

CDH NEWS

The MassGeneral Hospital *for* Children & Children's Hospital Boston
Congenital Diaphragmatic Hernia Study Newsletter

Meet Our Team!



Our dedicated staff ensures that the all aspects of the project run smoothly. **1. Dr. Donahoe**, Project Director & Principal Investigator, MassGeneral Hospital for Children **2. Dr. Wilson**, Principal Investigator, Children's Hospital Boston **3. Dr. Pober**, Geneticist. **4. Drs Longoni, Loscertales, Lage and Meaghan Russell, M.P.H., PhD(c)** Fellows **5. Dr. Maalouf**, Visiting Fellow **6. Caroline Coletti, M.S. & Anna Frangulov, B.S.** Coordinators

Project Update

From Principal Investigators Drs. Patricia Donahoe & Jay Wilson

This year we are gratified to be receiving our 8th year of funding from the National Institutes of Health (NIH) for our CDH study! To date, we have enrolled over 700 individuals with CDH and family members, both nationally and internationally! Our hope is to enroll over a 1000 patients. We continue to utilize a variety of cutting edge genetic and developmental approaches to better understand the causes of CDH. Your participation in this research study remains greatly valued and appreciated!

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CDH Resources

The Association of Congenital Diaphragmatic Research, Advocacy and Support (CHERUBS) (a support group for families of children with CDH):

<http://www.cherubs-cdh.org>

CHERUBS Australia: <http://au.geocities.com/ozcherubs/>

CHERUBS United Kingdom: <http://www.uk-cherubs.org.uk/>

Yahoo Listserv for CDH Families:
<http://health.groups.yahoo.com/group/Breathof-Hope/>

SHARE Pregnancy and Infant Loss Support, Inc. (early pregnancy loss, stillbirth, or infant death support):
<http://www.nationalshareoffice.com/>

The Compassionate Friends (non-profit organization providing resources and support for grief resolution):
<http://www.compassionatefriends.com/>

MUMS National Parent to Parent Network:
<http://www.netnet.net/mums>

The International Birth Defects Information Systems website:
<http://ibis-birthdefects.org/start/diaphern.htm>

How to Reach Us:

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Our patients spread the word...

With the help of one of our families, information about the CDH study was published in an overseas CHERUBS journal in England. As a result, we have enrolled more patients from England. Such help is very much appreciated and is certainly welcomed! Please do help us spread the word about our research in your community—every little effort helps this study and we hope that our findings will ultimately be of value to families like yours in the future!

Want to Share Your Family's Story?

Our patient's stories are a vital component of our newsletters. They are particularly helpful to newly diagnosed families. If you have a CDH story you would like to share with our families, please contact us for more details. We would love to publish it in our next newsletter!

DID YOU KNOW THESE FACTS??

- CDH is one of the most common, life-threatening birth defects in the United States.
- CDH without any other birth defects or genetic disorders is referred to as isolated CDH. About 60% of CDH cases are isolated CDH.
- Approximately 40% of CDH patients have what is known as complex CDH (or CDH+). These patients have other birth defects or genetic disorders in addition to CDH.
- The risk of having a child with CDH in the general population is 1 in 3000, or a 0.03% chance.
- After having a baby with isolated CDH the chance of having another child with CDH increases to 1 in 100, or 1%. In families where the baby has complex CDH, the chance of having another child with CDH can be harder to predict since it depends on the nature of the accompanying birth defects or genetic disorder.

HEARING LOSS IN PATIENTS WITH CDH

By Anna Frangulov, B.S., Brian Fligor, Sc.D., CCC-A & Barbara Pober, M.D.

Recent medical advancements have significantly improved survival for children born with CDH. As a result, understanding how to minimize or even prevent long-term complications for patients with severe CDH is more important than ever. Though outcomes for patients with CDH vary greatly, the most common complications involve breathing issues, feeding problems, delayed growth, low muscle tone and hearing loss. In our previous Newsletters we covered several of these topics. In this article, we will focus on hearing loss.

