Obesity in Children with Down Syndrome: Background and Recommendations for Management

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Children with Down syndrome have a higher risk for developing obesity. The primary care provider can assist the family in preventing or managing obesity by recognizing the physiological and behavioral factors that place children and adolescents with Down syndrome at increased risk to become obese, and establishing a screening and management plan early to prevent or treat excess weight gain. By using adapted strategies, the negative physiological and psychological outcomes associated with obesity may be lessened or avoided in this specific population.

Down syndrome, also called Trisomy 21, is a chromosomal disorder that results from genetic changes on chromosome 21. Almost 95% of cases are caused by nondisjunction, meaning the chromosome 21 pair fails to separate during cellular division (Nehring, 2010). The second cause results from translocation – when a chromosomal segment moves to a new position (Tolmie, 2002). The third chromosomal defect is mosaicism, which occurs when a mutation on chromosome 21 takes place later in cell division, resulting in affected and normal cells. This mosaic form of Down syndrome is thought to affect individuals less severely than nondisjunction or translocation (Leshin, 2000). Down syndrome is encountered in all racial and ethnic groups, and becomes more prevalent with advanced paternal and maternal age (Tolmie, 2002). The incidence is approximately 1 in 800 live births (Aitken, Crossley, & Spencer, 2002).

Down syndrome is characterized by a unique phenotype and associated with multiple congenital anomalies (Castiglia, 1998). Classic phenotypic findings include brachycephaly, epicanthal folds, palpebral fissures, macroglossia, neck skinfolds, clinodactyly, single transverse palmar creases, and widely spaced first and second toes (Nehring, 2010). Common congenital anomalies associated with Down syndrome include gastrointestinal tract anomalies (such as esophageal and duodenal atresia), tracheoesophageal fistula, pyloric stenosis, Hirshprung's disease, and imperforate anus (Tolmie, 2002). Fifty-eight percent of children with Down syndrome are born with cardiac defects. The most common defects include atrial septal defect, ventricular septal defect, and patent ductus arteriosus (Figueroa, Magaña, Pablos Hach, Jiménez, & Urbina, 2002).

Some degree of cognitive deficit occurs in all children with Down syndrome, although intellectual abilities vary greatly from being mildly affected to severely impaired (Nehring, 2010). Congenital or acquired hearing loss occurs in 45% to 75% of affected children and may be attributed to structural differences of the skull, eustachian tube dysfunction, or recurrent otitis media (Tolmie, 2002). Musculoskeletal differences include generalized hypotonia, joint laxity, and atlantoaxial instability of the spine (Nehring, 2010). Other associated problems in children with Down syndrome include growth retardation, altered immune function, celiac disease, seizure disorders, leukemia, skin conditions, visual problems, altered dentition, sleep-disordered breathing, and thyroid dysfunction (Nehring, 2010). In addition, all individuals with Down syndrome have a 4 to 16-fold increased risk of mortality due to venous thromboembolitic disorders, cardiovascular disease, and cerebrovascular disease (Hill et al., 2003).

Obesity in Children with Down Syndrome

A more recent concern for children born with Down syndrome is their tendency to become overweight and obese. Approximately 17.1% of children in the U.S. are obese (Ogden, Carroll et al., 2006). Some research suggests the number of children with Down syndrome who are obese approximates national trends (Cohen, 1999). However, the rate of obesity may be much higher than the general population; another study stated that up to 30% to 50% of children with Down syndrome are obese (Harris, Rosenberg, Jangda, O'Brien, & Gallagher, 2003). Fonesca, Amaral, Ribeiro, Beserra, and Guimaraes (2005) found children with Down syndrome had an increased risk for developing Type 2 diabetes mellitus due to their propensity for obesity and large amounts of abdominal fat stores. Because of the negative consequences associated with obesity, prevention and interventions in children with Down syndrome should be a major health priority. Although specific associated problems of Down syndrome, both physiological and behavioral, foster the development of obesity, identifying and addressing condition-specific problems in a prevention and management plan is essential when caring for the child with Down syndrome.

