About TOFS

TOFS offers patient-led support for adult TOFs and parents of TOF children. We provide mainly information and emotional support, via conferences, meetings, internet forums, newsletters, etc. TOFS does not offer medical support or advice, but we seek to share our membership's huge practical knowledge of living with the TOF condition.

The charity serves the TOF community throughout the UK and Ireland. It operates from a small office in Nottingham. It is linked with Europe's other OA TOF support groups.

We welcome interested healthcare professionals as members. TOFS will happily pass on healthcare professionals' enquiries to Doctors who are particularly knowledgeable about the TOF/OA condition for adults.

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

TOFS
St George's Centre,
91 Victoria Road, Netherfield,
Nottingham NG4 2NN
Tel:+44 (0)115 961 3092
Fax:+44 (0)115 961 3097
Email: info@tofs.org.uk Website: www.tofs.org.uk

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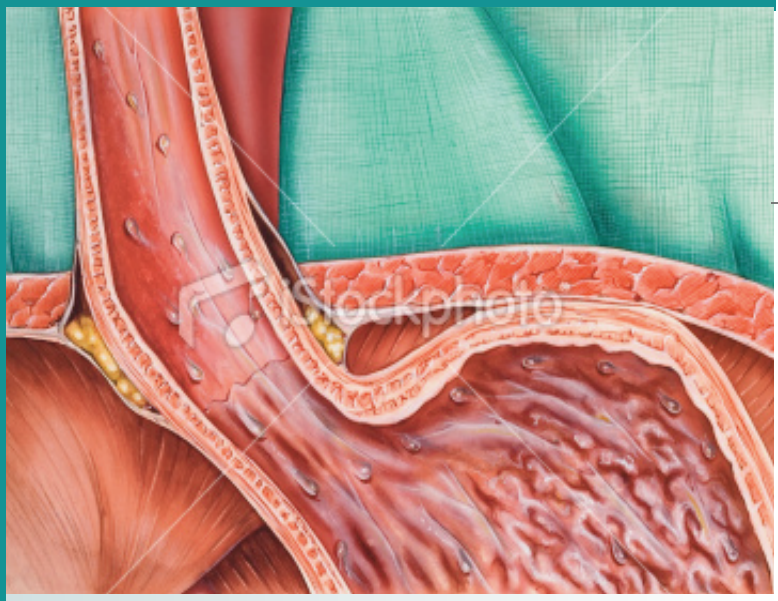
tofs
support for families
of children born unable to swallow

TOF/OA Adults – Health Issues

An introduction for generalist health professionals to the various chronic health problems which can afflict adults who were born with TOF/OA.



tofs
support for families
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Foreword

Most adults who were born with TOF/OA (adult TOFs) are able to follow an essentially or even wholly normal lifestyle.

However, a significant minority of adult TOFs experience at least one of the issues described in this leaflet. Fortunately, few adult TOFs suffer from multiple serious complications of the TOF /OA condition.

This leaflet provides information on some of the common health issues affecting adult TOF patients, their causes and initial treatments.

TOFS is grateful to members of the medical profession for their input to this leaflet.

The most well-known chromosomal defect associated with Oesophageal Atresia is **Down's Syndrome (Trisomy 21,** where there are three copies of chromosome 21 instead of the normal two) but also **Trisomy 13** and **Trisomy 18.** Where one of a couple has isolated TOF/OA, with no other anomalies, the risk of a child being born with TOF/OA appears to be low. Indeed, there appears to be little reason to expect the risk to be materially higher than the "normal population" 1 in 3500 rate.

Referral to a clinical Geneticist should enable a specific case to be thoroughly investigated, and appropriate counselling to be organised. Such referral could be particularly important in the complex cases, for instance a potential parent having TOF/OA and other abnormalities.

Long-term Outcomes

As noted elsewhere in this leaflet, most of those born with the TOF condition can look forward to an essentially normal adulthood.

So far, TOFS is not aware of any data which suggest that TOFs have a life expectancy different from that of the population as a whole. However the first successful TOF surgery was undertaken in 1947 in the UK and neonatal survival rates were low in the first few years. Consequently there are very few adult TOFs aged more than 50 years and little or no meaningful data about longevity.

