International 22q11.2 Foundation

Newsletter June 2021

Greetings from the International 22q11.2 Foundation. We hope you and your family are doing well. We are excited to bring you the latest issue of our newsletter with a new look!

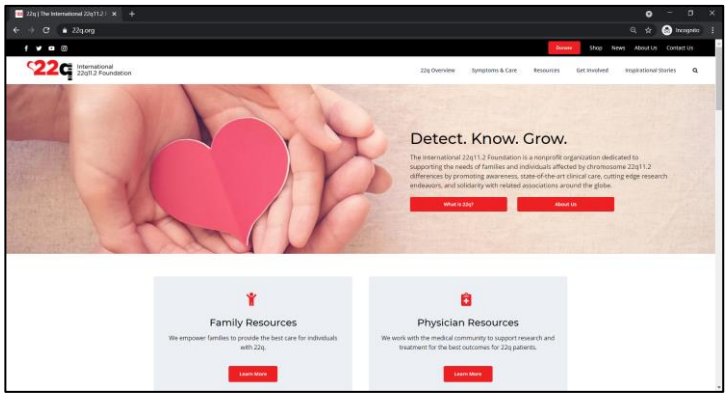
A quick shout out to our very own **Carol Cavana**, founding board member of the International 22q11.2 Foundation and 22q Mom -- for the countless hours she has spent on bringing our new and improved website to life!

From brainstorming the vision, working on content, coordinating experts and input, and working with the website editors to pull it all together Carol has been a tireless advocate for families affected by chromosome 22q11.2 differences everywhere!!!

Carol, thank you, thank you for all you do -- we truly appreciate you, and enormous thanks to your entire family, especially Louis, for sharing you with the world!

Come visit our NEW website!

The International 22q11.2 Foundation has a new look - established in 2003, and acting as the longest serving family organization in North America, our website has been updated and given a facelift, along with our logo, to continue to highlight the important partnerships we have cultivated over the past 18 years in bringing families, healthcare providers, and basic scientists together towards achieving our common goals.



We hope you'll love the new site as much as WE do – there are several new sections, and we think it is even easier for families to use and explore! Enormous thanks to our dedicated **board of directors and medical advisors**, as well as **321 Blink**, the **Scott Schultz team**, and **Michele** and **Chris** for always listening to our concerns, which are reflected in this outstanding website.

We look forward to welcoming you at:

www.22q.org

Quarterly Newsletters are back!

Be sure to sign up on our homepage so you don't miss any important information about 22q deletion and duplication.

Our new Logo

We put our hearts and souls into helping 22q communities around the globe – and not just during our 22q at the Zoo Worldwide Awareness Day Event. For our new logo, the **heart** symbolizes the compassion and love we bring to our mission every single day, while our **"q chromosome"** highlights our unparalleled relationship with world experts who tirelessly volunteer on our Executive and Medical Advisory Boards. We hope you like the logo as much as we do and we promise there is more to come.

A Successful 22q at the ZooM Event

THANK YOU to all our **International 22q11.2 Foundation** friends who made the **11th Annual 22q at the Zoo Worldwide Awareness Day** and **22q and ZooM** a great success!

A special SHOUT OUT to **Julie Wootton** and **Mark Tripp** from **Max Appeal** for spearheading the fun and games on ZooM, as well as to all of the healthcare professionals (**Madeline Chadehumbe** [Brain and Behavior], **Maria Mascarenhas** [Integrative Health], **Donna McDonald-McGinn** [Genetics and Same Name Campaign], **Ed Moss** [Education and Transition to Adulthood], and **Kate Sullivan** [Immunology and COVID-19]), special presenters (**Martin Kennedy** [Quizzo], **Tracy Heninghan** [Story Time], **Max Keir** [Sing Along], **Tessa Koller** [Learn to Draw], **Michael Weinberg** with a touching solo performance, **Chris** and his amazing critters at the **Dudley Zoo**, as well as the **Philadelphia Zoo**), **international 22q coordinators**, and all of the **22q families** who joined us on ZooM representing >20 countries around the globe (**Argentina, Australia, Belgium, Canada, Chile, Columbia, Denmark, England, Netherlands, New Zealand, Northern Ireland, Poland, Scotland, Serbia, South Africa, Spain, Sweden, and the USA**), numerous cities across the United States stretching from the Atlantic to Pacific Oceans and Canadian to Mexican borders, and >10 virtual 22q and ZooM sites, many in local languages, including Japanese! Continuing the tradition, **Maria Kamper** from the **Australia and New Zealand 22q Foundation** kicked us off in person in two locations, while many others were fortunate enough to gather in person due to confidence in a low risk of COVID-19 in their local communities.