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Physiological Mechanisms Associated with Obesity In Down Syndrome

Hypothyroidism. In young children, untreated hypothyroidism results in significant growth and developmental retardation, and is implicated in the development of obesity due to the slowing of the body's metabolic rate. Hypothyroidism is a common congenital or acquired condition in children with Down syndrome. Approximately 30% to 50% of school-aged children with Down syndrome have sub-clinical hypothyroidism due to acquired autoimmune dysfunction (Roizen, 1997).

Decreased basal metabolic rate. Basal metabolic rate refers to the amount of calories the body burns at rest. Although resting metabolic rate has long been researched in adults, Luke, Roizen, Sutton, and Schoeller (1994) were the first to use a controlled trial to study energy expenditure in children with Down syndrome. Their results indicate prepubescent euthyroid children with Down syndrome have a decreased resting metabolic rate compared to children without Down syndrome when corrected for movement. The underlying mechanism is likely to be an abnormality in cellular metabolism or in the composition of fat-free mass. These findings suggest that at rest, euthyroid children with Down syndrome burn fewer calories than their counterparts, contributing to the development of obesity.

More recently, Bauer et al. (2003) found energy expenditure differs in neonates with Down syndrome. These authors document neonates with Down syndrome expend 14% fewer calories compared to non-affected infants and have significantly decreased muscle tone. This suggests hypotonicity may be a factor that decreases resting energy expenditure (Bauer et al., 2003).

Increased leptin. Leptin is a hormone that plays an important role in regulating food intake by stimulating satiety and promoting energy homeostasis via energy expenditure. The hormone has been implicated in the development of obesity within the general population and may also be relevant for children with Down syndrome. Typically, increased levels of the hormone correlate with obesity because in excess, the body becomes desensitized to the hormone. As a result, the body responds poorly to leptin, and the individual experiences decreased satiety. Results from a study by Magni et al. (2004) show prepubertal children with Down syndrome have high levels of leptin and posit leptin as a potential factor in the development of obesity-increased leptin levels, which were positively correlated with increased bosdy mass index (BMI) and degree of adiposity (Magni et al., 2004).

Poor mastication. Children with Down syndrome have difficulty eating raw fruits and vegetables, and chewing firm or fibrous foods. While this subject has not been thoroughly investigated, Hennequin, Allison, Faulks, Orliaguet, and Feine (2005) assert that masticatory dysfunction may lead to nutritional deficiencies in individuals with Down syndrome. Difficulty masticating foods may also lead the child to eat softer foods, which are often higher in carbohydrates, sugars, fats, and cholesterol (Hennequin et al., 2005).

Behavioral Tendencies Associated with Obesity In Down Syndrome

According to a study by Whitt-Glover, O'Neill, and Stettler (2006), children with Down syndrome engage in less vigorous physical activity compared to their siblings. The study examined physical activity over seven days using accelerometers and found children with Down syndrome have similar levels of low-intensity and moderate activity compared to their siblings but perform significantly less vigorous-intensity activity. The children with Down syndrome also had a higher BMI compared to their siblings, leading the authors to assert the discrepancy in less vigorous-intensity activity could be a factor in the development of obesity (Whitt-Glover et al., 2006).

Other behavioral tendencies may play a role in the development of obesity. Negativity, impulsivity, oppositional behavior, inattention, disobedience, and noncompliance are behaviors that tend to surface as children with Down syndrome become older, and are particularly evident when they become frustrated (Jahromi, Gulsrud, & Kasari, 2008). Negative behavior may occur when the parent attempts to encourage healthy food choices or involve the child in physical activity, creating barriers that prevent necessary dietary and lifestyle changes.

Assessment of the Child With Down Syndrome

Pediatric practitioners must perform a comprehensive assessment on a child with Down syndrome to develop appropriate prevention and management plans tailored to the child's condition and level of associated problems.