However it would not be surprising if data can eventually be assembled indicating that TOF adults have a slightly reduced life expectancy. As noted above, there is some evidence that TOF patients are at elevated risk of Barrett's oesophagus and potentially oesophageal carcinoma. Regular endoscopy is sometimes suggested in view of this. Moreover many TOF patients have permanently damaged lungs, which may impact their life expectancy.

Genetics of TOF/OA

Adults who were born with TOF/OA are often understandably concerned that any children they might have could be born with TOF/OA.

Considerable research has been undertaken in recent years to try to identify a "cause" of TOF/OA. A number of environmental factors have been suggested (Vitamin A, alcohol abuse), though there appears to be little or no strong evidence of any such factor leading to TOF/OA. Where TOF/OA occurs in twins, most normally only one of the twins is affected, supporting the idea that there is often no "cause".

One specific research project has been examining the Genetics of Oesophageal Atresia. The current understanding is that TOF/OA is only rarely associated with a chromosomal defect or a specific genetic defect.

However there are some rare and complex syndromes which involve Oesophageal Atresia for which genetic defects have been identified. These include:

- **Feingold syndrome:** microcephaly (small head), digital anomalies (commonly affecting the 2nd and 5th fingers or the toes) and atresias of the gastro-intestinal tract, associated with defects in the gene N-MYC.
- **CHARGE syndrome:** Coloboma (a specific eye defect), Heart problems, Atresia choanae (a narrowing of the airways at the back of the nose), Retarded growth, Genital hypoplasia (defects in the sex organs) and Ear abnormalities, associated with defects in the gene CHD7.
- **Anopthalmia-Esophageal-Genital syndrome (AEG):** a rare condition involving the combination of Anophthalmia (no or very small eye(s)), Esophageal and Genital problems, associated with defects in the gene SOX2.

Introduction

TOF (when there is a conduit between the oesophagus and trachea) and OA (the oesophagus ending in a blind pouch) are congenital conditions that occur in about 1 in 3500 births. Close to 200 TOF/OA babies are born every year in the UK.

A typical GP might see a patient with the condition just a handful of times in a career, and in view of this the TOFS support group (working with acknowledged medical experts) has assembled this quasi-medical introduction to the condition, with especial relevance to adult TOFs.

As with people who suffer from other complex conditions, adults with TOF/OA sometimes need the input of several specialist medical disciplines. Gastro-intestinal and respiratory specialties are top of the list for TOF patients.

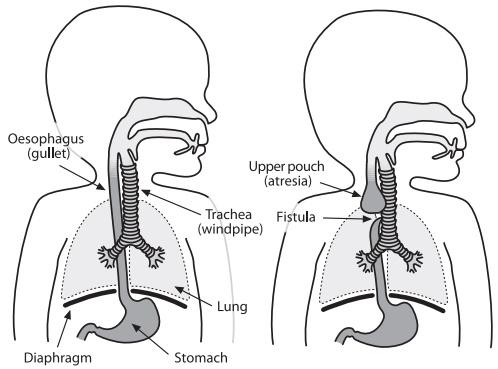
NOT just a paediatric issue

TOF and OA have until recently often been considered to be specifically paediatric conditions which are treated soon after birth. Moreover, it has commonly been thought that the problems which often arise following the surgical correction (such as Gastro-Oesophageal Reflux, oesophageal strictures and recurring chest infections) resolve during childhood.

Most babies born with TOF/OA do indeed grow up to lead essentially normal adult lives. However, as the population of adults born with OA and TOF has grown, (now estimated to be over 5000, very few of whom are yet aged over 55) it has become clear that many continue to have a number of medical and associated emotional issues as a result of the condition and their early surgery.

Childhood surgery and complications

TOF patients have normally had surgical repair within the first few days of life. Some 85% of TOF patients are born with a tracheo-oesophageal fistula and oesophageal atresia, with a lower pouch fistula.



Child without TOF/OA **Child with TOF/OA**

The aim of the surgery is to anastomose the ends of the oesophagus, and divide the fistula. Such surgery is generally done at one of approximately 30 specialist UK centres. Often it is now undertaken laparoscopically. When the gap between the ends of the oesophagus is too long for easy repair, more involved surgical techniques such as colonic interposition, gastric tube oesophagoplasty and gastric transposition are used.

TOF children often need further surgery as they grow. Oesophageal strictures at the site of the oesophageal join are probably the most common issue: dilatations are the usual treatment. Some patients can need repeated dilatations through their development.

