ANC200002_17-24.qxp 1/23/09 2:20 AM Page 17

CE 2.3 HOURS Continuing Education

Assessment and Care of the Newborn With Down Syndrome

Rachel Ranweiler, MS, NNP

ABSTRACT

Well-defined, distinctive phenotypic features and natural history characterize Down syndrome, the most frequent form of developmental disability caused by a microscopically demonstrable chromosomal aberration. Triplicate state or trisomy of all or a portion of chromosome 21 causes Down syndrome. There are 3 genetic mechanisms leading to Down syndrome, or trisomy 21: nondisjunction, translocation, and mosaic. With advances in prenatal testing techniques, the diagnosis can occur before birth. This article explores the embryology and pathogenesis of Down syndrome and its multisystemic effects on the newborn. Specific attention is paid to presentation, clinical features, physical assessment, and family support. Recurrence risk and genetic counseling are discussed including the relationship of the risks and benefits of performing diagnostic procedures. In addition, this article reviews the management of healthcare needs for newborns with Down syndrome and the implications for nursing care.

KEY WORDS: chromosomes, Down syndrome, neonate, trisomy 21

Down syndrome is named after John Langdon Down, who first described the syndrome in 1866. Down syndrome, or trisomy 21, is the most common genetic cause of intellectual disability and best known of the chromosome disorders.[1,2] *Smith's Recognizable Patterns of Human Malformation* provides an overall appraisal of Down syndrome with an incidence of 1 in 660 newborns, in all races and economic groups, making it the most common pattern of malformation in man.[1,3] According to the Centers for Disease Control and Prevention, in 2006, the incidence of Down syndrome was 1 in 733 live births.[4,5] Down syndrome is a cytogenetic disorder caused by an error in cell division that results in the presence of an additional third chromosome 21 or "trisomy 21" (Figure 1). Approximately 5500 infants are born with Down syndrome in the United States each year.[4,6,7] Early recognition of Down syndrome in the newborn is essential to provide appropriate care for the newborn and support for parents and family. Down syndrome is the most prevalent malformation syndrome caused by an abnormal chromosomal condition in newborns.[1,8] The syndrome results when there are 3 rather than 2 copies of chromosome 21 in all or most cells of the body. The additional genetic material alters the course of development and causes the characteristics associated with Down syndrome.

EMBRYOLOGY/PATHOGENESIS

Although the chromosomal basis of Down syndrome is clear, the reason for the chromosome abnormality is still poorly understood.[3,9,10] The vast majority (>90%) occurs secondary to meiotic nondisjunction, which leads to the presence of an extra chromosome 21 in all cells of the individual. In such cases, the extra chromosome originates in the development of either the egg (oogenesis) or the sperm (spermatogenesis). Normally during oogenesis or spermatogenesis, each parent contributes 23 chromosomes, one-half of each pair, to the developing zygote through a process of cell division called meiosis (Figure 2). During meiosis I, the chromosome number is reduced from 46 to 23 as the cell divides. These cells are referred to as haploid, having half the number of chromosomes. During meiosis II, each haploid cell divides its 2 DNA threads (chromatids) between daughter cells, producing additional gametes, each

Address correspondence to Rachel Ranweiler, MS, NNP, P/SL Medical Center, 1719 East 19th Ave, Denver, CO 80218; rachelehlers@msn.com.

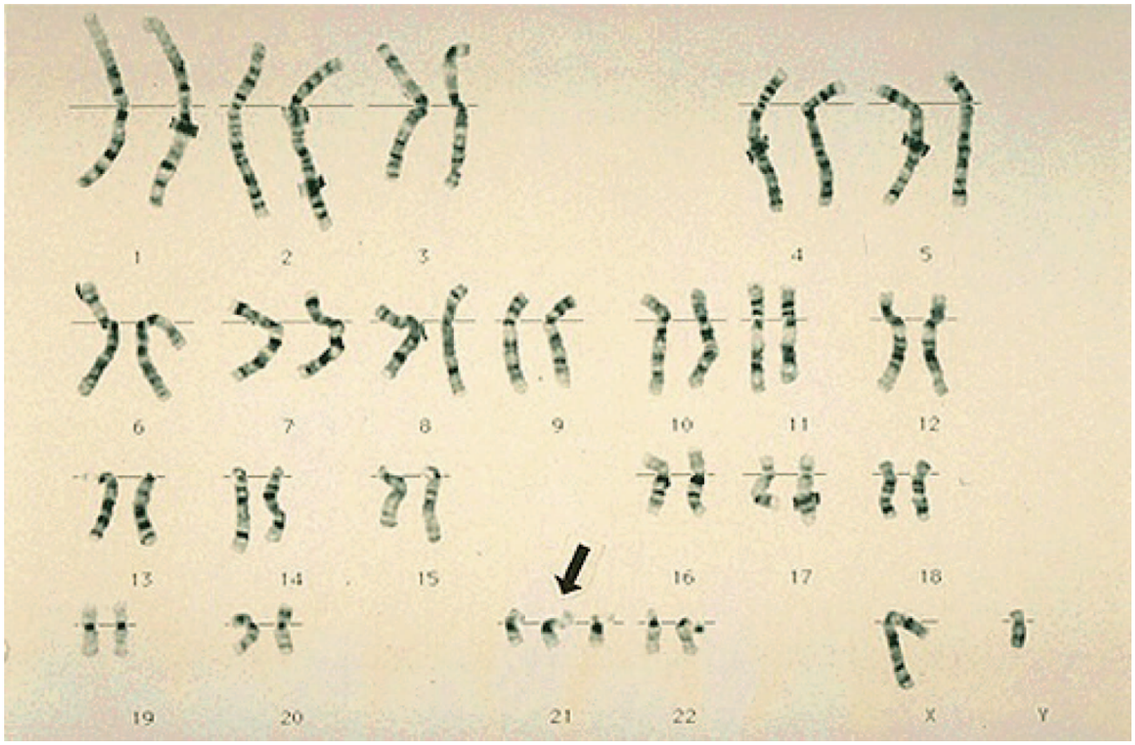
Author Affiliation: P/SL Medical Center, Denver, Colorado.