History

A complete history, including birth history, past medical history, laboratory results, surgical procedures, and hospitalizations, is important to have documented in the child's record. A thorough review of systems may assist in identifying current levels of activity, diet, behavioral problems, or health conditions requiring medical attention. As with all children, it is important to assess the child and the family's lifestyle patterns. A 24-hour dietary recall is one strategy to gain a basic understanding of eating habits. The dietary recall should include all food and beverages consumed by the child during a 24-hour period, excluding water (National Center for Health Statistics, 1994). In addition, asking the family to keep a food and activity journal for a week prior to the appointment may provide further insight into consumption trends and family patterns of activity. A family history of health risks should focus on factors such as high cholesterol, hypertension, heart disease, and diabetes mellitus.

Physical Examination

Key aspects of the physical examination will help a provider gauge changes in weight gain, recognize physiological contributions that cause obesity, identify risk factors, and decipher if the child may safely participate in physical activities to promote weight loss (see Table 1).

Measurements. The provider must establish baseline measurements for blood pressure, height, weight, and BMI. Blood pressure screening should be a routine part of care, and the child's blood pressure must be compared to age, height, and sex-based norms. Measurements for height and weight should be plotted on the National Center for Health Statistics growth charts specifically designed for children with Down syndrome. Growth charts for children with Down syndrome are adjusted to the reduced linear growth velocity due to congenital heart disease, hypothalamic dysfunction, thyroid disorder, and nutritional problems (Cohen 1999). By using both types of charts, the child's growth is compared to chronologic-age peers and Down syndrome peers (Nehring, 2010). The BMI should be calculated and plotted on a

Table 1.
Physical Assessment

Exam	Common Abnormal Findings in Children with Down Syndrome
Measurements	• BMI less than 5th percentile • BMI greater than 85th percentile
Neck	• Changes in size or consistency of the thyroid gland
Atlantoaxial Instability	• Weakness, neck pain, head tilt
Heart	• Mitral valve prolapse • Aortic regurgitation
Musculoskeletal System	• Hip instability • Patellofemoral instability • Deformities of the forefoot, pes planus, calcaneal valgus, pronation

Note: BMI = body mass index.

gender-specific BMI graph. A child with a BMI between the 85th and 95th percentile is considered overweight, while a BMI greater than the 95th percentile classifies the child as obese (Elliott, 2007).

Skin. Identifying skin changes that correspond to obesity is important and may indicate complications from being overweight. The practitioner should be alert to patterns of fat deposition, such as android (central fat deposition) versus gynoid (pear-shaped fat deposition), and note areas where striae are present. Additionally, it is essential to recognize signs of insulin resistance, which is common in overweight children with Down syndrome (Fonesca et al., 2005). Signs of insulin resistance include the appearance of acanthosis nigricans, which are dark, creased areas commonly located on the skin of the neck, axillae, elbows, knuckles, groin, and knees. If these changes are present, the child should be screened for fasting blood glucose and insulin levels. If the screening is abnormal, an oral glucose tolerance test (OGTT) should be ordered to screen for the presence of insulin resistance, impaired glucose tolerance, and diabetes mellitus.

Neck. It is important to examine the thyroid gland carefully for changes in size or consistency due to the high prevalence of hypothyroidism in children with Down syndrome. Thyroid problems may also manifest as weight gain, dry skin, or decreased height velocity (Cohen, 2006). The examination is often unremarkable; therefore, current recommendations include performing annual thyroid function screening tests by drawing levels of T3, T4, and TSH (American Academy of Pediatrics [AAP] Committee on Genetics, 2001). If hypothyroidism is detected based on childhood norms, the child should be referred to an endocrinologist for treatment with thyroxine to promote a euthyroid state and metabolic homeostasis.

Atlantoaxial instability. Between 10% to 30% of children with Down syndrome have atlantoaxial instability (Winell & Burke, 2003). Atlantoaxial instability is the result of laxity between the cervical vertebrae and is evident by an atlantoaldens interval of 4.5 mm or more between C1 and C2 on radiographs (Cohen, 2006). Children who have atlantoaxial instability are at greater risk of incurring spinal cord compression, injury, paralysis, and in some cases, death (Cohen, 2006). Discrepancy exists regarding the screening protocols used for atlantoaxial instability. Current guidelines recommend all children with Down syndrome have radiographic evaluation between 3 to 5 years of age (AAP Committee on Genetics, 2001). Primary care providers should confirm this X-ray has been completed and the results show no indication of instability prior to instituting a physical activity program for weight management or clearing the child for sports activities. If the child develops any new neck pain, weakness, head tilt, or abnormal gait, re-screening with another X-ray is advised.