Many TOF patients have had a stomach fundoplication during childhood, generally in order to control reflux. A fundoplication is often very effective against reflux but can lead to increased difficulties in swallowing food, and reduced

that have detrimental effects on psychological well-being and the ability to cope. Awareness on the part of the GP that this can be the case will help in the process of recommending hospitalisation or surgery; patients may require more than usual re-assurance that the treatment is necessary and that they will be properly cared for. However some patients become so accustomed to hospitalisation that they have no hesitation in accepting the recommended course of action. To repeat: the watchword is awareness.

2. Impact of chronic disease on mental health

Any long term illness increases the risk of depression. Chronic illness may limit a person's lifestyle and mobility compared with their peers, undermine self confidence and restrict hope for the future. Childhood health problems may have limited school attendance and achievement, and in turn employability and ability to work full or part time. The physical manifestations of the condition may also add to this. Embarrassment about the physical signs of the disease (the loud cough, bringing up mucus etc) may knock self confidence and cause reluctance to mingle with others.

3. Taking responsibility for one's own health

Taking responsibility for oneself as an adult in a medical context can be a quite daunting experience. The patient can sometimes be thrust into a one-on-one relationship with GP or other medical consultant where the previous experiences have all been with the support of one or more parent, who more often than not has acted as the primary interface with medical professionals on behalf of their child. The patient will suddenly be exposed to the realities of having to understand what is being said and having to frame the appropriate questions to their GP or other medical professional. This will test their own awareness of their condition, past treatment and experiences. It may soon become apparent to them that they still have much to learn about themselves in addition to the whole process of dealing with the Health Service, e.g. making appointments.

Symptoms
Coughing and spluttering on eating or drinking, symptoms of 'chest infections' which follow (actually aspiration episodes)

Action
Swallowing studies, seek advice from Speech and Language therapists

2. Damage to larynx, sinuses, teeth.

Causes
Reflux onto the larynx and into the upper airways can lead to symptoms outside of the GI and respiratory tract, and are known as extraoesophageal complications of GORD.

Symptoms
- Laryngeal: hoarse voice, chronic laryngitis, globus, recurrent throat clearing. More rarely, the risk of laryngeal stenosis and carcinoma is also raised.
- Sinuses: Chronic rhinosinovitis may be triggered by reflux into these areas.
- Teeth: dental erosions may result.

Treatment
This is as for GORD.

Psychological impact of TOF/OA

1. Impact of regular hospitalizations in childhood and adulthood

TOF patients are not unique, but like many people who are or have been frequently hospitalised, they can suffer ongoing anxiety about the prospect of re-hospitalisation and/or further surgical intervention.

Research has been conducted into the psychological effects of hospitalisation. The results have shown that hospitalisation may result in negative feelings

stomach volume, meaning that such patients can prefer to eat little and often.

Some TOF infants also will have undergone an Aortopexy. This procedure is used for instance in cases where the trachea tends to collapse as the patient attempts to breathe in.

Vacterl Association

TOF/OA is most commonly an isolated anomaly with in utero environmental rather than genetic triggers. It does, however, form part of a number of genetic syndromes, most commonly VACTERL. This is a constellation of anomalies:

V: Vertebral anomalies
A: Anal anomalies e.g. imperforate anus
C: Cardiac anomalies
T: Tracheal and
E: Esophageal anomalies
R: Renal anomalies
L: Limb malformations

The diagnosis requires 3 or more of the above anomalies. Although VACTERL is normally identified soon after the baby's birth (or even in utero) some older adults with TOF /OA were born before the association was recognised in 1968 and have since discovered that they qualify for the diagnosis. Cardiac and renal problems that were not detected at birth, as they were asymptomatic, have sometimes become apparent later on in the person's life. This is something to be considered if an adult with a history of TOF/OA develops symptoms that could be confused with a further deterioration of their respiratory status (deteriorating heart condition), or chronic lower back pain (for renal problems).

TOF/OA may also present in Down's Syndrome, Feingold syndrome, Edward's syndrome (trisomy 18) and CHARGE syndrome.

Ongoing Health Problems

Despite the repair of TOF/OA in infancy, remaining anatomical abnormalities, GORD, scarring and side effects from the original and subsequent surgeries may mean health problems can be life-long. Below we have detailed some of the common health issues affecting patients, their causes and initial treatments.

