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Treacher Collins Syndrome: Challenges for Anesthesia Care

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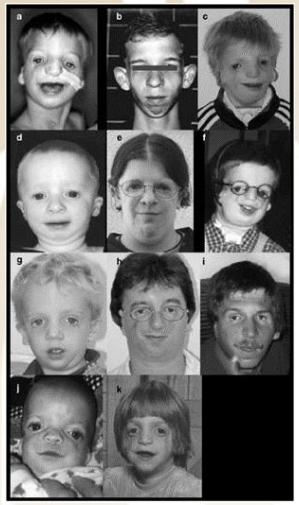
Introduction

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 disease and as a result his name is associated with the syndrome (Rashmi, Manjula, Balasubramanyam, Varsha, Aditi, 2014).

Treacher Collins syndrome occurs with a frequency of 1:40,000 to 1:70,000 births (Rashmi et al., 2014). Hallmark features of this disorder include "malar hypoplasia caused by hypoplasia of the zygomatic complex, mandibular hypoplasia, down slanting palpebral fissures, colobomata of the lower eyelids, and ear malformations often associated with bilateral conductive hearing loss" (Bowman et al., 2012). Although individuals affected with Treacher Collins syndrome can display a great deal of variability in expression of features, the syndrome is recognized as one of the most severe craniofacial malformation disorders (Sakai, Trainor, 2009). Disease symptoms can be so severe that death ensues before or immediately following birth while some cases of the syndrome are so mild they may initially go undiagnosed (Trainor, Dixon, Dixon, 2009).

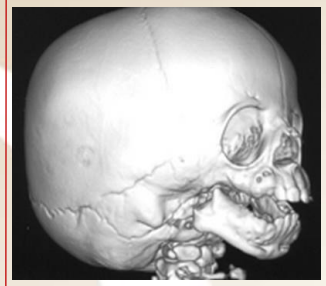
Individuals with TCS and a detected TCOF1 mutation (Katsanis, Jabs, 2012)



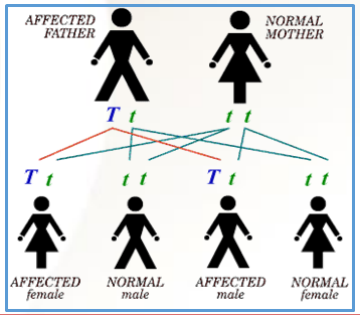
Pathology

Treacher Collins syndrome (TCS) is an inherited genetic disorder caused by mutation of one of three genes (Katsanis, Jabs, 2012). In most individuals, mutation of the TCOF1 gene is responsible (78%-93% of individuals with TCS) and in the remaining cases (8% of individuals with TCS) mutation of the POLR1C or POLR1D gene is the cause (Katsanis, Jabs, 2012). The majority of the time TCS is inherited in an autosomal dominate fashion with only about 1% of cases being inherited in an autosomal recessive pattern (Katsanis, Jabs, 2012). Clinical and radiographic findings assist in diagnosis of the syndrome (Katsanis, Jabs, 2012).

X-ray of Treacher Collins Syndrome
Image retrieved from http://syndrome pictures.com/treacher-collins- syndrome-pictures/



Possible combinations for offspring when one parent has TCS
Picture retrieved from http://www.treachercollins.co.uk/gene/genes.htm



Anesthesia Challenges

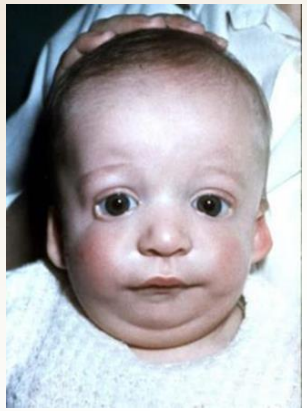
The craniofacial deformities which usually accompany this syndrome make it of special interest to anesthesia providers because of the risk for difficult airway management in patients undergoing surgery or other procedures requiring sedation. A high tendency for difficult intubation combined with difficult mask ventilation can easily create an airway emergency. Mallampati airway classification is one way in which anesthesia providers assess the degree of expected airway difficulty if intubation of the trachea is attempted (Vasudevan, Badhe, 2008). A class I airway is anticipated to be the easiest to manage and the degree of expected difficulty increases as the Mallampti score increases (Vasudevan, Badhe, 2008). Class III airways may be difficult to intubate while a class IV airway is expected to be difficult or even impossible to intubate (Vasudevan, Badhe, 2008).

In a 2012 study of pediatric patients with Treacher Collins syndrome it was found that approximately 80% of patients had a Mallampati airway score of class III or above (Hosking, Zoanetti, Carlyle, Anderson, Costi, 2012). The same study demonstrated an intubation fail rate of 5% with 123 pediatric cases of planned intubation (Hosking et al., 2012). Additionally, in most cases of Treacher Collins the degree of airway management difficulty increases with age which is the opposite of what happens in a patient without craniofacial malformations (Hosking et al., 2012).

A Study of Difficult Airway and Successful Anesthetic Management

A male infant is given a definitive diagnoses of TCS shortly after birth. The infant displays hypoplasia of the facial features with a prominent nose and downward slanting eyes. Micrognathia is present and the infant displays small malformed ears bilaterally in addition to sparse eyelashes. Distinctive facial abnormalities alerted the 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 provisions for follow up care the infant is discharged home.

2 month old boy with TCS
(Tolarova, 2016)



Parents of the infant are educated on the syndrome and provided with genetic counseling. As the child grows, care includes frequent visits with a primary care pediatrician as well as consultation with pediatric specialists. The child presents at the age of seven for reconstructive surgery of bilateral ears. The multi-specialty surgical team is skilled in managing patients with craniofacial malformations and plans carefully for the anesthetic care needed for the procedure. The team is aware that airway management during anesthesia for patients with TCS may be very challenging (Aggarwal, Sharma, Uttam, 2012).

It is decided that induction of anesthesia will be conducted via mask with inhalational agent and then endotracheal intubation will be attempted using a video laryngoscope. If intubation is unsuccessful the use of an intubating LMA will be employed. If both measures fail to provide a suitable airway for surgery then the patient will be awakened and consideration for tracheostomy will be made before surgery is attempted again. Successful fiberoptic nasal intubation has also been used in some cases of Treacher Collins (Tzu-Chiang et al., 2009). As a final precaution, the surgical team should be prepared to perform emergency tracheotomy or to initiate emergency 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, endotracheal intubation is successful using video laryngoscope and the surgery proceeds as planned.

Treatment

Treatment is based on the severity and manifestations of the disorder. Infants may require tracheostomy to protect the airway, hearing loss is common and craniofacial reconstruction may be necessary (Katsanis, Jabs, 2012). Individuals with TCS usually require care from multiple medical specialties such as craniofacial surgeons, speech therapists and orthodontists (Tolarova, 2016). When craniofacial repair is indicated, multiple surgeries are frequently necessary and should be conducted at the appropriate developmental age (Katsanis, Jabs, 2012). Cleft palate repair typically occurs at age 1-2 years, zygomatic and orbital reconstruction at age 5-7 and surgeries of the external ear are conducted sometime after age 6 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).

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 learning delays if hearing or vision deficits are not addressed (Tolarova, 2016). Genetic counseling is of great importance for affected individuals because each child of a patient with TCS will have a 50% chance of inheriting the mutation (Katsanis, Jabs, 2012). Unaffected parents of children with TCS may not be at increased risk of having a second child with the disorder because more than half of cases represent a new mutation (Tolarova, 2016).

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 those 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 positive patient outcomes.

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

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