Caring for Children with Down Syndrome and Their Families

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ABSTRACT

The overall purpose of this article is to provide pediatric nurses with the knowledge and motivation necessary to implement best clinical practice with children who have Down syndrome and their families. First, changes that have occurred in the care of children with Down syndrome are briefly reviewed. Next, recommendations concerning best clinical practice for children with Down syndrome are presented. Finally, implications for pediatric nurses practicing in an expanded role are discussed.

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Down syndrome, the most common chromosomal abnormality associated with mental retardation, occurs once in approximately every 600 to 800 live births (Hassold, 1999). According to the National Down Syndrome Society (2000), more than 350,000 persons in the United States have Down syndrome. Most persons with Down syndrome have an extra copy of chromosome 21 (Hattori et al., 2000). Down syndrome affects people of all ages, races, and socioeconomic levels (Pueschel, 1992).

Congenital abnormalities and diseases found in children with Down syndrome are the same as those that occur in the general population, but children with Down syndrome are affected more often and more severely with specific abnormalities and diseases than are typically developing children (Cohen, 1999). For example, children with Down syndrome are 10 to 30 times more likely to acquire leukemia than are children who do not have Down syndrome (Zipursky, Poon, & Doyle, 1992). Congenital heart disease occurs in 40% to 60% of children with Down syndrome, whereas only 8 of every 1000 persons in the general population have congenital heart disease (Freeman et al., 1998; Noonan, 1990). Children with Down syndrome have increased susceptibility to infections and a higher mortality rate from infectious disease than do children who do not have Down syndrome (Smith, 1995).

During the past few decades, fundamental changes have occurred in the care of children with Down syndrome (Pueschel, 2000a). These changes, which underscore the importance of the family and emphasize the need for health promotion and health protection activities, have helped persons with Down syndrome live longer and enjoy an improved quality of life (Nadel & Rosenthal, 1995). Unfortunately, not all children with Down syndrome receive this type of care. Some health care providers continue to follow outdated recommendations for the care of children with Down syndrome (Mayor, 1999; Spahis & Wilson, 1999; Van Riper, 1999a, 1999b). As a result, some children with Down syndrome experience needless health problems and a lower quality of life (Craze, Harrison, Wheatley, Hann, & Chessells, 1999; Rutter & Seyman, 1999). Although the denial of surgical intervention solely because of an infant's diagnosis of Down syndrome

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decreased substantially after the Baby Doe case in 1982, it still occurs (BBC News, 1998).

Pediatric nurses who are practicing in an expanded role are in an ideal position to help children with Down syndrome and their families benefit from the changes that have occurred in the care of children with Down syndrome. However, to help these children and their families, pediatric nurses need a working knowledge of health issues for children with Down syndrome and an awareness of recommendations concerning best clinical practice for children with Down syndrome.

The overall purpose of this article is to provide pediatric nurses with the knowledge and motivation necessary to implement best clinical practice with children who have Down syndrome and their families. First, changes that have occurred in the care of children with Down syndrome are briefly reviewed. Next, recommendations concerning best clinical practice for children with Down syndrome are presented. Finally, implications for pediatric nurses practicing in an expanded role are discussed.

CHANGES IN THE CARE OF CHILDREN WITH DOWN SYNDROME

Prior to the 1970s, children with Down syndrome were typically presented to their families as severely or profoundly retarded children "who will be a menace to society," "will never do anything," "will become a destructive force within the family," or "will have a negative influence on their brothers and sisters" (Pueschel, 1983, p. 1). Families were encouraged to relinquish the care of their children with Down syndrome and other disabilities to health care providers working in large, state-operated, residential institutions (Rynders, 1985). Once they were institutionalized, many of the children were deprived of all but the most elementary medical services (Pueschel, 2000a). Nutrition was substandard, immunizations were incomplete, infections were common, chronic health problems were treated inappropriately, and children with surgically correctable lesions were allowed to die (Pueschel, 1987). Professionals working in these institutions typically discouraged family members from visiting their children (Turnbull & Turnbull, 1990).