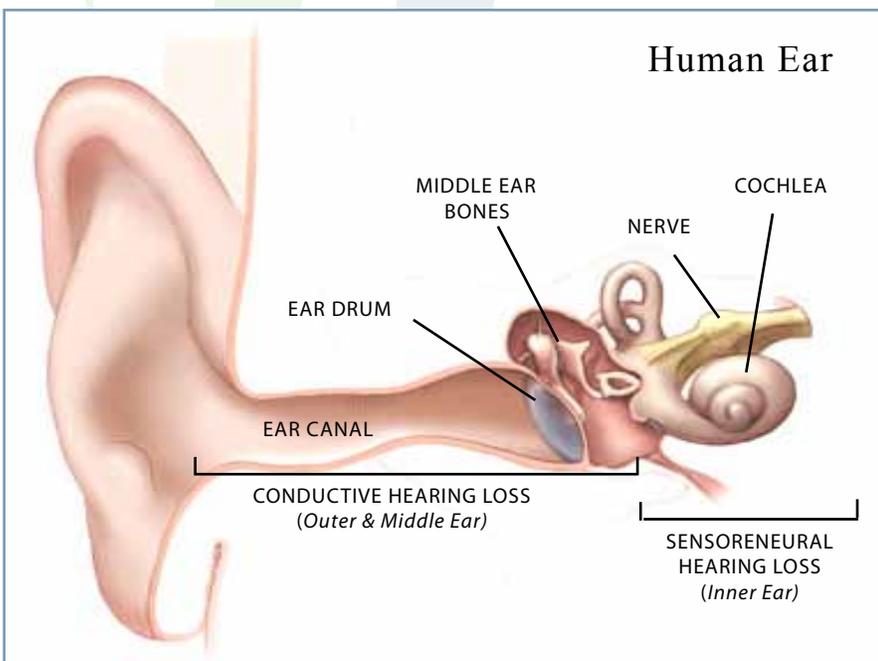
About Hearing Loss

Our ears have many components, all of which have to work well for us to hear sounds (see diagram). If one part of the ear does not function well, hearing loss may occur. However, not all hearing loss is the same. Hearing loss is classified into three types, according to the location and kind of problem:

Conductive - when something is not working in the outer or middle part of the ear (i.e. ear canal, ear drum, middle ear space or bones). More than 90% of conductive hearing loss in children is caused by frequent ear infections or middle ear fluid. Such conductive hearing loss is mostly temporary and is often is treated with an antibiotic, or in cases of chronic ear infections, surgical placement of “PE” tubes.

Sensorineural (SN) - when something is not working in the inner part of the ear (i.e. cochlea or the hearing nerve). In the general population, genetics (or family history) contributes to the cause of SN hearing loss in more than 60% of cases. Other causes may include head trauma, noise exposures, pre-natal or post-natal medications, infections and anatomical birth defects. Most SN hearing loss is permanent and has varied interventions which are specific to each individual case.

Mixed - a combination of both conductive and SN hearing loss



What Type of Hearing Loss Do Patients with CDH Have?

For the most part, patients with CDH who develop hearing loss have SN hearing loss. Children with more severe CDH complications are at a higher risk of developing SN hearing loss. It is also possible, however, that patients with CDH can develop conductive hearing loss, as may occur in any child who experiences frequent ear infections or middle ear fluid buildup.

How Common is Hearing Loss in Patients with CDH?

In the general population, SN hearing loss occurs in 0.3% of children. In other words, for every 1,000 births, three children develop SN hearing loss.

Among patients who stay in intensive care units (for reasons other than CDH), the rate of SN hearing loss is about ten times higher. Thus, for every 1,000 births there are 10-20 individuals with SN hearing loss, or about 1-2%.

In patients with CDH, SN hearing loss is even more frequent. Approximately half (50%) of patients with CDH who initially have normal hearing develop SN hearing loss.

“Approximately half (50%) of patients with CDH who initially have normal hearing develop SN hearing loss”

What Causes Hearing Loss in Patients with CDH?

To date, it is not exactly known what causes hearing loss in patients with CDH and why SN hearing loss rates are high in this population. However, some studies identify a few factors that put these patients at a greater risk. These factors include the need for:

- Longer stay in the intensive care unit (i.e. NICU)
- Higher doses and longer treatments with medications that are part of the care regimen for critically ill infants with CDH (including medications with side effects that are potentially damaging to the ear, etc)
- Prolonged use of breathing/ventilation machines (i.e. High Frequency Oxygenation)
- Use of heart and lung bypass machines (i.e. ECMO)

In addition to the factors above, family history may play a role as well. Since over 60% of SN hearing loss in the general population is genetic, it is possible that genetic changes may also contribute to developing hearing loss in patients with CDH.

