The lowdown on **Down syndrome**

Due to advances in healthcare and the older age of individuals with Down syndrome, it's becoming increasingly important for nurses to understand how to care for patients with this disorder.

By Amanda Perkins, MSN, RN

The leading cause of intellectual disability in the United States, Down syndrome is a disorder that affects approximately 400,000 Americans, according to the National Down Syndrome Society (NDSS). One out of every 800 babies is born with Down syndrome, according to the U.S. National Library of Medicine. The average lifespan of an individual with Down syndrome is 60 years; however, this can vary depending on associated conditions (see *Associated conditions*). Although 60 years old is relatively young, great improvements have been made regarding the longevity of people with Down syndrome. To put things in perspective, the average lifespan for a child with Down syndrome in 1910 was 9 years.

In this article, we discuss the pathophysiology of Down syndrome; the different types of the disorder; and its characteristics, diagnosis, risk factors, associated conditions, management, and nursing care.

What's happening in the cells?

Within the nuclei of the body's cells, all of our genetic material is stored. Rod-like structures called chromosomes are contained in the nucleus; under normal circumstances, the nucleus holds 46 chromosomes or 23 pairs, one set from each parent. Chromosomes are made up of genes that determine how the fetus forms in utero and how the baby grows after birth. They also influence physical characteristics, such as eye and hair color, development, and the probability of developing disease in the future.

Individuals with Down syndrome have a full or partial extra copy of chromosome 21 (see *Female karyotype showing Down syndrome*). Basically, these individuals have extra genetic material, causing changes in the body. Chromosome 21 is the smallest of the chromosomes, containing 200 to 300 genes responsible for making proteins that serve a variety of functions in the body.

Down syndrome can originate from the mother or father; however, only 5% of cases are linked to the father. Although Down syndrome can come from either parent, no one knows exactly why this disorder occurs or how many factors come into play. The chromosomal abnormalities seen with Down syndrome happen during cell division, regardless of whether the error occurred from the mother or father.

The most common risk factor for Down syndrome is increased maternal age, although it's important to note that 80% of children with Down syndrome are born to women younger than age 35, according to the NDSS. The higher incidence seen in mothers under age 35 is believed to be the result of increased births among younger women, according to the CDC. At age 20, the risk of having a child with Down syndrome is 1 out of every 2,000 births, according to the NDSS. The risk increases to 1 out of every 1,000 at age 28, 1 out of 450 at age 34, 1 out of 100 at age 40, and 1 out of 10 at age 49. The reason for this increased

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risk is unknown at this time. The only other identified risk factor is heredity, although this is extremely rare, affecting approximately 1% of all cases of Down syndrome (see *Risk of having multiple children with Down syndrome*).

Now, let's take a look at the types of Down syndrome, including the abnormalities that occur during development.

Three types

There are three different types of Down syndrome: trisomy 21 (nondisjunction), translocation, and mosaicism. You can't tell the difference between the types of Down syndrome based on outward appearance or behaviors. The only way to tell the difference is by looking at the individual's chromosomes.

Trisomy 21

Trisomy 21 is the most common type of Down syndrome, accounting for 95% of all cases, according to the NDSS. Trisomy 21 is caused by an error in cell division, referred to as nondisjunction, leading the individual to be born with an extra copy of chromosome 21. Under normal conditions, each cell will contain two copies of chromosome 21; the individual with trisomy 21 Down syndrome has cells that contain three copies of chromosome 21. This happens at conception when either the sperm or egg fails to separate. As the cells divide and multiply, the extra chromosome is repeated in every cell, resulting in trisomy 21 Down syndrome.

Translocation

This type of Down syndrome is rare, comprising only about 2% to 4% of all cases, according to the NDSS. With translocation Down syndrome, the long arm of chromosome 21 is attached to another chromosome, typically chromosome 14. The individual with translocation Down syndrome has 46 chromosomes but the genetic material of 47 chromosomes, caused by the adherence of a full or partial copy of chromosome 21.