Families who chose not to institutionalize their children with Down syndrome were expected to enroll them in specialized programs and comply with the decisions and recommendations of health care providers and other professionals (Turnbull & Turnbull, 1990). Professionals generally viewed themselves as "the experts" in the care of children with chronic conditions. The expertise of family members was seldom acknowledged (Johnson, McGonigel, & Kaufman, 1989). Interventions were based primarily on the needs of the child as perceived by professionals, rather than on the needs of both the child and the family as perceived by family members. Not only were families expected to accept the decisions and recommendations of professionals, they were expected to be appreciative recipients of services (Turnbull & Turnbull, 1987). Family members who questioned the type or amount of services being provided were usually viewed as resistant, uncooperative, or noncompliant (Petr & Barney, 1993).

At the present time, formal education for persons with Down syndrome usually starts during infancy and continues through high school.

The current philosophy of care for children with Down syndrome proposes that the most appropriate and humane place for care is in the context of the family (biological or adoptive) (Haslam & Milner, 1992). Although some health care providers still encourage parents to institutionalize their newborn with Down syndrome, the reality is that most of the large, state-operated residential institutions have been closed for 10 to 20 years (Mattheis, 1999a). In addition, the institutions that remain open are unlikely to care for infants. Moreover, currently a waiting list exists for families interested in adopting children with Down syndrome. Finally, the overall findings from 3 programs of research concerning families of children with Down syndrome do not provide support for the notion that the experience of raising a child with Down syndrome is a negative experience (Cunningham, 1996; Gath, 1990; Van Riper, 1999a). In fact, many families have described the experience as positive and growth-producing. One mother wrote, "Our entire family and marriage is stronger. It has changed our view of the world, our view of ourselves, and others. It has made us more giving and less selfish. It has drawn us closer to God. It has caused us to be more concerned about others who are different. It has shown us what we value in life—relationships—not power and wealth. It has made us more content to just be!" (Van Riper, 1999a, p. 4).

At the present time, formal education for persons with Down syndrome usually starts during infancy and continues through high school. Generally, parents are encouraged to enroll their infant with Down syndrome in some type of early intervention program as soon as the infant is medically stable (Cohen, 1999). Once children with Down syndrome reach school age, some are fully integrated into regular classrooms with typically developing peers, while others are partially mainstreamed into regular classrooms for specific classes and activities. Unfortunately, despite growing evidence that children with Down syndrome do better when they are educated in integrated classrooms in their neighborhood schools (Freeman & Hodapp, 2000), many children with Down syndrome are still being educated in special schools or segregated classrooms.

Following high school, most persons with Down syndrome will enter the workforce. For some, this will be paid employment. For others, it will be volunteer work. Currently, it is rather unusual for persons with Down syndrome to pursue postsecondary education, but this is likely to change. Pueschel (2000a), a well-known expert in Down syndrome and the father of a recently deceased young man with

Down syndrome, believes that "through effective planning and development of appropriate support, post-secondary educational programs can provide people with Down syndrome opportunities to pursue their dreams and goals" (pp. 10-11).

As with typically developing young adults, many young adults with Down syndrome will move out of the family home after they finish high school. Young adults with Down syndrome need greater ongoing support and guidance than that required by typically developing young adults. Therefore, most young adults with Down syndrome who move out the family home will move into small group homes or supervised apartments. Generally, a paid provider or a family member will provide ongoing support and guidance, especially with transportation, budgeting, bill paying, and problem solving about health care issues.

Now that most children with Down syndrome are being raised by family members, a family-centered approach to care should be the norm rather than the exception. In a family-centered approach to care, health care providers and other professionals acknowledge and respect the pivotal role of the family in the life of the child with special needs (Shelton, Jeppson, & Johnson, 1992). Families are helped to achieve their best possible condition for promoting the growth and development of all family members, not just the family member with the chronic condition (Haas, Gray, & McConnell, 1992). Whereas proponents of family-centered care emphasize the importance of family-provider collaboration, they respect the right of family members to choose the level and nature of their involvement with health care providers and other professionals (Shelton et al., 1992). In addition, proponents of family-centered care acknowledge that competing demands may limit the extent to which family members can become actively involved in managing their child's care.

BEST CLINICAL PRACTICE FOR CHILDREN WITH DOWN SYNDROME

Medical and surgical advances have helped to extend the average life span for persons with Down syndrome from 9 to 55 years and have made it possible for persons with Down syndrome to develop further than previously thought possible (Hayes et al., 1997). For example, vigilance in infancy and early surgical repair of cardiac defects have greatly improved the long-term outlook for persons with significant heart disease and Down syndrome (Amark & Sunnegardh, 1999). Children with Down syndrome and acute myeloid leukemia are now being treated successfully with intensive chemotherapy without the need for bone marrow transplants (Craze et al., 1999). Early diagnosis and treatment of recurrent otitis media and sleep apnea as a result of upper airway obstruction have resulted in improved hearing, language development, and social interaction (Strome & Strome, 1992).