The author has no significant ties, financial or otherwise, with any company that may have an interest in the publication of this educational activity.

Copyright © 2009 by the National Association of Neonatal Nurses.

ANC200002_17-24.qxp 1/23/09 2:20 AM Page 18

FIGURE 1.



The presence of an extra chromosome 21. Reprinted with permission from Milner and Herber.[2]

with 23 chromosomes. In spermatogenesis, the final product is 4 functional gametes (sperm). In oogenesis, 1 primary oocyte gives rise to 1 oocyte and 3 nonfunctional polar bodies.[3,9]

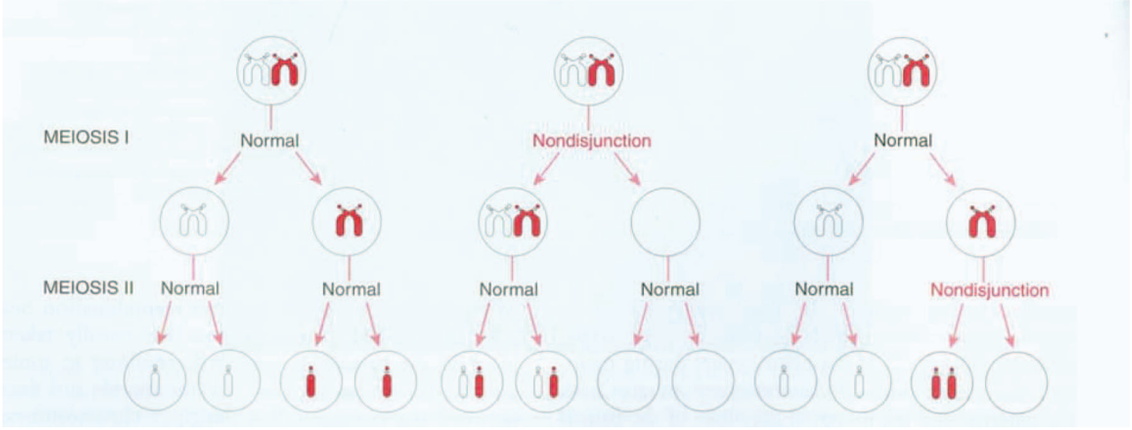
Nondisjunction refers to the failure of a pair of chromosomes to disjoin properly during one of the two meiotic divisions, usually meiosis I. The consequence of nondisjunction is a gamete with 24 chromosomes rather than 23 (Figure 2).[3]

The high percentage of all cases of trisomy 21 in which the abnormal gamete originated during maternal meiosis I suggests that something about maternal meiosis I, likely related to increased maternal age, is a correlating factor. It has been suggested that the older the oocyte, the greater the chance that the chromosomes will fail to disjoin correctly.[3,9,11] Consequently, when the egg and the sperm unite to form the fertilized egg, 3, rather than 2, chromosomes 21 are present. As the embryo develops, the extra chromosome is repeated in every cell. In this condition, nonfamilial trisomy 21, 3 copies of chromosome 21 are present in all cells of the individual. The risk of having a child with trisomy 21 increases with maternal age, especially after the age of 30 years.[3,9,11,12]