After a year of extraordinarily difficult times, with your help, once again we came together as a family - supporting one another, learning new things, making new friends, enjoying one another's company, just being silly with our children, and looking forward to better times ahead. So PLEASE mark your calendars and plan to join us next year as we celebrate the **12th Annual 22q at the Zoo – Worldwide Awareness Day** on **May 22, 2022**! We can't wait to continue the tradition!!!

The 22q at the Zoo event aims to **raise awareness** about chromosomal differences. It also gives families, friends, and professionals a chance to **socialize** and **network**.



Save the Date

The 12th Annual 22q at the Zoo - Worldwide Awareness Day®

will be held on

Sunday May 22, 2022

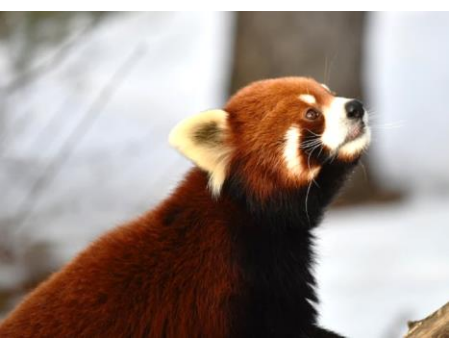
The International 22q11.2 Foundation, Inc., founders and organizers of this global event have selected the fourth Sunday of May (the 22nd) next year to bring the event full circle from its inaugural date on May 22, 2011!

We sincerely hope we will all finally be back in person together with our friends and families celebrating this most **AMAZING** 22q day…

22q at the ZOO WORLDWIDE AWARENESS day®

Questions?

Please email us at info@22q.org



Meet McKenna Grace

McKenna is the daughter of Robin, you may remember reading about Robin--our 22q nurse-- and McKenna in earlier feature stories. McKenna is now a thriving 4-year-old, but it was a rough start for her. McKenna was born April 2017 in Cincinnati, Ohio via a c-section at 39 weeks. Just as they were about to release McKenna from the hospital, the doctor making rounds told her parents that he did not like the sound of her heart murmur. At this point, they sent McKenna to Cincinnati Children's Hospital for an echo, where they discovered she had Truncus Arteriosus, an interrupted aortic arch, type B. Shortly thereafter, McKenna had a seizure, which was due to very low levels of calcium.

The medical professionals decided to do genetic testing and a few months later she was diagnosed with 2 chromosome deletions, 10q and 22q, with the 10q having no significant health issues and 22q being the source of her heart condition, as well as others. After this diagnosis, the geneticist counselor suggested her mom and dad be tested. After having to work with the insurance company several times, the genetic testing was approved and completed. The tests concluded that McKenna's father had 10q and her mother had 22q.

McKenna has overcome so much in her short 4 years of life. She has been through several heart surgeries and will require another in the future. She requires oxygen 24 hours a day due to the compression on her airway caused by her heart, and she has a G-tube. Having the G-tube doesn't require her to eat a special diet, only extra fluids, and although she eats everything by mouth, most of her fluids and some food, come through the tube. At the beginning of 2020, she was in the hospital for several weeks with the flu, but as she does with everything, she fought through the flu like a champ!

Despite some ongoing health issues, nothing stops her! McKenna will start face-to-face school in the Fall with an IEP and a nurse by her side. Speech has always been a challenge for her, but she knows sign language, and is currently in speech therapy, and can now almost say two complete words. She loves to play with her older sister Madison, baby dolls, and pretend. She also likes to play with the neighborhood kids and on a nice warm day, you can find her drawing with chalk and playing on her playground. As her mom says, "we try to stay healthy and thrive!"

What is her mom Robin's advice to other parents of children with the deletion or duplication? "The skies the limit, don't let 22q define you, YOU define 22q! It's one day at a time… and enjoy the little things."

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The Faces of Sunshine Calendar

The International 22q Foundation publishes a photo calendar every year and all proceeds benefit our Foundation. McKenna Grace's photo was featured on the May 2021 calendar.

For more information about the calendar and other campaigns, please visit our website:

www.22q.org
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The Cup of Wonderful

In honor of
Jeffrey "Ryan" Lanford (1978 – 2016)

Exciting news!

Cups for a Cause, ZPOTS has joined Juelerye and the International 22q11.2 Foundation in raising awareness in honor of Ryan Lanford.

The cup of wonderful is inspired by Ryan's love of the world; curiosity and his nickname "Mr. Wonderful". Proceeds will be given to the foundation to fuel our mission.

Learn more about Ryan here and get **YOUR** Cup of Wonderful today.

Remembering an Inspiring Advocate

Adam Aelick (1998 – 2020)
22q Advocate
Sudbury, Ontario, Canada

Adam Aelick was diagnosed with tetralogy of Fallot and 22q very early in his life. He endured multiple health challenges and surgeries as a child, but progressed with the help of therapy programs. In his teenage years, he dealt with academic and personal struggles. He switched to special education but re-integrated successfully into mainstream classes, graduating at the age of 19.