Although the primary emphasis of medical care for persons with Down syndrome continues to be the treatment of disease, increased attention has been allocated to health promotion and health protection (Castiglia, 1998; Cooley, 1995, 1999; Lovell & Saul, 1999; Saenz, 1999). In 1981, Dr Mary Coleman developed a Preventive Medicine Checklist that included information and recommendations specific to the health and developmental needs of persons with Down syndrome. The checklist has been revised a number of times, and most recently by the Down Syndrome Medical Interest Group (DSMIG) in 1999 (Cohen, 1999).

DSMIG was founded in 1994 with the express purpose of serving as a forum for professionals addressing aspects of medical care for persons with Down syndrome (Cohen, 1999). DSMIG promotes the highest quality of care for children and adults with Down syndrome by (a) fostering and providing professional and community education, (b) disseminating tools for clinical care and professional support (eg, health care guidelines), and (c) engaging in collaborative clinical research regarding issues related to the care of individuals with Down syndrome.

Included in the "Health Care Guidelines for Individuals With Down Syndrome: 1999 Revision" (Cohen, 1999) are current recommendations regarding best clinical practice for children with Down syndrome. The guidelines reflect contemporary health care standards and practices in the United States. The guidelines are based on the present level of knowledge concerning health care issues for persons with Down syndrome. Certain recommendations are clearly supported by empirical evidence (eg, evaluations for congenital heart disease), whereas others represent educated guesses by DSMIG members (eg, yearly screening for hypothyroidism). Further research is needed, and DSMIG members are committed to modifying the health care guidelines as new information becomes available.

The revised "Health Care Guidelines for Individuals With Down Syndrome" (Cohen, 1999) are available (*http://www.denison.edu/dsq/health99.shtml*) on the Web site for the *Down Syndrome Quarterly*, an interdisciplinary journal devoted to advancing the state of knowledge on Down syndrome. Therefore, only key recommendations will be addressed in this article.

Key Recommendations

Children with Down syndrome need the same immunizations and well-child care recommended for typically developing children by the American Academy of Pediatrics (Cohen, 1999). In addition, because of their increased risk for certain congenital abnormalities and diseases, children with Down syndrome need additional tests and evaluations (American Academy of Pediatrics, 1994).

Cardiac conditions. Dramatic changes have occurred in the diagnosis and treatment of children with Down syndrome and congenital heart disease (Reller & Morris, 1998). In the past, it was not uncommon for children with Down syndrome to be denied surgical intervention, even if their lesions were correctable (Amark & Sunnegardh, 1999). Today, surgical intervention is recommended for most children with Down syndrome who have correctable cardiac lesions. In children with Down syndrome, early increases in pulmonary vascular resistance tend to develop that reduce the left to right intracardiac shunt, minimize the heart murmur, and prevent respiratory problems and symptoms of heart failure (Cohen, 1999). Because of this, a serious cardiac defect may exist even if a murmur is not present. In children with Down syndrome who seem to be doing well clinically, serious pulmonary changes

may be developing. Therefore, it is recommended that all infants diagnosed with Down syndrome be promptly referred to a pediatric cardiologist for an evaluation that includes an echocardiogram (preferably before 3 months of age), then to a pediatric cardiac surgeon for early surgical repair if the infant has a correctable cardiac lesion (Clark, 1996). Children with Down syndrome who have congenital heart disease may need antibiotic prophylaxis prior to dental and surgical procedures (Buxton & Hunter, 1999).

Ears/audiology. It is estimated that more than 60% of children with Down syndrome have a hearing loss (Roizen, 1997). Because hearing plays a critical role in cognitive, social, and language development, it is recommended that all infants with Down syndrome have an objective measure of hearing (eg, auditory brain stem responses) performed at birth, if possible, or within the first 3 months of life (Nehring & Vessey, 2000). Then, a hearing evaluation should be done every 6 months until the child is 3 years old and yearly thereafter. Typical behavioral audiology requires a developmental age of 7 to 8 months. Therefore, when evaluating the hearing of a child with Down syndrome younger than 12 months of age, an objective measure of testing should be used. After 12 months of age, behavioral audiology may be appropriate.