Occasionally, chromosomes break, and pieces of one chromosome attach to another. This is called a translocation. Translocations can be balanced, in which case breakage and reunion occur between 2 chromosomes, but no critical genetic material is lost and individuals show no effects. They may be unbalanced, in which case part of one chromosome is lost or duplicated, resulting in missing or extra genetic material.[3] Approximately 3% to 5% of individuals with Down syndrome have cells containing 46 chromosomes but still have the features associated with the syndrome because they have the extra genetic material from an additional copy of all or part of chromosome 21.[3] It is simply attached to another chromosome. These cases are due to a translocation, either occurring de novo or passed from a balanced translocation-carrier parent, which becomes unbalanced and trisomic in the baby. In such situations, material from one chromosome 21 gets translocated onto another chromosome, either prior to conception or at conception. Typically, the translocated chromosome 21 attaches to another acrocentric chromosome (often chromosome 14), resulting in a Robertsonian translocation.[3,9,11] Cells from individuals with translocation trisomy 21 have the usual 2 chromosomes 21 but also have additional chromosome 21 material on the translocated chromosome. This overexpression of chromosome 21 results in features associated with Down syndrome. Down syndrome resulting from translocation shows no relation to maternal age but has a relatively high recurrence risk in families when

ANC200002_17-24.qxp 1/23/09 2:20 AM Page 19

FIGURE 2.

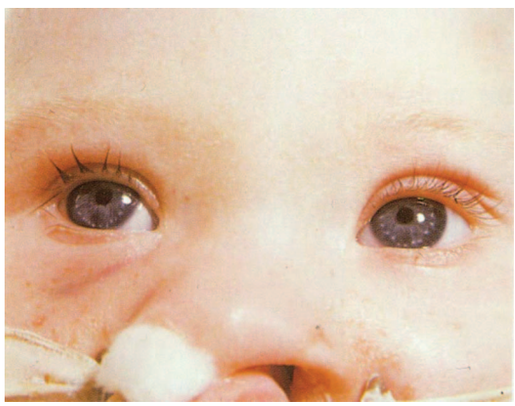


Meiosis and nondisjunction. Reprinted with permission from Nussbaum et al.[3]

a parent, especially the mother, is a carrier of the translocation.[3,9,11,13]

Mosaicism can occur in 2 ways. Occasionally, a nondisjunction can occur during mitosis, early in embryonic cell division. The other is that the zygote begins with a typical nondisjunction trisomy 21 but somehow the extra copy of 21 is lost or dropped in one or more cell lines, resulting in mosaicism. Mosaic trisomy 21 accounts for a small percentage of individuals with Down syndrome, most estimates conclude about 1%.[14-16] In this situation, the extra chromosome 21 is present in some, but not all, cells of the individual. An individual with mosaic trisomy 21 will have 46 chromosomes in some cells but will have 47 chromosomes in others. There is great variability in expression of the disorder among those with mosaic Down syndrome.[3,16] Depending on the proportion of cells that carry the additional chromosome 21, the phenotype may be milder than that of typical trisomy 21.[9,11]

FIGURE 3.



Common facial features of Down syndrome. Note flat nasal bridge, epicanthal folds, and Brushfield spots on the iris. Reprinted with permission from Milner and Herber.[2]

Clinical Presentation/Systemic Physical Examination

Down syndrome can usually be diagnosed at birth or shortly thereafter by its dysmorphic features, which vary among patients but nevertheless produce a distinctive phenotype.[4,10,17] Accurate assessment and description of external clinical features will assist in recognition of the newborn with Down syndrome and identify the need for further diagnostic evaluation.[6]

Begin with examination of growth and plot weight, length, and head circumference on a growth chart. Specialized growth charts are available for newborns, older infants, and children with Down syndrome. Growth parameters may be normal at birth, with decreased postnatal growth velocity being common.[9]

Examine the head, evaluating shape and fontanelles. Brachycephaly with a short and round skull with a flat occiput or mild microcephaly may be present. A large anterior fontanel or a third fontanel along the sagittal suture line may be noted. Hair may be fine and soft. The neck is usually short with loose skin at the nape. Ears are typically not low set but are smaller and may appear posteriorly rotated. To determine placement, imagine a line from the inner canthus to the outer canthus of the eye toward the ear. If the ear falls below this line, it is low set.[17,18]

Next, examine the facial features. The newborn with Down syndrome may have a flat facial profile including a small nose and a flat nasal bridge (Figure 3). Eyes may have an upward and outward slant with prominent epicanthal folds. Look for grayish white spots around the iris of the eye (Brushfield spots) (Figure 3). The tongue is typically of normal size but

ANC200002_17-24.qxp 1/23/09 2:20 AM Page 20

may appear large. Two important factors leading to the appearance of macroglossia are smaller oral cavities leading to a relative fit issue and the low tone allowing the jaw to hang slack and the tongue to protrude.[12,19]

Examine the extremities. Infants with Down syndrome typically have short fingers with broad, square hands and low-set thumbs that are separated more than usual from the second finger. Clinodactyly of the fifth finger may be seen. A single, deep transverse crease on the palm of the hand (simian crease) may be present on one or both hands (Figure 4). Examination of the feet will reveal a wide space with a plantar crease between the great toe and the second toe (Figure 5).

