[http://www.tofs.org.uk/index.php/what\_is\_tof\_oa/common\_problems](http://www.tofs.org.uk/)

# Common problems

Every child is unique and will be affected in different ways by TOF/OA. However, there are some common problems. These are:

## The “TOF cough”

Children with TOF/OA often have a loud, barking cough, known as the “TOF cough”. It’s caused by a floppiness (tracheomalacia) of part of the trachea (windpipe) and can get worse when a child has a cold or other respiratory problems. Whilst it can sound alarming, it doesn’t necessarily mean the child is ill.

Tracheomalacia and the TOF cough

*Content provided by Una M MacFadyen,   
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## The trachea

In the common type of TOF/OA there is a blind ending upper oesophagus or foodpipe (the atresia) and a connection between the lower end of the oesophagus and the trachea or windpipe (the fistula). Other forms of TOF/OA may be slightly different, but the effects on the trachea at the site of the fistula are the same:

1. the supporting cartilage (gristle) framework, which keeps the airway open, is not fully formed.
2. specialised lining cells (goblet cells and ciliated cells) are replaced by less specialised cells (squamous cells) that are less efficient at keeping the airway protected against infections.

In spite of these differences, in almost all cases the body recovers well so that there are no long term serious problems by the time the child has grown up.

## What is tracheomalacia?

Any problem with the cartilage framework which normally supports the trachea will mean that the wall making up the airway is softer than it should be. If there is a long section without cartilage, this can cause a ‘floppiness’ of the trachea, and this is called tracheomalacia.

### The ‘TOF cough’

Mucus is a sticky fluid which is found on the surfaces of the normal airway. Its main function is to trap dust particles in the air, before they travel deep into the lungs. The specialised lining cells of the airway have mechanisms both to produce this mucus, and then to transport it - together with the trapped dust particles - up the airway to the throat, where it is either coughed out or swallowed.

The mechanism is normally so efficient that most people hardly notice that they have mucus to clear and just automatically clear their throat from time to time.

In a TOF child, the gap in the specialised protective lining cells at the site of the fistula can make the clearance of mucus less efficient and/or the mucus drier than normal. The child may therefore have to cough quite hard in order to clear this dry and therefore extra-sticky mucus.

The ‘TOF cough’ results from the need to clear this dry mucus by coughing through a trachea with a slightly floppy section that makes the flow of air less smooth during the cough. Anything that increases mucus production will make the TOF cough worse, e.g. colds, liquids or food getting into the airways (aspiration), or asthma.

The noise of the TOF cough is quite characteristic and can be alarming to others. Most TOF families find their own ways of dealing with any comments which may arise, however it can often be useful to tell people who will be looking after a TOF child (friends, playgroups, schools) about the cough in advance. Letting them know that it is not distressing for the child (as is usually the case) and that it does not mean that the child is unwell in any way can prevent worry and embarrassment.

## How important is tracheomalacia?

Most babies who have had a TOF repair do not have major difficulties coping with tracheomalacia and it becomes less and less significant as they get older.

There are however a small number who have problems, which can be serious.

## When are problems worst?

The problems with the respiratory tract following TOF repair tend to be at their worst in the first two years.

This may be because the trachea of normal infants contains immature cartilage which is quite soft. The area of absent cartilage from the TOF trachea has a much greater effect when surrounded by a softer cartilage framework; when there has been more growth of the normal section of the trachea and the tracheal cartilage is firmer, the deficit is easier to cope with.

## Severe tracheomalacia

The infant with severe tracheomalacia usually becomes symptomatic around the age of 4-6 months when there may be an exaggeration of the TOF cough, excessive wheezing, or cyanosis (‘blue attacks’) during feeding.

In extreme cases, the infant may experience acute life-threatening episodes often called ‘near death episodes,’ when the baby seems to be choking and unable to breathe. The trachea actually collapses, so that no air can pass through it.

Because these extreme situations are so rare, many doctors have never witnessed one of these episodes; the child is in any case often perfectly well by the time they get to hospital or are seen by a doctor.

The story of a baby going blue with hard crying may suggest ‘breath holding attacks’ - which are relatively common. However, these tend to occur in older toddlers who are either angry or very upset, and happen at the end of a big breath in or out. The ‘near death episodes’ usually happen in the midst of a normal crying spell, and happen near the start of either breathing in or out (depending on where the tracheomalacia is). Observers who can describe these kinds of detail about episodes can help greatly in making a correct diagnosis.

