

March 2006

The Conformer

ican/ 1-800-580-ican

Message from the President

Hello Everyone, We have a tentative date for the next conference! It will be held in the Spring of 2007 at the Florida School for the blind. We will get more details out as soon as we have more information. We are open for suggestions from anyone who has ideas. **ican** is a family-run support group and we rely on all of you for input and assistance. **ican** is run on donations and membership fees therefore we ask all of you to help with fundraising ideas and events. We need everyone to help out with fundraising to make this conference a success. We look forward to hearing from you!

Sherry

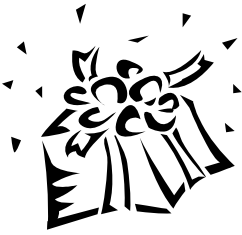
2007 Conference Plans Underway

We are looking forward to seeing everyone in the Spring of 2007. Our next conference will be held in SUNNY St. Augustine, Florida! The conference planning has just begun but we are in negotiations with the Florida School for the Blind to host part of the conference in their beautiful facility just minutes from Old Town St. Augustine. **We welcome any suggestions. If there is anything you would like to see at this conference, please let us know. After all, this is your conference too!**

Funds are currently being raised to set-up a conference scholarship program to assist families with travel costs. We want to make sure everyone can come to learn and connect with other families.

We will keep you informed as plans continue. You can check the website for updates as well.

See you in 2007!!!



Would you like to help ican? Then SHOP ON-LINE through iGive.com

That's right, go ahead and buy something for yourself from over 500 name-brand stores through www.iGive.com. First join through www.iGive.com and identify ican as the organization you want money donated to. Then shop in the mall at iGive.com. There is no cost to you! Urge your family and friends to join as well. Membership is free and your privacy is guaranteed.

This is an easy way to help raise much-needed funds to continue **ican's** mission.

FUNDRAISING CAMPAIGN: COOKBOOKS

Send in your favorite recipes:

ican's board members have been brainstorming on fundraising ideas so that **ican** can continue its mission to keep families informed and connected. In our last newsletter we included a fundraiser letter that we hope some of you have had the chance to use. We know for sure two of our members sent it out during Christmas and we have gotten excellent results. Our next idea is publishing a cookbook.

We have a very diverse group and the end result of this idea can be phenomenal. We ask that all of you help us by sending in some favorite recipes (the more the better). The book will include a kids' recipe section so if you have any recipes geared toward children please send them to us. You can email them to Nelly Gamino at Ngamino@tribune.com - subject: **ican** recipes or if you don't have internet access, mail them to:

ican, Albert Einstein Medical Center, 5501 Old York Rd, Genetics, Levy 2 West, Philadelphia, PA 19141

RECENT PUBLICATIONS

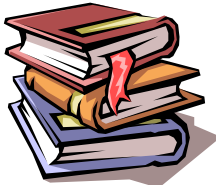
Identification of novel mutations in the SOX2 gene in patients with anophthalmia/microphthalmia. Terri L. Young, M.D., Jie Zhou, M.D., Ph.D., Tanya M. Bardakjian, M.S., C.G.C., Femida Kherani, M.D., James Katowitz, M.D., Nkecha Hughes, M.S., Lisa A. Schimmenti, M.D., Adele Schneider, M.D. Poster presentation at American Society of Human Genetics annual meeting in Salt Lake City October 2005. A2147

Anophthalmia/Microphthalmia Registry: Registry clinical data support extended work-up of individuals with anophthalmia/microphthalmia. A. Schneider, D. Mognillansky and T. Bardakjian. Poster presentation at American Society of Human Genetics annual meeting in Salt Lake City October 2005. A750.

Anophthalmia/Microphthalmia (A/M) Registry: Increased Prevalence of Multiple Births Noted in Children with A/M. T Bardakjian, K Hamman, D Dorsainville, R Keep, A Schneider. Poster Presentation at the National Society of Genetic Counselors, November 2005.

