

# ANESTHETIC CONSIDERATIONS FOR PATIENTS WITH DOWN SYNDROME

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*Patients with Down syndrome, or trisomy 21, present a unique set of anesthetic considerations to anesthesia providers. Down syndrome is the most prevalent genetic disorder worldwide and affects more than 1 in 800 live births. Patients with Down syndrome are afflicted with multiple congenital anomalies that affect almost all of their organ systems. Skillful management during the perioperative period is essential for a good outcome for patients with*

*multiple congenital abnormalities in the cardiopulmonary and musculoskeletal systems.*

*The purpose of this review is to describe clinically significant findings that are common to patients with Down syndrome. In addition to the presentation of clinical anomalies associated with trisomy 21, specific anesthetic considerations and interventions are reviewed.*

**Key words:** Anesthesia, Down syndrome, trisomy 21.

**T**risomy 21, also known as Down syndrome, is a genetic disorder that occurs in approximately 1 of every 800 live births.<sup>1</sup> John Langdon Down, a late 19th century English physician, earned recognition for discovering the syndrome because of his scholarly work published in 1866 that first offered an accurate description of a person afflicted with the syndrome.<sup>2</sup> Currently, Down syndrome affects more than 350,000 people in the United States.<sup>3</sup> Down syndrome represents the most frequent chromosomal abnormality and affects people of all ages, races, and economic levels. The purpose of this review is to describe clinically significant findings that are common to patients with Down syndrome.

## Pathophysiology

The normal human cell contains 46 chromosomes; however, in Down syndrome, the cell contains 47 chromosomes with the extra chromosome linked to chromosome 21, resulting in trisomy 21.<sup>4</sup> Trisomy 21 causes mental retardation in every case, but the degree is variable, ranging from severe to mild impairment with ability for independent living. Substantial literature exists suggesting potential causative factors for Down syndrome. Some of the suggested maternal factors include age older than 35 years, exposure to pesticides and electromagnetic fields, smoking, drinking, caffeine-containing drinks, thyroid autoimmunity, exposure to x-rays, and exposure to anesthetic agents. In addition to the developmental delays associated with Down syndrome, patients commonly have the following clinical sequelae: microcephaly and macroglossia, endocardial cushion defects and ventral septal

defects, duodenal atresia, atlantoaxial instability (AAI), and subglottic stenosis. The Table gives a summary of clinical findings and anesthetic considerations. As a consequence of the aforementioned clinical findings, patients with Down syndrome present a unique set of considerations and challenges for anesthesia providers.

## Review of the literature

An extensive literature review was performed using multiple search engines on the Internet, MEDLINE, and CINAHL. We identified potential sources, located the references, and screened the references for relevancy to anesthetic considerations for patients with Down syndrome. Extensive notes were taken, and inappropriate references were discarded. After all relevant references were reviewed, the sources were organized, analyzed, and integrated into this review. The articles were selected for inclusion in the topic review if they met one or more of the following criteria: (1) contained information about Down syndrome, (2) published in professional medical journals, and (3) included information on at least one of the health problems associated with Down syndrome.

## Atlantoaxial instability

Approximately 20% of patients with Down syndrome have ligamentous laxity of the atlantoaxial joint.<sup>5</sup> This condition may allow C1-C2 subluxation and predispose patients with Down syndrome to spinal cord injury. The potential subluxation of the C1-C2 vertebrae is the potential result of several causes. Laxity of the transverse ligament is one of the several causes of AAI. Hypoplasia, malformation, and absence of the

**Table. Anesthetic considerations for Down syndrome**

<b>System</b>	<b>Pathophysiology</b>	<b>Anesthetic considerations</b>
<b>Cardiac</b>		
<i>Acyanotic defects</i>	Complete atrioventricular defect; ventricular septal defect; atrial septal defect	Avoid increases in peripheral vascular resistance; avoid high SVR; use low $F_{IO_2}$ ; avoid low $ETCO_2$ ; antibiotics
<i>Cyanotic defect</i>	Tetralogy of Fallot	Normal to increased $F_{IO_2}$ ; maintain normal pH; keep $ETCO_2$ normal to low; keep SVR within normal limits; avoid large reductions; treatment of intraoperative tetralogy of Fallot spell: (1) fluids (10-20 mL/kg); (2) phenylephrine; (3) oxygen; (4) direct aortic compression via surgeon (if necessary)
<b>Pulmonary/airway</b>	Microcephaly; macroglossia; subglottic stenosis; sleep apnea/airway obstruction; respiratory tract infections	Consider smaller endotracheal tube; awake extubation; antibiotics
<b>Musculoskeletal</b>	Atlanto-occipital instability	Avoid neck flexion and extension during laryngoscopy; careful intraoperative positioning; review neck radiographs during preoperative assessment; consider using cervical collar intraoperatively (if demonstrated instability)
<b>Gastrointestinal</b>	Increased risk for gastroesophageal reflux disease; duodenal atresia common; tracheoesophageal fistula; Hirschprung disease; imperforate anus common	Possible rapid-sequence induction; awake intubation if severe gastrointestinal disturbance; gastroesophageal reflux disease prophylaxis preoperatively

