Care of the Patient with Down Syndrome

1.00 Contact Hour

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Outcomes

The purpose of this activity is to enable the learner to identify and aid in the management of patients with Down syndrome.

Objectives

By the end of this activity, the learner will:

- Define Down syndrome
- Identify characteristics of Down syndrome
- Discuss barriers to care and disability Acts
- Discuss the role of the Certified Nursing Assistant

Introduction

Down Syndrome is the most frequently seen congenital disorder in newborns. Although it is lifelong and there is no cure, these patients can live productive lives. Adults with this disease now live well into their 60's and even into their 70's. Many go to normal schools and are employed in part due to advances in medical care and the Americans with Disabilities Act. The services provided for these patients allow them more freedom than ever to live in their homes or with their families. If the disabilities are more severe, they may live in a group home or assisted living. Although they often suffer from co-morbid conditions, they can still maintain a good quality of life. Healthcare providers are an important part of the process regardless of where the patient lives or the disability's severity.

What is Down Syndrome

Genetics is responsible for the characteristics and traits that we all have. Genes carry a code that gives a person brown hair or blue eyes. This is accomplished through rod-like structures called chromosomes. A nucleus carries 23 pairs of chromosomes. A baby inherits 23 pairs of chromosomes from each parent for a total of 46. In Down syndrome, an extra copy of chromosome 21 appears, giving the baby 47 chromosomes. This extra chromosome alters some physical and intellectual characteristics of a person. The traits can vary in degree as well as severity. The exact

cause of the extra chromosome is not known. However, advanced maternal age seems to be a factor. Approximately one in every 690 babies born in the United States is diagnosed with Down syndrome (CDC, 2016), making it the most common genetic condition. About 400,000 Americans are diagnosed with Down syndrome today (CDC, 2016).

The characteristics and description of a person with Down syndrome was initially published in 1866 by an English physician, John Langdon Down (National Down Syndrome Society [NDSS], 2023). Even though others had recognized the characteristics of the syndrome before that, it was Down's published scholarly work of describing Down syndrome as a distinct entity from others that earned him the recognition as the "father" of the syndrome (NDSS, 2023). In 1959, Jerome Lejeune, a French physician, identified the disease as a chromosomal condition (NDSS, 2023). Lejeune noted that these individuals had 47 chromosomes instead of the normal 46. It was later determined that a duplication of chromosome 21, either a partial or complete copy, was associated with the cause of the syndrome. In the year 2000, scientists identified each of the approximately 329 genes on chromosome 21 and catalogued them (NDSS, 2023). This has opened up the opportunity for additional research to discover the reason behind the extra chromosome formation.

The only suggested association with the cause of Down syndrome is in women over the age of 35. A 35-year-old female has approximately a one in 350 chance of having a child with Down syndrome (CDC, 2016). This gradually increases to one in 100 at the age of 40, and by the age of 45, chances are one in 30 (CDC, 2016). Diagnosis is usually made at birth due to physical characteristics. There are two tests that can be performed before the birth of the child. A screening test that estimates the chances of the fetus having Down syndrome. This test does not tell whether or not the fetus actually has Down syndrome. However, a diagnostic test can provide a definitive diagnosis and be done before birth. Down syndrome is identified by the presence of certain physical characteristics at birth, such as low muscle tone, a single deep crease across the palm of the hand, a flattened facial profile, and upward-slanting eyes. However, these physical features may be present in babies without Down syndrome. A chromosomal analysis, called a karyotype, confirms the diagnosis after birth.

There are three types of Down syndrome conditions. However, it is difficult to tell the difference between the types without a chromosomal test as the symptoms are very similar. The first is trisomy 21, which accounts for approximately 95% of all cases of Down syndrome ("What is Down syndrome," 2012). In trisomy 21, each cell in the body has three separate copies of chromosome 21 instead of the normal two copies ("What is Down syndrome," 2012). The second type is mosaicism, which occurs when some cells contain the usual 46 chromosomes and some contain 47. This is the least common form of Down syndrome and accounts for approximately 1% of all cases ("What is Down syndrome," 2012). This form of Down syndrome has the least amount of physical and intellectual characteristics. Lastly, translocation type accounts for 4% of the population ("What is Down syndrome," 2012). The number of chromosomes remains at 46; however, an additional chromosome 21 is attached to another chromosome, usually chromosome 14. This abnormal attachment causes characteristics of Down syndrome.

