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. Critical 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 conditions (Rashmii, Manjula, Balasubramanyam, Varsha, Aditi, 2012).

Treacher Collins syndrome occurs in a frequency of 1:40,000 to 1:100,000 births (Rashmii et al., 2014). Hallmark features of this disorder include “malar hypoplasia caused by hypoplasia of the zygomatic bone, mandibular hypoplasia, down slanting palpebral fissures, colobomata of the eyelid, and ear malformations among others or facial” (Rossman et al., 2012). Although individuals affected with Treacher Collins syndrome can depict a great deal of variability in expression of features, the syndrome is recognized as one of the most severe craniofacial malformation disorders (Badhe, 2012). Ozoneic changes can be so severe that death ensues before immediately following birth while some cases of this syndrome are so mild they may initially go unnoticed (Troxler, Dixon, M. J. 2009).

Pathology

Treacher Collins syndrome (TCS) is an inherited genetic disorder caused by mutations of one of three genes (Katsuani, J., 2012). In most individuals, mutations of the TCOF1 gene (98%–99% with individuals with TCS and 3% in the remaining cases of TCS) cause the condition. Mutations of individuals with TCS mutation of the POLR1C or POLR1D gene is assumed (Katsuani, J., 2012). The majority of the TCS gene is inherited in an autosomal dominant fashion with about 1% of cases being inherited in an autosomal recessive pattern (Katsuani, J., 2012). Clinical and radiographic findings assist in diagnosis of the syndrome (Katsuani, J., 2012).

Anesthesia Challenges

The craniofacial deformities which usually accompany this syndrome make it of special concern to anesthesiologists. Patients with TCS require the airway to be evaluated for any malformations that may contribute to the difficulty of intubation (Vasudevan, Badhe, 2008). The degree of airway difficulty increases with age as the degree of airway hypoplasia increases (Katsuani, K., 2012). The majority of those cases with bilateral conductive hearing loss“ (Rashmii et al., 2014). Hallmark challenges to healthcare professionals. Individuals with TCS may be very challenging during anesthesia for patients 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 (Tolatov, 2016).

A Study of Difficult Airway and Successful Anesthetic Management

Treacher Collins Syndrome: Challenges for Anesthesia Care

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Treatment

Treatment is based on the severity and manifestations of the disorder. Infants may require tracheostomy to protect the airway but hearing loss is common with some cases requiring reconstructive surgery. It is the opposite of what happens in a patient with airway emergencies (Hosking et al., 2012).

The child presents at the age of seven for reconstructive surgery of bilateral ears. The multi-specialty surgical team is shifted 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, H., & Uttam C., 2012).

Possible combinations for offering when one parent has TCS


References


Additional Sources


Prognosis

Most individuals with TCS may mature to lead a normal life with appropriate medical care. Patients with TCS will usually display normal intelligence but may experience delays learning due to hearing or vision deficits are not addressed (Tolatov, 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 disorder (Katsuani, J., 2012). Infastrate, 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 (Tolatov, 2016).

Additional Sources


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