The attacks can be very distressing. Nonetheless, when the child passes out, their relaxed state helps to open up the airway, bringing recovery; gently pulling the tongue forward may help as a first aid measure. If the baby is unconscious and not breathing, blowing gently into the airway by mouth to nose-and-mouth respiration is the correct course of action.

## What other things happen?

A soft trachea can also be squashed from the outside. For example, a narrowed oesophagus can cause a ‘hold up’ to food (an obstruction), so that the upper oesophagus fills up and stretches so much that it pushes against the neighbouring trachea. Without a strong enough cartilage framework to keep it open, the trachea can become closed off until the oesophagus empties again.

The same situation can arise if the lower oesophagus fills up with refluxed stomach content. The latter episodes can be hard to explain as they may not seem to be linked to any reason for a breathing problem.

## Will my baby have problems?

Most TOF babies have some floppiness in the trachea, but the majority are not troubled by it and any problems improve with age. In assessing children, it is therefore not enough just to look for ‘floppiness’ – the oesophagus may actually be the origin of the symptoms.

Sometimes there may be more than one problem; in these cases it can be difficult to work out which is the most important, and then to decide on the best treatment.

## Tests for tracheomalacia

### Radiography

The diagnosis can be suspected on a lateral (side-on) radiograph of the neck taken either as the baby breathes in and out, which will show a collapsing trachea. A barium meal study is often useful to check for any oesophageal malfunction.

### Bronchoscopy

Bronchoscopy (looking into the airway via an endoscope) is the most reliable method of reaching a diagnosis.

### Respiratory function tests

Tests of the baby’s breathing can measure how much work the baby has to do in breathing in and out, but cannot give information about the length of the trachea which is abnormal.

## Treatment

Most TOF children do not require treatment for tracheomalacia. A few, however will require surgery, which aims to give the trachea extra support.

Tracheopexy and aortopexy are two such procedures; which is used depends on the individual baby. If the floppy part of the airway stretches down into the smaller airways (bronchi) then these procedures may not be so effective; in such cases, a tracheostomy tube may be inserted.

TOF children with severe tracheomalacia have a good chance of improving greatly with treatment and age.

## Respiratory problems, asthma and chest infections

Babies with TOF/OA can experience short periods when they find it hard to breathe, due to a floppy trachea. This is most likely to happen when a child is breathing heavily, for example when coughing or crying. Children usually grow out of this by the age of two and then only a minority of babies suffer severely.

Babies may also experience respiratory problems such as asthma and chest infections.  These are treated with inhalers or antibiotic treatments.

# Chest infections and ‘wheeze’

*Content provided by Una M MacFadyen,   
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## Infections

There is no evidence that TOF children have more respiratory infections, but they may have more trouble coughing enough to clear the airways of the extra mucus during normal infections.

## The cause of chest infections

Most childhood chest infections are caused by viruses. The body fights these by producing specific antibodies which endow the child with resistance to future attacks by the same virus. This defence system is called the immune system; TOF children have a normal immune system and make antibodies in the normal way.

Some viruses irritate the airways and cause them to become swollen and tight.

Viruses are not killed by antibiotics, so a doctor can only prescribe medications to help relieve the symptoms until the immune system overcomes the infection.

## Why do TOFs have problems?

TOF children have respiratory infections just like other children, however, problems may occur for two reasons:   
  
Firstly, the TOF child’s airway has a less efficient mechanism for clearing the extra mucus which is produced in any chest infection. This can allow mucus to settle in the lungs. Bacteria (germs) can gather in these local accumulations of mucus and cause a more serious infection to develop.

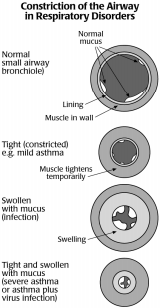
This is often called a ‘super-infection’ because it is on top of the first infection – not because there is anything very special about the germ involved.

Secondly, TOF children may be more sensitive to viruses causing swelling and tightening, making it hard to move air in and out. The effect is the same as is in asthma and responds to the same type of treatment.

## Antibiotics

Bacteria – such as those involved in a ‘superinfection’ – are killed by antibiotics, so if there are signs that a virus infection has not cleared naturally then antibiotics are recommended.Often antibiotics are given right from the start of respiratory infections in young TOFs because they are likely to have difficulty coughing up phlegm.