Most children with Down syndrome have very small ear canals, making it difficult to examine them properly with the instruments typically found in primary care offices and clinics (Cohen, 1999). Consequently, it may be necessary to refer the child to an ear, nose, and throat (ENT) specialist who can use a microscopic otoscope to visualize the tympanic membranes (Saenz, 1999). All children with an abnormal hearing evaluation and/or tympanogram need to be seen by an ENT specialist so that treatable causes of hearing loss (eg, ear infections, fluid in the middle ear) can be treated aggressively with antibiotics and tympanostomy tubes as indicated. For some children, hearing aids may be indicated (Nehring & Vessey, 2000).

Growth. Growth in people with Down syndrome is a complex issue (Roizen, 1997). At birth, infants with Down syndrome typically weigh less and are shorter than unaffected children (Palmer et al., 1992). After the age of 2 or 3 years, many children with Down syndrome experience an untoward weight gain that often persists throughout their lives (Prasher, 1995). In a study by Rubin, Rimmer, Chicoine, Braddock, and McGuire (1998) concerning a prevalence to be overweight in 283 adults with Down syndrome, 45% of the men and 56% of the women were considered to be overweight according to the criteria established in Healthy People 2000. Although many persons with Down syndrome are overweight, evidence exists that their caloric intake tends to be reduced (Luke, Sutton, Schoeller, & Roizen, 1996). The increased prevalence of obesity among persons with Down syndrome is most likely the result of a combination of factors, such as reduced activity level, lower energy expenditures, hormonal factors, and genetic factors (Luke, Roizen, Sutton, & Scholler, 1994; Pueschel, 2000b).

Inappropriate growth and excessive weight gain have ramifications for motor performance and social acceptance, and thus nutritional counseling at an early age and regular assessments of weight and height are needed (Nehring & Vessey, 2000; Pueschel, 2000b). Both standard and Down syndrome growth curves (available at *www.growthcharts.com*) should be used (Cohen, 1999). The Down syndrome growth curve reflects the unique growth patterns of persons with Down syndrome. The standard growth curve is needed to plot height for weight. Any significant drop in growth percentile on either the standard or Down syndrome growth curve should be investigated (Saenz, 1999). The most common reason for failure to thrive in children with Down syndrome is undiagnosed cardiac defects. Leukemia is another possible cause of failure to thrive. Hypothyroidism is a possible cause of unexplained weight gain.

Thyroid disease. The incidence of thyroid disease is significantly increased among persons with Down syndrome of all ages (Roizen, 1997). Normal thyroid hormone levels are necessary for growth and cognitive functioning (Cohen, 1999). Because symptoms of thyroid disease may mimic symptoms generally associated with Down syndrome (Saenz, 1999), annual screenings for thyroid disorders (by monitoring thyroid-stimulating hormone and T4 levels) are recommended (Roizen, 1997). In the revised health care guidelines (Cohen, 1999), an additional thyroid screening is recommended when the infant with Down syndrome is 6 months of age.

Gastrointestinal disorders. Children with Down syndrome have an increased incidence of various gastrointestinal disorders. Some of these disorders (eg, constipation, celiac disease) can be treated with dietary management, whereas others (eg, tracheo-esophageal fistula, pyloric stenosis, duodenal atresia, Hirschsprung's disease, and imperforate anus) require surgical intervention. As noted previously, it is no longer considered acceptable for children with Down syndrome to be denied surgical intervention solely because of quality of life concerns. Most health care providers now recognize that children with Down syndrome who have gastrointestinal defects have a right to, and deserve, the same medical and surgical interventions that are offered to nondisabled children with such defects (Pueschel, 2000b).

One of the additions to the revised health care guidelines was a recommendation for annual celiac disease screening beginning at 2 to 3 years of age (Cohen, 1999). Celiac disease occurs in 7% to 16% of persons with Down syndrome (Carlsson et al., 1998). Persons with celiac disease have a lifelong intolerance to dietary gluten (eg, wheat, rye, and oats), resulting in small bowel mucosal damage (Pueschel, 2000b). Screening for celiac disease is best accomplished using IgA antiendomysium antibodies (Pueschel et al, 1999). Positive results should be followed up with a jejunal biopsy. Symptoms of celiac disease (eg, poor weight gain, diarrhea, bloating, fatigue, and irritability) usually resolve once a gluten-free diet is instituted.

