Tracheoesophageal fistula in siblings - A rare occurrence

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Introduction: Esophageal atresia is one of the most common congenital malformations occurring in 1 in every 2000–4000 live births. Previously considered to be a fatal condition, it has now become an eminently treatable condition with survival limited only by the presence or absence of major cardiac malformations. For the majority of families, the condition is unique to one child which in itself is challenging. It is extremely rare to affect more than one sibling in any family. The risk of recurrence is 0.5-2% and rises to 20% if another sibling is affected.

Case description: We report on two siblings, born two years apart with Type C TOF who were treated at King Hamad University Hospital, Bahrain. The first sibling was born with a short gap esophageal atresia with a distal fistula and repaired shortly after birth in another institution. The second sibling was born with a more challenging long gap esophageal atresia also with a distal fistula. The fistula was ligated on the second day of life and a gastrostomy tube put in place for feeding; repair of the atresia took place 2 months later. Both patients underwent serial dilatations to overcome strictures but are both tolerating oral diets. The third sibling in the family was unaffected with TOF or any VACTREL association.

Conclusion: The etiology of TOF is multifactorial with genetic, environmental and unknown components. There were no known environmental factors which could have contributed to this rare occurrence, but the only factor of significance is consanguinity of the parents who are first cousins. On the other hand, genetic factors are responsible for 12% of cases, these are classified as Chromosomal mutations, Syndromic or Isolated. These siblings do not fall into any of the mentioned categories and it is therefore speculated that this occurrence is due to a sporadic mutation.

Take-home message: Esophageal atresia with or without fistula is a fairly common condition encountered by paediatric surgeons on a daily basis. Despite increased experience and advancements in management of complex cases, the etiology remains a mystery. Future research should focus on more in-depth genetic studies on the impact of co-consanguinity on sibling TOFs.

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GERD: A debated background of achalasia

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Achalasia is a primary esophageal motility disorder of unknown etiology, characterized by aperistalsis of the esophageal body and impaired lower esophageal sphincter (LES) relaxation. However achalasia is the best characterized esophageal motility disorder, its pathogenesis is still not entirely clarified. Available data suggest that the disease is multifactorial, involving hereditary, autoimmune and environmental factors, such as viral infections, but the exact initiating factors that may play a role in the development of the disease remain unclear. Our hypothesis is that one possible initial insult that leads to the development of achalasia can be the gastroesophageal reflux disease. This theory was first proposed by