Familial SOX2 anophthalmia syndrome with nonpenetrant parent. A Schneider, T. Bardakjian, M. Hummel, R. Keep, D. Dorsainville, L. Schimmenti, N. Hughes, J. Zhou, T. Young. Presentation at the American College of Medical Genetics Meeting, San Diego, CA March, 2006.

Order your Cookbook today!!



GIFT/DONATION FORM

Your gifts and donations help **ican** continue its mission of providing support and information to families dealing with anophthalmia or microphthalmia, educating medical professionals and encouraging and supporting research.

GIFT-HONOR-MEMORIAL

Gifts are a great way to let someone know how much you care. The listed person or family will receive an acknowledgment of your generosity without the specified amount.

Enclosed is my gift of \$_____

In Memory In Honor

of _____

Please acknowledge this gift to:

Name: _____

Address: _____

City: _____

State/ZIP: _____

All gifts are listed in the newsletter If you do not want it listed please mark the line below.

Please do not list in the newsletter

DONATIONS

We are a non-profit organization and rely on donations to achieve our mission. All gifts and donations are tax-deductible.

Enclosed is my donation of \$_____

Please send Receipt to:

Name: _____

Address: _____

City: _____

State/ZIP: _____

All donations are listed in the newsletter If you do not want it listed please mark the line below.

Please do not list in the newsletter

Donations Received

A special thank you to **Mrs. Merle Santerian** for her **\$5,000.00** donation in honor of the Varela family from Spain.

Additional donations made to **ican**:

David and Rita Kulik
John and Jennifer Justus
Margaret Fragale
John W. O'Neal

Milton and Sylvia Iralson
Carolyn Malatia and Sylvia Iralson
The Carr Family
Kevin Kelly and Associates

Please note that some of you may have made donations to the anophthalmia/research project directly. Those funds are not used by **ican** and therefore are not listed here.

RESEARCH UPDATE

ican

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e-mail:
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We're on the Web!
See us at:
www.
anophthalmia.org

The Genetics Research project has made some exciting discoveries in the past year. We have identified mutations in three children with bilateral anophthalmia and in a mom with no eye findings. The mutations were found in the gene called SOX2. SOX2 is a gene involved in eye development as well as other important systems.

The findings seen in the syndrome include but are not limited to: bilateral anophthalmia or microphthalmia, specific brain differences on MRI, delayed motor skills such as walking and delayed speech. Currently, these are the findings we have seen repeatedly with this mutation. However, we have just started testing so we may actually find a broader picture. Most cases of SOX2 are sporadic, meaning the mutation occurs for the first time in the individual with A/M with no family history. Research findings suggest that about 15% of individuals with bilateral anophthalmia or microphthalmia and 3% with unilateral have a mutation in the SOX2 gene. Based on the findings, a new syndrome termed "SOX2 anophthalmia syndrome" has been described.

Our recent analysis of 25 samples found 4 positive results. Three of the samples were from one family. Two sisters both have bilateral anophthalmia. The older sister has motor and speech delays as well as a seizure disorder. The younger sister is still a newborn therefore, development has not been assessed. Both parents are clinically normal. However, our protocol calls for us to test parents who have children with a mutation. Mom was found to have a mutation in SOX2 but has NO symptoms. This is only the second case reported of a SOX2 mutation in an unaffected person.

Through research we will continue to learn more about SOX2 and other genes associated with A/M. We do know that if a SOX2 mutation is identified both parents should also be tested. SOX2 has a relatively high positive rate in children with A/M, therefore current recommendations for individuals with A/M include testing for SOX2. **This is a new gene and most of your children have not been screened yet.**

If you are interested please contact us so we can help facilitate this testing. Both research and clinical testing are available. Contact Tanya Bardakjian, MS, Certified Genetic Counselor, Coordinator of A/M Research Project at 215-456-8722 or bardakjiant@einstein.edu

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