As the child gets older, antibiotics may not be required so much because the differences in the airway get less important.

Taking antibiotics does not stop the body’s own defences from fighting the virus and the child will build up immunity in the normal way. Most bacterial infections respond to the common antibiotics, but occasionally extra courses or wider acting (‘broad-spectrum’) antibiotics are required. This especially applies if an area of the lung has abnormally small airways, as with bronchomalacia or bronchiectasis (where the walls of small airways are weakened and stretched so that infection collects in tiny sacs deep in the lungs). In such cases chest physiotherapy helps to clear secretions from the lungs and antibiotics may be prescribed for an extended period (weeks or months) until the lungs are clear of signs of persisting infected phlegm.

## Sensitive or tight airways (‘wheeze’)

When the airways narrow from swelling or tightening, the child feels that he/she cannot breathe properly. This is very frightening and can be dangerous if so little air moves that the amount of oxygen reaching the blood is reduced. Prevention is therefore advisable:

1. protect the lungs by having the child immunised against whooping cough, Hib (Haemophilus influenzae), measles and, if advised by your doctor, against influenza and pneumococcus.
2. avoid situations that cause the problem. This may be easier said than done, but for example if you know the child is sensitive to animals then it is wise to avoid getting a furry pet.
3. one of the commonest causes of wheezing in young children (and also of more troublesome respiratory infections and ear infections) is being in an atmosphere where people smoke (passive smoking). Avoiding smoking in the home where ‘chesty’ children live is therefore important.

## Helping the child

Keep calm and know what to do to help. The muscles which tighten around the airways react to fear and anxiety; children are very sensitive to fear in adults, so if adults can know how to deal with any breathlessness it will give the child confidence.

Learn the basic treatment steps, what to give when and how to get help when needed, and you will be helping your child.

## Medications for wheeze

These fall into two main categories:

### Relievers (Bronchodilators)

These make the tight muscles around the airway relax and are most effective when inhaled directly into the lungs. Their action is quick, which ‘relieves’ both the breathlessness and the fear that goes with it. The effect wears off in four hours or less, so treatment may need repeating.

Various inhaler methods allow different ages of child to breathe in the medication. Several companies manufacture relievers, so there are a number of different names for the same kind of drug. Most in the UK come in a blue container, so this is the one to use when sudden tightness occurs.

The way to use relievers should be explained by a doctor or nurse who can make sure that the treatment plan makes sense both to the child and parents, and that when and how to get emergency help is fully understood.

Other relievers with a longer lasting action may be recommended for children with more troublesome symptoms.

### Preventers (Prophylaxis)

Because the airways are sensitive even when they are not tight, it is often preferable to use a regular treatment which helps them to withstand whatever irritates them. This both reduces the number of breathless attacks and makes reliever treatment more effective.

Preventers are administered in the same ways as relievers. There are two main types – sodium cromoglycate (Intal, in a red and white pack) and inhaled steroids (usually in a brown or orange pack, e.g. Pulmicort, Becotide, Aerobec).

Although the steroid preventers are related to the strong steroids that are taken by mouth for other conditions, the dose is so small when breathed into the lungs that they are much safer. They are not at all like the body building steroids that some athletes take against medical advice.

If the airways become very swollen, the response to relievers may not be adequate. The child may then need a short course of stronger steroids (e.g. prednisolone), taken by mouth. Your doctor will advise when this is required. When used in this short term manner, the risks of steroids are far less than the risks of being unable to breathe.

## Swallowing and getting food stuck

Children with TOF/OA often have abnormal swallowing mechanisms and will have to learn to cope with their particular feeding problems.  Some children will need to receive tube feeding to make sure they stay healthy. At first, many will need a special diet, but most will (with their support of their school) cope with school dinners or a packed lunch.

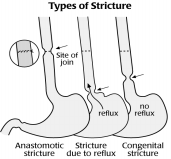
# Strictures

*Content provided by Mark D Stringer MS FRCS FRCP,   
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## What is a stricture?

A stricture is the commonest problem requiring surgical treatment after TOF repair.

It is a narrowing in the oesophagus, usually due to scarring at the join (the anastomosis) between the two ends of the oesophagus. This is called an anastomotic stricture. Occasionally, a stricture develops lower down in the oesophagus; in this area it is usually due to gastro-oesophageal reflux. Very rarely, children with TOF/ OA are born with a stricture in the lower oesophagus and this is congenital stricture.



