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VACTERL Association

by Tracy Reed

VACTERL Association. I remember hearing those words over the phone about a month after our fourth daughter Margaret was born. Maggie was born on a beautiful warm day in August, 2006. Like any child's birth, Maggie's was long anticipated and we couldn't wait to see the new member of our family. Little did my husband and I know that the day of Margaret's birth would begin a journey of twists and turns culminating in a word that we had never heard before: VACTERL.

The first thing I noticed after Maggie's birth was that she was missing a thumb on her right hand. As the tears flowed from my eyes, I thought, "She'll be okay, we have Shriners Hospital." What we weren't prepared for was when the nurse turned our baby over and showed us that she was born with no anal opening. Both my husband and I were in total shock. How could someone be born without an anus? I've watched Discovery Health and I've seen all sorts of "weird" things that you don't usually hear of everyday. Never once, though, did we ever hear of a person being born without an anus.

Following the hours after Maggie's birth, she was taken to a children's hospital three hours away to try to determine if she had any more problems. That started our journey into learning about our daughter's condition. It wasn't until a month later that we heard the words VACTERL or VATERS association.

VACTERL is an acronym for **V**ertebral, **A**nal, **C**ardiac, **T**racheal, **E**sophageal, **R**enal, and **L**imb. It is also known as VATER, VACTERLS, or VATERS association with the S standing for **S**ingle umbilical artery, another common anomaly in these children. VACTERL Association is basically a group of nonrandom physical birth defects that occur together and whose cause is not known at this time. While VACTERL can arise in tandem with some chromosomal disorders, at this time there is no known gene or set of genes that causes VACTERL, and its occurrence is probably due to a combination of factors. VACTERL Association applies to a child when there are at least three anomalies that fit into the VACTERL spectrum and genetic testing has ruled out other genetic diseases or syndromes. While some children are born with the full spectrum of congenital malformations, this is quite rare.

There are many different health issues one may face with a child who has VACTERL association. Apart from the defects themselves, children often have problems with growth and weight gain and may have multiple infections. While some have delayed development in the beginning due to many hospitalizations and illnesses, the majority of

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VACTERL children have normal intelligence. Other health issues occur as a result of individual defects.

Vertebral Anomalies

About seventy percent of children will be born with anomalies of their vertebral column. Most are benign, but they can contribute to other problems. Some vertebral issues that are common are hemivertebrae (half formed vertebrae), butterfly shaped vertebrae, fused vertebrae, missing vertebrae, tethered spinal cord, Chiari Malformation and scoliosis. For most children these anomalies cause few issues early in life, but scoliosis may become more significant later on.

Anal Anomalies

Another rarely talked about but profoundly important issue for children with VACTERL is Imperforate Anus. This is a topic all by itself, but in general, around fifty-five percent of children will be born with this defect, which ranges from having a misplaced anus to not having an anal opening at all. This defect is often the most difficult and shocking for parents to handle since it is such a taboo subject to talk about and also may result in significant problems related to stooling. This defect, however, is very serious and must be taken care of in the early days of life either though anoplasty (reconstruction of the anus), a pull-through procedure (colon is pulled through and reconnected to the anus), or colostomy (an opening in the abdomen that allows stool to be collected). Depending on the type of imperforate anus, correction may involve minimal surgery or full reconstruction through multiple surgeries.

Cardiac Anomalies

Cardiac issues are also common in VACTERL, and about seventy-five percent of VACTERL children are born with some type of cardiac condition. The spectrum can be endless but the more common defects are atrial septal defects, ventricular septal defects, or Tetralogy of Fallot. All children found to have VACTERL defects should be checked for cardiac problems regardless of whether a murmur is heard or not. Some children will require surgical repairs while minor defects may self-resolve or cause no ongoing problems.