Bolus obstruction of the oesophagus

Food can get stuck in the oesophagus. This problem is so common among TOFs that it is often known as a "stickie". It can happen for several reasons:

1. Poor oesophageal motility
2. Combination of poorly chewed food and a slightly narrowed oesophagus from surgical scarring
3. Stricture formation

Signs and symptoms

- Sensation of something being 'stuck'
- Inability to swallow fluids/ saliva
- Drooling due to above
- Wheeze/ stridor due to obstruction

Treatment

- Food may eventually dissolve with time, or fizzy drinks may help
- Removal of the lesion in hospital may be necessary
- If this is an increasing problem, the patient needs investigation for a possible stricture

- Aspiration pneumonia: The symptoms can appear suddenly and include shortness of breath, acute pain in the chest during inhalation or coughing and alternate shivering and sweating. The person may also feel very nauseous. This may resolve on its own, but may be severe enough to need hospital admission. Long term, this may lead to long term lung damage. Although aspiration pneumonia is generally associated with the inhalation of foreign objects or acidic liquid from the stomach, the gas that is released from the stomach during a bout of reflux is also acidic. When a person inhales this gaseous type of reflux, it can also irritate the lung membranes. If the aspiration has reached the lungs, abnormal sounds may be heard on auscultation. However, if the trachea is the main area affected, the lungs may sound clear, though stridor may be audible.
- Secondary infection: Any respiratory infection in TOF patients may be more severe whether viral or bacterial, due to the reactive airways caused by GORD. However, episodes of aspiration may be complicated by bacterial super-infections.
- Mucus collection : Rib stiffness, secondary to past thoracotomy, or other airway damage may reduce/ limit secretions being cleared, increasing the risk of infection. This can be countered in some patients by regular saline nebulisations, to keep the lungs moist and help counter the build-up of mucus.
- COPD/ Bronchiectasis: Uncontrolled reflux, recurrent aspiration and infections can all result long term in permanent lung damage.

ENT problems

1. Unsafe swallow - as well as the developmental anomaly that is TOF, the rest of the oropharynx may be abnormal, either in terms of the muscle reaction to swallowing or the nerves which control it. This can result in abnormalities in the oral or pharyngeal swallow in addition to the main abnormality in the oesophageal swallow.

sufferers to modify eating patterns to minimise symptoms. Small frequent meals, avoidance of simple sugars, milk and milk protein may be advised. Restriction of carbohydrates, with increase in proteins and fats may also help.

2. Medical - Acarbose interferes with carbohydrate absorption and can help patients with late dumping. Octreotide may also be of some use short term.

Respiratory problems in TOF adults

Causes

Reflux into the respiratory tract will result in a number of problems. Whilst acid suppression medication may make the reflux pH neutral, the reflux still persists and is still irritant to the airway linings. This may be either low grade and chronic microaspiration, causing airway irritability, chronic cough and increased mucus production, or larger amounts might be aspirated (typically at night or whilst bending over) leading to aspiration pneumonia and secondary bacterial infection.

In addition, like the oesophagus, the trachea may also have anatomical abnormalities. This may be from failure to develop (failure of C-shaped cartilage to form) resulting in tracheomalacia, which may be lifelong, or scarring or abnormal pouch or duct formation post surgery, causing tracheal stenosis.

Symptoms

- Hoarse brassy 'TOF' cough: the floppy trachea and scarring of the trachea results in an unusual seal like cough. This cough can be persistent and tiring, resulting in pulled rib muscles, airway bleeding, facial petechiae, interrupted sleep and, more long term, stress incontinence.
- Persistent sore throats and a hoarse voice: reflux onto the larynx and into the throat irritates and damages these areas. On examination, the back of the throat may look scarred and telangiectatic.

Gastro-oesophageal Reflux disease in TOFs

Causes

This condition is almost ubiquitous in TOFs due to abnormal development and motility of the oesophagus and the effects of surgery on it. In addition, the gastric sphincter is often ineffective as the vagus nerve may not have developed or been cut during surgery. This results in chronic and often severe lifelong reflux.

Symptoms

- Routine GORD symptoms, such as burning sensation in the oesophagus during and after eating, nausea and regurgitation, persistent sore throats, a metallic taste in the mouth, bad breath and a hoarse or croaky voice.
- Difficulty swallowing food.
- Choking and coughing during eating.
- Barratt's oesophagus- TOFs are at a higher risk of developing this.
- Oesophageal carcinoma- TOFs also have a higher risk of this.