Anastomotic strictures are more likely when the join-up of the oesophagus was difficult because of the wide gap between the two ends. If the join is under tension, the blood supply to the join site is impaired and it tends to heal as a tougher and more fibrous scar.

Strictures are believed to be worsened by gastro-oesophageal reflux; acid from the stomach damages the join in the oesophagus and leads to more scarring.

## Symptoms of stricture

Typically, symptoms develop within a few weeks or months after the baby’s oesophagus has been joined up. The baby may be slower to feed or may choke and splutter during the feed. This is not a ‘one-off’ episode, but happens with each feed. A stricture may become apparent for the first time when the baby is tried with solid food. Advice should be sought from the surgical unit.

Not all swallowing problems are due to strictures. We know that abnormal contractions of the oesophagus (called dysmotility) and difficulties with learning to feed are important causes of feeding problems in some TOF children.

In older children with a stricture, there may be difficulties swallowing lumpy food but it is important to remember that TOF children often have difficulties in this area.

If a child has previously coped well with a certain consistency of solid food and then starts to have problems with swallowing, this suggests a stricture.

### Dealing with obstructions

Occasionally, a piece of food may get stuck in your child’s oesophagus. This is called a bolus obstruction. It may be vomited back or remain stuck and cause distress. Once the child has calmed down, see if they are dribbling their saliva. If they are, then there is a complete blockage and you will need to seek expert medical attention.

If the child is not dribbling, try and get them to sip some water since food materials will often dissolve and pass through. If the child cannot tolerate liquids within an hour or two then you will need expert advice.

A food bolus obstruction can happen in an oesophagus with only minor narrowing and typically occurs with unchewed lumps of meat, apple, sausages, etc. However, a food bolus obstruction can also indicate a stricture that needs dilatation.

## Diagnosis of stricture

Stricture can be confirmed in two ways:

### Radiography

The child is given a drink of a safe liquid which shows up white on a radiograph. The dye (called a contrast material) is watched in motion as it goes down the oesophagus. Using X-rays like this is called fluoroscopy.

A stricture may be seen. Alternatively, there may be minimal narrowing in the oesophagus in which case the feeding difficulty has another cause such as oesophageal dysmotility.

### Endoscopy

This involves looking down the oesophagus with a telescope (called oesophagoscopy), a procedure which requires a short general anaesthetic. It is necessary in cases of persistent food bolus obstruction since the material can be removed using the endoscope.

Grasping forceps can be passed through the endoscope and the material withdrawn with the scope, or the food can be dissolved with squirts of water, or occasionally, the food bolus can be simply pushed on down the oesophagus into the stomach.

## Treatment of stricture

A stricture can be successfully treated by stretching it up (called a dilatation). This is nearly always done under anaesthetic but can be carried out in several ways.

Most commonly, the surgeon passes a telescope (called an endoscope) down through the mouth to the level of the stricture. Then a fine plastic rod is passed through the stricture. A series of gradually larger rods are then passed through until the stricture has been sufficiently dilated.

An alternative method is for an X-ray doctor (radiologist) to pass a thin flexible wire (known as a guide-wire) through the stricture whilst checking its position with a radiograph. Once the wire is safely through the stricture and into the stomach, a thin hollow plastic tube is passed over the wire and down into the oesophagus. This tube has a balloon on the end which can be inflated when it lies across the stricture. As the balloon is blown up, the stricture is dilated (called balloon dilatation).

When performed by experts both methods are safe and effective. The child is able to feed within a few hours and is often able to go home the same day or soon after. A chest radiograph may be done to check that there is no sign of a complication.

A very small number of strictures keep coming back despite repeated dilatations. In these circumstances any gastro-oesophageal reflux must be treated since the presence of stomach acid in the oesophagus may be aggravating the situation. Occasionally, further operative surgery may be needed.

Some surgeons try injecting the stricture scar tissue with steroids which have an anti-inflammatory effect. This injection can be carried out with a special needle through the endoscope. The idea is that when the stricture is then dilated it might heal with less scarring and therefore be less tight.

Other options include cutting out the stricture and rejoining the oesophagus or considering a major operation called oesophageal substitution.

### Complications of dilation

Complications are rare when the procedure is performed by experienced personnel.

