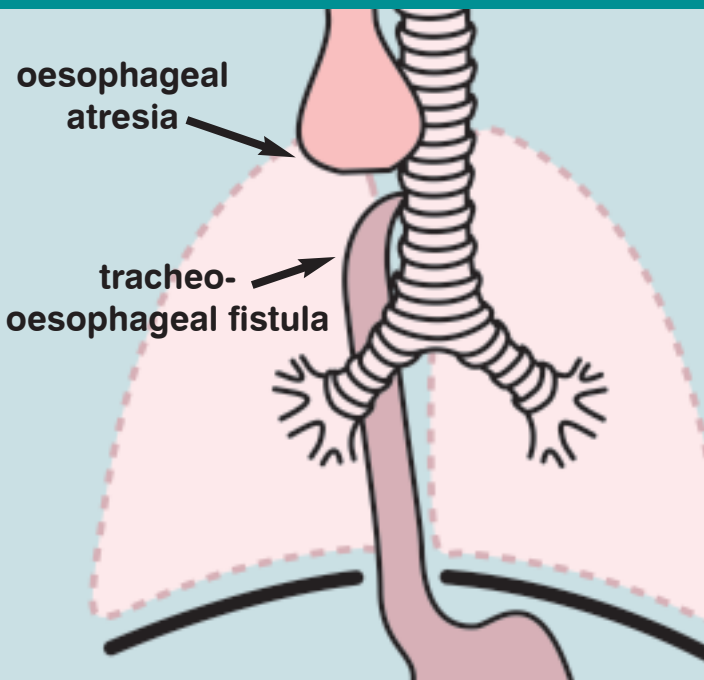


# Improving health outcomes for adults born with OA/TOF and VACTERL

*“OA/TOF is a birth defect with lifelong health consequences. Patients may continue to have swallowing and reflux problems in adulthood.*

*A smooth transition from paediatric to adult services is crucial, so that patients can see a consultant with a specialist OA/TOF interest.”*

Mr Nick Maynard, Senior Consultant Upper GI Surgeon,  
Oxford University Hospitals NHS Trust



lifelong support  
for those born  
unable to swallow

## Summary of content

The content for this guide has been written by Dr Caroline Love, a member of the pastoral support sub-committee in TOFS, and reviewed by members of a working group for adults born with OA/TOF, all of whom are part of the TOFS charity. It is an abridged version of a management handbook for primary care developed from the most recent research in this area, and available at [www.tofs.org.uk](http://www.tofs.org.uk) (from Spring '22). It was funded through numerous kind donations from members of the TOFS charity.

OA/TOF are rare, complex congenital conditions, and this pamphlet has been written to help health professionals understand, consider and signpost treatment to the appropriate specialist when needed. A GP, and other health-care professionals, may only see a patient with OA/TOF a handful of times in their career. Many adults born with OA/TOF still have ongoing sequelae from their childhood anomalies.

OA/TOF were once considered paediatric conditions, but as patients have grown into middle-aged adults, research consistently demonstrates that a repaired oesophagus and trachea do not function like an oesophagus and trachea that developed in normal continuity in utero.

Oesophageal nerve and muscle may not function normally, and dysphagia, gastro-oesophageal reflux disease (GORD) and laryngopharyngeal reflux (LPR) are common as a result, though not universal. Similarly, alongside the TOF formation, the C shaped cartilage may not form properly, causing tracheomalacia (TM), which can lead to ongoing respiratory problems. Many patients with these issues may not recognise these as ongoing symptoms of OA/TOF as they have never had a normal oesophagus and may have been led to believe it had been surgically "fixed".

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## **OA/TOF causes both airway and gastro-oesophageal ill-health.**

The combination of anatomical differences due to OA/TOF and reflux into the airways, oesophagus and oropharynx can cause a vicious circle of both respiratory and gastric symptoms. Steroids and antibiotics to ease breathing may have worsened the patient's gastro-oesophageal reflux. This may result in aspiration into the lungs, requiring further respiratory support and interventions. The cycle continues with the patient becoming increasingly fatigued and with little improvement to their respiratory function.

**The risk of Barrett's oesophagus in this group of patients is greater than in the general population from childhood onwards.**

## Oesophageal/GI symptoms

### Symptoms

- Heartburn
- Worsening of existing reflux symptoms
- Worsening swallowing difficulties/food sticking
- Regurgitation
- Choking when eating
- Chest pain
- Faintness, diarrhoea and nausea post eating
- Respiratory and ENT symptoms
- Weight loss
- Foam/bubbles in mouth

### Possible causes

- Reflux
- Dysmotility
- Oesophageal stricture
- Achalasia-like symptoms or high pressure lower oesophageal sphincter
- Eosinophilic oesophagitis
- Barrett's oesophagus
- Dumping syndrome

### Consider these investigations and treatments

- Full dose PPIs for four weeks, stepped down to the lowest possible dose to control symptoms (NICE guidelines for GORD)
- Consider adding H<sub>2</sub> antagonist and increasing the current dose of PPI for short-term relief until reflux has abated or until the patient has been reviewed by a gastroenterologist
- Oesophagogastroduodenoscopy
- Barium swallow
- Oesophageal manometry
- pH studies
- Refer to gastroenterology/GI surgeon with experience in OA
- OGD screen for Barrett's oesophagus every five to ten years.

