

November 2011

RDH12 Fund for Sight

Quarterly Newsletter

Focusing on a Cure

VOL. 1 ISSUE 1

*“I’m not telling you it is
going to be easy, I’m telling
you it is going to be worth
it.”*



*The RDH12 Fund for Sight
Focusing on the Cure*

www.rdh12.org

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This year, our goal is to raise \$250,000 for the next phase of research. We have raised about \$150,000 toward that goal so far. We hope to have the entire \$250,000 to Dr. Bennett at the University of Pennsylvania by March, 2012. Please consider donating or attending a fundraiser in your area to help us reach this goal. The only way our children have any hope for a cure is to raise the money.

In July, 2010, four families met at the Lebers Congenital Amaurosis (LCA) conference in Philadelphia. We immediately bonded over one thing we all had in common – young children with LCA caused by mutations in the same gene, RDH12. This conference was both an exhilarating and frustrating experience for each of us. We saw reports of new therapies being made available to some LCA patients that were changing their lives and giving them their vision back. What had looked like an assured future of blindness for each of our children didn't look like it was as certain as it once was. They didn't have to go blind. That is when we decided to form our RDH12 Fund for Sight. Today, a little over one year later, we are now 10 families and 12 children strong.



We have raised over \$220,000 for RDH12 research since 2010 thanks to the support of all of you. Our family and friends, and even sometimes strangers, are the reason that our children will have a chance to remain sighted people. There is no way to thank everyone enough for what has been done for our families. We hope that these quarterly newsletters will keep you up to date on what our fund has accomplished thanks to your wonderful donations. Awareness is key, money buys the research, and love for our children finds the cure.

Life is 10% what happens to you and 90% how you deal with it.



Feature Family

The Fiore Family



Bella is our oldest child. She is currently 6 years old, and she has a younger sister Ava and a younger brother Anthony who are almost 4 years old. We live in Syosset, NY, and most of our family live near by. When Bella was about 3 years old we started noticing some very peculiar things with her eyes. For one, she was very cautious about going down steps. Also, she was very cautious about transitioning from one step to another. For example, if she was on the lawn, and she came to the sidewalk, she would take that step with a lot of hesitation. In the spring of 2009, during an Easter egg hunt, she had a difficult time finding the eggs. At this point, knowing that Bella's mom has some pretty thick glasses and poor eyesight, we decided to take Bella to the eye doctor. Within 60 seconds of the eye doctor looking into her eyes he said "something is very wrong with her retinas." There was no family history of problems. The doctor gave us some opinions about what it could be – Leber's, retinitis pigmentosa, or even an infection.

This led us to make an appointment with the retina specialist. There was definitely some denial, and hoping that the doctor was mistaken. There was no way we could believe that our daughter was going to go blind. In July, 2009, we met with the retinal specialist, and after many tests he told us "your daughter has a retinal disease, it is mostly likely genetic. I think it is Lebers Congenital

Amaurosis, but it might not be". An ERG was performed which measured retinal function and Bella has basically none. She had very little vision. The doctor said there was no cure, and not much we could do. We had Bella genetically tested to confirm she really had LCA. The University of Iowa told us that they were 60% sure Bella had LCA but they wouldn't be sure until the genetic test came back. In May, 2010, while we were in Disney World, the University of Iowa called and said that Bella had RDH12 LCA. We were relieved to finally know the answer. We checked our other two children, and they are not affected. In the summer of 2010, we planned our first big fundraiser on Bella's behalf. We had a golf outing that raised over \$15,000. We met the other RDH12 families at the LCA conference in July 2010 and the RDH12 Fund for Sight was born.

We started a group called Bella's buddies and we hold two big fundraisers a year – a golf outing in the fall and a dinner dance in the spring. So far, we have been able to contribute about \$60,000 a year to our Fund.

Bella is in first grade now, and her eye sight has not gotten much worse. She is night blind and she doesn't have any peripheral vision. She is considered legally blind. She is a typical girl, and she is doing well, but her eyes are a little special.

We must be willing to let go of the life we have planned, so as to have the life that is waiting for us – joseph campbell

Meet our kids

*What is better than putting faces to a name?
Here are the sweet kids we work so hard for.*



Darius Hormozdyan: 3 years old from Virginia. Diagnosed just this past January. He attends day care, and he gets around on his own at this point without a cane. His vision is 20/260 in both eyes. He gets vision services from the school.



Ben Olsen: 5 years old from Illinois. He was diagnosed when he was 3 years old. Ben uses a cane to get around school and is learning braille. His vision is 20/100 in one eye and 20/4,000 in the other. He does not have peripheral vision and struggles with depth perception. He enjoys swim lessons and drum lessons. He can ride a bike and plays at the park.



Marie Zaknoun: 11 years old from Illinois. She was diagnosed at age 2. She is now in the 6th grade and has been learning braille since she was in 1st grade. She uses a cane for safety. She has an aide at school and her books are enlarged. She likes to ride her bike, swim, and gymnastics. She also loves going to the movies and bowling.



Aiden and Gabby Mann: Aiden is 7 years old and Gabby is 2 years old from Arizona. A brother and sister both affected. Aiden loves all things related to rocket shuttles and space. He has only 5% usual vision in one eye and light perception in the other. Gabby is in OT, speech and feeding therapy. She loves animals and blocks and baby dolls.



Bella Fiore: 6 years old from New York. She is night blind and doesn't have any peripheral vision. She has 20/50 vision in one eye and 20/150 in the other. She is learning braille and loves to sing and dance. She is involved in Daisy Scouts, soccer and swimming. She has a TVI 3 times a week at school and an orientation and movement specialist 1 time a week. She has all of her assignments enlarged.