Atlantoaxial instability (AAI). AAI, that is, increased mobility of the cervical spine at the level of the first and second vertebrae, is found in approximately 14% of all persons with Down syndrome (Cohen, 1998). The majority of persons with Down syndrome and AAI are asymptomatic, but approximately 1% of all persons with Down

syndrome have symptoms of AAI (eg, neck pain, torticollis, change of gait, and abnormal neurologic reflexes) when their spinal cord is compressed by the excessive mobility of the two vertebrae that form the atlantoaxial joint (Nehring & Vessey, 2000).

Routine screening for AAI is controversial (Cohen, 1998). Currently, DSMIG recommends screening all persons with Down syndrome for AAI between 3 to 5 years of age with lateral cervical radiographs in the neutral, flexed, and extended position and thereafter as needed for participation in Special Olympics (Cohen, 1999). Children with borderline or abnormal findings (ie, space between the posterior segment of the anterior arch of C1 and the anterior segment of odontoid process of C2 greater than 7 mm) should undergo a careful neurologic examination to rule out spinal cord compression. Neurologic imaging (computed tomography scan or magnetic resonance imaging) may be indicated. Significant change in a child's neurologic status necessitates further evaluation and possible treatment (ie, spinal fusion). Asymptomatic children with instability (5 to 7 mm) should be managed conservatively, with restriction only in sports that pose a risk for cervical spine injury (eg, football, gymnastics, and diving). Restricting all activities is not necessary. Also, repeat screenings at fixed intervals are no longer recommended because the value of repeat screenings has not yet been determined. DSMIG recommends (a) careful neurologic examinations of persons with Down syndrome, (b) immediate attention to symptoms of AAI, and (c) vigilance by ENT physicians and anesthesiologists during surgical procedures, which may hyperextend the neck (Cohen, 1999). Dentists also need to be aware of a positive history of AAI when they are manipulating the mouth of a child with Down syndrome (Desai, 1997).

Leukemia. In the past, many children with Down syndrome and leukemia received supportive care only (Stillar & Eatock, 1994), or they died from infections during the treatment (Creutzig et al., 1996). Although some health care providers are still reluctant to offer standard chemotherapy to children with Down syndrome, the degree of reluctance has decreased, and the outcomes for children with Down syndrome and leukemia have improved substantially (Craze et al., 1999). Findings from recent studies suggest that children with Down syndrome who have acute myeloid leukemia can be treated successfully with intensive chemotherapy without the need for bone marrow transplants (Craze et al., 1999). Recent findings also suggest that neonates with Down syndrome who have transient abnormal myelopoesis may achieve spontaneous remission (Zipursky, Brown, Christensen, & Doyle, 1999). Current recommendations are as follows: (a) neonates with Down syndrome in whom transient abnormal myelopoesis develops should be monitored closely because they may recover spontaneously and (b) children with Down syndrome and acute myeloid leukemia should receive standard intensive chemotherapy without bone marrow transplants (Cohen, 1999).

Alternative therapies—unconventional and controversial. Over the years, numerous unconventional and controversial therapies have been proposed for persons with Down syndrome. Typically, proponents of these alternative therapies claim the following: intellectual functioning will improve, facial characteristics will change, the number of infections will decrease, and/or the overall well-being of the person with Down syndrome will improve (eg, Whitaker, 1996). In most cases, these claims are based on anecdotal reports. To date, few of these claims have been supported in well-controlled studies (Cohen, 1999). In addition, there is growing concern that the use of these therapies may deplete existing family resources (eg, time, energy, and money). More importantly, some of these alternative therapies may actually harm persons with Down syndrome.

Van Dyke and Mattheis (1999) have divided the alternative therapies into 4 main categories: medical, physical, surgical, and educational. Some of the medical alternative therapies that have been used with persons who have Down syndrome include nutritional supplements, sicca cell treatment, and piracetam. Nutritional supplements are special mixtures of vitamins, minerals, amino acids, enzymes, and hormones. For more information on nutritional supplements for persons with Down syndrome, see Dr Len Leshin's Web site (*www.ds-health.com*). Sicca cell treatment or cell therapy involves the injection of freeze-dried fetal animal cells (usually from rabbits or sheep). Piracetam is a drug that is classified as a cerebral stimulant or nootropic. Existing research does not provide support for the use of any of these therapies with children who have Down syndrome. The use of megadoses of vitamins is of concern because of the potential adverse effects related to vitamin overdose, especially with the fat-soluble vitamins A and D. The use of piracetam with young children who have Down syndrome has not been adequately tested.

