International 22q11.2 Foundation

Newsletter September 2021

Greetings from the International 22q11.2 Foundation. We hope you and your family are doing well.

In this newsletter, you will find out our upcoming events: the Calendar Campaign and 22q Awareness Month this year. We will invite you to fill out a COVID-19 survey and provide you with our Medical Advisory Board's recommendation for vaccination.

You will get to read the inspirational story of Rhiannon "Rhys" Arjona and hear from Prof. Peter Scambler, who has been an instrumental researcher in heart conditions related to 22q. We are also very excited to tell you about a landmark study on the prevalence of 22q and share the need for eye care in individuals with 22q differ

Happy reading!

Faces of Sunshine Calendar — Calling for submissions

Since 2007, we have been showcasing the photos of our loved ones on **Faces of Sunshine** calendars every year. The calendar is a great way to show your 22q pride and makes a lovely gift. All proceeds go to our Foundation.

The 2022 calendar will be printed and sold on our website, and will display twelve months of "Faces of 22q". To enter an individual in the calendar, please email your photo(s) (maximum 2 photos per featured person) along with the signed consent form to info@22q.org **by November 1st**. The Foundation will make every effort to include each child or adult somewhere in the calendar. This is subject to the number of submissions and space available.

For more information, please visit the Calendar Campaign page of our website.

Cover from the 2021 calendar

Buy a birthday!

You can celebrate your loved one's special day by buying a birthday **by November 1st**. When you buy a birthday, the Foundation will include an acknowledgment (first name only) and a photo of your child or adult on their birthdate on the 2022 **Faces of Sunshine** calendar. The placement of a person's photo on a specific day happens on a first come first serve basis. If multiple individuals share a birthday, the Foundation will work with the families to find a suitable arrangement.

Quick Links

Calendar Campaign

Buy a Birthday

Pre-order your 2022 calendar

November — 22q Awareness Month

22q11.2 deletion and duplication syndromes are not well known conditions, even among medical professionals. Our goal during the 22q Awareness month is to let the world know more about the complexities that individuals with 22q differences have and the support they need.

Despite having multiple health and learning challenges, these amazing, beautiful children and adults overcome many obstacles to survive and thrive! We also remember the few that are lost far too soon. We want the world to know all our stories and to recognize the beauty within these individuals. This November, please remember to spread the word about 22q differences. Follow us on our website **www.22q.org** and share our posts on social media.

Would like to share your story for the 22q Awareness month in November? Please download this questionnaire and email your information to us at info@22q.org, along with two, high quality photos, and we will contact you for more information!

COVID-19 Susceptibility Study

Understanding COVID-19 in individuals with 22q differences

The Children's Hospital of Philadelphia (CHOP) has launched a study called 22qDS and COVID-19 Susceptibility. The goal is **to understand whether people with 22q11.2 deletion syndrome or 22q11.2 duplication syndrome are more susceptible to COVID-19 or have unique problems with the infection**.

If you would like to take part in the survey, please scan the QR code on the right or click the following link:

https://redcap.chop.edu/surveys/?s=LXMYC8X3NT

If the person with 22q11.2 and COVID is under the age of 18, the parent must complete the survey on the child's behalf.

If you have questions about this study, you may reach Dr. Kathleen Sullivan at (215) 590-1697.

The survey takes about 10 minutes to complete.

Thank you very much for your time.

To start the survey, please use the camera of your cell phone to scan the QR code above. You can also click the link to the survey website.

COVID-19 Information

Did you get your COVID-19 vaccines?

If you are aged 12 or older, please get vaccinated as soon as possible.

COVID-19 vaccines contain no virus capable of causing disease, so they are safe – and indeed recommended for people with immune deficiencies. If you are allergic to components of the vaccine, please check with your allergist for instructions.

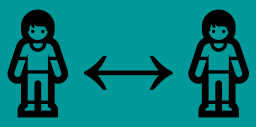
Our Medical Advisory Board's recommendations for Individuals who are eligible to receive COVID-19 vaccines:

All **individuals with 22q** should **receive the vaccine** - the two doses protect very well against the worst effects of the virus.

All **close contacts** (e.g., **immediate family members**) of individuals with 22q should also **receive the vaccine**, in order to protect the person in the family likely to be most in danger of getting very sick from the virus (that is, the person with 22q).

If you are concerned about receiving COVID-19 vaccines or have any questions about your health, please check with your healthcare provider directly.

To avoid getting sick from COVID-19 and spreading the virus:



Stay at least 6 feet (3 big steps) away from people

Get vaccinated

Wash your hands properly and frequently with soap and water OR use an alcohol-based sanitizer

Wear a mask

Why wear a mask?

