CHARGE SYNDROME: ESOPHAGEAL ATRESIA AND TRACHEOESOPHAGEAL FISTULA: FOR THE PHYSICIAN

Bryan D. Hall, M.D.
Dept. Pediatrics/Genetics, University of Kentucky Medical Center, 740 Limestone, Lexington KY 40536-0284 bdh@pop.uky.edu (859 257-5559

Meg Hefner, M.S., Genetic Counselor and Assistant Professor of Pediatrics, Division of Medical Genetics, St. Louis University School of Medicine, 1465 S. Grand Blvd., St. Louis, MO 63104
Hefnerma@slu.edu Meg@chargesyndrome.org (314)768-8730

TYPES AND FREQUENCY OF TRACHEO-ESOPHAGEAL ANOMALIES IN CHARGE:

Tracheo-esophageal fistula	20%* (or higher, often H-TEF is diagnosed late)
Esophageal atresia	15%

Related findings frequent in CHARGE which may affect treatment and/or management of tracheo-esophageal anomalies:

Polyhydramnios (prenatal)	frequent
Tracheomalacia	frequent
Laryngomalacia	30%
Gastro-esophageal reflux	50%
Esophageal dyskinesia	75%
Pharyngo-esophageal dysmotility	80%
Facial palsy	50%
Cleft palate	20%
Choanal atresia	30%
Cranial nerve IX/X anomalies	frequent
Abnormal Ba swallow	very frequent
Gastrostomy in patient w/o EA	36%
Fundoplication	frequent

DIAGNOSTIC TESTS:

- NG tube
- X-ray
- Barium swallow

The newborn with CHARGE who has TEF/EA will have copious oral secretions and a nasogastric tube will not pass into the stomach.

MEDICAL CONSEQUENCES OF FEATURES

Children with CHARGE and TEF/EA have higher mortality than other children with CHARGE. This is especially true when combined with choanal atresia and/or a heart defect.

Many individuals with CHARGE have abnormal esophagus motility and/or gastro-esophageal reflux. A significant percentage also has difficulties with suck and swallow resulting from cranial nerve anomalies. These problems further complicate feeding problems in these children. Many children need gastrostomy feeds for years.

MEDICAL MANAGEMENT AND CAVEATS

Treatment of EA/TEF in CHARGE may be similar to treatment of isolated EA/TEF. However, there are a number of special considerations in these cases:

1. Heart defects (present in 2/3 of children with CHARGE) may be complex. These may affect timing of surgery and overall stability of the patient.
2. Possible unexpected reaction to anesthesia. Some children with CHARGE are resistant to sedation, while others are slow to recover from anesthesia. Because of risks of anesthesia, it may be appropriate to combine surgical procedures in these children.
3. Laryngomalacia or tracheomalacia is common in CHARGE and can result in surgical or anesthetic complications. The mortality rate in children with this combination of features is high.
4. Cranial nerve palsies complicate post-operative feeding in a major way. Most importantly, apparent abnormalities of cranial nerves IX and X cause secretions and food to pool in the pharynx. Esophageal peristalsis is uncoordinated. Aspiration and reflux are common. This improves in weeks, months or years. A small number of children have permanent problems. Facial palsies are usually unilateral and may affect lip closure.

NON-MEDICAL MANAGEMENT

A feeding specialist is a must for children with CHARGE with TEF/EA. All of them will have feeding difficulties, which are likely to last for years. Transition from gastrostomy feed to oral feeding can take years, as can transition from pureed to solid and liquid foods. Growth may be impaired by poor caloric intake.

If a patient has CHARGE, TEF/EA is likely to be only one of a number of serious medical problems for that child. It is critical to talk to the parents about the whole child. An orofacial team which includes ENT, audiology, and feeding specialists can be a great help to parents because they have experience with feeding difficulties, PE tubes, and ear infections, all of which are common in CHARGE. Communication with other specialists following the child is critical to effective care of these children. These children often have multiple medical appointments every month or even every week, so coordination of medical appointments is appreciated by parents.