Mosaicism

Individuals with mosaicism, or mosaic Down syndrome, have some cells with 46 chromosomes and some with 47. This form of Down syndrome is extremely rare, accounting for only 1% of all cases, according to the NDSS. This type of Down syndrome results in multidivision after fertilization, leading to two cell lineages. Basically, the tissues and organs of the individual with mosaicism have some cells with the normal number of chromosomes and some with an extra chromosome 21. These individuals tend to have fewer characteristics of Down syndrome than those with trisomy 21 and translocation, although no case is identical. It's believed that the characteristics seen

Associated conditions

There are a number of conditions associated with Down syndrome. The most common are:

- Alzheimer disease
- heart defects
- malignancies
- seizures
- hematologic disorders
- thyroid disease
- hypodontia/delayed dental eruption
- otitis media
- obstructive sleep apnea
- vision problems
- hearing problems
- gastrointestinal problems.

Individuals with Down syndrome not only have a greater risk of Alzheimer disease, but they're also at an increased risk for early-onset Alzheimer disease. For individuals with Down syndrome, the risk of Alzheimer disease increases by 70% after age 50. Around age 50, individuals with Down syndrome start to show cognitive decline.

Two common heart defects seen in babies born with Down syndrome are congenital heart disease (up to 50%) and ventricular septal defect (up to 35%).

Individuals born with Down syndrome have an increased incidence of malignancies, including leukemia and solid tumors. These patients are also at an increased risk for duodenal stenosis, imperforate anus, celiac disease, and Hirschsprung disease.

Lastly, a small percentage of individuals born with Down syndrome will also have autism spectrum disorder.

with mosaicism vary depending on how many cells are affected.

When working with patients with Down syndrome, it's important to understand that they'll have varying degrees of physical abnormalities, as well as cognitive deficits. Delayed development and behavioral problems are common in individuals with Down syndrome. The features and characteristics associated with Down syndrome include:

- learning disabilities
- speech and language delays
- speech that's difficult to understand
- physical abnormalities, such as small stature; small chin; short neck; small ears; slanted eyes; poor muscle tone; flat nasal bridge; single palmar crease; small mouth; short fingers; and a protruding, large tongue.

Screening and diagnosis

Diagnosis of Down syndrome can occur pre- or postnatally using screening and diagnostic tests. Both screening and diagnostic tests aren't able to tell the full impact of developmental delays or complications associated with Down syndrome. What's the difference between screening and diagnostic tests? Screening provides an estimate of the chances of the fetus having Down syndrome, whereas diagnostic tests provide a definitive diagnosis.

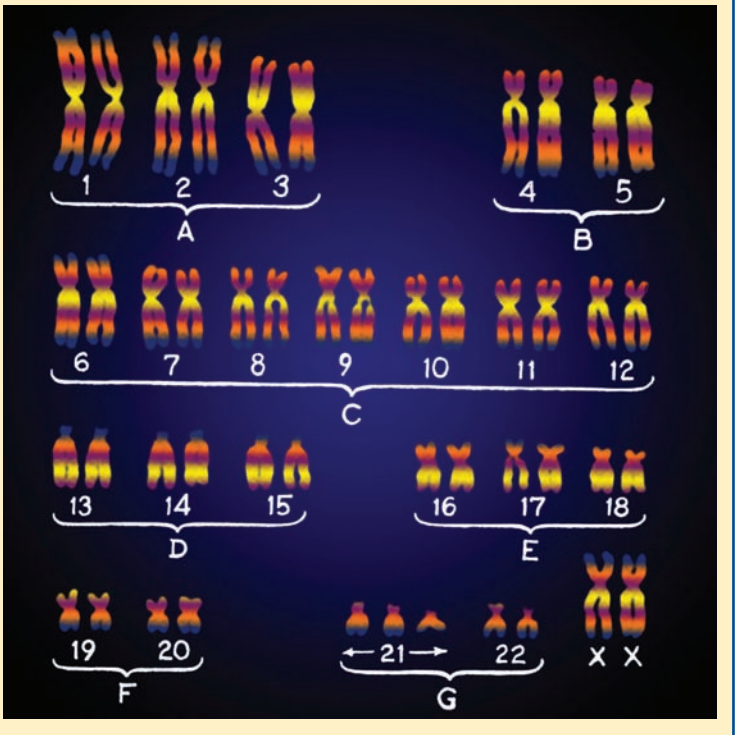
Screening tests, such as blood tests and ultrasound, help determine if diagnostic tests are needed. It's important to complete screening tests before diagnostic tests because these tests are safer for both the mother and fetus. Although not definitive, the blood tests used for screening are becoming more accurate; newer blood tests are able to detect chromosomal material from the fetus circulating in the blood. Blood tests are typically carried out in the first trimester for women who have an increased risk of having a baby with Down syndrome.

