party payers, such as Medicaid, the Civilian Health and Medical Program of Uniformed Services, and other insurance companies for managed care arrangements. Pediatricians should also provide continuity of care to mitigate the adverse long-term physical, developmental, educational, and psychosocial consequences of genetic and other chronic conditions.

CARE COORDINATION

Ongoing care often involves the services of a multidisciplinary team of health care professionals, which may include other medical specialists and surgical subspecialists, nutritionists, genetic counselors, public health and school nurses, physical therapists, occupational therapists, speech therapists, audiologists, psychologists, and social workers. Services the team should provide include enhancement of coping skills, educational planning with the local school district, and access to care coordination and respite services. To fulfill their important growing role in managed care systems, pediatricians must become familiar with the skills offered by many disciplines and the role each professional plays in the care of the patient, and then they should help coordinate services for families so the needs of the patient are met in a seamless, cost-effective fashion.

TRANSITION

Increased vigilance by the pediatrician may be necessary during key periods of transition when new and sometimes difficult adjustments must be made by the child and family. Examples include the start of school, a job, or an intervention program; the changes faced during puberty and adolescence; and
living independently as an adult when health may deteriorate and continuity of care is potentially compromised. The pediatrician should focus special attention on the family when an infant enters an early intervention program, when a child begins school, when a child is hospitalized, as changes of puberty and adolescence occur, and during the child’s transition into the world of higher education, employment, and independent adult living. The process of changing physicians—from pediatrician to an adult health care provider—may itself be difficult for the patient and family.

COMMUNITY-BASED SERVICES

The pediatrician should know which community service programs are available for all children (eg, parks and recreation and libraries) in addition to important special public and private programs that support children with special health care needs. Working with the family and social services providers, the pediatrician can help ensure that the family receives the important services and information provided by voluntary agencies (eg, the Arc [formerly the Association for Retarded Citizens], United Cerebral Palsy Association, Cystic Fibrosis Foundation, Crohn’s and Colitis Foundation of America, Alliance of Genetic Support Groups, and National Organization for Rare Diseases), publicly sponsored programs (eg, early intervention programs, special education and related services, Supplemental Security Income, and mandated services of the state health departments through Title V of the Social Security Act), and patient information networks. To maintain current knowledge about services available in the public and private sectors, pediatricians may keep in contact with the local chapter or national office of the American Academy of Pediatrics or their state Office of Services for Children With Special Health Care Needs, or they may develop a relationship with a developmental pediatrics or chronic illness program at a local medical center.

COMPREHENSIVE SERVICES

Children with special health care needs, especially those with severe mental retardation, multiple disabilities, or vision, hearing, or neuromotor impairment, may have less access than their peers to needed general preventive health care services because of their family’s financial or socioeconomic status, the limited availability of child care services, limited access to transportation, and the severity of the disability. These children and their families may have difficulty locating health care providers who have sufficient experience with or training in the complex nature of these disabilities to provide them with necessary services. Such children and families may therefore need even greater attention from their pediatricians to ensure that they receive timely and appropriate developmental screening and evaluation, immunizations, sex education, and other counseling services.

In the last few years, impressive progress has been made in genetic services with the introduction of formidable new tools for screening, diagnosis, and treatment of a wide variety of inherited conditions. Because this progress will continue, pediatricians must stay abreast of the scientific advances while retaining their perspective on the art of the daily practice of medicine to meet the needs of children with genetic or other chronic health conditions and the needs of their families.

REFERENCES

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