Treacher Collins Syndrome: Challenges for Anesthesia Care

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Treacher Collins Syndrome: Challenges for Anesthesia Care
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A Study of Difficult Airway and Successful Anesthetic Management

Introduction
Treacher Collins syndrome is a rare congenital disorder that poses many challenges to healthcare professionals. 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 (Katsanis, 2012). Balasubramanyam, Varsha, Aditi, et al. (2012). Treacher Collins syndrome occurs with a frequency of 1:40,000 to 1:70,000 births (Raath et al., 2014). Hallmark features of this disorder include “stare hypoplasia caused by hypoplasia of the hypoplastic maxilla, mandible dysplasia, downslanting palpebral fissures, contractures of the lower eyelids, ear malformations often associated with conduction hearing loss” (Brown et al., 2012). Although individuals affected with Treacher Collins syndrome can disrupt a great deal of variability in expression of features, the syndrome is recognized as one of the most severe craniofacial malformation disorders (Sakai, Tramer, 2000). Lesions can be so severe that death occurs before immediately following birth while some cases of the syndrome are so mild they may initially go unnoticed (Troner, Dixon, 2009).

Pathology
Treacher Collins syndrome (TCS) is an inherited genetic disorder caused by mutation of one of three genes (Katsanis, 2012). In most individuals, the mutation is of the TCOF1 gene (79%-99% with TCS and individuals with TCS) and in the remaining cases, the mutation is of the POLR1C gene in the causative (Katsanis, 2012). The majority of the time TCS is inherited in an autosomal recessive pattern (Vasudevan, Balasubramanyam, Varsha, Aditi, et al. 2012). Clinical and radiographic findings assist in diagnosis of the syndrome (Katsanis, 2012).

Anesthesia Challenges
The craniofacial deformities which usually accompany this syndrome make it of special interest to anesthesiologists. Patients with TCS may have a difficult airway and may require care from multiple medical specialists (Katsanis, S.H., & Jabs, E.W. 2012). Although the degree of expected difficulty increases the degree of expected difficulty increases the Mallampati score increases (Vasudevan, Balasubramanyam, Varsha, Aditi, et al. 2012). In a 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 (Skroding, Zaouetti, Courti, Andersen, et al. 2012). The same study demonstrated a Mallampati score of 5% with 123 pediatric cases of planned sedation. In addition, in most cases of Treacher Collins the degree of airway management difficulty increases with age and severity of the disorder (Skroding, Zaouetti, Courti, Andersen, et al. 2012). Treatment
Treatment is based on the severity and manifestations of the disorder. Infants may require tracheostomy to protect the airway before leaving the hospital. Reconstructions in conjunction with speech therapists and orthodontists may be necessary (Katsanis, Jabs, 2012). Dental issues requiring care from multiple medical specialties such as oral surgeons, orthodontists, speech therapists and orthodontists (Tolarova, 2016). When craniofacial repair is indicated, it is necessary to define the specific developmental age (Katsanis, Jabs, 2012). In patients with TCS aged 3 years, key surgical decisions can be made (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).

Possible combinations for offspring when one parent has TCS and a detected mutation of the TCS gene: 5% of cases being inherited from one parent (78% of cases being inherited from the other parent (Tolarova, 2016). Possible combinations for offspring when one parent has TCS: 5% of cases being inherited from one parent (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). Informed 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).

Prognosis
TCG 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 various techniques which may present life threatening complications can lead to better outcomes for patients. Although understanding the disorder is valuable to all healthcare providers, it is especially important for those providing anesthesia care. The ability to prepare for anticipated and anticipate potential problems can help ensure successful care and positive patient outcomes.

References
Balasubramanyam, Varsha, Aditi, et al. (2012). Treacher Collins syndrome (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 various techniques which may present life threatening complications can lead to better outcomes for patients. Although understanding the disorder is valuable to all healthcare providers, it is especially important for those providing anesthesia care. The ability to prepare for anticipated and anticipate potential problems can help ensure successful care and positive patient outcomes.

Additional Sources

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