“If hearing loss develops, it is typically not present at birth.”

Finally, it is possible that several factors, rather than just one factor, may combine together and lead to the high occurrence of hearing loss in patients with CDH.

Time Course and Severity of Hearing Loss in Patients with CDH

Little is known about exactly when SN hearing loss begins in patients with CDH. Most studies show, however, that if hearing loss develops, it typically is not present at birth but is usually detected within the first 3 years of life; in some individual cases, the hearing loss sets in after age 3 years. Once SN hearing loss occurs, it remains stable in some CDH patients, while for others it worsens. The severity of SN hearing loss also varies significantly from patient to patient. That is, some patients develop slight hearing loss, whereas others may not hear at all. Families need to work closely with an

“Since patients with CDH are at a higher risk for developing SN hearing loss, ongoing hearing evaluations by an audiologist until late childhood are crucial.”

audiologist to provide the best intervention and management should hearing loss be detected in their child.

PARENT'S CORNER

FREED'S FAMILY STORY

By Candace Freed

Your family is coming over on Sunday to do WHAT?" I can still hear my friend's voice screeching through my cell phone. "We're having a blood draw party at my house." I know it may not be the typical end to a family reunion weekend, but it was how my family- the Freed family of Cleveland, Ohio, spent one sweltering Sunday this August.



Candace's 30th Birthday

Growing up as a child I never realized how unusual the incidence of CDH was in my family. Actually, I didn't know what CDH was at all, even though I underwent surgery to correct a Right-CDH that presented when I was 9 months old. I knew that I had a scar on my stomach and I had heard tales of the doctors who had fixed my "tummy" when I was a baby. When I was almost 5 years old my brother,

Things to Remember

Normal hearing is very important for speech, social and emotional development. Since patients with CDH are at a higher risk for developing SN hearing loss, ongoing hearing evaluations by an audiologist until late childhood are crucial. Early detection and management of hearing loss optimizes social interaction, education, as well as language acquisition. ■

References

Masumoto K, Nagata K, Uesugi T, Yamada T, Taguchi T. Risk factors for sensorineural hearing loss in survivors with severe congenital diaphragmatic hernia. *Eur J Pediatr.* 2007 166: 607-612.

Morini F, Capolupo I, Masi R, Maria PR, Locatelli, M, Corchia C, Bagolan P. Hearing impairment in congenital diaphragmatic hernia: the inaudible and noiseless foot of time *J Pediatr Surg.* 2008 43:380-4.

Hearing Loss Resources

National Institute on Deafness and Other Communicative Disorders (NIDCD) - Offers overviews on the many causes of hearing loss.

<http://www.nidcd.nih.gov/health/hearing/>

Self Help for Hard of Hearing People (SHHH) - Info on technology for battling hearing loss and news from the world of medicine on hearing loss developments.

<http://www.shhh.org/>

BUPA: Hearing Loss - British health website has important information for all on sound and the ear, the causes of hearing loss, deafness in children, and diagnosing and treating hearing loss.

http://hcd2.bupa.co.uk/fact_sheets/html/Hearing_Loss.html

Hearing Loss Web - Site is dedicated to people who have hearing loss but who are not members of the traditional deaf community.

<http://www.hearinglossweb.com/>

Raising Deaf Kids - Information on learning and communicating with hearing loss, and general information on hearing loss.

www.raisingdeafkids.org

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Charlie as a baby

Charlie, was born with a Left-CDH. He was immediately transported via big green machine

to Rainbow Babies and Children's Hospital, which thankfully was only about 15 miles away. Doctors did not seem hopeful. I knew that my baby brother was very sick, but I had no idea of the severity of his condition. I thought that G-tubes and IV pumps in the nursery were standard issue. Didn't everyone's baby brother stay in the hospital for 3 months after they were born? Weren't there in-home nurses in everyone's houses? Who didn't cut holes in their baby's sleepers so that the leads for their heart and breathing monitors could come through? I remember more than once being roused from sleep by the

"It's not that I didn't love my brother. Medical problems and hospitals became as commonplace to me as watching Sesame Street."

sights and sounds of an ambulance carting off my little brother. As long as he didn't mess with my toys, I didn't care.