SVR indicates systemic vascular resistance;  $F_{IO_2}$ , fraction of inspired oxygen;  $ETCO_2$ , end-tidal carbon dioxide.

odontoid process are other causes that predispose patients to the C1-C2 instability.<sup>6</sup>

In 1984, the American Academy of Pediatrics issued its first position statement on atlantoaxial instability in Down syndrome.<sup>7</sup> This position statement suggested that several precautions be taken when caring for patients with Down syndrome. All children with Down syndrome who want to participate in sports must have a cervical spine x-ray. When the distance on x-ray between the atlas (first vertebra) and odontoid process (second vertebra) is more than 4.5 mm, restriction on sports is advised. Repeated x-rays are not indicated for children with Down syndrome who have previously had normal x-rays. Persons with atlantoaxial subluxation or dislocation and neurologic signs should be restricted from all strenuous activities. Persons with Down syndrome who have no evidence of AAI may participate in all sports.<sup>7</sup>

For the aforementioned reasons, maneuvers such as positioning the head and neck during anesthetic management may place the spinal cord at risk if ligamentary instability is present. Hence, pediatric radiologists recommend that patients with Down syndrome

undergo a radiologic evaluation of the cervical spine before undergoing surgical procedures requiring anesthesia.<sup>5</sup> In addition to neck radiographs, assessing for excessive laxity of other joints, such as the finger, thumb, elbow, and knee, tends to correlate well with the presence of atlanto-occipital dislocation.<sup>6</sup>

In the preoperative period, it is imperative that anesthesia providers be aware of and understand the neurologic manifestations of atlantoaxial instability. A focused neurologic assessment accompanied by thorough documentation of findings on the anesthetic record is a high priority. Although most of the population with Down syndrome is asymptomatic with atlantoaxial instability, a small subset of patients has signs and symptoms that anesthesia providers should note. Signs and symptoms of AAI, if present, may include a positive Babinski sign, hyperactive deep tendon reflexes, ankle clonus, muscle weakness, increased muscle tone, neck discomfort, abnormal gait, and difficulty walking.<sup>7</sup> Such signs and symptoms often remain relatively stable for months or years; occasionally they progress to paralysis or, rarely, death.<sup>8</sup> In addition to a thorough preoperative assess-

ment, it is advisable to inform parents and/or caretakers of the risks associated with the manipulation of the head and neck required for anesthetic management. Preoperative care should include notifying parents and caretakers of patients with Down syndrome that neurologic deficit and injury may occur and require further therapy.

Due to the risks associated with Down syndrome, perioperative care needs to focus on the likelihood of cervical instability in patients with and without symptoms of AAI. Great care must be taken to maintain the neck in a neutral position. This may be accomplished by placement of a soft collar after induction of anesthesia to avoid extreme neck flexion, extension, and rotation. Leaving the collar in place may also serve as a valuable reminder to all caring for the patient during the perioperative and postoperative period that cervical instability may exist. If the preoperative examination or radiologic examination reveal AAI, the child should be referred to an orthopedic surgeon or neurosurgeon for further evaluation. The further evaluation should address whether stabilization of the cervical spine is necessary before any other surgery is undertaken.<sup>8</sup> If AAI is found, all elective procedures should be postponed until further evaluation of its severity can be made.

### **Tracheal stenosis**

Another concern to the anesthetic management of patients with Down syndrome is tracheal stenosis. Tracheal stenosis is a more common finding in children with Down syndrome. Tracheal stenosis presents anesthesia providers with additional and heightened concerns regarding complicated intubation and increased airway narrowing after intubation or bronchoscopy. Congenital tracheal stenosis is characterized by the absence of the membranous portion of the trachea and fusion of the posterior aspect of the tracheal cartilage.<sup>9</sup> "Tracheal stenosis may result in generalized hypoplasia of the entire trachea, segmental stenosis, or funnel shaped narrowing with a tapered distal trachea."<sup>9</sup> Clinical sequelae as they relate to tracheal stenosis may include respiratory distress, stridor, croup, bronchiolitis, cyanotic events, wheezing, and failure to thrive. Radiographs are useful in identifying stenosis; however, confirmation usually is provided by direct endoscopy. The possibility exists that all children with Down syndrome have a smaller subglottic area than children without Down syndrome.<sup>10</sup> According to Kobel et al<sup>6</sup> in a study of 100 patients during a 2-year period, of patients with Down syndrome whose records were reviewed, 23% had been intubated with smaller tubes than would be expected for their age.

