Antenatal diagnosis of TOF/OA

Content provided by JAS Dickson FRCS FRCSE FRCPCH, Consultant Paediatric Surgeon, and Dr Alan Sprigg, Sheffield Children's Hospital.

How can TOF/OA be detected before birth?

Because of the widespread use of prenatal ultrasound scanning, some TOF babies are now detected before they are born.

What does the scan show?

Two signs that the radiologist doing the scan looks for are as follows:

EXCESS FLUID IN THE WOMB

The inability of the baby to swallow may lead to excess fluid collecting in the womb in 20-25% of babies. This is called hydramnios or polyhydramnios.

SMALL OR ABSENT STOMACH

In babies with no fistula from the lower oesophagus to the windpipe, the stomach is also very small, there being nothing passing into it to encourage it to stretch and grow. This has to be noted on several examinations to be significant.

The combination of excess fluid around the baby and a small or undetectable stomach on a prenatal ultrasound scan is therefore strong presumptive evidence that the baby has this rare form of oesophageal atresia with tracheo-oesophageal fistula.

So if these findings are present on my scan, does this mean that my baby is a TOF?

And if they aren't, that he/she is not?

No! these findings are not as clear cut as they seem since there are various factors which can confuse the interpretation:

THE INABILITY TO SWALLOW

This is also seen in severe cases of cleft lip (hare lip) and palate – but these conditions can usually be picked up on the scan.

An inability to swallow is also seen with other conditions, such as brain problems, which can not be seen on a scan. Other than as an exceptional case, TOF is not associated with brain problems.

FLUID IN THE STOMACH

The presence of a distal tracheo-oesophageal fistula allows fluid a route to the stomach by providing a way for it to travel from the trachea to the oesophagus and on to the stomach, bypassing the atresia. These pregnancies therefore do not show polyhydramnios.

Are there any other ways of detecting TOF prior to birth?

The level of alpha feto protein (used to detect spina bifida and abdominal wall defects) in the mother's blood may also be slightly raised if the baby has TOF.

Under these circumstances, an amniocentesis test may be carried out, to exclude the possibility of a chromosomal (genetic) defect which may be incompatible with the baby's survival; examples of such defects include Trisomy 13, 18 and 21. If these are present the mother is given the option of an elective termination of the pregnancy.

Otherwise, the pregnancy should be allowed to continue to term and the baby delivered by the normal vaginal route.

What are the chances of detecting TOF prior to birth?

Overall it is not possible to put a mathematical figure on the predictive value of these signs.

In the commonest form of oesophageal atresia with tracheo-oesophageal fistula, even in the presence of polyhydramnios, it is not possible to make a definite antenatal diagnosis by conventional methods.

Sometimes the scan will miss a TOF child, sometimes TOF is suspected but the baby is born normal – and sometimes the scanner gets it right: he or she suspects TOF, and the baby is born with TOF.

This information has been written for the parents of TOF children by TOFS (Tracheo-Oesophageal Fistula Support) – helping children born unable to swallow.

If you have any feedback on this leaflet, please use our leaflets feedback form which is available from either the TOFS office our web site.

TOFS relies on money from membership fees, voluntary donations and other sources of charitable income to fund its activities.

Web site
www.tofs.org.uk
Address
TOFS,
St George's Centre,
91 Victoria Road,
Netherfield,
Nottingham NG4 2NN
Telephone
0115 961 3092
Fax
0115 961 3097
Email
info@tofs.org.uk

TOFS does not offer specific medical advice to parents. We work only in a supportive role, offering emotional and practical support to meet the needs of parents and providing a source of information which complements that given by the specialist hospital.

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"We were advised there was a possibility our baby would have a tracheo-oesophageal fistula. The advice given to me over the telephone by TOFS was very, very helpful and we very much appreciated receiving a lot of literature on the subject. We were, as a result, able to ask the right questions and to the right people, which put our mind at rest at that stage. Fortunately, we now have a very healthy baby boy, with no fistula to report."

Related leaflets from TOFS which you might like to read:

1. What is TOF/OA?
2. The news that your baby is a TOF
3. What causes TOF?
4. Conditions occurring with TOF/OA
5. VACTERL: an overview

These are all available from the TOFS web site (www.tofs.org.uk) or from TOFS office.

TOFS also publishes a book, 'The TOF Child,' which is suitable for both parents and medical professionals. Details are available from TOFS.

What should I do if my scan indicates I may be having a TOF child?

This news will be a big shock for you, and if you are in this situation now you must be feeling extremely anxious. This is normal; everybody wants things to go perfectly when they have a baby and you have every reason to be worried.

However, there are things you can do which may help to put your mind at rest ...

FIND OUT ABOUT TOF AND OA

Understanding more about these conditions will help you to know what you might expect to happen in the future.

You will find that with modern techniques and nursing care, TOF children can be expected to do very well. Although the early years with a TOF child can, for some families, be challenging, any problems will generally disappear as the child grows. Adults who were born with TOF/OA may prefer to eat with a drink to hand to aid swallowing, and may have reflux or occasional swallowing problems, but they are not otherwise affected by the condition and live full and active lives. Very few babies die from TOF, unless they have other major problems, for example if they are very premature or have a serious heart defect.

TALKING HELPS

Share your feelings and don't be afraid to ask questions.

TOFS is here to help you: if you wish we can put you in touch with some TOF parents, so that you can discuss what the condition means to them.

Might there be other problems?

A proportion of TOF children are also born with other problems, most commonly those described in VACTERL syndrome which includes vertebral (spinal), anal (related to the back passage), cardiac (heart), renal (kidney) and limb (usually the hand and/or forearm) anomalies.

VACTERL children have three or more of these anomalies ,many of which can be detected on a scan.

There is a wide range of problems within each part of VACTERL. Some are very minor and will not affect the child at all, whilst others may be serious – even life-threatening. Your consultant will have checked for the major problems during your scan.

What difference does this make to the birth?

Knowing that your baby may be born with TOF means that you may be asked to consider having your baby at or near a hospital with experience in the treatment of TOF/OA.

If the mother has polyhydramnios, regular repeat scans may be carried out to check on the baby's growth and development. The doctor's will also want to monitor the mother's health and comfort level, since the excess fluid can become uncomfortable.

Being prepared will help everybody to be ready to give your baby the very best care and, because you will know more about what is happening, you won't be having to deal with the news that your child is a TOF at the same time as dealing with the surgery and post-operative care.

We wish you well and hope that you will keep us informed about your baby.