Craniosacral therapy and massage therapy are examples of alternative physical therapies (Van Dyke & Mattheis, 1999). Proponents of craniosacral therapy believe that the body's ability to heal itself can be restored through manipulation of the craniosacral system. Massage therapy involves gentle tactile stimulation of the body. To date, there have been no research studies demonstrating the effectiveness of these therapies with persons who have Down syndrome.

Alternative surgical therapies include cosmetic procedures to alter the facial appearance of persons with Down syndrome (Van Dyke & Mattheis, 1999). Possible alternations include reduction in tongue size, enlargement of nose and chin, and changes to the eyes and face. Currently, facial plastic surgery for persons with Down syndrome is very controversial (Cohen, 1999). People in favor of the surgery claim that it will help the person with Down syndrome speak more clearly and be better accepted by society. Studies to date do not provide support for these arguments.

Facilitative communication is an example of an alternative educational therapy (Van Dyke & Mattheis, 1999). Facilitative communication is a technique in which a "facilitator" provides physical support to the arms or hands of the individual with Down syndrome so that the person can communicate by pointing to symbols and words, or by using a keyboard (Cohen, 1999). Recent studies have not provided support for the use of this method.

At this time, the revised Health Care Guidelines for Individuals with Down

BOX 1 Red flags to watch for in evaluating alternative therapies

1. "Pseudo-science" claims and theories are presented as science, but they are not supported by research
2. Advocates of the therapy refuse to collaborate on objective research
3. Advocates of the therapy report that "there are no adverse effects to worry about"
4. Time passes and no new information appears concerning research results or possible adverse effects of the therapy
5. Information regarding the therapy is presented in closed meetings—questions are either not allowed or ignored
6. When questioned about the therapy, advocates of the therapy become aggressive and make personal attacks on their critics
7. Guilt and fear are used to promote the therapy (eg, "If you don't do this, you will never forgive yourself and your child will never catch up")
8. The alternative therapy is very expensive
9. "Before" and "after" photographs are presented as support of the alternative therapy

Data from Mattheis, 1999b.

Syndrome (1999 Revision) does not include specific recommendations concerning the use of unconventional and controversial therapies with persons who have Down syndrome (Cohen, 1999). However, considering the amount of time currently being spent discussing these therapies during health care visits and support group meetings, it appears that the need for such recommendations is increasing rapidly. Currently, members of DSMIG are in the process of carefully evaluating existing findings concerning these therapies (eg, anecdotal reports, descriptive information, and the results of well-controlled studies). Philip Mattheis (1999b), a member of DSMIG, has listed 9 "red flags" to watch for when evaluating alternative therapies (Box 1).

BOX 2 Resources for families of children with Down syndrome

National organizations

March of Dimes, *www.modimes.org,* (888)MODIMES

National Down Syndrome Society, *www.ndss.org,* (800) 221-4602

National Association for Down Syndrome, *www.nads.org*

National Down Syndrome Congress, (800) 232-6372

Books

Berube, M. (1998). *Life as we know it: A family, a father, and an exceptional child.* New York: Vintage.

Kingsley, J., & Levitz, M. (1994). *Count us in: Growing up with Down syndrome.* New York: Harcourt Brace Jovanovich.

Kumin, L. (1994). *Communication skills for children with Down syndrome: A guide for parents.* Bethesda: Woodbine House.

Meyer, D. (1997). *Views from our shoes: Growing up with a brother or sister with special needs.* Bethesda: Woodbine House.

Stray-Gunderson, K. (1995). *Babies with Down syndrome: A new parents guide* (2nd Ed.). Bethesda: Woodbine House.

Trainer, M. (1991). *Differences in common: Straight talk on mental retardation, Down syndrome, and life.* Rockville, MD: Woodbine House.

Winders, P. (1997). *Gross motor skills in children with Down syndrome: A guide for parents and professionals.* Bethesda: Woodbine House.

