PRIMEVIEW

OESOPHAGEAL ATRESIA

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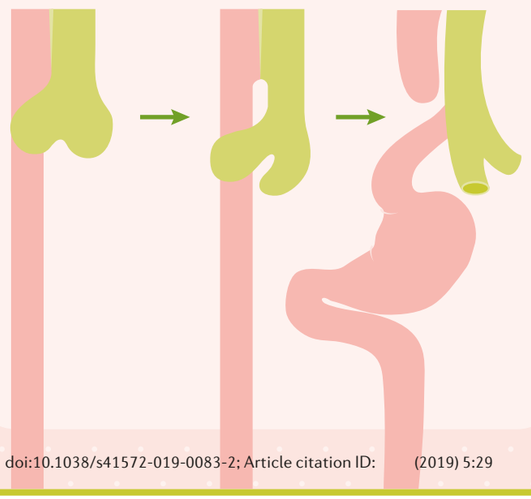
Oesophageal atresia (EA) is a congenital abnormality of the oesophagus that requires surgical reconstruction soon after birth. Oesophageal dysfunction, associated with numerous co-morbidities, occurs in those born with EA.

MECHANISMS

EA is thought to arise as a result of abnormal embryonic development of the foregut, resulting in a 'disrupted' oesophagus. The exact mechanism of separation of the embryonic foregut into the oesophagus and trachea has not yet been verified, but three morphological models have attempted to explain the process. Of these models, the watershed model — which describes the growth of foregut tissue at both sides, with new tissue becoming either trachea or oesophagus — is the most widely accepted model, although little evidence supports any one model. Several genes and pathways seem to be essential for foregut compartmentalization into oesophagus and trachea, but their specific roles are poorly understood. However, study of mouse embryos (normal and treated with chemicals such as adriamycin to induce a similar defect to EA) is actively being sought to better understand the processes involved.



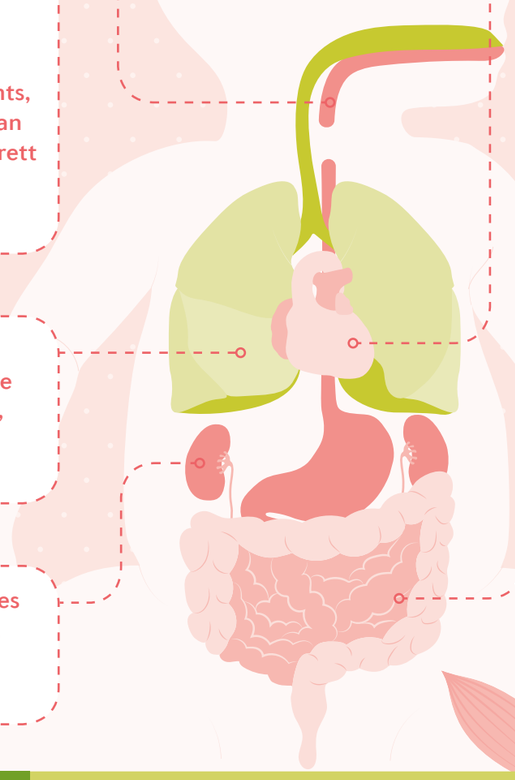
EPIDEMIOLOGY

EA is the most common congenital abnormality of the oesophagus, with many also having a tracheo-oesophageal fistula (TEF). People born with EA often have associated birth defects or other anomalies, with vertebral, anorectal, cardiac, TEF, renal, radial and/or limb (VACTERL) association being common.

Oesophageal motility is often disordered in patients born with EA, leading to delayed oesophageal clearance. Coupled with gastro-oesophageal reflux disease, which is also common in these patients, chronic inflammation can develop, leading to Barrett oesophagus and even adenocarcinoma.

Respiratory anomalies are common and include laryngotracheomalacia, vocal cord paresis and subglottic stenosis.

Genitourinary anomalies include renal agenesis, cystic kidneys and ureteral anomalies.



Cardiovascular anomalies occur in 29% of patients born with EA, including tetralogy of Fallot and atrial and ventral septa defects.

Gastrointestinal anomalies occur in 16% of patients born with EA and include anorectal malformations, duodenal atresia and intestinal malrotation.

Musculoskeletal anomalies include vertebral/rib anomalies and limb reduction deficiencies.

DIAGNOSIS

Most patients with EA are diagnosed after birth. A neonate with EA usually produces bubbly saliva, has signs of respiratory distress and shows distress during the first feeding attempt. The diagnosis can be confirmed when it is impossible to position a nasogastric catheter in the stomach. Assessment of co-occurring anomalies and post-surgical co-morbidities is essential to optimize care.

Long-gap EA can be defined using various descriptions, and is generally considered the most difficult to repair

MANAGEMENT

EA is treated surgically to join the proximal and distal oesophageal pouches and, if present, separate the TEF. Early postoperative complications include anastomotic leak, oesophageal stricture (narrowing) and recurrence of the TEF, but strategies are available to address these issues. Importantly, a multidisciplinary team comprising a paediatric surgeon, gastroenterologist, pulmonologist, otolaryngologist, clinical geneticist, speech pathologist, physiotherapist and/or dietician should be consulted if needed throughout the life of the patient to address the co-morbidities associated with EA.

OUTLOOK

Given that EA is a rare condition, it is unsurprising that many questions abound — from how the anomaly forms in utero to how best to manage patients. Current efforts aim to set up registries of patients for research purposes, for example, to use exome sequencing for molecular characterization of patients born with EA. Additionally trials are needed to compare and evaluate different surgical techniques (such as open versus thoracoscopic) and different medical treatments for GERD in this population and to address the respiratory complications.

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