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Treacher Collins Syndrome: Challenges for Anesthesia Care

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**Introduction**

Treacher Collins syndrome is a rare congenital disorder that poses many challenges to healthcare professionals. British ophthalmologist Edward Treacher Collins was the first to publish a paper detailing the features of the disorder and as a result his name is associated with the condition (Barnes, Marshall, Balasubramaniam, Varma, Aditi, 2014).

**Pathology**

Treacher Collins syndrome (TCS) is an inherited genetic disorder caused by mutations of one of three genes (Katsanis, Jabs, 2012). In most individuals, mutations of the TCOF1 gene (80%) result in Treacher Collins syndrome (TCS). In the remaining 20% of cases, mutations of the PGRN or POLR3C gene are observed (Katsanis, Jabs, 2012). The majority of the time, TCS is inherited in an autosomal dominant fashion and it affects both males and females (Katsanis, Jabs, 2012). Clinical and radiographic findings assist in diagnosis of the syndrome (Katsanis, Jabs, 2012).

**Anesthesia Challenges**

The craniofacial deformities which usually accompany this syndrome make it of special interest to anesthesia providers because of the risk for difficult airway management in patients undergoing surgery or other procedures requiring intubation. Difficult intubations combined with difficult mask ventilation can easily create an airway emergency (Hosking, 2014). Many anesthesia providers assess the degree of expected airway difficulty if intubation of the trachea is attempted (Vanadeven, Balasubramaniam, Varma, Aditi, 2008). A class 1 airway is anticipated to be easiest to manage and the degree of expected difficulty increases if the Mallampati score increases (Vanadeven, Balasubramaniam, Varma, Aditi, 2008). It was found that approximately 80% of patients had a Mallampati airway score of class I or above (Hosking, Zeenatti, Gayte, Anderson, Costi, 2012). The same study demonstrated an incidence of 6% of 123 pediatric cases of planned surgery requiring intubation (Hosking, Zeenatti, Gayte, Anderson, Costi, 2012). Additionally, in most cases of Treacher Collins the degree of airway management difficulty combined with the need for reconstructive surgery makes the airway the opposite of what happens in a patient with a normal airway for reconstructive malformations (Hosking et al., 2012).


A male infant is given a definitive diagnosis of TCS shortly after birth. The infant displays hypoplasia of the facial features with a prominent nose and downswept slanting eyes. Micrognathia is present and the infant displays small malformed ears bilaterally in addition to sparse scalp hair. Distinct facial abnormalities alert medical staff to the possibility of TCS and further testing revealed a mutation of the TCOF1 gene. The infant is able to maintain a patent airway and initial feeding difficulties are not severe enough to pose concerns for malnutrition or aspiration. After an extended observation period and 3 months of age the infant is discharged home.

**2 month old boy with TCS**

(Tolatova, 2016)

**Prognosis**

Most individuals with TCS can mature to lead a normal life with appropriate medical care. Patients with TCS will usually display normal intelligence but may experience delays of hearing or vision deficits if not addressed (Tolatova, 2016). Genetic counseling is of great importance for affected individuals because each child of a parent with TCS will have a 50% chance of inheriting the disorder (Katsanis, Jabs, 2012). Intrauterine ultrasound should be prepared to perform emergency tracheostomy, cricothyroidotomy with jet ventilation if an airway emergency ensues and the patient is unable to be successfully ventilated or awakened (Mittal, 2015). In this case, emergency airway management was attempted using video laryngoscopy and the surgery proceeded as planned.

**Conclusion**

TCS is a rare disorder which healthcare providers may never encounter during their career. However, understanding the unique features of the syndrome and knowledge of the characteristics which may present life threatening complications can lead to better outcomes for patients. Although understanding the disorder is valuable for all healthcare providers, it is especially important for those providing anesthesia care. The ability to prepare for difficult airway management and anticipate potential problems can help ensure successful care and patient outcomes.

**Treatment**

Treatment is based on the severity and manifestations of the disorder. Infants may require tracheostomy to protect the airway because it is compromised with speech and reconstructive surgery may be necessary (Katsanis, Jabs, 2012). Infants who require tracheostomy usually require care from multiple medical specialties including craniofacial surgeons, speech therapists and orthodontists (Tolatova, 2016). When craniofacial repair is indicated, it is usually performed frequently over time in order to follow developmental age (Katsanis, Jabs, 2012). Micrognathia is corrected at age 1 years, and then computer reconstruction at age 5-7 and surgery of the craniofacial unit is performed at sometimes age 4 years (Katsanis, Jabs, 2012). The exact location on the gene where the mutation occurs can affect the expression and severity of the disorder (Katsanis, Jabs, 2012).

**References**


Chiang et al., 2009). As a final precaution, the surgical team should be prepared to perform emergency tracheostomy, cricothyroidotomy with jet ventilation if an airway emergency ensues and the patient is unable to be successfully ventilated or awakened (Mittal, 2015). In this case, emergency airway management was attempted using video laryngoscopy and the surgery proceeded as planned.

**Additional Sources**


**Possible combinations for offering when one parent has TCS**

Picture retrieved from http://dlassoappictures.com/treacher-collins-syndrome**