Tracheo-**E**sophageal Anomalies

The T and E defects of VACTERL usually occur in tandem, typically in the form of a **T**racheal **E**sophageal Fistula and **E**sophageal Atresia, though other defects of the trachea or esophagus may be present. Esophageal atresia occurs when the stomach and esophagus do not connect with each other, while a fistula is an opening between the trachea and esophagus that should not be there. Children born with these defects typically require surgical repair of the trachea and esophagus, and can have serious ongoing gastric reflux disease that may require fundoplication surgery or a feeding tube. Some children also have significant respiratory issues, including tracheomalacia (a floppy trachea). Around seventy percent of children born with VACTERL will have these birth defects.

Renal Anomalies

Renal issues are also commonly found in children with VACTERL. Approximately fifty percent of children are affected, and one can find a large spectrum of kidney and urological problems. These defects can be severe, such as incomplete formation of one or both kidneys or obstruction of outflow of the urine, or more minor, like kidney reflux (backflow of urine). If these defects are corrected early in life, kidney failure may be prevented. Some children may require surgery, medication, or cathing to deal with these anomalies.

Limb Anomalies

Limb issues vary widely, but affect about seventy percent of children. Some children have extra fingers or toes, fused digits, missing digits, clubbed feet or hands, or forearm abnormalities. Many can be corrected surgically, while others require ongoing adaptations throughout life.

Our daughter Maggie was born with the V, A, C, R and L of VACTERL. The one thing that brings peace of mind to me is that we live in such a day that Maggie has a wonderful chance at having a good quality of life. Maggie has had four surgeries thus far, two major and two minor, though we are far from done. Because she was so stable at birth, she had her first surgery, an anoplasty to create a new anus, at two days old. She also had a tethered cord spine release, which has helped significantly with her development since her tethered spinal cord seemed to affect many areas of her body. In addition, she has had a frenuloplasty to release a tongue tie and ear tubes placed for chronic ear infections. Maggie is due to have her anal prolapse repaired and both my husband and I are considering pollicization surgery, in which the index finger is moved over to create a thumb.

Maggie also takes many medications. Though she was not born with the TE of VACTERL, she still suffers from silent reflux and takes Prevacid and Pepcid. She also has multiple food allergies and is on an elemental formula called Elecare that she takes orally. We do breathing treatments when needed for asthma due to allergies and she is on a prophylactic antibiotic to prevent infection from kidney reflux.

Treatments will differ from child to child since children with VACTERL are all very unique, resulting in different health problems. The number of surgeries varies widely among children born with VACTERL. Some can have as few as three surgeries to fix defects, while others have as many as 60 or more, depending on the extent of the defects.

While the defects of VACTERL never go away, through surgery, treatment, and acceptance they become more manageable. VACTERL is not consistent from child to child, and there never is a constant with ongoing surgeries and treatments. The only thing that remains constant for a child like Maggie with VACTERL association is the never-ending love of her parents.

Margaret 'Maggie' Reed was born August 8, 2006 and is one of 4 daughters born to Tim and Tracy Reed of Corry, Pennsylvania. We are a homeschooling family that loves to learn, enjoy life to the fullest and meet other families. You can reach Tracy at sonlightuzer4ever@yahoo.com

Further Resources:

- Pull Thru Network: www.pullthrunetwork.org
- Congenital Heart Defects Support: http://groups.yahoo.com/group/congenitalheartdefectsupport
- IA Parents (Imperforate Anus): http://groups.yahoo.com/group/IA-parents
- LMC-TCS (tethered cord group): http://groups.yahoo.com/group/LMC-TCS
- VATER Connection: www.thevaterconnection.org
- VACTERL Association (in the UK): www.vacterl-association.org.uk/index.php
- VACTERL Network: http://www.vacterlnetwork.org/
- Congenital Scoliosis Support: http://groups.yahoo.com/group/CongenitalScoliosisSupport
- Helping Hands Foundation (upper limb differences): www.helpinghandsgroup.org
- On the Other Hand (information about hand differences): www.ontheotherhand.org
- Limb Differences.org (for both upper and lower limb differences): www.limbdifferences.org
- TEF/VATER: www.tefvater.org
- TOF/OA (in the UK): www.tofs.org.uk/index.php
- EA/TEF Family Support Connection: http://www.eatef.org/index.html