Treatment

1. Lifestyle measures: avoiding very acidic and spicy foods, caffeine and chocolate generally and particularly before bedtime. Giving up smoking will often help, as will sleeping in a bed which has the head end higher and losing weight (if necessary).

2. Medication: Proton pump inhibitors (lansoprazole, omeprazole etc) and histamine receptor blockers (ranitidine, cimetidine) are first line medications, though may be needed at higher doses or given more frequently, e.g. with each meal, to control symptoms adequately. Gastroprokinetics, like metoclopromide and domperidone, increase gastric emptying, thus reducing the amount of stomach contents available to reflux. Azithromycin has also been shown to improve oesophageal motility. In addition, products like Gaviscon and other gastric coating agents may reduce symptoms, but are unlikely to be sufficient on their own.

3. Surgical: A Nissen's fundoplication may be considered to strengthen the gastric sphincter. However, many patients will have had this in childhood already. It is also inappropriate in patients with oesophageal dysmotility as this will exacerbate, rather than help their problems, slowing food entering the stomach. Reflux may persist, however, post fundoplication, and the patient will be left unable to vomit post operation.

NICE guidelines suggest that patients under age 55 without 'alarm symptoms' can be treated for dyspepsia without gastroscopic investigation. This seems inadequate for adult TOFs: a number of long-term outcome studies have shown that Barrett's oesophagus is far more prevalent amongst adult TOFs than the normal population. The literature is moving toward a consensus that there is a need for guidelines for long-term endoscopic surveillance of adult TOFs. One paper suggests regular endoscopies every 5 years.

Dumping syndrome

Causes

Dumping syndrome is usually associated with people who have had surgery to bypass the stomach or to restrict the size of the stomach. The type of procedures that people with OA have undergone to repair the atresia and consequent reflux can also lead to the rapid gastric emptying that characterises dumping syndrome. Rapid gastric emptying is when food passes through the stomach and into the small bowel so fast that it is not properly digested. The partially undigested food is rapidly absorbed by the duodenum which leads to a large volume of fluid being released into the bowel and the bowel contractions strengthen.

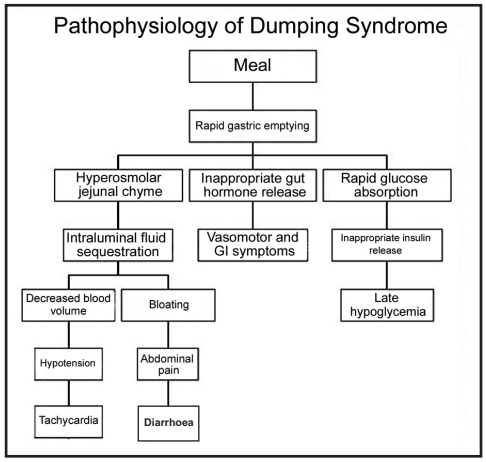
Symptoms

1. Early - may start during or shortly after a meal

- Sudden (and at times uncontrollable) diarrhoea, nausea, stomach cramps, a fast heart rate, feeling faint or dizzy, sweaty and lethargic. Before any of these become apparent the person may be surprised to feel their mouth drying so fast that it seems that saliva is being sucked back into the body.

2. Late - occurs 2 to 3 hours after a meal

- Sudden loss of energy, sleepiness, heavy perspiration, a rapid heart rate, shakiness, an urgent need to eat, visual disturbances, slurred speech and mental confusion. Many of these symptoms relate to the hypoglycaemic state that may occur in late dumping, as is characterised in the flow chart below.



Author: Alan BR Thomson, MD; Chief Editor: Julian Katz, MD.
http://emedicine.medscape.com/article/173594-overview#a0104

Complications

The symptoms of the early form can lead to severe weight loss, anaemia and malnutrition. The embarrassment and discomfort caused by the urgent diarrhoea and cramps can lead some people to avoid eating to prevent the symptoms and they may also restrict physical and social activities so they can remain close to a toilet. Symptoms relating to the late form of dumping can be very frightening for the sufferer and may also be dangerous if the blood sugar level falls very low.

Treatment

1. Lifestyle - referral to a dietician may allow