Focusing on a Cure

RDH12
Fund for Sight

Issue 3, August 2012

Our group is up to 14 families and 18 children. We have grown by leaps and bounds in the last two years.

To donate, please visit

www.rdh12.org

Our organization serves all children/people affected by RDH12 Lebers Congenital Amaurosis.

Address:
OUR PO BOX is:
PO Box 161481
Boiling Springs, SC 29316

a message about the foundation

Since our March 2012 newsletter, we have grown a little larger, and have now raised $600,000 for RDH12 research in less than two years.. Thanks to a very generous gift of $250,000 from one of our supporters, we took a giant step toward a cure for our children. Our researcher, Dr. Debra Thompson, has received her first donation from our group, and has begun work on the safety study for RDH12. By March, 2013, we hope to have raised another $150,000 to prepare for our entrance into clinical trials.

a message to our supporters

We would not be where we are without all of you. If you have donated time, love, good thoughts, or money this last year, we thank you. When we started this journey, we were taking a leap of faith. We weren't sure if we would succeed. And our supporters have shown us that we can, and that we have. As we continue to grow, we hope that you are proud of what you have helped accomplish!

What our kids are up to this summer

Bella

Bella is in a summer reading/math program, as well as a summer camp at the town pool. Her eye sight has remained stable. In the summer she enjoys the pool, the beach, swimming, camping and playing with her friends. They went to Myrtle Beach in July, and are going to Hershey Park in August.

Raphael

Raphael will be entering primary school in the fall and is now 6 years old. This summer he has tried tennis and golf. He takes swimming classes and judo classes (and has earned his yellow belt), and is in summer camps during the day. He is taking a vacation to Cyprus with his family this month.

Rose

She is 7 and has completed first grade. She has started cane and braille training. She loves swimming, singing, playing outdoors, and vacationing in the Smoky Mountains.

Joanna

She is 6 and will enter 1st grade in the fall. She is starting can and braille training. She loves drawing, swimming, and playing dress up in the summer.

Celine

She is now 2 ½ years old and enjoys reading books, drawing, swimming, and playing with her siblings.

Darius

This picture was taken when he and his family visited a safari animal park in June. In the summer, Darius likes to ride his bike and go swimming.

What our kids are up to this summer

Bill and Lily

Both kids are enjoying summer camp at the South Carolina School for the Deaf and Blind. They have been swimming almost every day and playing with their neighborhood friends.

Ben

In the summer ben enjoys swimming, baseball, bike rides and trips to the park

Finley

Finley finished Kindergarten and is getting ready for first grade. In the summer, Finley enjoys swimming, riding her bike, playing outside, and going to camp. We have taken a few day trips this summer to the zoo, local historical sites, and Six Flags.

Abigail

Abigail loves to swim, ride her bike, and go camping in the summer. This summer she has been to Myrtle Beach, NYC to visit the American Girl doll store, and going to amusement parks.

What our kids are up to this summer

Aiden

Aiden will be entering 3rd grade this coming fall and will be in the gifted program. He competed in the national braille challenge and placed in the top third for the country. He takes guitar and drum lessons. He is very interested in rockets and space.

Gabby

Gabby turned 3 in May and will be starting a special needs preschool in a few weeks. She loves music, singing, and dancing. She has been working on self feeding and has been seizure free since May! She spends most of her summer in the pool.

Cecilia

NEW FAMILY! Meet Cecilia from New York. Her family just joined our group in late spring, and we are so glad they did. Cecilia is six years old and just finished Kindergarten. She has 20/80 vision right now and is doing great in school. Her favorite summer activity is swimming. She enjoys riding her bike and going on bike rides. She is a member of the U8 soccer team and has fun playing.

Brooke

NEW FAMILY! Meet Brooke from Kansas. Her family joined our group a few weeks ago, and we are glad they found us. Brooke is 5 years old, and is getting ready for Kindergarten.

***Our other kids are busy having a fun summer, but know that Maria, Stijin, and Julie and well and happy*!

Instead of giving myself reasons why I can't, I give myself reasons why I can"

Meet the Kinney Family

We are the Kinney family from Ohio. We have three children, David, age 13, Andrew, age 11 and Abigail, age 8. Abigail's story began when she was 18 months old. I noticed that something was unusual about her vision. She did not look directly at me, but appeared to look past me. I expressed my concern to her pediatrician who then referred us to a pediatric ophthalmologist. He determined that her retina was not normal and recommended that she be seen by a retinal specialist. At this time, we began seeing a doctor at the Cole Eye Institute at the Cleveland Clinic in Cleveland, Ohio. Here, they performed an ERG, electroretinogram, which measures the electrical responses of various cell types in the retina. Abigail's response was determined to be minimal. Her blood was sent to the Carver Lab in Iowa to confirm what the doctor had suspected – LCA, Leber's Congenital Amaurosis.

This was a very difficult time for our family. We were heartbroken as we learned the devastating effects of this disease and because it is progressive, we have no way of knowing how long Abigail will retain what sight she now has.

