

What's New with 22q?

July 6—10

2012 Biennial Meeting Lands in Orlando

Eight reasons why you don't want to miss the 8th Biennial 22q Conference...1) to learn about available treatment...2) to discover educational strategies...3) to hear clinicians share the latest in research...4) to bond with other families who live what YOU live...5) to put faces with names in the world of 22q...6) to attend the 22q Barbeque Disney Character Dinner For All ...7) to soak up the magic of *Disney*...8) to learn, share and bond!

The conference, entitled *Detection * Care * Cure – The Magic of Making a Difference*, will indeed be magical as it is set in the most inviting of settings – *Disney's Yacht and Beach Club Resorts* in Lake Buena Vista, Florida, USA.

This superb conference, sponsored and organized by *The International 22q Foundation, Inc.*, will feature presentations from top clinicians and scientists from across the globe who devote themselves to study of the 22q syndrome. They will cover the medical, psychosocial, speech, language, and cognitive characteristics of the diagnosis with an emphasis on treatment.

Congratulations to the 22q Foundation, which is working hard to plan and present the meeting. Visit 22qconference.com for more information.

SEE YOU IN ORLANDO!!!!!!!!!!!!!!!!!!!!!!

22q At The Zoo a Global Success

For the second year in a row, zoos around the world witnessed an extraordinary gathering of 22q families. The *2nd Annual 22q at the Zoo! Worldwide Awareness Day* took place on Sunday, April 22 spanning more than 12 countries on four continents, including 55 cities. An estimated 15,000 participants attended. See pages 4, 5, 6 and 7 for more photos and details.



Philadelphia, Pennsylvania, USA

**Save the Date – 3rd Annual Zoo Day –
May 19, 2013**

Vital funds needed to prove 22q case

Federal Committee Requires Data Before Recognizing Need for 22q Newborn Screen-

Collaborating organizations including The International 22q Foundation, Max Appeal, and the Dempster Family Foundation, are launching a rapid fund raising campaign to fund a study to generate pilot data in support of the need for newborn screening for 22q11.2.

The campaign is in response to a challenge by the US Secretary of Health's Committee on Heritable Disor-

ders and Newborn Screening that contended there is no evidence to support the approval of an application to add 22q screening to the current federally mandated list of newborn screening studies.

In an historic moment on January 26, groups from the United States, Canada, and the UK rallied support for

(Continued on page 2)

2012 Calendar Contest Winners are Picture Perfect Perfection

Congratulations to the 2012 Calendar Contest winners. In addition to those selected to represent the individual months and cover, we were happy to be able to include all the children in some way in this year's calendar. An independent marketing agency picked the winners, a big job since we received the largest number of contestant photos in the contest's history.

Proceeds from calendar sales support the Foundation's mission. Order calendars for yourself, friends, family, and co-workers and for your

child's doctor, teacher or therapist — a great way to get the word out about 22q. Thanks to all for participating.

Our winners are: **January:** Teddy from Missouri, USA; **February:** Carley from Ontario, Canada; **March:** Dominic from New York, USA and Dylan from Georgia, USA; **April:** Clare from Ontario, Canada and Connor from Florida, USA; **May:** Amy from Virginia, USA; Tyler from South Carolina, USA; Zachary from California, USA; and Haley from Utah, USA; **June:** Cayden from North Carolina, USA and

Alex from Massachusetts, USA; **July:** Leenah Rose from Pennsylvania, USA and Lilly from Queensland, Australia; **August:** Tyler from Georgia, USA; **September:** Cassie from Kansas, USA; **October:** Charles from Pennsylvania, USA; Holly from Ohio, USA; and Slade from Louisiana, USA. **November:** Skylar from Mississippi, USA; **December:** Ethan from New Jersey, USA. **Calendar Cover:** Elise from Missouri, USA; Jaxon from Ontario, Canada; Lauren from Arkansas, USA; and Zachary from Missouri, USA.

Delegates Advocate for Funding

(Continued from page 1)

newborn screening for 22q11.2 before the Committee. They represented The Children's Hospital of Philadelphia, The University of Toronto, The International 22q Foundation, Max Appeal, Children's Hospital of Wisconsin, and The Ryan and Jenny Dempster Foundation.

Pilot Data To Be Resubmitted

They discovered that the Committee, prior to any public comments or proponent group presentation, had predetermined that the application to add 22q screening be denied. The Committee cited the lack of pilot study evidence to support the proposal. However, following a convincing presentation by the Newborn Screening Proponents, the Committee agreed to re-hear the proposal once such pilot data is submitted.