Ultrasound is useful because it allows the examiner to check for markers or characteristics that are commonly seen in fetuses with Down syndrome. When using ultrasound to aid in the diagnosis of Down syndrome, the test is most reliable at 14 to 24 weeks' gestation. In rare instances, ultrasound can give false-negative or false-positive results.

A variety of diagnostic tests can be used during pregnancy, but the most common are amniocentesis and chorionic villus sampling (CVS). Amniocentesis is typically carried out in the second trimester, around 15 to 20 weeks' gestation, and CVS is done in the first trimester, around 9 to 14 weeks' gestation. Amniocentesis and CVS are very reliable tests (almost 100% accurate), but they do carry an increased risk of miscarriage. Keep in mind that although the risk is small,

Female karyotype showing Down syndrome

Color-enhanced karyotype for a female with Down syndrome, characterized by an extra copy of genetic material on chromosome 21.



Biophoto Associates / Science Source

with only 1% of women being affected, it may not seem like a small risk to the parents.

It's recommended that infants with Down syndrome undergo genetic testing with a complete chromosomal analysis if genetic testing wasn't performed prenatally. Karyotype is a chromosomal analysis that involves obtaining a blood sample and examining the infant's cells. Due to the fact that children born with Down syndrome have a high risk of congenital heart disease, it's also recommended that neonatal echocardiography be completed within the first month, along with a completed blood cell count to check thyroid functioning.

Care across the continuum

The management of Down syndrome can be difficult because the severity of signs and symptoms, along with complications, can vary widely by patient. This variability makes it challenging to provide consistent care. It's important to meet the patient's physical and emotional needs throughout his or her lifespan. Hearing screens, ophthalmic evaluations, and thyroid tests should be completed because these individuals are at high risk for problems with hearing, vision, and thyroid function. By the time the child is 4 years old, he or she should undergo a sleep study to assess for sleep apnea. If the patient with Down syndrome presents with gastrointestinal problems, poor growth, anemia, and/or behavioral issues, he or she should be tested for celiac disease.

Behavioral issues are common in patients with Down syndrome, including attention problems, obsessive-compulsive behavior, stubbornness, and tantrums. It's important to address these behaviors in childhood because they can persist into adulthood and become more difficult to manage. Negative behaviors that may be seen with Down syndrome include attention seeking, immediate gratification, and communication difficulties. However, individuals with Down syndrome tend to have prosocial behaviors and, as a result, share easily, display patience, stay on task, and are interactive when in situations involving groups. These behaviors may not be observed when the individual is outside the group environment.

Your role

Now, let's discuss the nurse's role in caring for the child and adult with Down syndrome, as well as the patient's family.

Caring for the child and family

Emotional support may be necessary for families because they may experience shock and grief upon learning the diagnosis. Always maintain a nonjudgmental attitude and provide emotional support as needed. Keep in mind that parents may go through the grieving process, which includes denial, anger, bargaining, depression, and acceptance. These parents may need to grieve the loss of the baby they were planning to have and accept the child that they had. At first, they may only think about the challenges and obstacles that the child will face. They may also be concerned about the increased risk of associated health complications. Education and support during this period of time are essential.

Risk of having multiple children with Down syndrome

When caring for parents of a child with Down syndrome, they may ask questions about the chances of having another baby with this disorder. With trisomy 21, the chances of having another baby with Down syndrome is 1 out of 100, according to the NDSS. Of the three types of Down syndrome, translocation is the only type with a hereditary component. With inherited translocation, the parent will have a rearrangement of genetic material between two chromosomes, one of which is chromosome 21. Known as balanced translocation, the parent will be unaffected, without health complications. Although the parent is unaffected, this translocation can be passed on to a child and become unbalanced, leading to Down syndrome. With translocation, the chances are 3% if the father is the carrier and 10% to 15% if the mother is the carrier, according to the NDSS. Due to the fact that mosaic Down syndrome is so rare, there's inadequate evidence on the risks of having multiple children with this type.

Provide education both pre- and postnatally regarding the importance of watching for and reporting slow feeding, choking, spitting up, and abnormal stools because these signs and symptoms can be indicative of associated complications.