Heart. All children with Down syndrome should receive screening at birth for congenital cardiac defects. Most significant defects will be surgically corrected during the first year of life. Individuals with Down syndrome are prone to additional cardiac problems later in life. The incidence of mitral valve prolapse increases in the teenage years, and manifests on examination as a late systolic crescendo-decrescendo murmur with a mid-systolic click heard best at the apex. Aortic regurgitation is a blowing, decrescendo diastolic murmur heard best at the third left intercostal space, radiating to the left sternal border (Cable, 1997). If any abnormalities are found on the cardiac examination, the child should be referred to a cardiologist for further evaluation that may include an electrocardiogram (EKG) and echocardiogram. The cardiologist will determine if the child is cleared to participate in vigorous physical activity. The Special Olympics organization suggests screening for the presence of mitral valve prolapse and aortic regurgitation prior to any participation in sports-related physical activity (Platt, 2001).

Musculoskeletal system. Joints must be thoroughly examined due to the common finding of joint laxity in children with Down syndrome. Special care should be taken while examining the hips, knees, and feet (Winell & Burke, 2003). Examining the hips for stability is necessary through 10 years of age because children with Down syndrome sometimes have instability that leads to recurrent dislocations, which in severe cases cause loss of ambulation (Hresko, McCarthy, & Goldberg, 1993). Knee strength, flexibility, and range of motion should be noted when screening for patellofemoral instability, which is known to occur in children with Down syndrome (Winell & Burke, 2003). The feet should also be carefully examined because 90% of children with Down syndrome have deformities of the forefoot, 60% have flat feet, 24% have calcaneal valgus, 22% have knee valgus, and 16% have pronated flat feet (Concolino, Pasquzzi, Capalbo, Sinopoli, & Strisciuglio, 2006). Properly fitting shoes, and in some cases, orthotics will enhance foot comfort and encourage physical activity. It is also important to note children with Down syndrome who are obese will likely place more strain on their musculoskeletal systems due to their weight; therefore, screening carefully for musculoskeletal abnormalities and joint laxity is imperative to prevent unnecessary injury.

Laboratory Assessment

Laboratory evaluation will contribute to the assessment of the child with Down syndrome who is obese

Table 2.
Laboratory Assessment

BMI	Risk Factors*	Laboratory Tests
85th to 95th percentile	No	None indicated
85th to 95th percentile	Yes	Lipids, AST, ALT, fasting glucose
Greater than 95th percentile	No	Lipids, AST, ALT, fasting glucose
Greater than 95th percentile	Yes	Lipids, AST, ALT, fasting glucose

Note: BMI = body mass index, AST = amino-transferase, ALT = alanine amino-transferase.

* Risk factors include a family history of obesity-related illness, elevated blood pressure, or high cholesterol.

(see Table 2). As stated previously, thyroid function tests should be performed annually (AAP Committee on Genetics, 2001) due to the high incidence of hypothyroidism in children with Down syndrome. At this time, there are no specific recommendations for laboratory assessment of obesity in children with Down syndrome, but the protocols developed by Krebs et al. (2007) for the general pediatric population should be applied. When a child's BMI is between the 85th percentile and the 94th percentile with no risk factors, a fasting lipid panel should be ordered. However, if the child's BMI is between the 85th percentile and the 94th percentile with risk factors, such as family history of obesity-related illness, elevated blood pressure, or high cholesterol, the provider should order a fasting lipid panel, aspartate amino-transferase (AST) and alanine amino-transferase (ALT) levels, and a fasting glucose level (Krebs et al., 2007). When the child's BMI exceeds the 95th percentile, the previous tests should be ordered regardless of the family's risk factors (Krebs et al., 2007). If laboratory results and physical findings indicate possible diabetes mellitus, an oral glucose tolerance test measuring both glucose and insulin levels should be ordered (Krebs et al., 2007). A local obesity or endocrine clinic should be consulted if the provider is unsure which tests to perform or how to interpret abnormal results.