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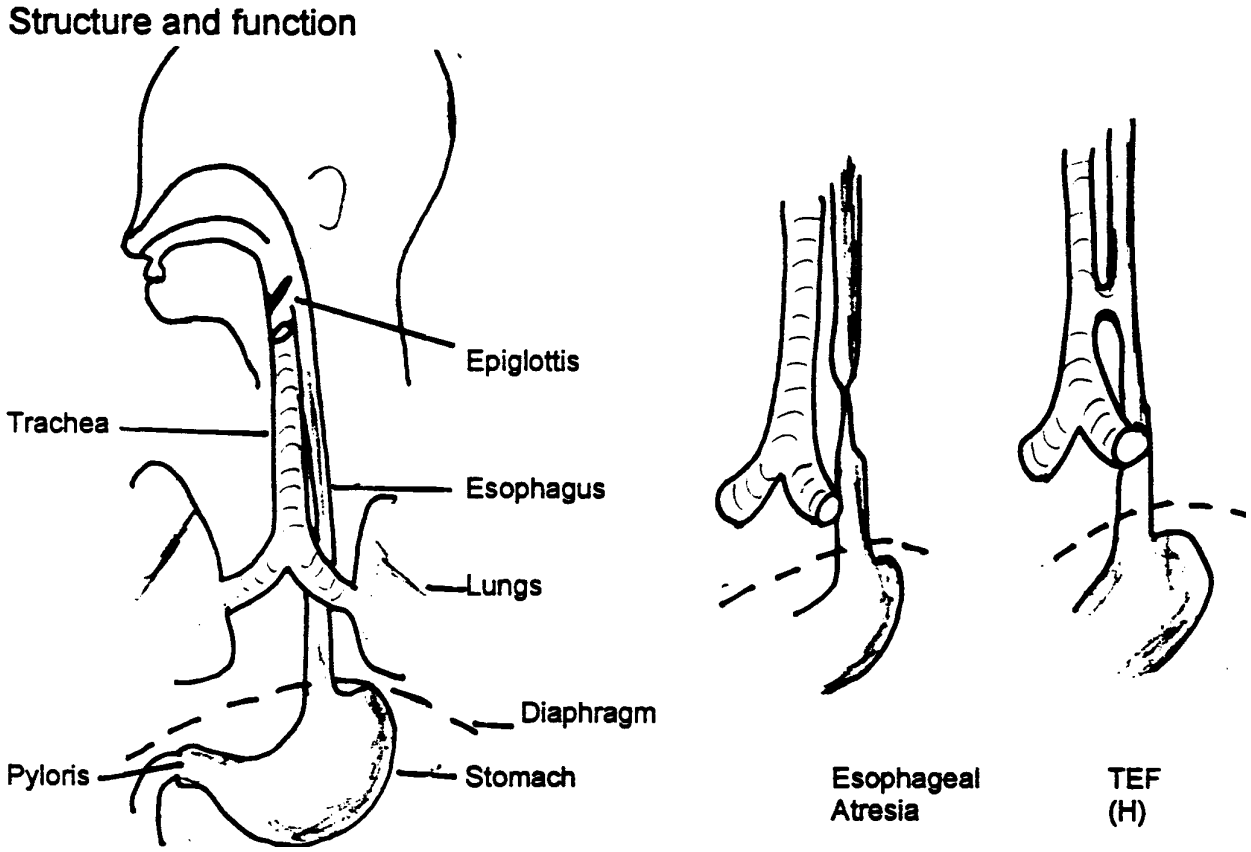
CHARGE SYNDROME: ESOPHAGEAL ATRESIA (EA) AND TRACHEOESOPHAGEAL FISTULA (TEF): PARENT INFORMATION

Meg Hefner, M.S., Genetic Counselor and Assistant Professor of Pediatrics, Division of Medical Genetics, St. Louis University School of Medicine, 1465 S. Grand Blvd., St. Louis, MO 63104
Hefnerma@slu.edu Meg@chargesyndrome.org (314)768-8730



The **epiglottis** is a small flap in the back of the throat. The purpose of the epiglottis is to ensure that food goes down the esophagus and air goes down the trachea.

The **trachea** (windpipe) is the tube leading from the back of the throat to the lungs for breathing. The trachea runs along side the esophagus and then splits in two along either side of the esophagus, leading to the two lungs. The trachea is surrounded by rings of cartilage and muscle.

The **esophagus** is the tube leading from the back of the throat to the stomach. It is surrounded by a coat of muscle, which help the food move down into the stomach. Food normally moves down the esophagus into the stomach. Normally, when food or saliva is swallowed, waves of movement in the esophagus (peristalsis) propel the food into the stomach. The opening from the stomach to the intestines is called the **pyloris.** Swallowing normally begins in the first three months of prenatal development.