Regardless of the type, the extra chromosome 21 is the culprit in the cells. This extra chromosome changes the genetic makeup enough in the development of the individual to cause characteristics associated with the disease ("What is Down syndrome," 2012). Changes caused by the extra chromosome create both mental and physical challenges for the person. Typically, the patient has

a mild to moderate low IQ, and children typically reach developmental milestones at a slower pace. Physical symptoms seen are a flattened face, particularly the bridge of the nose, eyes that slant upward, a short neck, small ears, smaller hands and feet, a single crease on the palm of the hand called a Palmer crease, a small pinky finger that can curve toward the thumb, poor muscle tone, loose joints, and short stature (CDC, 2016) Although these signs are telling, it is still important to observe behavior and cognition.

It is essential to remember that children and adults with Down syndrome experience developmental delays. However, most children are in regular classrooms, and most adults can work in a structured environment. Beyond the developmental delays, many have medical complications. Approximately 40% have congenital heart defects ("Facts about Down syndrome," 2015). Some heart conditions require surgery, while others only require monitoring. Additionally, there tends to be a higher incidence of infections, respiratory issues, vision, hearing, and thyroid issues (Sorentino & Remmert, 2016).

Services

Typically, children born and diagnosed with Down syndrome need support and services from the community and state. Most can learn self-care skills but may need help in other areas. This may include assistive devices, surgery, drug therapy, physical therapy, occupational therapy, speech therapy, and recreational therapy. They may also need hearing and vision aids. Where some can live independently, other patients live in group homes or long-term care facilities. Since Down syndrome is considered a disability, the patient does have the protection of their rights through the Americans with Disabilities Act of 1990, the Developmental Disabilities Assistance, and the Bill of Rights Act in 2000 (Sorentino & Remmert, 2016). The Americans with Disabilities Act prohibits discrimination against those with disabilities. The Developmental Disability Assistance and Bill of Rights Act also protects the rights of those with disabilities to have access to state, federal, and community services (DD Act, 2011).
Furthermore, The Individuals with Disabilities Education Act is based on the need for all disabled children to receive education, whether that be in a regular classroom or special programs. This may include behavioral programs, hearing impairments and visual programs, learning programs, or physical disabilities. An individualized educational plan may also be completed for a child in an educational setting to make sure they receive education fit for their needs.

Barriers to Care

Patients with intellectual disabilities, such as Down syndrome, have health problems that are higher than the general population. Health inequalities and disparities such as housing, poverty, unemployment, communication problems, behaviors related to poor diet, poor exercise, and poor access to mainstream healthcare can be seen. Empowerment can be important for the patient as well as for the family to target the needs of the disabled person (Naaldenberg). Although some health disparities and inequalities are modifiable, they still put the patient at a disadvantage for healthcare. Healthy People 2020 outlines public health outcomes that incorporate a reduction of these barriers. Healthy People targets health interventions around the needs of a disabled population related to the quality of life and health (Krahn & Fox, 2014) with the goal of reducing and eliminating as many barriers in health inequality as possible. The program also helps practitioners to plan for disparities they may see in various populations and locations. The

healthcare provider's role is important to help alleviate as many disparities and inequalities as possible.

Role of the CNA

When caring for a patient with Down syndrome, it is important to remember that the patient may struggle with speech, learning, mobility, and self-care needs. These needs may be extensive, especially in the home. The patient still has the right to make choices if able and to be treated with dignity and respect. Always provide quality care with a positive attitude. When working with the child, keep in mind that the parents may be grieving and struggling with how to handle all of the complications associated with the disease. It is important to emphasize resources such as child healthcare professionals, group counseling, and literature to help the family learn and deal with the child's problems. Be sure to help promote independence and self-help skills such as self-feeding. Some areas that should be observed are socialization skills, verbal skills, activities of daily living, preventative health, nutrition, and behaviors.