Anesthesia providers should be aware of the interventions necessary to care for a patient with Down syndrome with coexisting subglottic stenosis or occult subglottic stenosis. During the preoperative period, special care should be taken with respiratory assessment. Auscultation over the 4 lung fields and the trachea is necessary and beneficial to selecting the appropriate airway modalities for effective airway management. It is recommended that a large selection of endotracheal tubes be prepared for intubation and that anesthetists carefully select the endotracheal tube that will allow a leak during positive-pressure ventilation to avoid postoperative complications. It is also advisable that anesthesia providers have a selection of emergency airway modalities available, such as fiberoptic equipment, intubating laryngeal mask airway, flexible bougie, and emergency surgical airway necessities. As a result of subglottic stenosis in addition to other clinical sequelae, awake intubation often is recommended. During the postoperative period, special care should be taken to assess the patient for adequate ventilation due to the increased possibility of postintubation croup and airway swelling.

### **Cardiac complications**

Cardiac abnormalities are very common in patients with Down syndrome. Thus, it is of utmost importance that anesthesia providers be aware of such cardiac abnormalities. The clinical manifestations of cardiac anomalies often are not found until the patient is placed in a compromising clinical situation such as anesthesia. It is imperative that the patient undergo a thorough preanesthetic assessment that includes extensive cardiac examination before any procedure requiring anesthesia. Although many cardiac abnormalities are associated with Down syndrome, some are more prevalent than others.

As many as 12% of patients with Down syndrome have cardiac lesions that are clinically apparent.<sup>10</sup> In addition, as many as 60% of autopsy studies indicate the presence of cardiac lesions in patients with Down syndrome.<sup>10</sup> It is important for anesthesia providers to recognize that the incidence of specific cardiac lesions associated with Down syndrome is different and more significant than the in rest of the population. The specificity of the cardiac abnormality presents a unique set of anesthetic considerations when preparing for and providing anesthetic care to a client with trisomy 21.

The prevalence of each cardiac abnormality is variable. Endocardial cushion defect is the most common lesion, occurring in about 40% of detected lesions.<sup>11</sup> Ventricular septal defect and complete atrioventricu-

lar defect occur in 30% to 60% of all patients with Down syndrome.<sup>11</sup> Patent ductus arteriosus (12%) and tetralogy of Fallot (8%) occur less frequently but are also of significance.<sup>10</sup> Although many children with Down syndrome undergo repair of cardiac defects repaired, there is an increased incidence of mitral valve prolapse and aortic regurgitation in adolescents and adults with Down syndrome.<sup>11</sup>

It is important to note that all of the aforementioned cardiac lesions, with the exception of tetralogy of Fallot, can result in increased blood flow to the pulmonary vasculature. An exacerbation of the symptoms associated with these cardiac lesions may occur as a consequence of coexisting pulmonary vascular disease. The literature supports an increased tendency for children with Down syndrome to develop pulmonary vascular disease independent of preexisting cardiac anomalies.<sup>11</sup> Many children with Down syndrome undergo surgical correction of cardiac lesions. Despite a relatively high success rate in the correction of cardiac abnormalities, current literature suggests an increase in morbidity and mortality associated with the surgical correction of cardiac lesions, presumably due to recurrent infections and an increased incidence of pulmonary hypertension.<sup>12</sup>

As part of the preanesthetic evaluation of a patient with Down syndrome, it is important to note whether any surgical correction or other invasive procedures have been performed for previously or currently existing cardiac abnormalities. Many patients with Down syndrome are admitted for surgery with general anesthesia after successful repair of cardiac lesions. Some patients may be asymptomatic, whereas others may have residual defects that may limit their activities and increase their anesthetic risk.<sup>10</sup> Conduction disturbances are common following repair of atrioventricular fistula, tetralogy of Fallot, and ventricular septal defects. In particular, atrial rhythm anomalies are quite common following repair of transposition of the great vessels. It is important for anesthesia providers to recognize that residual symptomatic defects after the repair of cardiac abnormalities occur not only in the immediate postoperative period, but also throughout the patient's lifespan.