Teach the following techniques for addressing negative behaviors:

- follow a schedule
- offer choices
- associate difficult tasks with enjoyable tasks
- collaborate
- teach appropriate responses
- reinforce positive behaviors
- don't reward problematic behaviors
- provide simple, clear, and concise instructions.

As the child ages, his or her parents may need to think about guardianship/power of attorney. Parents may also have questions about who will care for their grown child. Educate families about how to plan for the future. For example, as their child becomes an adult, it may be appropriate for him or her to find employment, depending on the degree of cognitive delay. Teaching families about available resources and support systems is beneficial and a necessary component of care.

Remember, don't assume that everyone with Down syndrome will behave or act a certain way. These individuals may need additional services, such as speech, physical, and occupational therapy. It's also important to be aware that these children may need extra assistance at school.

Caring for adult patients

It's essential that we understand how to effectively care for patients with Down syndrome in the healthcare setting. Hospital admissions are more common in individuals with intellectual disabilities, such as those with Down syndrome. Research has shown that many healthcare professionals have a lack of knowledge about caring for patients with disabilities; as a result, these patients tend to receive a decreased level of care in the healthcare setting.

Healthcare services should be flexible to best meet the patient's needs. Providing fair and equitable treatment should be of utmost importance. This means that we may need to adjust our care to accommodate the specialized needs of the patient with Down syndrome.

cheat sheet

Common characteristics of Down syndrome

- Learning disabilities
- Speech and language delays
- Speech that's difficult to understand
- Physical abnormalities
  - small stature
  - small chin
  - short neck
  - small ears
  - slanted eyes
  - poor muscle tone
  - flat nasal bridge
  - single palmar crease
  - small mouth
  - short fingers
  - protruding, large tongue

When caring for a patient with Down syndrome:

- ensure safe discharges by involving both the patients and his or her caregivers
- acknowledge the caregivers' expertise and use them as a resource
- communicate directly with the patient
- use alternative forms of communication when needed, such as photographs, picture books, and so on
- assess for nonverbal cues indicating pain, discomfort, anxiety, and so on
- conduct a baseline assessment to gain a thorough understanding of the patient's abilities
- don't make assumptions about the patient's quality of life.

Patient-centered and prepared

Due to the fact that Down syndrome is the leading cause of intellectual disability in the United States, it's essential that nurses understand the disorder and how to care for patients with Down syndrome. Make sure that education is in place at your facility regarding the care of individuals with disabilities. It's our responsibility to understand the

on the web

CDC: www.cdc.gov/ncbddd/birthdefects/downsyndrome.html

Mayo Clinic: www.mayoclinic.org/diseases-conditions/down-syndrome/basics/definition/con-20020948

MedlinePlus: https://medlineplus.gov/downsyndrome.html

National Association for Down Syndrome: www.nads.org

National Down Syndrome Society: www.ndss.org

National Human Genome Institute: https://www.genome.gov/19517824/learning-about-down-syndrome/

World Down Syndrome Day: https://worlddownsyndromeday.org/

legislation in our area and stay current on best practices for providing care to these patients. ■

REFERENCES

Asim A, Kumar A, Muthuswamy S, Jain S, Agarwal S. Down syndrome: an insight of the disease. *J Biomed Sci.* 2015;22:41.

Blair J. Care adjustments for people with learning disabilities in hospitals. *Nurs Manag (Harrow).* 2011;18(8):21-24.

CDC. Facts about Down syndrome. www.cdc.gov/ncbddd/birthdefects/downsyndrome.html.

Kessling A, Sawtell M. The genetics of Down's syndrome. www.intellectualdisability.info/diagnosis/articles/the-genetics-of-downs-syndrome.

Khatri S, Carlisle JB. Down syndrome: primary physicians and parents partner in care. http://contemporarypediatrics.modernmedicine.com/contemporary-pediatrics/news/down-syndrome-primary-physicians-and-parents-partner-care.

Kyle G. Caring for a child with Down's syndrome in the acute care setting. *Nurs Child Young People.* 2012;24(4):18-22.

National Down Syndrome Society. What is Down syndrome? www.ndss.org/down-syndrome/what-is-down-syndrome.

U.S. National Library of Medicine. Chromosome 21. https://ghr.nlm.nih.gov/chromosome/21#sourceforpage.

U.S. National Library of Medicine. Down syndrome. https://ghr.nlm.nih.gov/condition/down-syndrome#sourcesforpage.

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