Other resources

Dr Len Leshim's Comprehensive Web site: *www.ds-health.com*

Down Syndrome Quarterly, www.denison.edu/dsq

IMPLICATIONS FOR PEDIATRIC NURSES PRACTICING IN AN EXPANDED ROLE

The incidence of Down syndrome is high enough that most pediatric nurses are likely to care for a number of children with Down syndrome during the course of their careers. Conversely, the occurrence is rare enough that only a minority of pediatric nurses will have extensive experience in caring for children with Down syndrome. Therefore, it is essential that pediatric nurses make use of established guidelines concerning the health care of persons with Down syndrome. Moreover, it is crucial that the guidelines used are those that have been developed and periodically revised by health care providers with expertise in the care of persons with Down syndrome.

Pediatric nurses practicing in an expanded role have the power to change both the nature and the quality of the care that children with Down syndrome and their families receive. For example, they can help shift the focus of care from disease management to health promotion and health protection. They can make sure that family members and other health care providers working with the family not only have access to, but follow, the Health Care Guidelines for Individuals with Down Syndrome: 1999 Revision. They can include a flow chart, such as the Down Syndrome Health Care Guidelines (1999 Revision) Record Sheet (Cohen, 1999) in the front of the child's medical record for rapid consultation (see Figure). They can refer children with Down syndrome and their families to local early intervention/infant stimulation programs.

Although there is growing evidence that many families are adapting well to the ongoing challenges associated with raising a child with Down syndrome, evidence also exists that some families are having a difficult time (Cunningham, 1996; Van Riper, 1999a, 1999b). For example, in a recent study concerning families of children with Down syndrome, one mother wrote, "We as a family, I feel, are doing very well. We try to be individuals and yet keep a close family relationship. We are all interested in each other's goals, talents, and challenges. We try to take an active part in these interests when we can. This means lots of juggling but we make it work. The key is connecting as a family and communicating" (Van Riper, 1999a, p. 3).

In contrast, another mother wrote, "Our family has been hanging from a string since N's birth. We have financially struggled, mentally struggled.... We (my husband and I) don't talk much. Our daughter had to grow up a little faster than others. She doesn't see me much lately. Trying to get

Name: _____

Birthdate: _____

DOWN SYNDROME HEALTH CARE GUIDELINES (1999 Revision) RECORD SHEET

AGE, in years

Medical Issues	At Birth or at Diagnosis	6 mo	1	1 1/2	2	2 1/2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20-29
Karoytype & Genetic Counseling		■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■
Usual Preventive Care																								
Cardiology	Echo	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■
Audiologic Evaluation	ABR or OAE																							
Ophthalmologic Evaluation	Red reflex			■		■																		
Thyroid (TSH & T_4)	State screening			■		■																		
Nutrition																								
Dental Exam [1]		■	■	■	■																			
Celiac Screening [2]		■	■	■		■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■
Parent Support																								
Developmental & Educational Services	Early Intervention																							
Neck X-rays & Neurological Exam [3]		■	■	■	■	■	X-Ray																	
Pelvic exam [4]		■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■					
Assess Contraceptive Need [4]		■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■					
Pneumococcal Vaccine		■	■	■		■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■	■

Instructions: Perform indicated exam/screening and record date in blank spaces.

[1]Begin Dental Exams at 2 years of age, and continue every 6 months thereafter.

[2]IgA antiendomysium antibodies and total IgA.

[3]Cervical spine x-rays: flexion, neutral and extension, between 3-5 years of age. Repeat as needed for Special Olympics participation. Neurological examination at each visit.

[4]If sexually active

FIGURE Down Syndrome Health Care Guidelines (1999 Revision) Record Sheet. (Reprinted from Health Care Guidelines for Individuals With Down Syndrome: 1999 Revision [Down Syndrome Preventive Medical Check List], published in *Down Syndrome Quarterly* [Volume 4, Number 3, September 1999, pp. 1-16.] Complete Guidelines also available at *www.denison.edu/dsq/health99.shtml*. Reprinted, duplicated, and/or transmitted with permission of the Editor. Information concerning publication policy or subscriptions may be obtained by contacting Dr Samuel J. Thios, Editor, Denison University, Granville, OH 43023 [e-mail: thios@denison.edu].)

enough money to pay a couple months worth of bills.... We've stayed married so far and kept our house—this is a major accomplishment" (Van Riper, 1999a, p. 3).