The most worrying one is splitting the oesophagus which causes it to leak (called an oesophageal perforation). This is recognised on the chest radiograph or if the child is unwell after dilatation. Oesophageal perforation can be dangerous and requires prompt treatment with antibiotics, fluids and sometimes a chest drain. This kind of treatment would allow most to heal up. Only rarely would an operation be required for this complication.

### Other considerations

Any child born with a cardiac (heart) abnormality ought to receive antibiotics before the dilatation to minimise any chance of an infection settling in the heart.

### What if it doesn’t work?

Anastomotic strictures usually get better after one or two dilatations but occasionally, several stretches are necessary.

If the stricture keeps coming back, it might be because there is a particularly dense scar at the join, it may be because gastro-oesophageal reflux is making the stricture worse, or it may be because there is a congenital stricture. These problems all require separate solutions which may involve surgery.

## Reflux

Children with TOF/OA often experience gastro-oesophageal reflux (GOR). This is where the acidic stomach contents pass back into the lower oesophagus, causing pain and often reluctance to eat. Reflux can usually be treated using a combination of practical measures and prescribed medications. Occasionally it requires further surgery.

# Gastro-oesophageal reflux

*Content provided by Mark D Stringer MS FRCS FRCP,   
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## What is reflux?

In gastro-oesophageal reflux (GOR), as the stomach empties into the bowel (the duodenum), part of its contents are also squeezed back into the oesophagus. Since the stomach contents are acidic, irritating acid passes into the oesophagus.

The lower oesophagus has features that normally prevent GOR causing problems. These are not strongly developed in babies, which explains their tendency to vomit; this reduces during the first months or years as the antireflux mechanisms get stronger.

In TOF babies, GOR is even more common. This is partly because the oesophagus has not developed normally and partly because repair of the OA often pulls the junction between the oesophagus and stomach upwards, weakening the antireflux mechanisms. TOF babies therefore vomit more easily than normal babies.

GOR is more likely to be a problem if there was a wide gap between the oesophageal ends and the join was tight.

## Symptoms of reflux

Because all healthy babies have a tendency to GOR, the issue is whether GOR is causing a problem in your baby – rather than whether GOR is occurring at all.

Reflux causes frequent vomiting after feeds. This is not the small mouthfuls of vomit (‘possets’) seen in all babies, but the vomiting of large amounts of the feed. This can happen straight after a feed or right up until the next feed. If GOR is severe, the baby may have difficulty gaining weight.

The oesophagus may become sore from the acid (the adult equivalent of which is heartburn) leading to irritability and poor feeding. In some cases, bleeding from the oesophagus causes anaemia or signs of blood in the vomit (haematemesis). Strictures can also be made worse.

Rarely, reflux can happen so quickly that it leads to the baby inhaling vomit, leading to a chest infection or difficulty with breathing. In severe cases, the baby may temporarily stop breathing (called ‘apnoea’).

Most TOF babies have mild reflux, which gets better either by itself or with medicines, but a few have severe reflux which needs treatment.

## Diagnosis of reflux

Listening to the history often gives the doctor clues as to whether GOR is causing problems. It can however be difficult to tell whether a TOF baby’s feeding problems are due to GOR, a stricture or another problem.

### Radiography

In this method the child swallows some dye (barium contrast material) in the radiography department. If the child has GOR, as the stomach contracts to empty, the dye is also seen travelling up the oesophagus. The severity of reflux can be assessed by how much dye passes upwards and whether it just enters the lower oesophagus or passes right up to the throat.

### pH Study

This involves passing a fine tube through your child’s nose down into the lower oesophagus. A radiograph is sometimes needed to check the position of the tube. The tip of the tube has a sensor which measures the acidity (or pH) and records it on a small computer. The pH study usually runs for 24 hours, during which the child can eat and drink and live a fairly normal life within the constraints of the equipment (as carried by the boy shown here).   
  
Any antireflux medicines must be stopped before the test to avoid a false reading.

Normal children have a little acid in the lower oesophagus for brief periods only (less than 5-10% of the time) but children with significant GOR have acid present for much longer.

### Endoscopy

Persistent gastro-oesophageal reflux causes the lining of the lower oesophagus to be inflamed (oesophagitis) and this can be seen by endoscopy (oesophagoscopy).

