

Spring 2004

ican

International children's anophthalmia network

www.anophthalmia.org

# The Conformer

ican/ 1-800-580-ican/ican@anophthalmia.org

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## Message from the President

### **Hello and welcome to new members!**

We are looking forward to seeing everyone in the Spring of 2005 in Philadelphia. Plans are underway to have another successful conference. Please remember to let us know if you have any suggestions for the conference.

As you will see in this issue-fundraising for our cause is now essential. **ican** will not survive without more donations-please help us in any way you can! We are counting on all of you to help.

There are exciting new research developments which we hope will take us further than we were able to before to understand the cause of A/M. We know that research can take time and we hope the new process will help provide more of us with results.

Thank you to all of those who help **ican** and we hope you enjoy this issue. A special thank you to Pat Renfranz who put this issue of The Conformer together.

## Fundraising: A Priority

**ican** is expanding daily. We must commit to increase our income in order to continue to provide support to families as well as to enhance research projects. **ican** funds can assist families in need and enable important research projects to determine the cause of A/M.

**You can help!** Call or write your local Lions or Lioness Club and ask to speak at their meeting. We have done this at several branches around Philadelphia and successfully raised money. This group is very supportive of our efforts. The discussion can be as simple as telling your family story and a brief overview from **ican** (take a brochure or the web pages with you). It will only take a few hours of your time and the money raised will significantly further research. We cannot do this alone. We need all parents to get involved! If you are willing to do this contact:

Tanya Bardakjian at 215-456-8726 and she will help you plan the talk.

## Melatonin might help sleep disorders

Melatonin is a hormone produced by the pineal gland in the brain. Several studies suggest that it can help regulate sleep patterns. The level of melatonin produced by the gland is tightly regulated by light: When the sun sets or when we turn off the lights at night, the level of melatonin increases, and that helps us sleep. When the sun rises or we turn on the lights, melatonin levels drop.

Blind adults and children, especially those with no light perception, are more likely to suffer from sleep disturbances than sighted adults, probably because melatonin production is not regulated by light. Anecdotally, a number of children in **ican** families suffer from sleep problems – unfortunately, this makes sense, because children with bilateral anophthalmia don't have the means to perceive light, and hence the regulation of sleep cycles by melatonin is likely to be disturbed.

A number of recent studies suggest that treatment of adults or children with sleep disorders with melatonin

can improve sleep patterns. In one study, children with various neurological issues or blindness who had sleep pattern disturbance were treated with melatonin. Most of the children slept longer and more continuously when treated with melatonin; the treatment was most effective in the visually-impaired children. In another study, a group of blind adults received melatonin, and the vast majority developed normal sleep patterns.

Little is known about the safety of melatonin treatment in children or about the long-term effectiveness of treatment. If your child appears to be suffering from sleep disturbance, it is critical that you talk with your pediatrician. You might ask for a referral to a sleep center, where specialists can assess the situation and make recommendations.

For more information, go to

[www.ChildHealthMonitor.org](http://www.ChildHealthMonitor.org)

[www.nlm.nih.gov/medlineplus/sleepdisorders.html](http://www.nlm.nih.gov/medlineplus/sleepdisorders.html)

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“Blind people have more sleep disorders than sighted people...People with no light perception have a higher incidence and a greater severity of sleep disorders than blind people with some degree of light perception...”

Charles A. Czeisler, M.D. Ph.D., speaking to a convention of the American Council of the Blind in 2002

## ican Conference 2005: See You in Philadelphia

Plans are underway for the next **ican** conference. It will be held in Philadelphia in the Spring of **2005** in Philadelphia. We need volunteers to assist us in planning the conference. Please consider helping. Distance is not a factor. We are looking for suggestions for topics, speakers and activities as well as with conference logistics.

If you are willing to participate in any way, big or small you can contact:

Tanya Bardakjian  
215-456-8726  
[bardakjian@einstein.edu](mailto:bardakjian@einstein.edu)

***Let's work together to make this conference a huge success!!!!***

## Feature Family- The Hughes

My name is Patrick John Hughes. My son, Patrick Henry Hughes, was born March 10, 1988. Here is a recollection of his first 16 years.....

Patrick Henry Hughes was born March 10, 1988, after a perfect pregnancy. Patrick's mom followed all the rules for expectant mothers. No alcohol, no tobacco, no caffeine, a healthy diet. She had an office job for a major brokerage firm, and our home was as clean as we could make it while each of us were growing our careers. We did make a trip to Hawaii about 8 to 10 weeks into the pregnancy. I often wonder if our tour of the pineapple fields or something else there had an effect.

The delivery room was hectic (they all are). The first word we recall hearing from the staff was "anomalies". To me, he just looked like a newborn baby, covered in muck and crumpled up. I recall shortly after the delivery a doctor pulling me aside and offering to pray with me. At this point, I still wasn't sure what we were praying about - pretty much convinced that nothing could be as wrong as first indications said they were.