By taking the time to discover how families define and manage the experience of raising a child with Down syndrome, pediatric nurses can tailor the plan of care so that it is based on the unique values, beliefs, strengths, and resources of the family. Some families may view the experience as a never-ending tragedy, while other families may view it as a positive, growth-producing experience. The way in which a family defines its situation will influence all aspects of the child's care. For example, it will influence whether family members want to take an active role in promoting and protecting the child's health. In addition, it will influence the type of interventions family members are willing to allow for their child. It will also influence the family's ability to use its strengths and resources to successfully manage the ongoing challenges associated with raising a child with Down syndrome. Depending on how they define their situation, a family with very limited resources may actually be more successful in managing the challenges than a family with abundant resources.

Pediatric nurses need to be aware of possible resources for families of children with Down syndrome. Examples of possible resources are shown in Box 2. Not all families will benefit from, or want, the same resources. For example, most parents of children with Down syndrome find it helpful to talk with other parents of children with Down syndrome. Some parents may want to get involved with a local support group or attend the annual conference for a national organization like the National Down Syndrome Congress. In contrast, other parents may find it easier to use the Internet to "chat" with other parents.

Recent advances in genetics will have a dramatic impact on the care of children with Down syndrome. For example, the complete sequencing of chromosome 21 (Hattori et al., 2000) will help scientists to better understand the pathogenesis of diseases commonly associated with Down syndrome and may lead to new therapeutic approaches. The recently developed chromosome 21 gene catalogue (Hattori et al., 2000) will open new avenues for deci-

phering the molecular bases of Down syndrome. Pediatric nurses need to be aware of the ongoing research concerning children with Down syndrome, and they need to incorporate significant research findings into their practice. In addition, pediatric nurses need to get more involved in planning and conducting research concerning children with Down syndrome and their families. Most importantly, pediatric nurses need to remember that children with Down syndrome are first and foremost children. Therefore, like other children, they deserve nothing less than state-of-the-science, family-centered care.

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LITERATURE REVIEW

Berenson, A. R., Chacko, M. R., Wiemann, C. M., Mishaw, C. O., Friedrich, W. N., & Grady, J. J. (2000). A case-control study of anatomic changes resulting from sexual abuse. *American Journal of Obstetrics & Gynecology, 182,* 820-834.

This study by Berenson et al. will be remembered as a classic research investigation that compared genital characteristics of sexually abused and nonabused prepubertal females (ages 3 to 8 years) using photographic documentation of the subjects' genital anatomy. These researchers used a comparative case-study design that controlled for many key variables. The steps used to control for age, race, pubertal stage (breast development), and examiner bias between the two populations differentiate this study from earlier studies seeking to identify genital findings in female pediatric patients who were sexually abused. All of these variables have been problematic in interpreting the results from prior studies. However, strict screening criteria for inclusion as either a child victim of sexual abuse or a child without a prior history were established in the study by Berenson et al.

The abused population (n = 192) was composed of children with a history of digital or penile penetration who were referred to a specialized child protective clinic. The researchers developed the Digital/Penile Vulvar Penetration Rating Scale and used this tool as an indicator to judge the likelihood of sexual penetration. Similarly, the nonabused children (n = 200) were screened using several interview criteria and the Child Sexual Behavior Inventory. They were patients seen at a university-affiliated pediatric clinic.

The three medical examiners who performed the genital examinations were trained so that standard physical examination techniques and photography views were consistent. Film processing, the type of gloves used, and the number of slides taken were also standardized. After the film was developed, the examiners separately reviewed all slides and identified genital findings but did not know whether the slide was from an abused or nonabused child.

Vaginal discharge was the only significant finding observed more frequently in abused than in nonabused prepubertal females. Other findings such as hymenal bumps or tags, friability, external ridges, increased vascularity, and bands did not differentiate the groups. Hymenal perforations, transactions, and deep notches were observed only in children (n = 4) with a history of abuse.

Data from this study will help pediatric health care professionals determine whether anatomic findings in a young child's genital area are the result of traumatic abuse or represent normal anatomy or variants of normal that are not necessarily associated with sexual abuse. This article presents a thoughtfully planned out and carefully controlled study. After reading about this study, the research interest of pediatric nurse practitioners should be sparked to embark on their own research investigations to answer the patient care questions that intrigue each of us.

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