These delegates presented compelling arguments to the Federal Committee: Back Row: Michelle Breedlove-Sells, The Ryan and Jenny Dempster Foundation; Carol Cavana, The International 22q11.2 Foundation; Anne Bassett, MD, The University of Toronto; Sheila Kambin, MD, The International 22q11.2 Foundation; Aoy Mitchell, PhD, Children's Hospital of Wisconsin. Front Row: John Routes, MD, Children's Hospital of Wisconsin; Donna McDonald-McGinn, MS, CGC, Children's Hospital of Philadelphia; Julie Wootton, UK Max Appeal. Also present but not pictured was Stu Berger, MD, Children's Hospital of Wisconsin.

New Name Reflects Expanded Reach to 22q Families

The International 22q11.2 Deletion Syndrome Foundation, Inc. is changing its name to The International 22q11.2 Foundation, Inc. Why? Because we are expanding our services to include all individuals and families affected by abnormalities of chromosome 22q11.2, including those with smaller deletions, those with changes in genes within the region such as TBX1, and those with the 22q11.2 duplication syndrome. All families with these diagnoses, as well as those with the deletion syndrome, are encouraged to participate in all events, especially the 8th Biennial International 22q11.2 Conference from July 6-10 at *Disney's Yacht and Beach Club Resorts* in Lake Buena Vista, Florida, USA.

Good Showing for No Show Ball

Thank you to the family members, friends and donors who had a great time "not showing up" at the *No Show Valentine's Ball*. This fundraiser was a great success. While supporting our Ball our donors loved the idea of not having to attend an event. The event raised \$4,000.

Dragonfly Forest Session: July 12-17

Dragonfly Forest, a camp located near Philadelphia, Pennsylvania, USA, will welcome kids ages 7 to 14 with 22q11.2 syndrome during the week of July 12-17. The camp combines the therapeutic impact of camp with a state-of-the-art facility and an innovative program staffed by medical specialty professionals. The International 22q11.2 Foundation, Inc. is proud to join The 22q and You Center at Children's Hospital and The Dempster Family Foundation to support this special camping session. Check the 22q website for more information.

Awareness Campaign On Track

The Tell 22 about 22q Campaign is in full swing. Since its kick-off in September 2011, we have raised over \$10,000 toward the campaign goal of \$222,222. Students from Maryland, USA and New Jersey, USA raised funds through Bar and Bat Mitzvah service projects while spreading awareness. Spread the word. *Tell 22 about 22q!*

Golf Outing Raises \$12,000

Congratulations to Progressive Payment Solutions and Jonathan Kramer on the First Annual Charity/Networking Golf Outing held on May 21 in New Jersey, USA. Though the weather did not cooperate, the 75 registered golfers enjoyed a wonderful breakfast at the New Jersey National Golf Club and proudly supported our Foundation. The event raised more than \$12,000.

Are you a parent who would reach out to another family? Often all that is required is a sympathetic ear or a comforting email to let another parent know he or she is not alone. Please complete the *parent network form* on the "Helping Families" tab at the 22q website to be added to the list of contacts for new families seeking support.

What's New With 22q is published for families affected by the 22q11.2 syndrome, which is caused by a missing or extra piece of genetic material on chromosome 22. Symptoms of the condition include heart anomalies, immune deficiency, cleft palate, feeding problems, speech delays, learning disabilities, and social/emotional issues. Learn more at www.22q.org.

Our Mission

The mission of the 22q Foundation is to improve the quality of life for individuals affected by 22q11.2 syndrome through family and professional partnerships.



**The International 22q11.2
Foundation, Inc.**

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Los Angeles, California, USA

22q At The Zoo! Worldwide Awareness Day 2012 Attracts 20,000 ???

This very special event gave families touched by 22q issues a chance to socialize, network and raise the public profile of the common but poorly recognized conditions of the 22q syndrome. The international event kicked off at 11am in Sydney, Australia and continued westward around the world. See a complete list of the participating zoos on page 6.



Barcelona, Spain



Fort Worth, Texas, USA



Miami, Florida, USA



Atlanta, Georgia, USA



Sacramento, California, USA



Cincinnati, Ohio, USA



South Africa

**Please see
pages 6 and
7 for more
photos of
22q At
The Zoo!
Worldwide
Awareness
Day 2012.**



Spain



St. Louis, Missouri, USA



Philadelphia, Pennsylvania, USA



Houston, Texas, USA

22q At The Zoo! Worldwide Awareness Day 2012 Locations

U.S. Locations

Abilene, Texas
Asheboro, North Carolina
Atlanta, Georgia
Austin, Texas
Boston, Massachusetts
Bridgeport, Connecticut
Cincinnati, Ohio
Columbus, Ohio
Dallas, Texas
Denver, Colorado
Evansville, Indiana
Fort Worth, Texas
Houston, Texas
Kansas City, Kansas
Knoxville, Tennessee
Miami, Florida
New Orleans, Louisiana

Omaha, Nebraska

Philadelphia, Pennsylvania
Portland, Oregon
Sacramento, California
San Diego, California
Staten Island, New York
St. Louis, Missouri
St. Paul, Minnesota
Washington, D.C.