Website: <http://www.bellas-buddies.com/>

Meet our Kids



Abigail Kinney: 7 years old from Ohio. She is in the 2nd grade and enjoys tap, jazz, tumbling and ballet. She participates in Girl Scouts. She attends Sunday School and is a member of her church choir. Abigail was diagnosed in 2007 when she was 3 years old. Her vision is about 20/250 in one eye and 20/350 in the other. She has no peripheral vision. She uses a CCTV that enlarges her work at school and she has an aide who teaches her braille an hour each day.

Website: www.abigails-angels.com



Bill and Lily Pryor: Bill is 7 years old and Lily is 5 years old from South Carolina. A brother and sister, both affected. Bill enjoys football and both children did karate last summer. Night vision is an issue for both of them as is bright sunlight. Bill and Lily have their homework enlarged. They are both in beginning braille instruction and have a vision instructor once a week.



Raphael Deboeck: 5 ½ years old from Belgium. He was diagnosed in June 2009 when he was almost 3 ½ years old. He is now in Kindergarten and has an aide. He does not have any peripheral vision and is night blind. He enjoys swimming and running. He also loves legos and watching movies. Raphael's parents developed our European sister fund: The Candle in the Dark Vision Research Fund".

Website: <http://www.candleinthedark.be/>



Finley Pletcher: 5 years old from Massachusetts. She was diagnosed in July 2009 at 3 years old. She has 20/200 vision in both eyes. She has braille instruction 3 times a week, has an aide at school, and is also learning to use a cane. Finley is missing her central vision. She can see at night, but that is significantly diminished recently. She plays soccer, and is learning to play the piano. She loves to swim, play with dolls, and ride her bike.

Websites: www.finleyfighters.com
www.pletcher5journey.blogspot.com

Turn your face to the sun, and the shadows fall behind you.

www.rdh12.org

Dr. Bennett Update

Thanks to a grant by the RDH12 Fund for Sight to Dr. Jean Bennett at the University of Pennsylvania the first step in the development of a gene therapy has been successfully taken. Dr. Bennett's lab has built a virus that expressed the gene RDH12. It is this virus that will eventually be used to infect the cells of the retina in our children in order to give them back the gene that they are missing. If the cells of the retina can start to produce the RDH12 they are currently missing, it is highly likely that the progression to blindness will be stopped. But for now, we have the virus and Dr. Bennett's lab has shown that isolated cells grown in a petri dish can be infected by the virus to express our gene.

The next step is to show that virus can correct a vision deficit in an animal model of our children's disease. To do this, Dr. Bennett's lab has established a colony of mice, that like our children, are missing RDH12. Right now the lab is working out the best method to measure the impact of the gene therapy in these mice. Once they know that, they can begin testing the treatment and demonstrating how it might work in the children of RDH12-LCA.



Dr. Debra Thompson



Dr. Jean Bennett

At the same time that Dr. Bennett is progressing the RDH12 Fund for Sight effort, we received some very exciting news. The foundation Fighting Blindness awarded \$8.2 million in grants for the development of new gene therapies for LCA. Among those awards was a grant to Dr. Debra Thompson at the University of Michigan for the development of gene therapy in RDH12. Dr. Bennett has already reached out to Dr. Thompson about the possibility of collaborating on the development of the RDH12 gene therapy, including being open to providing Dr. Thompson with the virus that her lab built. This means that the work of RDH12 Fund for Sight and Dr. Bennett would significantly jump start the FFB-funded work thus speeding the delivery of the cure we are all hoping for. Now that the RDH12 effort has received the major backing of FFB, we have even more reason to believe we will be successful.

In March, 2011, the RDH12 families went to the University of Pennsylvania to present Dr. Bennett a check for \$70,000 to help cover the first phase of research. And in March, 2012, we hope to return to her again with our \$250,000 check so that she can start the second phase of research which puts us one step closer to the cure we desire for our children.



Upcoming Fundraisers

And recent past fundraisers

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Upcoming Fundraisers

Pennsylvania:

Connellsville area:

Christmas cookie sale: December 3-4, 2011. Our non-profit will be picked up by a local business.

Dinners in the dark weekend. February, 2012. Time and exact date TBA.

Massachusetts:

Littleton area:

Dinner in the Dark weekend. January, 2012. Time and date TBA.

Fundraising dinner – March, 2012. Time and date TBA.

New York:

New Hyde Park:

Dinner dance and Chinese Auction. Saturday, March 24, 2012. 7pm at the New Hyde Park Knights of Columbus.

Europe:

Gala concert in Namur - November 4th, 2011
November 8th, 2011 – Candle in the Dark (our European sister fund) will receive a check for \$2831 from the Innerwheel Deinze

Candle in the Dark Gala concert in Brussels – January 23, 2012

Please consider coming to one of the events listed above. If you would like to host a fundraiser for a family in your area, please contact that family with the details. We appreciate your support.

Fundraisers since August, 2011

Pennsylvania:

August 20, 2011- Bike/Walk/Run. Raised \$15,000.

Ohio:

September, 2011 – Chicken grill and Chinese raffle. Raised \$7,000

September, 2011 - Gospel Sing for Sight. Raised \$2800

October, 2011 – bake sale raised \$407

New York:

October, 2011 – Golf Outing. Raised \$28,000

Virginia:

Dr. Tahmtan Hormozdyaran - \$10,000 raised.

On going fundraisers:

Ohio

Wristband and bracelet sales

Pennsylvania and Massachusetts:

Wristband, bracelet, and braille product sales

We thank everyone who participated in these fundraisers to help us get closer to that \$250,000 goal we have this year. We could not do it without your support.

There is no telling how many miles you will have to run while chasing a dream.