Problems associated with CHARGE

Esophageal atresia (EA) refers to an esophagus which ends blindly before reaching the stomach. It is often found along with tracheo-esophageal fistula. Often, polyhydramnios (excessive amniotic fluid) was noted during pregnancy. Prenatally, the fetus normally swallows and recycles amniotic fluid. If the fetus cannot swallow due to EA, polyhydramnios can result. Also, the stomach may not be visible by ultrasound exam because it is not filled with amniotic fluid.

Once the baby is born, he or she will often have copious frothy secretions pouring out of their mouths which require constant suctioning until the EA is treated. A baby with EA cannot eat by mouth (food can't get to the stomach) until the atresia has been surgically corrected.

Tracheo-esophageal fistula (TEF)
A fistula is a connection. TEF is a connection between the esophagus and the trachea. This connection allows food (from the esophagus) to enter the lungs (aspiration). Food in the lungs can cause pneumonia (referred to as aspiration pneumonia), which can be very serious. If there is a TEF with EA, all the food taken in will end up in the lungs, as the esophagus does not connect to the stomach, but ends blindly.

The hardest type of TEF to diagnose is **H-type TEF**, where the esophagus does not end blindly, but there is a fistula or connection between the trachea and the esophagus. In H-type TEF, air can enter the stomach (from the trachea) and food can enter the lungs. Air in the stomach can cause bloating of the baby's abdomen and can make the baby very uncomfortable

Symptoms of TEF/EA
If you had polyhydramnios or the fetal stomach could not be seen on ultrasound, TEF/EA may have been suspected during your pregnancy. After a baby is born, esophageal atresia may be suspected in the first few days if the baby doesn't tolerate feeding (chokes and spits), doesn't seem to swallow saliva (froths at the mouth), seems to aspirate the milk into his or her lungs, or has abdominal distention (bloated tummy).

H-type TEF may not be recognized until much later. The typical symptoms are choking with feeding, excessive gas in the stomach or intestines, and **frequent aspiration pneumonias.**

By now, you probably already know enough about CHARGE to recognize that many of these symptoms can also be caused by other problems associated with CHARGE. Choanal atresia, cleft palate, reflux, and cranial nerve abnormalities can all interfere with breathing and eating. Infants with CHARGE may not be fed right away due to other problems. And so on. This means that TEF and associated problems may not be diagnosed as quickly or as thoroughly in infants with CHARGE.

Diagnosis of TEF/EA
Esophageal atresia is usually diagnosed by placing a tube either down the nose or through the mouth, down the throat and into the stomach. An X-ray is taken to document that the tube is in the esophagus (not the trachea) and that it has reached the stomach. If the baby has EA, instead of ending up in the stomach, the tube will curl up in the blind-ending pouch. If there is a tracheo-esophageal fistula (which is very common with EA), the tube may end up in the lungs.

Other tests, which might be done to diagnose or confirm the diagnosis, are bronchoscopy or esophagoscopy. Barium swallow may be needed to diagnose an H-type TEF, where the esophagus is attached to the stomach, but there is a connection to the trachea.

Treatment
EA and TEF can be surgically repaired. Before the repair and while it is healing, the baby will need to be fed by a gastrostomy tube or button. This is a tube or opening which goes directly into the baby's stomach, bypassing the esophagus. How long the baby will require the G-tube is variable. Many children with CHARGE are fed by g-tube for years (see below and Feeding section)

Occasionally, EA will be repaired and yet a small H-type TEF is not recognized. The remaining TEF can still cause multiple aspiration pneumonias. A barium swallow test might be helpful in diagnosing the TEF.

Outcome and complications:
Even after surgical correction, there can be some leakage at the site of the reattachment. The esophagus can tighten up. If this happens, it can be treated by dilating the esophagus. Many children with CHARGE have swallowing difficulties, gastroesophageal reflux, recurrent pneumonias, and poor growth. Some of this may be due to TEF/EA, but much of it may be due to other complications of CHARGE (see FEEDING section). Once the baby has recovered from surgery, oral feeding might be tried. However, individuals with CHARGE typically have other features which complicate oral feeding. Be careful not to push it!