Overall, muscular hypotonia is seen in approximately 80% of newborns with Down syndrome.[20-23] Other general physical findings may include short stature, hyperflexibility of joints, and dysplasia of the pelvis with a narrow acetabular angle. Cutis marmorata (marbling of skin) is common.[17]

Listen to the chest for the presence of a murmur or abnormal heart sounds. Congenital heart defects are seen in approximately 40% of infants with Down syndrome.[3,11,16] Examine the newborn's peripheral pulses, color, and work of breathing for other evidence of congenital heart disease.

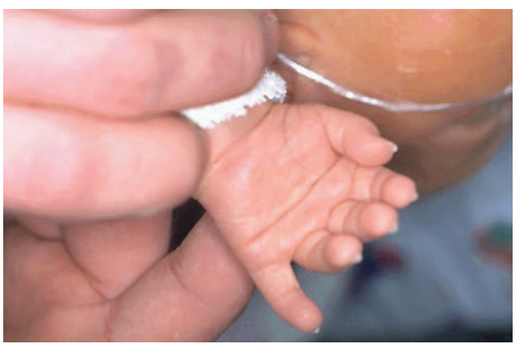
Examine the abdomen for presence of bowel sounds, masses, and organomegaly. Distension and/or the presence of bilious emesis may indicate a bowel obstruction. Check to ensure that the newborn has a patent anus.

Other Clinical Findings

Numerous other birth defects and medical disorders have been described in association with Down syndrome. The extra chromosome found in Down syndrome interferes with mental development and causes developmental delays. Average IQs can range from 25 to 70s.[20]

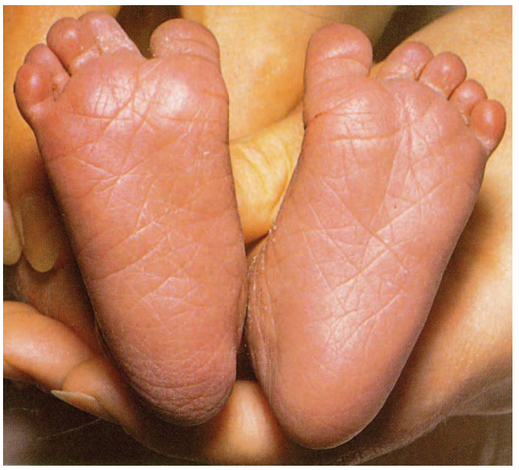
Congenital heart disease occurs in 40% to 45% of infants with Down syndrome.[14,16] The most common cardiac abnormalities are endocardial cushion defects (atrioventricular canal), ventricular septal defects, atrial septal defects, and patent ductus arteriosus.[22] An echocardiogram should be produced for all infants to rule out the presence of congenital heart disease, even in the absence of a cardiac murmur or other symptoms of heart disease. Gastrointestinal abnormalities also occur more commonly, in particular, duodenal atresia, tracheoesophageal fistula, omphalocele, pyloric stenosis, and Hirschsprung's disease occurring in 10% to 15% of those with Down syndrome.[1,3,24]

FIGURE 4.



Single palmer crease. Reprinted with permission from Nussbaum et al.[3]

FIGURE 5.



Widely separated first and second toes. Reprinted with permission from Milner and Herber.[2]

Congenital hypothyroidism, characterized by a reduced basal metabolism, an enlargement of the thyroid gland, and disturbances in the autonomic nervous system, can occur more frequently in babies with Down syndrome. Follow-up for development of hypothyroidism is essential. Several other known medical conditions are more prevalent among those with Down syndrome including vision disorders such as cataract and hearing loss because the external ear and the bones of the middle and inner ear may develop differently.[17,18,20,21,24,25] More significantly, the factor leading to high rates of hearing loss in individuals with Down syndrome is conductive loss related to frequent otitis media.[25] Atlantoaxial instability, an infrequent but serious complication related to Down syndrome, is a malformation of the upper part of the spine located under the base of the skull, which can cause spinal cord compression if not treated properly.[17,20]

Approximately 1 in 150 children with Down syndrome develops acute lymphoblastic leukemia.[9] Transient myeloproliferative disorder associated with spleenomegaly and hepatic fibrosis may occur at birth and usually resolves spontaneously in the first year of life.[3,11] Neonatal thrombocytopenia is also associated with Down syndrome.[8]