As the hours passed, and the doctors examined Patrick further, the information came trickling in. Patrick's eyes were mostly not there, and his arms and legs were unable to straighten out all the way, as well as the limbs being abnormally short. His hips were not in socket, though that seemed to be something a little more common. Six months later, surgery would determine that they couldn't be put back in socket because there were no femoral heads to put into a socket which hadn't formed.

Because Patrick was jaundiced, his mother and he spent an extra few days in the hospital. These extra days probably gave the doctors more time to determine what other anomalies Patrick may be hiding beneath the surface. Fortunately, to this date, there are no more. Sometimes I worry that another problem will arise. There was back surgery in 1999 to correct scoliosis from years of wheelchair use, but, other than the wheelchair and the blindness, Patrick is probably the most healthy of any of our family. Never ill, always in a good mood, and an unusually good disposition all of the time.

The first couple of years after Patrick's birth, we spent many days traveling to different doctors in different cities trying to find out what caused

Patrick's condition - officially known as bilateral anophthalmia with pterygium syndrome and congenital bilateral hip dysplasia (now that we are use to Patrick and know him, I think it sounds worse than it is). One of the doctors I remember most was a geneticist who took Patrick's case history to a world wide convention of his peers trying to locate another case like Patrick's. There was none.

During these early years, Patrick spent days with his paternal grandparents while his mother and I chased our careers. Patrick had various braces on his arms and legs during these years as doctors tried their best to straighten his limbs and much as possible without surgery, which became less and less of an option as we all learned more about Patrick. These years were great for Patrick and for our relationship with my parents. We all grew much closer because of Patrick. And to this day, Patrick is the greatest ambassador of goodwill we have ever known. Through Patrick, neighbors have become friends faster, and we have met hundreds, if not thousands, of celebrities and well-wishers through Patrick - fans of Patrick that we would never have known had it not been for this special young man!

I first placed Patrick on a piano when he was an infant. There were times when I knew Patrick was fed, changed, and should be happy, but, as with most babies, he just seemed to want to cry. I found that if I laid him on our piano and played, he would immediately be quiet. To this day I wonder if he enjoyed it, or if it just scared him to death. Who knows?

At nine months old, I sat a high chair at the piano and let Patrick touch the keys. Taking the tray off the chair enabled Patrick to play while keeping him supported in the back and on the sides. And he loved it! We did simple listen and play exercises, where I would play notes and he would find them. Patrick was never one to bang on the piano. He always seemed to be studying which keys made what sounds. By the time he was two years old, his mother and I could work around the house or prepare dinner, and listen to Patrick play his Sesame Street tapes, complete with narration (though spoken like a two year old) and simple melodic phrases on the piano.

The piano would become a great tool for Patrick. He has performed thousands of times during his years, beginning when he was just 4 years old, playing for **con't on page 5**

## Sources of Braille Books for Children

*Children with significant visual impairment often use Braille as their medium for reading and writing. However, it can be difficult to obtain children's books embossed in Braille. Here are some sources of embossed books :*

### National Braille Press:

[www.nbp.org/](http://www.nbp.org/)

Children's Braille Book Club features a new print/Braille book every month. They also have a large selection of books in Braille for kids. To encourage Braille literacy, NBP also sponsors a Braille Book Bag program in conjunction with Seedlings Braille Books for Children. Each bag contains:

- An age-appropriate print/Braille book for three age groups: birth-3, 4-5, and 6-7 in English or Spanish;
- A Braille primer for sighted parents entitled [Just Enough to Know Better](#);
- A colorful print/Braille place mat;
- Print/Braille bookmarks;
- [Because Books Matter](#), a guide for parents on why and how to read books with their young blind child;
- A gift coupon redeemable for another print/Braille book or Braille/large print playing cards.

Phone: (617) 266-6160  
Toll-free: (888) 965-8965

### Seedlings Braille Books for Children:

[www.seedlings.org/](http://www.seedlings.org/)

Seedlings provides high quality, low cost braille books for children, with nearly 600 titles from which to choose. They offer

- Print-Braille-and-Picture Books
- Print-and-Braille Books (Easy-readers where the print and Braille words are matched line for line. No pictures.)
- Braille Books-Fiction and nonfiction

Phone: (734) 427-8552  
Toll-free: (800) 777-8552

### Braille Institute:

[www.brailleinstitute.org](http://www.brailleinstitute.org)

Dots for Tots program: multisensory story books in print and Braille designed for visually impaired preschoolers. Each kit includes

- A popular picture book with grade 1 Braille overlays
- A descriptive audiotape of the story
- Hard-plastic toys that represent characters or objects in the story.

Free books subscription program: Popular books for kids 5 to 18 embossed in Braille.

Toll-free: 1-800-BRAILLE (272-4553)

### American Printing House for the Blind:

[www.aph.org](http://www.aph.org)

APH has a number of products designed to promote literacy in children of all ages, as well as numerous other items to help blind children learn. They offer the *On the Way to Literacy* series of storybooks (print and Braille, with tactile illustrations). They also offer a good number of books that combines Braille with conventionally published, illustrated books for children.