It's not that I didn't love my brother. Medical problems and hospitals became as commonplace to me as

watching Sesame Street. My father's younger brother Terry was a R-CDH survivor. He made headlines and tugged at the heartstrings of our local newspapers, who affectionately dubbed him "the baby who couldn't cry". Born in 1964, his case was groundbreaking. His was one of the first known cases of successful CDH repair. It's a miracle that he survived. He's had a host of abdominal and intestinal problems since then, but to me he is just Uncle Terry. Years later, another of my

"People in a family helped each other out even when it wasn't convenient. Families didn't keep score."

father's brothers, Uncle Larry, fathered a baby named David who passed away due to complications of CDH and heart defects. It was very sad, but again, I was young and CDH seemed, in my 10-year-old mind, akin to the common cold.

The adage "Ignorance is Bliss" should have been written about me. I was happy, well-adjusted, and remarkably lucky that my uncle, my brother and even myself were alive. Concurrent with my belief that CDH and breathing treatments were "normal" was a strong sense of family that had been instilled in me at a very young age. Families did anything they could for each other and never complained. People in a family helped each other out even when it wasn't convenient. Families didn't keep score. My mother's cousin and her husband opened up their home to us so that we



Charlie 25 years old



Candace's Family. Mom & dad top left, Charlie top right, Candace with her husband bottom

could be a little closer to the hospital. They also took care of me while my parents, who could only afford one car, took shifts at the hospital while juggling bills, housework, and my father's demanding job. My mom's cousin still chides me with stories of my dramatic wailing for my mother. Both sets of grandparents took turns watching me. Uncle Terry's wife would babysit for my brother and me when my parents needed a break. I never remember my parents telling me that they were sad, overwhelmed, tired, frustrated or that they were struggling to make ends meet. I remember my mom making turkeys out of exam gloves during our countless hours of waiting for doctors. I remember my dad taking me to the hospital cafeteria for custard. I remember my grandparents taking me on elevator rides. I remember the nurses sneaking me into the PICU to see my brother, clad in a scrub cap and gown that swallowed me up. I remember Uncle Terry playing games with me and making me laugh when I was at my grandparents' house. They carefully filtered all difficult news away from me, keeping me safe in my ignorance, replacing it with hope, love and bliss. I guess whoever wrote the adage was right.

Now that I'm a mother myself and 30 years old (gasp!), my blissful ignorance has turned into thankful respect. My husband and I are blessed with two beautiful healthy boys. Noah is 3 and Henry is 21 months. I struggle to see the blissful side of things sometimes. I'm constantly running from one place to another, working on a Master's Degree in Sociology while raising kids and trying to have some semblance of a relationship with my husband, family and friends. I let the small things get to me. The more I learn and piece together about my family's medical history, the more I am amazed. My brother is turning 26 next week and will be married in April 2010. My Uncle Terry is a

"Now that I'm a mother myself and 30 years old...my blissful ignorance has turned into thankful respect. "

happy 44-year-old man with a wife, two grown children and a daughter-in-law. I am healthy, too. My only CDH-related problems have been scar tissue growth and I have frequent hiccups. As I've grown up, I began to feel extremely guilty about having caused my parents such problems. I felt guilty because other CDH survivors didn't fare as well as I have, and many didn't survive at all. My brother has had far more medical problems than I have. He only has one fully formed lung. He has scoliosis as a result of his CDH. He's undergone 24 surgeries. He has a learning disability. Charlie

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even contracted Hepatitis C as a result of a blood transfusion he received in 1983, which was made necessary because of the CDH repair surgery.

I decided to do what I could to help further research and funding for CDH. I joined email lists. I read research studies. I proudly wore my turquoise bracelet on March 31st in support of CDH Awareness Day. I became a supporter of CDH research on Facebook. It all felt so superficial to me. I wanted to make a real difference. How could I help people who were going through the unthinkable with their children when I escaped relatively unscathed?