ANC200002_17-24.qxp 1/23/09 2:20 AM Page 21

Diagnostic Studies/Risk Factors

Down syndrome can be detected prenatally by cytogenetic studies. Karyotyping is necessary for confirmation, for providing a basis for genetic counseling, and for determining the recurrence risk. In about 95% of all patients, Down syndrome involves trisomy for chromosome 21, resulting from meiotic nondisjunction of the chromosome 21 pair.[1,3,4] The meiotic error responsible for the trisomy usually occurs during maternal meiosis (90% of the time) but can also occur in paternal meiosis (10% of the time).[3,11]

To provide accurate recurrence risk information to parents, it must be determined whether the child with Down syndrome has a translocation. If a translocation is present, parents should be offered chromosome analysis to determine whether one of them is a carrier of a balanced translocation as this may increase the risk for recurrence and may also have implications for other family members[1,27-30] Recurrence risk for the rare translocation carrier parent will depend on the type of translocation and the sex of the parent.[1,28] This is because 1 of the 2 parents may be a balanced carrier of the translocation. The translocation occurs when a piece of chromosome 21 becomes attached to another chromosome, often number 14, during cell division. If the resulting reproductive cells receive the chromosome with the piece of chromosome 21 attached and retain the chromosome 21 that lost a section, then the reproductive cells contain the normal or balanced amount of chromosome 21.

If the translocation is de novo, that is, neither parent has a balanced translocation, then there is no increased risk of recurrence. The translocation is de novo about half the time. If the mother carries a balanced translocation (typically on chromosome 14), then the risk for future pregnancies is about 10% to 15%. If the father carries the balanced translocation, the risk is about 3% to 5%. Rarely, the translocation involves both of the chromosomes 21. In this circumstance, the carrier parent of the 21/21 translocation would have a 100% risk of recurrence. Genetic counseling should be offered to all parents of a child with Down syndrome for review of the chromosomal cause of their specific recurrence risks. The genetic counselor can also determine whether any other family members may also be at increased risk for having a child with Down syndrome or for miscarriage due to the possibility of an inherited translocation.

Researchers have established that the likelihood that a reproductive cell will contain an extra copy of chromosome 21 increases dramatically as a woman ages.[3,9,11,28] Although younger mothers have a much lower risk, their birthrate is so much higher that more than half of the mothers of babies with Down syndrome are younger than 35 years.[9] The relationship of Down syndrome incidence to mother's age is as follows: 15 to 29 years, 1:1500; 30 to 34 years, 1:800; 35 to 39 years, 1:270; 40 to 44 years, 1:100; and more than 45 years, 1:50.[1] The risk of Down syndrome due to translocation or mosaic trisomy is unrelated to maternal age. The paternal age appears to have no influence on the risk.[1,28] The probability that another child with Down syndrome will be born in a subsequent pregnancy is somewhat in dispute. Some authors state that the risk is about 1%, regardless of maternal age, whereas others state that the risk is about 1% plus maternal age–related risk.[3,7] Although a low figure, it is enough to justify amniocenteses for chromosome studies on any future pregnancy.

Prenatal screening for Down syndrome includes maternal serum screening. Serum markers for Down syndrome include maternal serum alpha-fetoprotein, chorionic gonadotropin, and unconjugated estriol. While these measurements are not a definitive test for Down syndrome, a lower maternal serum alpha-fetoprotein value, a lower unconjugated estriol level, and an elevated chorionic gonadotropin level, on average, suggest an increased likelihood of a fetus with Down syndrome and additional diagnostic testing may be desired.

Prenatal diagnostic tests can be performed to determine the occurrence of Down syndrome. Most commonly, an amniocentesis in performed. Amniocentesis, done during the 14th to 18th weeks of gestation, is the removal and analysis of a small sample of fetal cells from the amniotic fluid. In addition, diagnostic ultrasound can reveal fetal malformations before birth. Increased nuchal translucency as seen by ultrasound is associated with trisomy 21. The cause of the transient fluid accumulation in the neck is unknown although the appearance at birth is usually normal.[29]

Nursing Care

Implications for nursing care, when presented with an infant exhibiting the clinical manifestations of Down syndrome, include providing a thorough, systematic, head-to-toe physical assessment of the newborn and offering information and support as needed.[31,32] The birth of an infant with a congenital anomaly or the birth of an infant who is acutely ill elicits feelings of loss, guilt, and confusion for parents. Nurses must expect grief reactions and help the family cope with the crisis. However, it is important to note that not all parents respond the same, and the diagnosis of Down syndrome may not be seen as a crisis for all new parents. Strategies to help parents cope include support for early contact between the parents and the infant and explanations with factual information of the infant's condition and plan of care. It is important that the information include a good

