

Information for GPs about TOF, OA and VACTERL

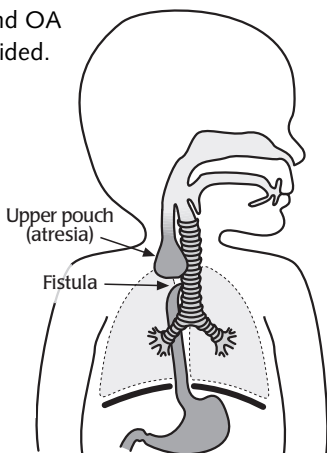
This leaflet contains information about tracheo-oesophageal fistula (TOF) and oesophageal atresia (OA) for general practitioners and other members of the primary healthcare team.

BELOW: the most common combination of TOF and OA

Basic information about VACTERL association, of which TOF and OA may be features, is also provided.

Although these conditions are not particularly rare, it is likely that most GPs will only see one or two TOF children in a lifetime of practice.

We hope this booklet will help to make the experience a successful one for you.



A leaflet provided courtesy of TOFS (Tracheo-Oesophageal Fistula Support) – helping children born unable to swallow, with thanks to Quilter and Co. Ltd. (www.quilter.co.uk) for help with printing costs



TOFS

How to use this booklet

The page opposite offers the reader a very brief overview of tracheo-oesophageal fistula and oesophageal fistula. Once you've read that, either take the rest page by page or select from the table of contents on the back cover.

Text on a coloured background was written by volunteers with experience of TOF children through their work with the support group TOFS, while that printed on white was prepared in collaboration with medical professionals working in the larger surgical centres treating TOF children.

To inspire you to read on, take a quick look at this review of the content by Penny Seymour, neonatal surgical outreach sister at the Leeds General Infirmary ...

“This booklet is excellent and will, I am sure, prove a particularly useful addition to the collection. A copy should ideally be sent to the GP, health visitor and community nurse of every baby discharged home following a TOF/OA repair.”

Introduction

Tracheo-oesophageal fistula (TOF) with oesophageal atresia (OA) is seen in about 1 in 3,500 births. The term ‘TOF’ has become widely used although OA/TOF is more accurate.

Surgery is carried out in the first days of life by a paediatric surgeon. Mortalities are now mostly attributable to any associated cardiac anomalies or prematurity, but morbidity and quality of life are important to address. Problems commonly seen in clinical follow-up include gastro-oesophageal reflux, oesophageal stricture, feeding difficulties and respiratory symptoms. These problems will mostly resolve during the first years of life – however, long term problems are currently poorly documented and life long oesophageal follow-up by endoscopy is wise.

The impact on the family should not be underestimated. Parents often find infancy a stressful period; feeding can be slow and tiring, a barking cough often draws attention to the child in public and mothers often blame themselves and worry about recurrence risks.

Parents may not appreciate that their GP or health visitor may never have seen a ‘TOF child’ before, and can find the lack of local knowledge frustrating. Some have found themselves being asked to justify medications the hospital have prescribed, or having to point out that the problem they want help with is the child’s swollen knee and not the gastrostomy tube which has inadvertently caught the doctor’s attention!

Text prepared in collaboration with Anthony Lander, consultant paediatric surgeon, Birmingham Children’s Hospital.

Diagnosis in the neonate

Very rarely a GP might see a baby with OA after a home birth or following hospital discharge after only a very short stay. The neonate with OA is typically a ‘mucousy’ baby who is unable to swallow saliva and who may have a degree of respiratory difficulty, especially on attempting to feed. Milk or saliva collects in the upper pouch and is vomited, with the risk of inhalation. Sometimes there is a blueness of the fingertips and a distended abdomen – if air travels through the TOF to reach the stomach.

85% of cases have OA with a lower pouch fistula (*see cover picture*). In other cases the fistula may be attached to the upper pouch, there may be no fistula, or indeed there may be two fistulae.

Diagnosis is confirmed by passing a stiff tube (which will not coil up) down the oesophagus; with OA, the tube will be held up in the upper pouch. Radiography and bronchoscopy can further refine the diagnosis.

The exception is the isolated TOF, which can be a cause of feeding difficulties and/or recurrent respiratory problems of varying severity. In some cases, this has continued over a period of years before a diagnosis has been made.

Antenatal diagnosis

Oesophageal atresia may be suspected where there is polyhydramnios or an absent foetal stomach image. Parents should be offered as much information as they want, knowing the diagnosis is only suspected and will need either to be confirmed or ruled out after birth.