Anything that the child needs to learn should be broken down into the smallest components or baby steps to help them digest the information (Hockenberry & Wilson, 2014).

Socializing can be difficult for the patient with Down syndrome. Role-playing and positive reinforcement can be particularly helpful (Hockeberry & Wilson, 2014). Teach and encourage parents to show the child socially acceptable behaviors such as waving goodbye, saying hello, responding to their name, and greeting visitors. It may be helpful to expose the child to strangers so that they can practice to enhance their self-esteem (Hockenberry & Wilson, 2014). Preschool programs can be helpful and encourage social interactions. Educating the family is very important. If there are cognitive impairments, the child or adult may not be able to recognize cues from others. This can be frustrating to the patient as well as the family. One strategy is to practice exaggerated cues to help the patient recognize the behaviors of others and respond appropriately. Short-term memory may also be a problem. Explaining directions in simple, short sentences, one step at a time may be helpful.

Remember that verbal skills may be delayed more than other physical skills. In order to speak, hearing and interpretation of sounds should be checked to make sure the patient can hear properly. Assistive devices may be needed to help the process. Sometimes, a picture board may be used to help the child express their feelings and thoughts (Hockenberry & Wilson, 2014), or children and adults may also learn sign language.

When working with a person with Down syndrome, observations should include noticing if the patient is irritable, unresponsive to contact, abnormal eye contact during feeding, gross motor problems, decreased alertness to voice cues, or difficulties with activities of daily living. If any of these areas show change, the nurse should be alerted.

Be sure to assist the family with the prevention of other problems. Discuss the need for modifications as part of the daily routine to aid in patient care. For example, the joints on a baby or toddler may be limp, and the child may sag in the parent’s arms. Encourage parents to swaddle the newborn tightly and hold the toddler close, supporting their head and spine (Perry, Hockenberry, Lowdermilk, & Wilson, 2014). Observe for respiratory problems due to decreased muscle tone. An underdeveloped nasal bone may lead to inadequate draining, stuffiness, and dry mouth. The child may also be at risk for upper respiratory infections (Hockenberry & Wilson, 2014). Be sure the

parents know how to suction the nasal passages properly if needed. Suggest the parents rinse the child's mouth after eating and sit the child upright for feedings. Also, if the patient needs to be fed, give adequate time for chewing. Make sure the mouth is empty before adding more food. Using a long, straight-handled spoon to push toward the back of the mouth will prevent the tongue from pushing the food back out (Hockenberry & Wilson, 2014). Note that mouth breathers have a difficult time swallowing. Observe for gagging or choking. Food should be eaten slowly, and they should take small bites. Fluids should be consumed slowly as well. Dysphagia is often a concern due to the patient's short neck. This can increase the risk of aspiration pneumonia (Herron-Foster & Bustos, 2014). Be sure to educate the parents or patient on the importance of taking the time to eat meals. Additionally, the Down syndrome patient often has problems with gastric motility, such as gastric reflux or gastrointestinal upset (Herron-Foster & Bustos, 2014). Observe for nonverbal pain cues and use a rating scale for pain levels. Be sure to note all food intake.

Because of poor muscle tone, gastric motility may be slowed, causing constipation. Make sure fiber is a regular part of the diet. Eating habits should be carefully observed and documented. Height and weight measures should be monitored regularly to check for obesity and high BMIs (Perry, Hockenberry, Lowdermilk, & Wilson, 2014).

The child may grow more slowly and not reach developmental milestones on time. Keep a growth chart to measure height and weight regularly. Skin care is also important. Be sure to check the skin for cracks or reddened areas. Be sure to apply lotion when necessary and use sunscreen when outside (Perry, Hockenberry, Lowdermilk & Wilson, 2014). Check bony prominences, such as heels and elbows, for red areas or sores (Herron-Foster & Bustos, 2014). Furthermore, check skin folds for rashes. Be sure the area is washed and patted dry. Apply topical barrier creams if indicated by the treatment plan. If any areas look to be of potential concern, report to the nurse for additional treatment planning. Do not use any perfumed or alcohol-based products, as this can cause irritation (Herron-Foster & Bustos, 2014)

Recreational activities are also important. Not only do these activities help with muscle tone, balance, and coordination, but they also help to manage weight. Furthermore, activities in the community can help with socialization skills (Hockenberry & Wilson, 2014). Ensure the activities are within the child's or adult's limits and will not cause undue stress on joints and weak muscles.