# Respiratory

## Symptoms

- Chronic cough/TOF cough
- Coughing associated with eating
- Difficulty clearing secretions
- Recurrent chest infections
- Slow recovery from chest infections
- Wheeze that is aggravated by or doesn't respond to bronchodilators
- Sleep difficulties (apnoea, nocturnal cough, nocturnal choking)

## Possible causes

- Tracheomalacia
- Dysphagia and aspiration due to airway reflux
- Bronchiectasis — the British Thoracic Society guidelines on bronchiectasis include OA/TOF as a causative condition
- Undiagnosed airway defects eg laryngeal cleft
- Recurrent fistula

## Consider these investigations

- Chest X-ray
- Bronchoscopy
- Pulmonary function tests
- Oesophageal manometry
- Consider referral to respiratory physician with OA/TOF experience

## Suggested treatment

- Patients may benefit from enteric coated steroids and antibiotics, inhaled medication via an aero chamber
- Improve secretion clearance - respiratory physiotherapy, mucolytics
- Treat GORD - PPIs, pro-motility drugs, review existing medication
- Treat airway inflammation - inhaled steroids or oral steroids help some
- Oral antibiotics for infection, may need at least a two-week course
- **Influenza and pneumococcal vaccination recommended**
- Consider prophylactic antibiotics
- Consider SaLT and/or dietitian referral

# Nutrition

## Assess

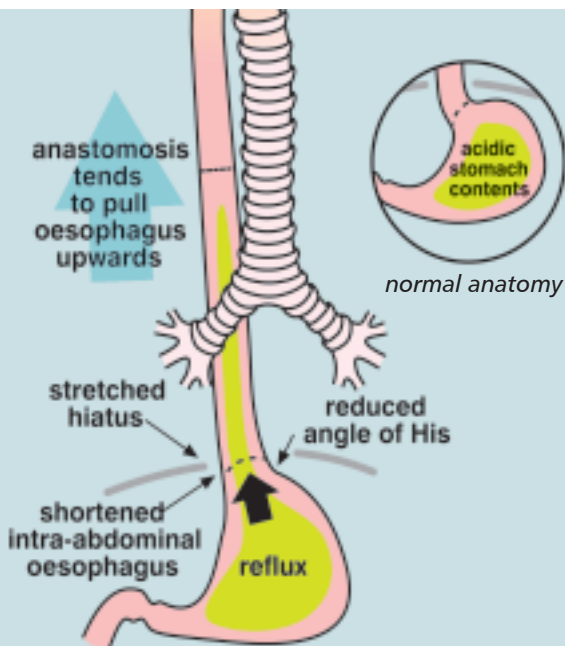
- Weight and BMI
- Calorie intake — is this optimum?
- Vitamin D
- Ferritin, haemoglobin, iron studies

## Possible nutritional issues

- Malnutrition especially in long-gap OA
- Vitamin D deficiency
- Iron deficiency
- Other micronutrient deficiency, related to long-term use of PPIs

## Consider

- Dietitian referral
- Gastroenterology referral with special interest in OA/TOF
- Correction of nutritional deficiencies
- DEXA scan to monitor bone density
- Iron infusion is often needed to correct iron deficiency as acid suppression inhibits oral absorption



# Dumping syndrome

This occurs when undigested food passes too quickly from the stomach into the small intestine. A proportion of those born with OATOF may experience this abnormal gastric function because of surgery to the oesophagus and/or stomach.

## Symptoms

**Early onset** —  
presenting shortly after  
a meal:

- Nausea
- Vomiting
- Palpitations
- Sweating
- Bloating
- Cramping
- Diarrhoea
- Dizziness
- Fatigue

**Late onset** —  
occurring one to three  
hours after a meal:

- Hypoglycaemia  
(low blood sugar)
- Weakness
- Confusion
- Hunger
- Sweating
- Dizziness

## Consider these investigations

- Symptom based questionnaire
- Oral glucose tolerance test — gold standard investigation
- Gastric emptying study

## Consider this management

- Dietary modification and dietitian referral is first line
- Referral to a gastroenterologist
- Acarbose — this is more useful in late dumping
- Somatostatin analogues

## Complications

- Weight loss
- Anaemia
- Malnutrition

# ENT problems

## Symptoms

- Hoarseness
- Voice change
- Cough
- Throat clearing
- Postnasal drip
- Facial fullness/pain
- Loss of smell
- Bad breath

## Possible causes

- Laryngeal cleft
- Vocal cord paralysis
- Laryngopharyngeal reflux
- Chronic sinusitis
- Dental loss

## Consider this management

- Treat reflux with PPI, lifestyle measures, Gaviscon Advance® (sodium alginate and potassium bicarbonate in suspension)
- Swallow assessment — consider referral to speech and language therapist
- Consider laryngoscopy if not settling

## Surgical and anaesthetic risks

**Patients with OA/TOF are high-risk surgical patients due to:**

1. Vocal cord paralysis post-surgery
2. Unsafe swallow
3. Severe GORD and dysmotility
4. Tracheobronchomalacia
5. Adhesions post-surgery
6. Diagnosed or undiagnosed spinal anomalies due to VACTERL



**VACTERL** — is a group of birth defects that occur together non-randomly, and affect multiple parts of the body. It occurs in between 1:10,000 - 40,000 births. Over 60% of those born with VACTERL have OA and/or TOF, and they form the E (the American spelling of Esophageal) and T of the acronym\*.