ANC200002_17-24.qxp 1/23/09 2:20 AM Page 22

balance of the challenges as well as the possible positives such as the love, joys, and rewards that can come from raising any child. New parents are often given nothing but the details of everything that can go wrong. Parents also need reasons to be hopeful about their child. Anecdotal information about particular individuals with Down syndrome, information about early intervention, and the opportunity to contact other parents of children with Down syndrome are helpful. The lines of communication must be kept open to reinforce information that the family may have trouble processing and to assist the family in responding to its grief. If diagnosed prenatally, consultation with those previously involved with the family and notification to the primary care provider for immediate follow-up and support to the family can be an anticipated nursing intervention. In addition, providing the family with written information on local and national support groups can be beneficial. The nurse may offer to call a local support group on behalf of the parents. Many parents may want this support but find it difficult to make the call themselves.[17,32]

The passage in October 2008 of the Prenatally and Postnatally Diagnosis Conditions Awareness Act recognizes the need for physicians and other healthcare professionals to educate families who receive a prenatal or postnatal diagnosis of Down syndrome and other conditions. The bill authorizes the federal government, through the Department of Health and Human Services, to providing funding for grants to provide "up to date, scientific information about life expectancy, clinical course, intellectual and functional development, and prenatal and postnatal treatment options."[4] It will also aid in creating support services for families, such as Web sites, information clearinghouse, adoption registries, and parent support groups, and for providing physicians and other healthcare providers with current, evidence-based information about Down syndrome and other conditions.[4,26]

Fundamental changes have occurred in the past few decades to promote the best clinical practice for children with Down syndrome. Last revised in 1999, the Down Syndrome Health Care Guidelines Record Sheet provides a preventative medical checklist that includes information and recommendations specific to the health and developmental needs of persons with Down syndrome.[14,33,34] Current practices in providing care to those with Down syndrome include primary emphasis of treatment of disease with increased attention allocated to health promotion and protection. The American Academy of Pediatrics recommends the same immunizations and well-child care for those children with Down syndrome as for a typically developing child and because of their increased risk for certain congenital abnormalities and diseases, children with Down syndrome should be further examined and tested accordingly. There have been significant changes impacting the lives of individuals with Down syndrome, not only medical changes but also changes in education practices and community inclusion.[4-10]

The ongoing research and progress associated with Down syndrome should be incorporated into nursing care and practice. Most importantly, nursing care should emphasize that children with Down syndrome are children and, therefore, like other children, they deserve state-of-the-art science and technology as well as family-centered care.[35,36] In addition, nursing care includes the awareness that genetic differences in the disorder exist. Recognition of these variations will help ensure that patients receive the appropriate care and that families with children who have Down syndrome can identify the risk to future offspring.

Medical Management

Medical care for infants with Down syndrome should include the same well-baby care that other children receive during the first years of life. Attention to heart, digestive, orthopedic, or other medical conditions identified during the neonatal period should continue to be monitored. The optimal management of the newborn with a genetic condition includes a multidisciplinary team approach with participation of pediatric specialists. Children with Down syndrome show varying degrees of developmental delays. Language and motor delay become more pronounced during the second year of life.[9,10,18,20] Physical therapy and speech therapy are important for optimizing language and motor development. The degree of central hypotonia is correlated to some degree with later cognitive and motor development.[1]

Children with Down syndrome present a unique circumstance to healthcare providers. In summary, the practice guidelines set forth to serve as a guideline for those caring for individuals with Down syndrome should be followed. *Part I: Clinical Practice Guidelines for Children With Down Syndrome From Birth to 12 Years*[14] offers a conclusive approach to the care of young children with Down syndrome. The guidelines review the importance of generating a thorough health history, taking a physical examination, and the anticipatory guidance piece of caring for those with Down syndrome.[14] In addition, primary healthcare has become essential to helping individuals with Down syndrome thrive and have longer, more productive lives.

Prognosis

The mortality for children with Down syndrome varies greatly with the severity of associated malformations. Individuals with Down syndrome have

ANC200002_17-24.qxp 1/23/09 2:20 AM Page 23

developmental disabilities, with IQs usually ranging from 25 to 70.[17,21] By 40 years of age, individuals with Down syndrome have an increased likelihood to develop symptoms that are nearly identical to those of Alzheimer's disease.[9,17,21] With Down syndrome, there is an increased incidence of neonatal death.[17] Statistics state that three-fourths of fetuses known to have Down syndrome are spontaneously aborted or stillborn and 40% of those born with Down syndrome and a congenital heart disease die in less than 5 years from infection. Survivors at 5 years have a mean survival of 45 years, 20% of infants born with Down syndrome die during their first 10 years of life, and for those who survive beyond 10 years, the average life expectancy is now about 60 years.[11,12,17,33] The outcome for patients with duodenal atresia depends more on the severity of these associated anomalies and the ease with which they can be corrected than on the surgical management of the obstruction itself.[17,24] The understanding of the mechanism of abnormal cardiogenesis in children with Down syndrome centers around collagen type VI and its role in the pathogenesis of heart defects.[23]