### Milk scans

This technique is also known as gastro- oesophageal scintigraphy and involves the baby taking a small amount of radioactive material by mouth, followed by a milk feed; the baby is then scanned by a special camera. The test does not require sedation and the radiation dose is less than that involved with radiography. Radioactivity in the oesophagus indicates GOR but there is none of the detail seen with a barium swallow.

## Treatment of reflux

This depends on the severity of the reflux and how much trouble it is causing.

Mild GOR, which is probably present in all TOF infants (and many otherwise healthy babies), tends to improve spontaneously with age and often gets a little better when the baby is able to wean on to more solid food. Simple measures that are helpful include changing the position of your child (posturing) and, in babies, thickening the milk.

### Posturing

GOR tends to be worse when lying flat and therefore a gentle raise of the head of baby’s cot can be useful. This can be done by putting a pillow or folded blanket under the mattress to create a gentle head-up slope. Never attempt to let your baby sleep directly on a pillow which could be dangerous. During the day, keeping your baby propped up in a chair (but not slumped over) can help prevent reflux. Changing the nappy before feeds makes vomiting less likely than doing so when his/her tummy is full.

### Milk feed thickeners

There are many types of milk thickeners:

Carobel, Nestargel - add just before a feed; thickens it instantly.

Thixo-D, Thick and Easy - can be added just before a feed or mixed in advance when feeds are prepared and stored. Gaviscon - powder which can be added to infant formulas.

Feeding your baby with slightly smaller volumes of milk given at more frequent intervals may also be helpful.

### Antireflux medicines

In general, antireflux medicines either reduce the severity of the reflux by improving the downward movement (i.e. motility) of the oesophagus and stomach, or by reducing acidity so that the reflux is less damaging to the oesophageal lining.

These are some commonly used drugs:

Acid lowering drugs - cimetidine (Tagamet ®) and ranitidine (Zantac ®) are the drugs most frequently used to lower the stomach’s acidity. Omeprazole (Losec ®) is a more powerful acid- lowering drug.

Gaviscon contains antacids and tends to float on top of the feed, thickening it.

Any medicine can have side effects but these are not known to be common with these drugs. Any unusual reaction to a medicine should always be reported to your doctor and the drug should be stopped, at least temporarily.

Occasionally, a child may get diarrhoea with cisapride, develop unusual move- ments or a rash – in which case, stop the medicine and seek medical advice. Medication often has to be continued for many months. It can be gradually removed when the reflux has improved.

In some children, reflux is persistent but mild, causing occasional heartburn or discomfort but never severe enough to need surgery. In these patients, antireflux medicines need to be used occasionally or only when they have symptoms.

Sometimes children do not get better with the medicines or have major problems such as repeated stricturing, chest infections from overspill of refluxed material into the lungs, persistent severe oesophagitis or inadequate weight gain. In these children antireflux surgery has to be considered, most commonly the Nissen fundoplication procedure.

# VACTERL - an overview

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## What is VACTERL?

The term ‘VACTERL’ describes a group of anomalies which often occur together in newborn babies. It is an acronym for:

Vertebral (spinal) defects   
Anorectal atresia (failure of the anus and lower end of the gut to form)   
Cardiac (heart) defects   
Tracheo-oesophageal fistula with or without Esophageal atresia (American spelling of ‘oesophageal’)   
Renal (kidney) anomalies   
Limb defects.

VACTERL was once simply ‘VATER’ but the longer term is now preferred, since it includes cardiac defects – which over 70% of these children have – and acknowledges that the limb problems are not only in the radius bone of the forearm. ‘VATER’ was originally described in the mid 1970’s, so children born before this date may not have been screened for these problems. Other synonyms for VACTERL are Kaufman syndrome, PIV and PIAVA.

## What is a VACTERL child?

To qualify as a ‘VACTERL’ child, three of the seven components mentioned above must be present. There may also be other characteristics which occur more frequently in affected children than the rest of the population; these include ear abnormalities, genital anomalies, cleft lip and/or palate, thumb abnormalities, various changes in gut development and in the fetus, and the presence of a single artery in the umbilical cord (normally there are two).

No one baby is likely to display all these features and no two individuals are likely to be affected in exactly the same way.

## What causes VACTERL?

For every 6,250 births, one child will have VACTERL, making it relatively rare. Large numbers of patients are required to scientifically study any medical condition, so the small numbers of children with VACTERL, and their wide variety of features, has made research difficult. Consequently, little is known.