In June 2007, when Abigail was almost three years old, I received a call from the Carver Lab which confirmed Abigail's initial diagnosis of LCA with a RDH12 gene mutation. The doctor said research is progressing, but there had been no new research developments of this specific gene. We continually kept searching, reading, and learning information about LCA, realizing that the RDH12 gene mutation was very rare. In October 2010, I joined forces with other families who had formed the RDH12 Fund for Sight. This organization raises funds to support research and development of a cure for LCA caused by RDH12. Through this group's efforts, the researcher, Dr. Jean Bennett, of the University of Pennsylvania, has agreed to begin work on a gene therapy treatment.

But funding this research takes a lot of money. I could no longer just sit back and do nothing. I had to become involved because my child's eyesight was at stake. So I joined the other families and began fundraising efforts. Our fundraising name is "Abigail's Angels". At first I didn't know where to begin, but the problem was quickly resolved. When others learned of Abigail's condition and what funds were needed for the research, they became a godsend. Her school held a PJ/movie day fundraiser; my friends organized a kids' dance and a rock-a-thon; my husband's work held a Jean day/raffle; my church family held chicken grill dinners, a Chinese raffle, a Trash-to-Treasure sales, a Gospel Sing for Sight concert, and our first Abigail's Angels Charity Walk is set for this September. Local Bible Schools have also donated their collections to our fund as well as numerous donations from caring family and friends. I can't thank everyone enough for all of their love and dedication to Abigail. They are truly "Abigail's Angels".

Abigail spends one hour per day learning braille. She has a CCTV at her desk which zooms in on anything that needs enlarged. Her eyes take longer than normal to adjust to changing light. She wears sunglasses outside to enable her eyes to adjust easier. Abigail has two older brothers, both of which do not have this gene mutation. She loves to sing, dance, swim, ride her bike, and go camping. She participates in tap, jazz, tumbling, and ballet and is a member of a Brownie troop. She also attends Sunday School and is a member of the children's choir. She is a very happy little girl.

Every day that Abigail can see is another day of learning. We never know if one day she will wake up to a world of darkness. We pray that a cure will be available for Abigail and all who have this terrible disease.

The Kinney family (left to right): David, Abigail, Keith, Andrew, Lori

Update from Dr. Debra Thompson

The RDH12 Fund for Sight has been fortunate to be able to partner with some of the leading researchers in the field of gene therapy and LCA. Through our funding of Dr. Jean Bennett, we were able to see the creation of a viral construct sufficient to produce the RDH12 protein in cells that previously did not express RDH12. This was a critical first step in bringing a gene therapy to reality for our kids.

Now we have to extend our research funding to Dr. Debra Thompson of the University of Michigan, one of the preeminent researchers in the area of RDH12 biology. Independently, Dr. Thompson produced her own gene therapy for RDH12 LCA. With funding from the RDH12 Fund for Sight, Dr. Thompson is extending this work to demonstrate the safety of conducting gene therapy for RDH12 LCA. In her first set of experiments made possible with our funding, Dr. Thompson treated mice lacking the RDH12 gene with the gene therapy. She was able to demonstrate that the injection of the gene therapy into the eyes of these mice did not have a detrimental effect on the visual function of these animals as measured by an electroretinogram.

Furthermore, the level of RDH12 expressed by the injected virus in the mouse retinas did not result in changes at the molecular level of important components of the visual signaling pathway. This is a critical step for the development of the gene therapy. It helps to establish that there is minimal risk of unexpected negative consequences associated with restoring RDH12 to a retina that has developed without it.

The next step for Dr. Thompson’s work is to confirm in the mouse that lacks RDH12 that gene therapy has can correct the defect in these animals at the biochemical level. This research is providing us with the necessary first data from an intact, living organism for RDH12 gene therapy. The research is progressing, the cure is progressing, and the achievement our ultimate goal progress with it.

There is no better way to thank God for your sight than by giving a helping hand to someone in the dark." - Helen Keller

Upcoming Fundraisers and Recent Fundraisers

RECENT Fundraisers

Pennsylvania

June, 2012 - Pancake Breakfast raised $1460. Thank you to New Brighton's Interact Club.

Ohio

Spring, 2012 - Starkdale church events totaling $2293

Spring, 2012 - Hills Elementary School basket raffle - $901.10

New York

June 9th - Eye Love Cecilia yard sale and volleyball tournament - $12,003

Massachusetts

May 2012 - Bunco night - $1000
July 2012 - Bunco Night - $500
July 2012 - Swim for Fin - $1400

Europe (Candle in the Dark)

Raphael's 3 Aunts Donated $5500

Upcoming Fundraisers

Pennsylvania

August 25, 2012- Finley's Fighters 5K/10K in Connellsville, PA

Ohio

September 22nd - Abigail's Angels charity walk in Wintersville, OH

New York

September 27th 4th annual Bella's Buddies Golf Outing, Long Island

November, 2012 - TBA at Tug Hill Vineyard in Lowville, NY

Massachusetts

September 2012 - Bunco Night
October 2012 - Spaghetti dinner, Acton, MA
November 2012 - Bunco Night

Europe (Candle in the Dark

October 21st - Festival Concert

Generous Donor

This past June, a personal donation to "Eye Love Cecilia's" fundraising team was made from a close family member for the RDH12 Fund for Sight. That donation was **$250,000.** We would like to thank this generous family member for helping us take a giant leap toward our goal for a cure.

"Never doubt that a small group of thoughtful, committed, citizens can change the world. Indeed, it is the only thing that ever has."
— Margaret Mead