Children with Down syndrome have several expected developmental and physical challenges. These include poor physical growth and delayed development with achieving milestones such as gross and fine motor skills, speech, and secondary sex characteristics. They may have frequent respiratory tract infections or episodes of otitis media. Children with Down syndrome may also exhibit transient leukemoid reactions and polycythemia in the neonatal period and have a lifelong increased risk for leukemia, thyroid disorders, premature senility, and aging. Ophthalmologic abnormalities include strabismus, cataracts, glaucoma, and refraction abnormalities. Atlantoaxial instability may also be seen.[11,17]

Characterized by deficient growth in infancy and early childhood, children with Down syndrome are less likely to remain at a given percentile level on the growth charts.[19] Deviations commonly occur between 9 and 24 months.[6,8,10,20] Specific growth charts for children with Down syndrome have been developed and are standardized. These charts were derived from a study, which was based on a longitudinal study of 90 children with Down syndrome conducted at the Developmental Evaluation Clinic at Boston Children's Hospital.[6]

Ethical issues surrounding the diagnosis of Down syndrome become evident in the neonatal period. Since 1989, the US Commission on Civil Rights has stated that withholding care on the basis of the diagnosis of Down syndrome is not acceptable and cannot be supported ethically or legally.[17,31] Before the mid-1970s, it was commonplace to institutionalize newborns with Down syndrome. Improved quality of life for these individuals comes with early care and proper management of health issues including cardiac, gastrointestinal, hearing, vision, and thyroid. Intervention including early educational intervention and support for families and the individual can enhance life skills and optimize education, employment, and community life opportunities for those with Down syndrome.[9,11,17]

Recent advances in mapping the human genome will dramatically impact the care of children with Down syndrome. Further studies related to chromosome 21 will provide a better understanding of the pathogenesis of diseases commonly associated with Down syndrome and may lead to advanced therapeutic management. Innovations in genetic analysis have made it possible to determine the specific regions of chromosome 21 that cause the typical features of Down syndrome. The National Down Syndrome Society is currently sponsoring research on the causes of Down syndrome.[4] In conclusion, the genetic analysis of chromosomes, particularly chromosome 21, has led to new perspectives on diseases. The ability to identify and predict the outcome of a particular individual given the genetic makeup offers healthcare providers the opportunity to develop individual unique approaches to care.[15,16,19,35]

References

1. Jones KL. Down syndrome. *Smith's Recognizable Patterns of Human Malformations*. 6th ed. Philadelphia, PA: WB Saunders; 2006:7-9.
2. Milner RDG, Herber SM. *Color Atlas of the Newborn*. Oradell, NJ: Medical Economics Company Inc; 1894:99-101.
3. Nussbaum RL, McInnes RR, Willard HF. Clinical cytogenetics: disorders of the autosomes and the sex chromosomes. *Thompson & Thompson Genetics in Medicine*. 6th ed. Philadelphia, PA: WB Saunders; 2004:157-162.
4. National Down Syndrome Society. www.ndss.org. Published 2005. Accessed October 27, 2008.
5. National Association for Down Syndrome. www.nads.org. Published 2008. Accessed November 2, 2008.
6. Cronk CE, Pueschel SM, Crocker AM. Growth of children with Down syndrome: birth to age 3 years. *Pediatrics*. 1978;61:564-568.
7. Saenz RB. Primary care of infants and young children with Down syndrome. *Am Fam Phys*. 1999;59:381-390.
8. Mile High Down Syndrome Association. www.mhdsa.org. Published 2007. Accessed September 18, 2007.
9. Saitta SC, Zackai EH. Specific chromosome disorders in newborns. In: Taeusch HW, Ballard RA, Gleason CA, eds. *Avery's Diseases of the Newborn*. 8th ed. Philadelphia, PA: Elsevier Saunders; 2005:204-207.
10. American Academy of Pediatrics Committee on Genetics. Health supervision for children with Down syndrome. *Pediatrics*. 2001;107:442-448.
11. Jorde LB. Genes and genetic diseases. In: McCance KL, Huether SE, eds. *Pathophysiology: The Biologic Basis for Disease in Adults and Children*. 5th ed. St Louis, MO: Elsevier Mosby; 2006:123-156.
12. Pillitteri A. Nursing care of the child born with a physical or developmental challenge. *Maternal & Child Health Nursing Care of the Childbearing & Childrearing Family*. 5th ed. Philadelphia, PA: Lippincott Williams & Wilkins; 2004:1219-1220.
13. Moore KL, Persaud TVN. Human birth defects. In: *The Developing Human Clinically Oriented Embryology*. 7th ed. Philadelphia, PA: Saunders; 2003:161-165.
14. Van Cleve SN, Cohen W. Part I: clinical practice guidelines for children with Down syndrome from birth to 12 years. *J Pediatr Health Care*. 2006;20:47-54.
15. Online Mendelian Inheritance in Man. Johns Hopkins University. www.ncbi.nlm.nih.gov/OMIM. Published 2006. Accessed October 19, 2008.
16. Korenberg J, Bradley C, Disteche C. Down syndrome: molecular mapping of congenital heart disease and duodenal stenosis. *Am J Hum Genet*. 1992;50:294-302.
17. Hernandez JA, Glass SM. Physical assessment of the newborn. In: Thureen PJ, Deacon J, Hernandez JA, Hall DM, eds. *Assessment and Care of the Well Newborn*. 2nd ed. St Louis, MO: Elsevier Saunders; 2005:119-172.
18. Haubrich K. Assessment and management of the auditory system. In: Kenner C, Lott JW, eds. *Comprehensive Neonatal Nursing: A Physiological Perspective*. 3rd ed. St Louis, MO: Elsevier Saunders; 2003:733-737.
19. Pastva M, Corwin EJ, Morin K. Down syndrome with an unusual etiology: case report and review. *J Am Acad Nurse Pract*. 2004;16:244-250.