Toll-free: (800) 223-1839

### American Action Fund:

[www.actionfund.org/](http://www.actionfund.org/)

The AAF philosophy is that "Blind kids want the same things that sighted kids want," including reading the newest popular books. They sponsor a Free Braille Books Program, which provides children a new book every month from a popular children's reading series. The titles published every month are the same titles that are available in bookstores everywhere.

Phone: (410) 659-9314 ext. 361

## Membership Dues

In January, you should have received an invoice from **ican** asking for the Membership dues. The fee is \$25.00 per family. This covers expenses for newsletters, mailings, the 1-800 number and the website. In the past we have not been billing regularly for the annual dues. However, due to increasing services and the costs associated with them we must be diligent about dues collections. We will be sending an invoice at the end of every year. Please help us continue to provide support to you and other families by paying your dues. Please note that if

you cannot afford the dues simply contact us and we will make every effort to provide you with member privileges

Contact us by calling  
1-800-580-ican or e-mail  
ican@anophthalmia.org

## The Hughes Family con't

people attending charitable campaign kick-offs that I would speak at about raising a blind, disabled child and the obstacles and successes we ran into. These benefits were mainly for Metro United Way, and a local telethon called the Crusade for Children. However, he now also performs for VSA International, our local school system, and many other groups, helping to motivate people to donate funds to all these worthwhile causes.

Over the years, Patrick has become an exceptional pianist. He currently studies with a Julliard grad, and is going to play this summer in Washington, D.C. He plays Mozart, Bach, Beethoven, Brahms, and many others flawlessly. He has performed twice on the stage of the Grand Ole Opry (he is a lover of Country Music), as well as in Florida, California, New York, Indiana, Kentucky, Tennessee, and a couple of other states I might be leaving out.

Patrick also has a great voice. He is an all-state choral tenor, and has performed with his school choirs in New Orleans and in the Spring of 2004 will perform in Quito, Ecuador. His talents also include the trumpet. Patrick has played in All-State Band, Eastern Kentucky University Honors Band, as well as All-County Band and plays in his school's pep band and concert band. And did I mention, while handling all of these musical functions, Patrick has maintained a 4.0 GPA since the 6th grade. He is now a sophomore.

Patrick's list of awards, honors, and accomplishments is worth mentioning. Besides the above listed performances and accomplishments, Patrick was an official Olympic Torchbearer in 2001. He has received Presidential Academic awards from two different Presidents (1999 and 2002). He speaks fluent Spanish and was selected to appear in Who's Who Among American High School students as a freshman. He performed on the Maury Povich Show, the University of Louisville's Coach Rick Pitino show, and has sang with country music stars Faith Hill, Lonestar, Chad Brock, Pam Tillis, Bryan White, Lane Brody, and many, many others.

At this point in time, Patrick Henry hopes to attend college in Nashville, Tennessee, thus putting him close to the heart of Country Music. He wants to be a Country Music star. His fallback ideas involve being an ambassador to a Spanish speaking country, and/or an interpreter of Spanish. His major will either be musically related or involve the study of foreign language. Because of Patrick's anomalies, some personal assistance is needed, which means his mother or I will probably have to attend college with Patrick - both a chore and a pleasure. This latter fact makes a local university a distinct possibility as well as a Nashville based University. We'll see.

With Patrick, you never know....

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.org

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We're on the Web!

See us at:

**www.anophthalmia  
.org**

## Research Update

The DNA research project has been successful thus far, with some mutations identified in a few patients. However, the researchers feel that the amount of DNA obtained from the cheek swabs was not enough to perform all the analysis warranted. Therefore, we will be collecting **blood** samples from all interested/willing parties.

Lymphoblastic cell lines will be established. This is a technical term to describe a source of DNA that is unlimited. This means that many researchers interested in A/M can access the DNA (all samples are anonymous) and complete the analysis instead of having partially tested samples because of a lack of DNA. Also, as new information or genes are discovered, the DNA can be retested for the new discoveries. You will be receiving detailed information about this new procedure shortly, once all funding has been approved. We hope you will consider providing us with a sample which would be invaluable to the further research into the causes of A/M. If you have any questions please contact

Tanya Bardakjian,MS,CGC  
Coordinator, A/M Research Project  
215-456-8726 [bardakjiant@einstein.edu](mailto:bardakjiant@einstein.edu)

### **Association of A/M and Tracheoesophageal atresia (TEF)**

Recently Dr. Adele Schneider and Tanya Bardakjian identified 4 cases of this association (A/M and TEF) from the A/M Registry. This is a rare association with only nine other cases reported in the medical literature. An article has been written about these cases and has been submitted for publication to The American Journal of Medical Genetics. Also, we have been contacted by a researcher who feels he has identified a gene that leads to A/M and TEF. If you or your child has these two conditions, with or without other findings please contact Tanya Bardakjian for inclusion in the study. This information will be very helpful to research the causes of these birth differences.

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