Text prepared in collaboration with Anthony Lander, consultant paediatric surgeon, Birmingham Children’s Hospital.

Surgical repair

The aim of surgery is to divide the fistula and to anastomose (join) the ends of the oesophagus. This is possible in the vast majority of patients. Survival is expected in almost 90%, with most of the deaths due to associated major heart defects or to extreme prematurity.

Where there is a significant gap between the two ends of the oesophagus it may be possible to perform the anastomosis under tension, after which mechanical ventilation may be necessary for around five days so that healing can occur.

In other circumstances, the timing of the anastomosis may have to be delayed (up to twelve weeks or even longer), during which the infant is fed by a gastrostomy while the distance between the ends of the oesophagus decreases.

In a small minority of patients it is impossible to retain the oesophagus because the distance between the two ends is too large or due to complications following anastomosis; an oesophageal substitution is then required. There are three methods – colonic interposition, gastric tube oesophagoplasty and gastric transposition (currently the favoured method due to the lower complication rate). In the time period before this surgery (around 6–9 months of age) it will almost certainly be necessary to perform a cervical oesophagostomy in addition to the gastrostomy. The oesophagostomy serves to drain saliva (which would otherwise enter the air passages causing pneumonia) and allows the introduction of ‘sham feeding’ so that the child experiences the action of swallowing while being fed by gastrostomy.

Text prepared in collaboration with Prof Lewis Spitz, Institute of Child Health and Great Ormond Street Hospital for Sick Children, London.

Parental counselling

Parents' initial reactions to the birth of a baby who is found to have problems will inevitably differ. Although it is likely that the local practice may not be directly involved with the family until some weeks after surgery, the following paragraph may be useful to surgery staff ...

Reports from parents whose local practice told them (however well-meaningly) that 'the worst was over' once the initial surgery had been carried out, only to later find that their baby suffered complications and may even have required further surgery, indicate that their resentment of such false reassurances tends to remain in their minds for some time. Whilst compassion is welcome, an 'honest confession of ignorance' – perhaps tactfully combined with an endorsement of the care their baby is receiving – may be preferable to a 'guesstimate' about future outcome. Previous experiences of TOF children may be useful, but should not provide the basis for a prediction about other individuals.

Discharge from hospital

Bringing a baby home is a worrying time for TOF parents, even though the hospital will have made every attempt to ensure that adequate preparations have been made.

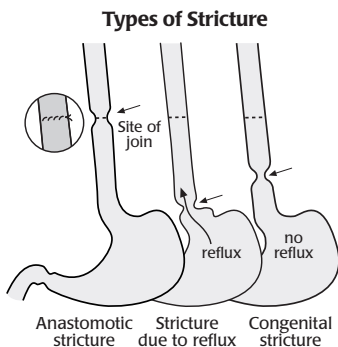
Freedom from worries about supplies of drugs or other equipment offers considerable security, and a practice which has been well-informed by the hospital and is able to offer the parents suitable support will also help.

Parents may appreciate social contact with the practice at this time – confidential reports and other correspondence between professionals 'behind the scenes' being no substitute for a friendly personal exchange.

Strictures

Oesophageal strictures are probably the most frequent reason for further surgical consultation after TOF repair, occurring in about a third of babies and particularly in the first months after surgery.

Parents will often have been alerted to the possibility of a stricture by the hospital, and should be encouraged to consult hospital staff if their child experiences recurrent or progressive difficulties feeding.



Dilatations are carried out under general anaesthetic, either by an interventional radiologist using a balloon catheter or by the surgeon with bougies. Repeated dilatations may be necessary – or occasionally, if a stricture is particularly resistant, surgical resection may be required.

A few centres have used conscious dilatations for older children who 'self-dilate' using a mercury-filled bougie tube, but this is uncommon and obviously requires consent and considerable cooperation on the part of the individual.

Text prepared in collaboration with Paul Losty, consultant paediatric surgeon at Alder Hey Children's Hospital, Liverpool.

Gastro-oesophageal reflux

Reflux is a common occurrence in newborns. For most babies this condition improves with age or requires simple measures such as feed thickeners, antacids or acid blocking medication to control vomiting and/or oesophagitis.

In patients born with OA/TOF, reflux can be a troublesome symptom which is more resistant to conventional medical management. Anatomically, the gastro-oesophageal reflux barrier is distorted and to some extent defective due to shortening of the abdominal oesophagus as a result of the surgical repair.