ANC200002_17-24.qxp 1/23/09 2:20 AM Page 24

20. Holditch-Davis D, Blackburn ST, Vandenberg K. Newborn and infant neurobehavioral development. In: Kenner C, Lott JW, eds. *Comprehensive Neonatal Nursing: A Physiologic Perspective*. 3rd ed. St Louis, MO; 2003:272-273.
21. Sadowski SL. Cardiovascular disorders. In: Verklan MT, Walden M, eds. *Core Curriculum for Neonatal Intensive Care Nursing*. 3rd ed. St Louis, MO: Elsevier Saunders; 2004:584-597.
22. Lott JW. Assessment and management of the cardiovascular system. In: Kenner C, Lott JW, eds. *Comprehensive Neonatal Nursing: A Physiologic Perspective*. 3rd ed. St Louis, MO: Elsevier Saunders; 2003:382-383.
23. Zahka KG. The cardiovascular system: causes and associations. In: Martin RJ, Fanaroff AA, Walsh MC, eds. *Neonatal-Perinatal Medicine Diseases of the Fetus and Infant*. 8th ed. Philadelphia, PA: Mosby Elsevier; 2006:1202-1205.
24. Watson RL. Gastrointestinal disorders. In: Verklan MT, Walden M, eds. *Core Curriculum for Neonatal Intensive Care Nursing*. 3rd ed. St Louis, MO: Elsevier Saunders; 2004:643-654.
25. Mozzoni DS, Ackley RS, Nash DJ. Abnormal pinna type and hearing loss correlations in Down's syndrome. *J Intellect Disabil Res*. 1994;38:549-560.
26. Sanders C, Kleinert HL, Free T, et al. Caring for children with intellectual and developmental disabilities: virtual patient instruction improves students' knowledge and comfort level. *J Pediatr Nurs*. 2007;22:457-466.
27. Skotko B. Mothers of children with Down syndrome reflect on their postnatal support. *Pediatrics*. 2005;115:64-77.
28. Bianchi DW. Prenatal genetic diagnosis. In: Taeusch HW, Ballard RA, Gleason CA, eds. *Avery's Diseases of the Newborn*. 8th ed. Philadelphia, PA: Elsevier Saunders; 2005:186-191.
29. Judge NE, Morrison SC. Perinatal ultrasound. In: Martin RJ, Fanaroff AA, Walsh MC, eds. *Neonatal-Perinatal Medicine Diseases of the Fetus and Infant*. 8th ed. Philadelphia, PA: Mosby Elsevier; 2006:141-164.
30. Landry N. Uncomplicated antepartum and intrapartum care. In: Verklan MT, Walden M, eds. *Core Curriculum for Neonatal Intensive Care Nursing*. 3rd ed. St Louis, MO: Elsevier Saunders; 2004:3-20.
31. Developmental Disabilities Nurses Association. Published 2004. www.ddna.org. Accessed October 12, 2008.
32. National Institute of Child Health and Human Development. Published 2007. www.nichd.nih.gov/health/topics/down_syndrome.cfm. Accessed October 15, 2008.
33. Hockenberry MJ. The child with cognitive, sensory, or communication impairment. In: Wilson D, ed. *Wong's Nursing Care of Infants and Children*. Philadelphia, PA: Mosby Elsevier; 2000:987-993.
34. Van Riper M, Cohen W. Caring for children with Down syndrome and their families. *J Pediatr Health Care*. 2001;123-131.
35. Sears M. In: Soper KL, ed. *Mothers Reflect on How Children With Down Syndrome Enrich Their Lives*. Bethesda, MD: Woodbine House Inc; 2007.
36. Veach SA. Down's syndrome helping the special parents of a special infant. *Nursing*. 1983;13:42-43.

For more than 24 additional continuing education articles related to neonatal, go to NursingCenter.com/CE.