**Vertebral anomalies** — think osteoarthritis, undiagnosed anomalies (particularly scoliosis), chronic pain.

**Anogenital anomalies/cloaca** — ask about chronic constipation, incontinence (urine and faeces), sexual function. One in Five Thousand Foundation is a worldwide resource for the Imperforate Anus/Anoctal malformation community: [www.onein5000foundation.org](http://www.onein5000foundation.org).

**Cardiac defects** — Is there cardiology follow-up to prevent presenting as an emergency arrhythmia? Consider long-term lung, kidney, peripheral vascular complications. The British Heart Foundation provides support for those with heart conditions, and has a support group for young adults with heart diagnoses and an information section for adults with CHD: [www.bhf.org.uk](http://www.bhf.org.uk).

**Tracheal** — See rest of pamphlet.

**Esophageal** — See rest of pamphlet.

**Renal** — Recurrent UTIs and kidney stones can be signs of undiagnosed renal anomalies; known renal anomalies can deteriorate over time. Kidney Care UK is a useful resource and has a Facebook group for young adults with kidney disease: [www.kidneycareuk.org](http://www.kidneycareuk.org).

**Limb anomalies** — Think chronic pain and osteoarthritis. REACH is a useful support for those with upper limb anomalies: [www.reach.org.uk](http://www.reach.org.uk).

\*NORD - National Organisation for Rare Diseases is: <https://rarediseases.org/for-patients-and-families/information-resources/rare-disease-information>.

## **Important to note!**

# **Mental health in adults born with OA/TOF**

Whilst many will cope well with the ongoing physical health problems and complications related to OA/TOF, for some, these difficulties can affect quality of life and psychological wellbeing. Some adults born with OA/TOF report increased levels of anxiety and depression, along with trauma responses to medical interventions and hospital admissions.

## **OA/TOF related issues impacting wellbeing**

### ***in adulthood***

- Difficulties accessing healthcare professionals experienced in OA/TOF.
- Previous negative experiences due to lack of understanding of OA/TOF.
- Negative body image due to surgical scars and impact on relationships.
- Auditory manifestations of OA/TOF, such as the 'TOF cough', choking episodes and throat clearing can contribute to the development of anxiety in social situations.
- Difficulties with eating, including anxiety about eating in public.
- Difficulties in communication, due to vocal cord damage or respiratory disease.

### ***in childhood***

- Having a child with complex health problems can impact on parental mental health, wellbeing and attachment within the family.
- Multiple admissions, traumatic experiences and painful procedures in medical settings may contribute to the development of anxiety about health care professionals and hospitals.
- Bullying in childhood due to the condition.

## Top tips from OA/TOF patients

- It is important to be aware of the possibility of past negative experiences with health care professionals and be mindful of the impact this may have on the doctor patient interactions.
- It can be helpful to acknowledge OA/TOF as a rare disease and to reassure the patient by saying, “I don’t know about OA/TOF but I’m prepared to listen and work with you to help”.

## References:

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Hill A, Sullivan A, Chalmers J, De Soyza A, Elborn S, Floto A, et al. British Thoracic Society Guideline for bronchiectasis in adults. *Thorax.* 2019 Jan;74(Suppl 1):1–69.

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## Additional resources

The position paper published in October 2021 entitled: "Towards a Holistic Model for the treatment of those born with Oesophageal Atresia (OA) from diagnosis to transition and adult care" is written from a patient perspective, reflecting on patients as "experts by experience".

This document represents the fundamental ethos of the TOFS charity: to promote and support lifelong care for those born with OA/TOF. The charity endorses the author's proposal of the provision of centralised services to enable children and adults to achieve the highest standard of care. Read the paper in full here: <https://tofs.org.uk/news/2021/10/tofs-issues-recommendations-to-the-nhs-for-oa-treatment.aspx>

A full reference guide for health professionals treating adults born with OA/TOF will be available at [www.tofs.org.uk](http://www.tofs.org.uk) from Spring 2022.

Useful video resources available here:  
<https://tofs.org.uk/videos.aspx>

Adult TOF Facebook group, endorsed by TOFS, visit:  
<https://www.facebook.com/groups/79491291650/>

**International Dysphagia Diet Standardisation Initiative** – A global initiative to improve the lives of people living with dysphagia. Further resources available via the website/app:  
<https://iddsi.org/> OR **IDDSI app on the App Store/Google Play.**

For additional copies of this booklet, or for more information about TOFS, please contact:

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