Anesthesia providers have an important role and commitment to minimizing the anesthetic and surgical risk of a patient who undergoes anesthesia. Because of the link between increased morbidity and mortality and the high occurrence of infections in patients with Down syndrome,<sup>12</sup> in collaboration with the surgical team, appropriate antibiotics should be administered during the perioperative period for patients undergoing surgical repair of cardiac lesions.<sup>12</sup> Prophylactic

antibiotics are indicated before any surgical procedure in some patients with Down syndrome, even if they have undergone previous successful repair of cardiac lesions. In addition, and as a general rule, patients who have undergone aortic valvotomy, resection of coarctation of the aorta, pulmonary valvotomy, correction of tetralogy of Fallot, or any valve replacement should receive antibiotics.<sup>10</sup>

In addition to reducing the morbidity and mortality of associated cardiac risks and infections by administering antibiotics and using strict aseptic technique, it is important to emphasize the need for a complete and thorough cardiac assessment. If any doubt exists about the patient's current cardiac status, elective cases should be delayed for referral to an appropriate cardiac provider.

### **Respiratory complications**

Respiratory problems often are encountered in children with Down syndrome. Upper and lower respiratory problems are frequent in this subset of the population. Lower airway problems have been linked to hypotonia, relative obesity, cardiac disease, small upper airways, small lower airways, and a degree of pulmonary hypoplasia.<sup>12</sup> Congenital anomalies of the lower airways are common and are strongly linked with cardiovascular anomalies.<sup>13</sup> The anomalies of the lower airways include stenotic anomalies, anomalies that cause airway collapse, tracheoesophageal fistula, and branching anomalies.<sup>13</sup> Vascular compression can also cause compression of the large airways. The innominate artery often can be aberrant in patients with Down syndrome, which can cause compression of airway structures.<sup>13</sup> Factors affecting the upper airways include hypotonia, obesity, midface hypoplasia, relative glossoptosis, small upper airway volume (approximately two thirds of the normal volume), increased secretions, and excessively large tonsils and adenoids.<sup>13</sup>

Obstructive airway disease has been recognized as a significant problem for children and adults with Down syndrome. Symptoms include snoring, unusual sleeping positions, increased fatigue during the day, reappearance of napping in older children, or behavior change.<sup>13</sup> During the preoperative period, individuals with these symptoms should be evaluated completely via a detailed history and physical assessment with regard to tonsillar size and airway structure evaluation.

Pulmonary vascular disease is increased in the population with Down syndrome. The risk of pulmonary hypertension and the development of Eisenmenger heart disease in children with Down syndrome are

accelerated compared with children without Down syndrome.<sup>13</sup> Eisenmenger syndrome exists when left to right intracardiac shunt is reversed as a result of increased pulmonary resistance that is equal to or greater than systemic vascular resistance.<sup>11</sup> This finding has been linked with pulmonary hypoplasia. Children with Down syndrome often have a decreased number of alveoli, which means that the size of their pulmonary vasculature is decreased, resulting in increased risk of pulmonary problems. This finding, in association with the common finding of obstructive sleep apnea, is thought to lead to increased pulmonary vascular disease in this population. Strict attention must be given to preoperative assessment of patients with Down syndrome with regard to their respiratory status. A thorough respiratory assessment is paramount to a successful intraoperative course.

### Other organ systems

Several other organ systems are affected by Down syndrome. Gastroesophageal reflux disease is more prevalent in this population. Some symptoms that should be assessed preoperatively are the presence of vomiting, esophagitis (which can be associated with chest pain, anemia, and irritability), and respiratory symptoms such as apnea, coughing, wheezing, and aspiration pneumonia.<sup>13</sup> Patients with Down syndrome have some degree of immune dysfunction, and children with Down syndrome have a developmentally different immune system. This affects their cellular and humoral immunity, and it is thought that this can lead to an increased rate of infection.<sup>13</sup> It also is important to note that patients with Down syndrome can have significant hearing loss and eye and vision problems.

### Conclusion

There are myriad clinically significant findings that anesthesia providers may encounter when providing anesthesia to patients with Down syndrome. With the relatively high prevalence of Down syndrome in the general population, the likelihood of contact with a patient with the syndrome is high in the anesthesia community. With a sufficient knowledge base of the

significant health problems that can accompany Down syndrome, anesthesia providers can avoid complications that may be encountered during the perioperative period. Perhaps in the future, further research will expand on the existing knowledge base for trisomy 21.

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