International Locations

Sydney, Australia
Vienna, Austria
Toronto, Canada
Bristol, England
Chester, England
Edinburgh, England
Flamingoland (near Leeds), England

Whipsnade (London), England

Bremen, Germany
Hamburg, Germany
Hannover, Germany
Leipzig, Germany
Munich, Germany
Nurnberg, Germany
Stuttgart, Germany
Wuppertal, Germany
Utrecht, Holland
Dublin, Ireland
Falconara Marittima, Italy
Kimberley, South Africa
Barcelona, Spain
Madrid, Spain
Murcia, Spain
Valencia, Spain



Dublin, Ireland

22q At The Zoo! Worldwide Awareness Day 2012 A Wild Success



Spain



Whipsnade, UK



Asheboro, NC



Spain

Same Name Campaign strives to eliminate confusion among families, professionals

The Foundation is pleased to continue the *Same Name Campaign*. This is an educational effort to unite the 22q community and empower those affected by promoting use of the name currently recognized by the standard chromosome naming system.

One of the Foundation's primary goals is to increase awareness about the 22q11.2 syndrome among professionals and the general public. A major barrier to awareness is the use of several names for what we now know to be the same condition.

This is due in part to the way our understanding of the syndrome evolved. Older terms include DiGeorge syndrome (DGS), velo-cardio-facial syndrome (VCFS), conotruncal anomaly face syndrome (CTAF), Opitz G/BBB syndrome, and Cayler cardiofacial syndrome. Those with these diagnoses all have the same underlying condition: the 22q11.2 syndrome. In fact, genetically, there is no detectable difference in the microdeletions found in people with VCFS versus those with DGS or the other related syndromes.

Why Do We Need A Campaign?

Having a syndrome called by several names has confused both parents and professionals. Parents of a baby diagnosed with DiGeorge syndrome may be frustrated by the lack of information about DGS, not realizing that VCFS and 22q resources (including this Foundation) also pertain to their child. Health care providers may not access all available information if they search for literature under an older name. Finally, having several names splinters precious resources, hampering a united approach to research and support.

Hopefully, this campaign will result in fewer people newly diagnosed with VCFS, DGS, CTAF or other outdated names. The campaign will include:

- Presenting to professionals involved in diagnosis at national and international meetings.
- Distributing a descriptive postcard to genetic counselors and geneticists throughout the U.S. and to new families contacting the Foundation.
- Working toward consensus among professional and support organizations. We already have endorsement of the Dempster Family Founda-

tion, Max Appeal, and The Colorado c22c Group.

- Encouraging publishers of journals to adopt consistent standards for naming the syndrome.
- Linking our website to related groups indefinitely, regardless of what name they use, to facilitate information searches.

How You Can Help

Make the switch to 22q, even if you are used to using older names. We understand families who have lived with a diagnosis of VCFS, DGS, CTAF or a related condition may find it difficult to switch to a different name.

Support the aims of the *Same Name Campaign*. When you describe yourself or your child, be sure to mention the 22q11.2 syndrome.

Educate medical professionals you encounter about the effort to adopt a single name. They represent a crucial gateway to a correct diagnosis name.

Support the Foundation's work. Your donation helps us fund this and other initiatives.

Download the Foundation's *Same Name Campaign* slide presentation. Show it to all who will listen.

Dempster Family Foundation to kick off Awareness Mobile Bus Tour in Orlando

The Dempster Foundation Tour Bus helps bring exposure to a community where an individual or group is arranging grass roots events to raise awareness or funds for 22q. Tour stops are limited to one a month and are arranged on geography and pre-scheduled dates. They can include baseball games, Special Olympics, Children's Museums, Zoo's, amusement parks, events such as Walks, Runs, Races, large community gatherings as well as visits to Hospitals with 22q clinics or personnel dedicated to children with the syndrome.

Are you planning a 22q Family Get-Together, Awareness Event or Fundraiser? The mobile tour could visit your town? Email Michelle Breedlove-Sells, 22qbus@gmail.com.