Despite the difficulties he faced, he always encouraged people to "**think positive**". He had many followers on social media and was part of many online 22q support groups, where he shared both his successes and struggles.

"Think positive."

- Adam Aelick

On April 16, 2020, Adam gained his wings after a cardiac arrest. In memory of Adam's far-reaching and inspiring work, the International 22q11.2 Foundation has established the **Adam Aelick Advocacy Award**. The award honors patients and family members who make a significant impact in advocacy about 22q11.2 deletion and/or duplication syndromes. To find out more about Adam, this award, and to donate, please visit our Adam Aelick Advocacy Award page.

Meet our Medical Director

Prof. Anne Bassett, MD, FRCPC
Chair of Medical Advisory Board
International 22q11.2 Foundation

Prof. Anne Bassett is the Director of the Dalglish Family 22q Clinic at the Toronto General Hospital and the Dalglish Chair in 22q11.2 Deletion Syndrome. A Professor at the University of Toronto and Director of the Clinical Genetics Research Program at the Centre for Addiction and Mental Health, she is a world-renowned expert in adults with 22q11.2 deletions and in the genetics of complex conditions.

Prof. Bassett is the Medical Director for the International 22q Foundation, and a Founding Member of the 22q11.2 Society. She is a leader in international studies of, and in developing international clinical practice guidelines for, 22q11.2 deletion syndrome. Prof. Bassett has been providing exemplary care to adults with 22q, and their families and caregivers, for over 25 years.

"The patient's needs always come first."

- Prof. Anne Bassett

COVID-19 Vaccination

Did you get your COVID-19 vaccines?

If you are aged 12 or older, please get your COVID-19 vaccines as soon as possible.

COVID-19 vaccines are **safe** and **effective** in preventing serious illness. They have been tested rigorously according to the standards of the Food and Drug Administration. Millions of people in USA and other countries have received their COVID vaccines.

If you are allergic to polyethylene glycol (PEG) or polysorbate, or if you have concerns about getting the vaccines, please check with your healthcare provider before booking your appointment.

Reliable information about COVID-19 vaccines

Center for Disease Control (CDC)
Vaccines for COVID-19

The New England Journal of Medicine
COVID-19 Vaccines – Frequently Asked Questions

Government of Canada
Vaccine Safety, Concerns and Possible Side Effects

European Medicines Agency
COVID-19 vaccines: key facts

The International 22q11.2 Foundation
COVID-19 Series for Individuals with 22q11.2 Differences

Immune Deficiency Foundation
COVID-19 Videos

Research Corner

Variable Immune Deficiency Related to Deletion Size in Chromosome 22q11.2 Deletion Syndrome

Blaine Crowley, Melanie Ruffner, Donna M. McDonald McGinn, and Kathleen E. Sullivan
Am J Med Genet A. 2018; 176(10): 2082–2086. doi:10.1002/ajmg.a.38597.

This report contains findings about both deletions and duplications in 22q11.2. A summary is provided here.

The thymus is an organ in the immune system, and it is where T cells mature. T cells are a type of white blood cells that play many important roles in the fights against infections.

This report first provides a review about immune deficiencies in individuals with 22q. A hypoplastic (under-developed) thymus and the resulting low number of T cells are common in young children with 22q. In some adults with 22q, T cells no longer work well enough, and antibody functions that depend on T cells also decline as a result.

The authors then report newly uncovered relationships between 22q11.2 status and T cell count. Individuals whose 22q11.2 deletions include the *TBX1* gene had a lower number of some types of T cells, while those with a 22q11.2 duplication had a relatively similar T cell count compared to people with no 22q differences. This report also provides an overview of autoimmune diseases and atopy (the tendency to develop heightened immune responses to common allergens) in individuals with 22q.

Two authors of this report are members of the Medical Advisory Board of our Foundation. Donna McDonald McGinn (left) is the Program Director of the "22q and You" Center at The Children's Hospital of Philadelphia (CHOP). Dr. Kathleen Sullivan (right) is the Division Chief of Allergy and Immunology at CHOP.

The full article is available on the website of the *American Journal of Medical Genetics*, but a purchase is required. You can also download the author manuscript for free on PubMed Central.

Upcoming Conference

The **12th Biennial International 22q11.2 Conference** has been rescheduled for **2022.** This conference will take place **in-person** in **Croatia**. Please save the dates.

Family Meeting

- **June 26 (Sunday) and 27 (Monday)**

International Brain and Behaviour Consortium (IBBC) and Network of the Americas

- June 28 (Tuesday)

Professional Meeting

- June 29 (Wednesday) to July 1 (Friday)

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