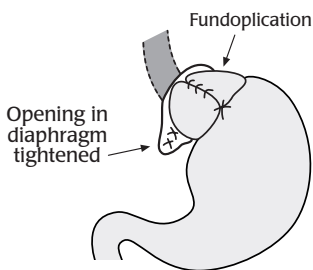
Symptoms and signs of reflux in OA/TOF patients include vomiting (although this is by no means always a feature), failure to thrive, feeding difficulties such as choking (as a result of peptic oesophageal strictures) or pain and discomfort during feeds, recurrent wheezing or chest infections. On occasions aspiration may provoke life-threatening apnoea spells.

Investigations and treatment are best achieved by prompt referral to the paediatric surgical centre which cared for the child.

About half will require surgery in the form of fundoplication to control refractory symptoms, the most popular operation being the Nissen procedure (*see diagram*).

Surgery generally achieves good results. On occasion however the procedure may need to be 're-done' as signs of failure can evolve over time.

The fundus of the stomach is wrapped around the lower oesophagus, creating a valve to prevent reflux



Feeding the TOF child

For parents, the psychological impact of not being able to feed their child normally is enormous. Most TOF children will experience some difficulties with feeding, which will improve with age – however even TOF adults may experience occasional problems, often preferring to eat with a drink to hand.

Problems may be of a physical nature (stricture formation – *see page 7/8*), functional (oesophageal dysmotility is not uncommon) or of a psychological origin (as a result of previous experience, e.g. pain, discomfort or often simply because a child has not learnt to eat or to feel hungry following tube feeding).

Support and guidance from the hospital should therefore be forthcoming, and adequate arrangements be in place for assistance should the child experience an oesophageal obstruction which does not resolve itself within a few hours of eating. Frequently food impaction is seen in these cases in association with a stricture that requires removal by oesophagoscopy under general anaesthesia. This can often be combined with a dilatation procedure (*see page 7*) under the same anaesthetic.

Text prepared in collaboration with Paul Losty, consultant paediatric surgeon at Alder Hey Children's Hospital, Liverpool.

Membership of TOFS offers parents a useful outlet for their anxieties and frustrations. The role of self-help groups in assisting families to adjust to medical problems has been proven time and time again; frequently the support of others who have seen their own TOF child through the difficult early years can be of enormous benefit.

The TOF cough

An area of tracheomalacia (often referred to as ‘floppy trachea’) remains after a TOF repair, and this results in the so-called ‘TOF cough’. This has a characteristic ‘honking’ or ‘barking’ nature and can prove intrusive in social situations.

Respiratory infections

Reduced mucociliary action at the site of the TOF repair, combined with a degree of tracheomalacia, means that TOF children may find it hard to remove the increased mucus load during a respiratory virus infection. This increases the chances of bacterial superinfection, and routine administration of antibiotics at the start of respiratory symptoms may be beneficial.

In fact, the airways of TOF patients may actually be more sensitive to viral infection. Many TOF children show asthma-like symptoms which respond to bronchodilator therapy.

The development and/or progression of chronic lung changes needs to be avoided if at all possible, and TOF children are often put onto long term antibiotic cover to protect their lungs.

Avoidance of exacerbating conditions such as passive smoking and furry pets is wise. Some children benefit from chest physiotherapy, most usually co-ordinated by the hospital.

Tracheomalacia

For most TOF children, the influence of any tracheomalacia becomes less with age.

Some children however experience more severe symptoms. The TOF cough becomes exaggerated, and excessive wheezing or cyanosis occur during feeding. More rarely, the infant may experience acute life-threatening episodes.

Other possible manifestations of tracheomalacia result from compression of the trachea from either refluxed stomach contents or oesophageal obstructions.

Stridor and recession may be marked in certain positions e.g. flat on the back, so positioning can be important for these patients.

Severe tracheomalacia in TOF children will usually be dealt with by a paediatric surgeon, although some centres refer to an ENT specialist.

Text prepared in collaboration with Prof Rosalind Smyth, chest physician at Alder Hey Children's Hospital, Liverpool.

Child development

Many TOF children visit hospitals on a regular basis, either as an outpatient or to be hospitalised for diagnosis or treatment.

Most will not make too much fuss about this, however some TOF parents report that their child has become terrified of hospitals (especially of injections) and the family may be at a loss for what to do about it. Often the surgeon will be able to refer the child to a psychologist within the hospital, however a GP may also be able to locate a source of behavioural therapy (*note that parents sent to an analytical/family therapist for this type of problem have not found the experience useful*).

Parents often worry about their child's sense of 'differentness' – especially when around their peer group. This may be a problem however older TOFs do not usually recall being aware of their 'uniqueness' in a problematic way – children are more resilient than we think! The teenage or early adult TOF may, however, go through a period where they may desire or find it useful to 'revisit' their medical history.