Fast forward to my cell phone conversation with my friend. I read about the genetic CDH study that Boston Children's Hospital and Massachusetts General Hospital are conducting and I surged with

"...I wanted to make a real difference..."

excitement. I couldn't wait to help discover genetic information about CDH and eventually further the treatment, awareness and possibly prevention of what seemed to me an obvious condition that turns out isn't as publicized as I had thought. I contacted Meaghan Russell and I knew immediately that this study was for our family. Meaghan and all of the

people involved with the study have been so wonderful and so helpful to us. Meaghan, Dr. Barbara Pober, and Meaghan's mother, Alison, came to our house along with a phlebotomist on Sunday August 9th and we had what my

"...I was able to be thankful for ... the gift that my family has for facing tough times armed with a sense of humor and a huge helping of love."

friends jokingly call "the blood party". We were able to collect genetic samples and medical histories from 19 members of my family. We spent the entire day together laughing and reminiscing about our family while being united for the cause that has affected our family more than any other. Once again, we didn't focus on the negative aspects of CDH. We joked while we underwent head and finger measurements. We awarded the honorary title of "Biggest Freed Head". We ate doughnuts and pizza. While we contributed to this wonderful cause, we were able to strengthen our family ties and reconnect with family members that we hadn't seen in months. In the midst of what could be an anxiety-provoking needle stick, I was transported back to my 5-year-old blissful ignorance. I was able to be thankful for my family and their health, my own health, and the gift that my family has for facing tough times armed with a sense of humor and a huge helping of love. Now that's bliss. ■

FREED'S FAMILY STORY

By Terry Freed

I was born in May of 1964 in Berea, Ohio without a diaphragm on the right side. I do not remember a lot until I was 13 years old. I was told that at birth I could not cry. I had my first surgery when I was 2 days old, and I kept going in and out of the hospital for many procedures related to CDH.



Terry as a baby

“...I was told that at birth I could not cry.”

When I was 13, I was in rushed to the hospital yet again. It turns out that my small intestines were bleeding internally. I underwent another long surgery where doctors removed 80% of my small intestines. After waking up from the procedure, I remember asking my dad if I had died. I thought I had a dream where I was dead and I could see myself lying on a table and doctors working on me. Well, according to my father, I did stop breathing on the operative table and had to be revitalized back to life. So it was not a dream! I was in the hospital for 3 months recovering from that surgery! When I finally got back out of the hospital, I felt like I was different. I could not play sports like I used to, I could not take gym in school and my parents were always looking out for me. I remember just playing around

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with my brothers or neighbors and my dad and mom would say, “don’t hurt him, don’t bump him, be careful not

“...After waking up from the procedure, I remember asking my dad if I had died.”

to cause him to fall down.” I felt like my parents were overprotecting me.

When I got older I was teased a lot from kids at school because I was not like a “normal” kid. I had trouble gaining weight and always looked sickly. Kids said you “will never have a date with a girl because they will see your scars and run away”. This made me wear 2 shirts at times!

As I grew older I tried more things on my own, like sports, to prove I

“I can remember getting hit and getting the wind knocked out of me but I would play it off just so I could be “normal.”



Terry's Family. Terry with his son (top row), daughter & wife (bottom row)

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was just like everyone else. I can remember getting hit and getting the wind knocked out of me but I would play it off just so I could try and be "normal."

My last major surgery concerning CDH was in 1992 when I had problems breathing. They found out that the diaphragm they built inside me broke open (reherniated) and my intestines were in my chest. They went in and fixed it with a patch.



Terry today

Today, I still have problems with my scar tissue, blockages, and at times kidney stones and stomach problems. These remind me that I am bit different. However, I am now able in every other way! I finally gained weight! I got married in 1984, and despite our worries, we have 2 healthy children. I believe that sharing my story gives hope to other people with CDH. Even though I've gone through a lot, you can see I've turned out OK!!! ■

"Thank you" to the Freed Family!!

Our words of "thanks" to the entire Freed Family cannot begin to convey the extent of our gratitude for their amazing efforts in support of CDH research! Candace and all her relatives are simply remarkable!! They have already gone through so much, yet they selflessly gave of their precious time, resources, and privacy in an effort that may eventually help others. Meeting so many wonderful people was awe-inspiring and makes us work even harder to achieve our goal, that of identifying genes responsible for CDH with the eventual hope of developing new treatments in the future.