Management

Physical Activity

Providers can help children with Down syndrome prevent excess weight gain and promote weight loss by encouraging increased vigorous activity. Increasing participation in sustained vigorous physical activity prevents obesity and its negative health implications (Whitt-Glover et al., 2006). Ordoñez, Rosety, and Rosety-Rodriguez (2006) enrolled 22 overweight and obese adolescent males with Down syndrome in an exercise program consisting of three, one-hour aerobic exercise sessions weekly for three months. The reduction in mean value of fat mass was statistically significant (31.8% ± 3.7% to 26% ± 2.3%) by the end of the study.

Helping children become more active begins by counseling families to limit sedentary activities and identify any barriers to increased activity. Suggestions for activity include using exercise balls, going on family outings, and modifying traditional games, such as kickball, to increase movement and participation. Other ideas consist of prompting families and children to obtain gym memberships, attend fun exercise classes, play active video games (such as "Dance Dance Revolution®"), attend summer camp, and join sports teams with local youth leagues or the Special Olympics. Sports offered by the Special Olympics programs are year-round and include swimming, basketball, figure skating, cycling, skiing, tennis, and softball. To initiate involvement in this program, visit the Special Olympics' Web site for more information (www.specialolympics.org). Ultimately, the health care provider, child, and family must create a plan to increase activity together, or compliance will be minimal.

When making recommendations to families, the child's developmental status in addition to cardiac and musculoskeletal issues associated with Down syndrome must be taken into account. For example, teens with Down syndrome report they prefer to participate in activities, such as jogging or yoga, that do not require ability-matched teammates or opponents (Menear, 2007). These activities allow teens to enjoy the physical and social benefits of exercise without being concerned about performance or competition. Teens with Down syndrome may also be less interested in participating in "special" or adapted activities because they desire to be part of a peer group and not feel different (Menear, 2007). Whatever the activity and age of the child, the most important consideration is that the child has fun. If the child does not enjoy the activity, exercising will become a stressor and a battle for the family instead of an opportunity for the child to have fun, relate to others, and build new skills.

The child should be adequately screened for orthopedic anomalies and cardiac conditions prior to vigorous physical activity. Any musculoskeletal problems in children with Down syndrome must be addressed prior to participation in athletics to prevent musculoskeletal injury. If a child is found to have cervical instability, the family must be counseled regarding the importance of abstaining from contact sports, such as hockey, soccer, and gymnastics (Platt, 2001). Parents should also be counseled to ensure their child wears well-fitting shoes to avoid complications of foot anomalies; the use of proper footwear, such as lace-up sneakers with arch support or properly fitted cleats and skates, may prevent more serious injuries resulting in the need for surgical intervention and termination of the exercise program (Concolino et al., 2006; Platt, 2001). Additionally, if the child displays patellofemoral instability on examination, he or she should wear a patellar sleeve while participating in physical activity to prevent injury (Winell & Burke, 2003).

While most children now receive complete cardiac corrective surgery early in life and have excellent cardiac function, if the child or adolescent has a cardiac condition, clearance from a cardiologist must be obtained. The child, family, and coaches should be educated to recognize warnings of distress, such as palpitations, syncope, lightheadedness, and dyspnea. Attention to these warning signs may avoid potential episodes of cardiac arrest or cerebrovascular accidents.

Although costly, consulting with exercise physiologists may provide information about community programs or help develop individualized home exercise plans for families who are unable to commit to more structured activities. Increased physical activity is more likely to be sustained if it is fun and enables the child to socialize with peers in a non-competitive manner.

