

# 4Q 2014



## 4Q 2014 Newsletter for One Breath, One Hope Inc.

### Holiday Giving Opportunity!

The holidays are here, and with them come the joy of giving.

One Breath, One Hope and Boston Children's Hospitals' PCD Center have been fostering a wonderful relationship over the last year. The staff at the PCD center are amazing and we are so happy to be able to work with them to help the rare disease community.



**Boston Children's Hospital**  
Until every child is well™

This year, OBOH would like to give the gift of hope to some of the brave patients that the PCD Center serves. We will be packing up small drawstring backpacks filled with a warm blanket for those cool hospital rooms, a game or book, a bottle of water, and a small treat. These bags will then be passed out to patients requiring hours or even days of treatment or testing at the PCD center. With a gift to One Breath, One Hope of \$25, we will send one of these bags, in your name or that of a loved one, to a child who needs to know they are not alone in their fight.

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To make your tax deductible gift, please mail a check along with the name you would like listed on the bag, or email [info@onebreathonehope.org](mailto:info@onebreathonehope.org) with the information and donate through PayPal on our website at [www.onebreathonehope.org](http://www.onebreathonehope.org)

Thank you for choosing the gift of HOPE!



## Happy Holidays From OBOH

On behalf of the Board at One Breath, One Hope I would like to wish you and your families a very happy holiday season. It has been a wonderful time for u at OBOH and we are so grateful for all the support we have had over the past year. We became an official organization, received our 501c3 non profit status, and were able to host some great events and meet some amazing people. We would not be here without the efforts of our OBOH family and want to thank you for your continued involvement in our cause to help the rare disease community. We hope that during these holiday times, you take the opportunity to spend precious moments with your loved ones, indulge in holiday cookies, and remember that you are always in our thoughts and hearts.

**Meghan Montana**

**CEO - One Breath, One Hope Inc.**

## Smile!

We are very excited to announce that we have a new way to donate to OBOH that is easier than ever! We have become a member of the Amazon Smile program. This means that any time you shop on Amazon through the Amazon Smile site, a portion of your purchases will go to One Breath, One Hope Inc. automatically when you choose us as your designated charity! Simply go to the link below, or visit our website, click on the Amazon Smile icon and do your shopping. When you checkout, .05% of your purchase, every time you shop through Amazon Smile, will go to OBOH!

<http://smile.amazon.com/ch/46-4223599>

## Relief Report

Because of your generous support of our organization, OBOH was able to host a great Harvest Pie Bake Off and Fundraiser in October. It was a fun filled day of games, prizes, food, and PIE! Thank you to all who attended, all who gave their time to help, and to all who generously donated. We could not have done it without your support. Thank you!

We would also like to thank those making “In Memoriam” gifts that help support the rare disease community. We remember Jake Mahon, Sharon Mahon, James Simler, Nancy McKenna, Vincent Guarniere, and Rita Chaghatzbanian. Our thoughts are with their families.



# In The Spotlight...



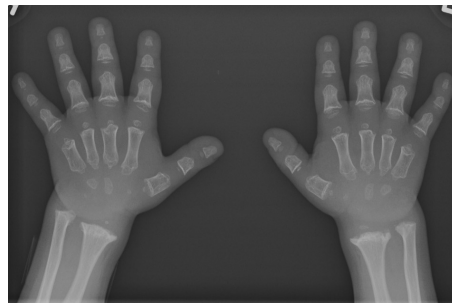
Meet Jamison! Jamison has a rare type of dwarfism called pseudoachondroplasia (sooo-doe-A-KON-droe-plays-eee-uh). Dwarfism occurs in about 1 out of 10,000 births, but pseudoachondroplasia occurs in about 1 out of 30,000 and is usually diagnosed in children around age 2. Pseudoachondroplasia is caused by a mutation in the cartilage oligomeric matrix protein or COMP gene. This rare disease causes the bones to grow slowly and the cartilage at the tips of the bones to not harden the way a normal bone does. This causes pain in the joints and leads commonly to osteoarthritis. Most children with the disease have to go through bone alignment surgeries and hip and/or knee replacements as they grow into their 20's.

Jamison was an averaged sized baby when he was born and even wore clothes about one size larger than what was for his age until he was about 18 months old. His parents started to notice his growth rate to slow early on, but it took until age 5 for the pediatrician to really give the issue some attention. He had basically fallen off the growth chart and the discrepancy between Jamison's height and the average growth curve was growing.

At that time and his parents took him to some local specialists: an orthopedist, two geneticists, an endocrinologist, physical therapists, and the pain and palliative medicine team. It was determined that Jamison had dwarfism, but the kind of dwarfism could still not be determined. Jamison and his parents travelled to New York City and saw a team of dwarfism experts who eventually found the mutated gene. It is a mutation shared with just ONE other (known) individual in the world. The family also travelled to Washington D.C. to attend the Little People of America (LPA) convention where they saw additional orthopedists and geneticist who could still not shed any light on the diagnosis. However, while there, they found and connected with invaluable resources for knowledge and support in the LPA group. In the meantime Jamison's parents contacted the researcher in England who knew the one other man with this mutated gene. He referred to Jamison's case as "classic pseudoachondroplasia". At the urging of LPA friends, they took Jamison to the top orthopedic expert in dwarfism in Delaware and here is where they finally received a confirmed diagnosis of pseudoachondroplasia.



Top:  
Bones of an average  
6 year old.  
Bottom:  
Jamison's bones at  
age 6.



The process for diagnosis was long and difficult as is true for most rare diseases. Jamison's parents were faced with more questions than answers and the struggle of being their child's only advocate. They have struggled with finding doctors, sorting through treatments, dealing with insurance issues and the social stigmas that come with being "different". They have travelled hundreds of miles and put their child through endless tests, all while trying to stay in a positive mind-frame and live in the present, not fearing for the future. Jamison and his family have a long journey ahead of them, but they have hope that the quote from Henry David Thoreau is true: "If one advances confidently in the direction of his dreams, and endeavors to live the life which he has imagined, he will meet with success unexpected in common hours."

If you or your child would like to be featured on our "In The Spotlight" section for the next newsletter, please email [info@onebreathonehope.org](mailto:info@onebreathonehope.org)!

## Quarterly Connecticut Support Group!

Do you live in the Connecticut area? Would you like to have an opportunity to meet and talk with other PCD and Rare Disease families? OBOH would like to start hosting a quarterly support group that will meet at different locations around the state. Please let us know if you would be interested in joining this group to have an opportunity to chat with people who know exactly what you are going through.

Email [info@onebreathonehope.org](mailto:info@onebreathonehope.org) to be added to our list!



## **One Breath, One Hope Inc.**

6 Huntington Ridge Rd.  
Wallingford, CT 06492  
\*New Address\*

Phone: 203-640-2386

E-mail: [info@onebreathonehope.org](mailto:info@onebreathonehope.org)

## ***Our Mission:***

**One Breath, One Hope Inc. is a Connecticut based non-profit organization serving the PCD (Primary Ciliary Dyskinesia) and rare disease community by providing financial support for treatments, advocating for medical research, and helping families get in touch with the information and support groups they need.**

## ***Visit Us Online...***

Website: [www.onebreathonehope.org](http://www.onebreathonehope.org)

Friend us on Facebook: One Breath, One Hope Inc.

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