Associated anomalies

Tracheo-oesophageal fistula is associated with other congenital anomalies, most commonly atresias elsewhere in the GI tract (e.g. duodenal atresia, anorectal atresia).

TOF and OA may also be part of a wider range of anomalies, for example CHARGE (Coloboma, Heart defects, Atresia of the choanae, Retarded growth, Genital hypoplasia and Ear anomalies/deafness) or VACTERL.

VACTERL

This term is an acronym for Vertebral, Anal, Cardiac, Tracheal, Esophageal, Renal and Limb anomalies. To 'qualify' as VACTERL a baby should have three or more such anomalies.

Some 40% of TOF patients have other anomalies, although these may not be immediately apparent (e.g. unilateral renal agenesis, defects of vertebral formation or segmentation).

Where a baby has multiple significant anomalies – for example OA/TOF, VSD, anorectal atresia, radial dysplasia, scoliosis due to congenital vertebral defects – the whole family is put under enormous strain. The child may need to undergo many investigations and treatments with a range of medical specialists, and may have to endure repeated hospitalisations. High levels of support are required in such situations.

VACTERL was previously known as Say-Gerald or Kaufman syndrome, PIA or PIAVA; current terminology also includes VATER/VACTERLS (S for Single umbilical artery) or ARTICLE. VACTERL is also associated with cleft lip/palate and ear anomalies/deafness.

Text prepared in collaboration with Leela Kapila, consultant paediatric surgeon, Nottingham.

Genetic counselling

Only very rarely is oesophageal atresia associated with a chromosomal defect e.g. Down's syndrome (Trisomy 21) or Edward's syndrome (Trisomy 18) or a specific genetic defect (Treacher-Collins syndrome).

There have also been reports of environmental factors (vitamin A, alcohol abuse), however for the vast majority of cases these conditions are considered as 'one-off occurrences' with no identifiable cause.

Parents are naturally concerned about the possibility of recurrence. Where the OA/TOF is isolated, with no other anomalies, the risk of recurrence is generally around 1%, and where there are multiple anomalies (e.g. VACTERL) the risk increases to around 3% – however referral to a clinical geneticist will enable a more thorough investigation of the individual case and allow access to appropriate counselling.

Research is under way in various centres to determine the cause of OA/TOF and VACTERL, so current advice may need to be revised in years to come.

Text prepared in collaboration with Prof Lewis Spitz, Institute of Child Health and Great Ormond Street Hospital for Sick Children, London.

Long term outcome

The results of long term follow-up studies are reassuring. Whilst complications are at their worst during the first two years, they become less common after five. By early adolescence, most patients with oesophageal atresia have learned to cope with the disordered motility of the oesophagus and can live a near-normal lifestyle.

Children should however be followed up until the age of 16 years to ensure adequate growth.

In recent years, concerns have been raised about the risks of oesophageal carcinoma in adult TOFs, and practitioners should be aware of the risks of long term reflux and Barrett's oesophageal changes. It should also be noted that some patients have developed gastro-oesophageal reflux (*see page 8*) in later life in spite of the fact that they did not suffer from this in early childhood.

Text prepared in collaboration with Prof Lewis Spitz, Institute of Child Health and Great Ormond Street Hospital for Sick Children, London.

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Further information

The charity TOFS offers support and information to the families and carers of babies born with tracheo-oesophageal fistula, oesophageal atresia and related conditions.

The group enables families to benefit from the friendship of other parents who have experienced the particular stresses of caring for a TOF child – as well as the joy when problems have been overcome.

Membership offers access to a range of informational leaflets – which are also freely available on our web site (www.tofs.org.uk) and our book *The TOF Child*, a quarterly newsletter *Chew* and attendance at our biennial Conference (for both parents and medical professionals).

TOFS also has a small fund to help parents experiencing financial strain; we can lend various items of equipment (e.g. food blenders and apnoea monitors) and we fund research into the cause and treatment of these conditions.

Whilst the majority of our activities are directed towards children and their parents, TOFS is also keen to encourage medical professionals to take out a membership.

For further information about TOF, OA or VACTERL, or about the charity TOFS, please contact the TOFS national office (*contact details on back cover*) or visit our web site at www.tofs.org.uk.

TOFS is reliant on members' fundraising and voluntary/trust donations to continue its activities; we are therefore constantly appreciative of funds raised and received.

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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

Web site: www.tofs.org.uk

Registered Charity no. 327735



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