Table 3.
Interventions

Physical Activity	• Decrease sedentary behaviors • Increase physical activity – Gym membership – Special Olympics – Yoga – Dance – Traditional games – Active video games – Sports leagues • Consultation with exercise physiologist
Nutrition	• Limit caloric intake • Decreased control over feeding practices • Consultation with nutritionist

Table 4.
Helpful Web Sites

The Special Olympics	http://specialolympics.org
National Down Syndrome Society	http://www.ndss.org
Center for Disease Control on Obesity	http://www.cdc.gov/obesity/index.html
American Academy of Pediatrics on Obesity	www.aap.org/obesity
United States Department of Agriculture	http://www.mypyramid.gov

Nutrition

Children with Down syndrome should eat a balanced diet with vitamin and mineral supplementation (Luke, Sutton, Schoeller, & Roizen, 1996). Caloric restriction to prevent obesity or promote weight loss is recommended due to their decreased metabolic rate (Roizen, 2002). Limiting portion sizes or identifying consumption of foods with hidden sugar, such as cereals and beverages, serve as basic strategies to reduce caloric intake. Encouraging families to limit fast food dining to once per month is also an effective way to decrease the amount of calories consumed.

Although caloric restriction may be a key factor, parents must be warned against the tendency to control their child's feeding practices. The provider should encourage parents to talk about how they perceive their child's weight, assess their concern, and counsel them to avoid pressuring children to eat during mealtimes. Parents should also be educated about chewing difficulties in children with Down syndrome and provided with ideas of healthy, soft food alternatives, such as yogurt, steamed vegetables, and pureed fruits (Hennequin et al., 2005). Finally, parents should be discouraged from using foods as rewards or punishments.

Nutritionists may assist with family education. They serve as excellent resources regarding appropriate portion sizes, child-friendly food products, and healthy snacks. After talking with families, nutritionists may help to establish a meal plan and assist in identifying ways of altering maladaptive feeding behaviors or negativism in the child. In children with Down syndrome who also have celiac disease, a nutritionist is essential to assure the elimination of gluten from the child's diet (Allen, 2004).

Provider Roles

Research

Very little research has been conducted on obesity in children with Down syndrome, the role physical anomalies play in the development of obesity, behavioral and psychosocial aspects that place children at risk for excessive weight gain, or the efficacy of interventions to prevent obesity. Additional research examining successful weight-loss interventions used in the general pediatric population and their application to children with Down syndrome and their families should be conducted. Interventions, such as dancing, intensive dietary education, and specially designed camps, have been employed to manage obesity in children without Down syndrome, but no such interventions have been used for children with Down syndrome.

Community Involvement

In addition to research, providers may also become active in the community by volunteering and raising awareness of issues surrounding obesity in children with Down syndrome. One potential avenue for this work may be collaborating with existing local or national organizations, such as the Special Olympics or the National Down Syndrome Society. Providers may also serve as political advocates, addressing obesity issues pertaining to the Down syndrome community and the general population. These issues range from mandating physical education and nutrition courses, applying certain food taxes, or requesting healthier food options in schools.

Conclusion

Children with Down syndrome have rates of obesity as high, if not higher, than the general population. Research indicates children with Down syndrome develop obesity due to a variety of physiological mechanisms and behavioral tendencies. Because of the unique characteristics of the syn-

drome that contribute to the increased risk of developing obesity, nurses and other primary care providers must learn how to appropriately assess children with Down syndrome to identify abnormalities and ensure safe participation in sports activity. Taking pertinent histories, performing focused physical examinations, and ordering indicated laboratory tests serve as crucial aspects of quality patient care.

Exercise and nutrition-based interventions are important in preventing and reducing excess weight gain, but interventions should be individually tailored to the child with Down syndrome. Exercise recommendations consist of increasing movement through a variety of activities suitable to the child's physical and developmental abilities. Nutritional recommendations include decreasing caloric intake and reducing parental control over feeding practices. When counseling the family on both topics, it is important to address any barriers or stressors relating to diet and exercise. Together, the provider and family must problem solve to construct an agreeable plan. In particularly complex situations, providers should employ the help of other individuals, such as nutritionists, in the plan of care.

For a summary of interventions, see Table 3, and helpful Web sites can be found in Table 4. By working with families and collaborating with other health care professionals, children with Down syndrome will have an increased chance of living a healthier adult life.

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