Nineteen (19!) members of the Freed family donated their blood for research. What are we doing with these precious samples? We are using two cutting edge genetic methods to search for a changed gene that is shared among those with CDH. Their genetic patterns may be very instructive and have much to teach us. But for right now -- many thanks to all of you!

~CDH Study Team

KID'S CORNER

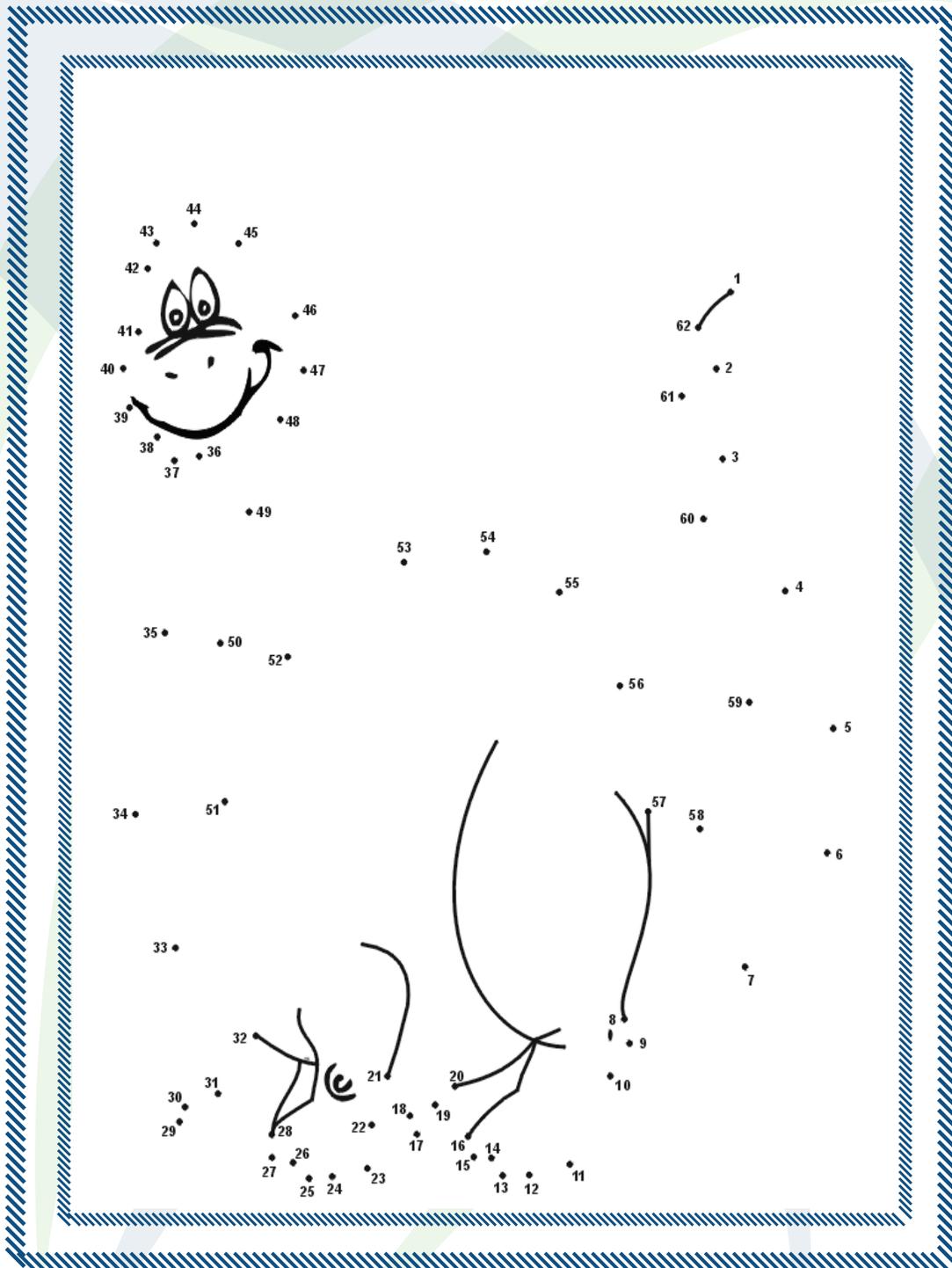
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Lets Count & Connect the Dots!

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