Growth of the fetus in the womb is enormously complex; in spite of the massive scientific efforts aimed at understanding the intricate mechanisms involved, many pieces of the jigsaw are still missing.

The cause of VACTERL is unclear. One theory suggests that cells are disrupted at an early stage of development. There are only three types of cell in the three-week- old fetus, one of which is called a mesodermal cell. These cells go on to form the gut wall (including the oesophagus), the kidneys and bone (including the spine and the skeleton of the limbs). In principle, a change in a mesodermal cell could result in a change in any of these body parts, which would help explain the wide variety of abnormalities present in VACTERL.

Efforts have been made to identify agents, such as drugs, which could adversely affect these cells, but so far nothing has been proved.

Chromosomes are the inherited structures of DNA which carry the genes to determine everything about an individual. Two chromosomal abnormalities (the more common of which is Edwards syndrome) may result in the features of VACTERL – two rare cases in which the cause of VACTERL is known. In order to exclude this, the chromosomes of all VACTERL babies are examined.

## Can VACTERL be detected before birth?

The regular use of ultrasound to examine the fetus is greatly increasing the number of abnormalities detected before birth.

Many VACTERL babies will not be picked up during a routine scan, but in pregnancies which merit detailed ultrasound investigation it is more likely that skeletal abnormalities, kidney defects and a single umbilical artery will be seen.

Perhaps, as ultra-sound scanning continues to improve, ante-natal diagnosis of VACTERL will become the norm.

## What are the exact problems in VACTERL?

### Vertebral abnormalities

Vertebrae are flat bones which are stacked on top of each other to form the spine. If they are deformed, an abnormal spinal shape may result (‘scoliosis’ or ‘kyphosis’). Abnormal bone development may also be accompanied by abnormalities in the associated muscles and nerves; treatment depends on the severity of the deformity.

### Anorectal atresia

This is a broad term which describes a range of abnormalities. At one extreme, children have an intact bowel with a blind end (‘low imperforate anus’). At the other extreme, the bowel stops quite some distance short of what should be the anal opening, and there are often abnormal connections between the bowel and the bladder or vagina.

### Cardiac abnormalities

‘Ventricular septal defect’ (VSD) is the commonest type of cardiac abnormality, accounting for more than three-quarters of all cases. It is a hole in the wall that separates the two large chambers of the heart. Consequently, the heart does not function efficiently. The child is symptom-free if the hole is small, but in more severe cases children can become breathless and fail to thrive. The defect may close spontaneously but sometimes surgery is required.

There are many other heart defects which may occur in isolation or accompany a VSD.

### Tracheo-oesophageal fistula with oesophageal atresia

VACTERL children are classed as TOFs because this is a feature of the condition, but their problems and treatments differ due to their multiple abnormalities.

### Renal anomalies

These fall into two main categories; total failure of one or both kidneys to form, and ‘other’ anomalies.

Absence of both kidneys has sinister implications and may be detected ante- natally by ultrasound. A single absent kidney is however entirely symptomless, because the remaining kidney can fully compensate for the deficiency.

The ‘other’ category is vast, including a large number of different problems. These are not always associated with malfunction, but problems may arise if the kidney cells do not work efficiently or if kidney infections result.

### Limb abnormalities

The forearm (the part of the arm between the elbow and wrist) contains two bones, the ulna and radius. Partial or total failure of the radius (and the muscles which attach to it) to develop is frequently seen in VACTERL children. Characteristically this causes the hand to lie at a right-angle to the forearm; usually the thumb is also malformed or absent. Splints, or even surgery, may be required to correct the defect.

Limb abnormalities are not always restricted to the forearm – the feet or legs can also be affected.

## The VACTERL Child

Hospitals are a second home for VACTERL children. They receive intensive medical attention, both to treat the immediate problems and to monitor their progress. This involves frequent consultations with a variety of specialist doctors, in addition to intensive general health surveillance, such as regular growth checks.

The impact of such prolonged hospital- isation should not be underestimated, although VACTERL children have comparable intelligence to their peers.

Ultimately, the future of an affected child will be determined by the nature of their initial problems and the success of their treatment. Residual problems may cause long-term disability, the severity of which will vary between individuals and depends on the number and type of defects.

In the majority of cases, continued medical support will be required, but this does not prevent a VACTERL child from leading an active, independent and fulfilled life.