Following a nutrition plan is also important. The patient should eat three meals daily, including the food groups (Perry, Hockenberry, Lowdermilk, & Wilson, 2014). Self-management techniques can include a food diary to monitor calories and sodium intake if heart conditions are present. Check the treatment plan or discuss options with the nurse.

When working with children and adults, check for proper oral hygiene. The patient may be unable to brush effectively due to decreased motor skills or cognitive impairments (Herron-Foster & Bustos, 2014). They made need instructions on how to brush and floss effectively. There are also several products available that can help the patient with their oral hygiene. Be sure to check the treatment plan for aids or talk to the nurse if you notice problems.

Safety is also a concern. The patient may have poor neck control or be generally weak. Protecting the neck and proper transferring of bedridden patients is essential. Logrolling techniques can also be used, as a two-person lift or equipment designed to lift patients may need to be used

(Hockenberry & Wilson, 2014). Other medical equipment may be used as well to help prevent falls. Examples are wheelchairs, shower chairs, or walkers.

In older adults, arthritis may be an issue. Special pillows or rolled towels may be needed to support the neck (Herron-Foster & Bustos, 2014). Additionally, a flat towel may be a better alternative than a pillow so the neck is not hyper-flexed. Wedges may also help to alleviate back and hip pain. Document any pain and notify the nurse if the pain is new or has worsened.

Prevention of illness is also important. Be sure to teach the patient to wash hands thoroughly and to stay away from people who are ill. When teaching the patient, breaking up the task into steps may be necessary. Use simple, short sentences and, if possible, ask the patient to repeat each step back to you (Herron-Foster & Bustos, 2014).

Working with patients who are diagnosed with Down syndrome can be challenging. It is important to remember that their physical and mental growth rate may be delayed. Helping the patient and family make good choices and provide a safe environment is a particular duty of the certified nursing assistant. If any changes occur in the patient's mood or behavior or if there are new problems that develop, report to the nurse for additional treatment planning. Caring for the Down syndrome patient can be very rewarding. Patience is a key factor in helping these patients have a good quality of life.

Case one:

Susan and John are bringing home their newborn son, who has been diagnosed with Down syndrome. You have been assigned to go into the home and help the family adjust to the child's needs. Susan, the mother, is doting and very good at holding and caring for the new baby. However, John, the father, is visibly upset and does not want to hold the baby. Susan feels that John does not like the baby and is sorry they decided to continue the pregnancy after finding out the baby may have Down syndrome. What can you do to help this family?

Most likely, John is grieving. Helping John learn about Down syndrome and how to help his child will be most effective. You can start with talking about the disease and offering written education materials. Help John learn how to hold the baby so that the infant's body and head, which are weak, are supported. He may also be afraid of hurting the baby or not holding him properly. Teach both parents how to feed, swaddle, and hold the infant. Check-in on them regularly and make sure they are following up with support services.

Case Two:

Sara is a 22-year-old who has been your patient for the last six months. She is diagnosed with Down syndrome and has limited capabilities. She is not able to speak and is confined to a wheelchair. Sara lives at home with her mother and father, who are her caregivers. The parents feel Sara needs to be with people other than them all the time. The parents have found a community center where Sara could interact with other patients with Down syndrome. They are asking how to prepare Sara for this change. What are some suggestions you can give the parents?

First, assess Sara's ability to respond to you as an outsider. If she responds well, having other people over to the house might be a good idea for Sara to meet first. Sara needs to be comfortable with strangers approaching her. Does Sara know how to wave goodbye or hello? Does she recognize her name? These are some areas to start working on and assess. It is also important to assess Sara's means of communication.

Can she use pictures to communicate her needs? The parents will have to ensure Sara's new caregivers understand and accept her needs. Socializing is a good activity and will promote self-esteem.

Implicit Bias Statement

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