Bill Nye explains in his video about masks and COVID-19

Meet Rhiannon

September 8, 2021

My name is Rhiannon “Rhys” Arjona, I am 18 years old, I was diagnosed when I was 3 days old with Tetralogy of Fallot and other clear-cut signs of 22q. As for difficulties, I had heart surgery when I was 3 months old since I would turn purple due to my Tetralogy of Fallot, I over heat quite easily since I can’t regulate body temperature well, I have learning differences (I prefer the term differences instead of disabilities) in math, reading, and writing and I also have a lot of back and neck pain.

My school years went relatively well all things considered, I got really lucky and had great support from my teachers and parents. I went to a public school for the first 3 years but then switched since it wasn’t meeting my IEP needs, they would pull me out of class to get help and then they got mad at me for ‘skipping class’, even though 2 - 3 of my teachers were really wonderful and absolutely amazing. After that I went to a homeschool hybrid public school, so my mom was around the building when I attended classes as well as other students’ parents. I really liked that school and the people there they really helped me overcome my fear of sharing my writings and actually asking for help when I needed it, I was still bad though. I got to be a teacher assistant for some other students with IEPs in a history and Spanish class and found out that I was really good at it.

I joined theater in freshman year and even though I wasn’t able to do a lot of the productions as an actor I always tried to help with work parties and building sets as well as ushering. I learned so much from my high school theater teacher and really owe him a lot for being a safe and fun person to talk to. Theater became my main hobby and pastime in middle school and I am really glad that I stuck with it since I feel like I learned a lot about myself from playing someone else, and understanding how my 22q worked for me in general.

I took Japanese in high school and it was one of the hardest things I did since I have bad phonemic answers but I was persistent about it because my sensei was absolutely amazing. Her class quickly became my absolute favorite to be in. Learning Japanese was hard for me since a lot of the sounds sounded the same, my sensei encouraged me to keep going with it. It took me a lot longer to learn and I failed almost every test the first time I took it but the more I put my mind to it and the longer I did it,

(This story continues on the next page)

Would you like to share your story like Rhiannon did?

We are looking for stories for our November Awareness Campaign.

We all need inspirations every now and then. If you or someone you know is living and thriving with 22q, please share your story! Please download this questionnaire and email your information to us at info@22q.org, along with two, high quality photos, and we will contact you for more information!

Rhiannon “Rhys” Arjona

Meet Rhiannon (Continued)

things started to make sense, I learned Japanese and am now able to have speaking conversations. I am really proud of myself for sticking with it and continuing to learn. I had to get a lot of extra help from my dad and my sensei but it really all payed off. And I learned more than just Japanese in that class, my sensei helped me see that if you wanted to learn something even if it takes a lot longer than someone else you just have to try and try again, and have to learn from your mistakes to get better at them. That helped me so much in my education journey with having 22q. It's really how I overcome my challenges.

I don't really do anything specific to help keep myself healthy; I just try to eat healthy foods.

Some of the accomplishments that I am the most proud of are: Graduating high school with a very good GPA, taking 3 years of Japanese, getting inducted in to the International Thespian Society, getting into my dream college and of course being a big sister to Rhonwen, my 3-year-old sister.

My advice for someone with 22q is know you're not the only person out there, that there are other people going through the same or similar issues as you. Thinking that you're alone only makes things worse, but knowing that there are others can help you see the bigger picture. As my Sensei taught me, "If you want to learn something keep going, it might be hard for you but you have to keep moving and learn from your mistakes to get better. It might take longer but it isn't a race. If you fail a test, try again." I am going to University of Alaska Southeast, one of my goals is to get a degree in something to do in history. My goals are to work on an animated show with Dan Povedmire and Swampy Marsh, the creators of my two favorite animated TV shows. I would also like to meet Jennifer Lee and Rick Riordan and I would like to work as a writer for Disney one day or become a teacher.

I run an account for 22q advocacy on Tik-Tok called Redfor22q. I make videos sharing my experiences. I also hope to hit 20K someday.

Thank you very much to Rhiannon for sharing her story. We wish her all the best!

For more stories of individuals with 22q differences, please visit:

https://22q.org/inspirational-stories/

We appreciate your financial support!

Our mission: to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.

At the International 22q11.2 Foundation, we help families that need important resources and information to meet the needs of loved ones with 22q.

We work with top medical experts from around the world to build our understanding of 22q, improve treatment options for families, and conduct the necessary research for longer, healthier lives for patients.

Your support makes a difference!

Please visit the **Donate** page on our website for information on donating online, by mail, or via other methods.

Please also **Shop** on our webstore.

For more information, please visit our website at www.22q.org or email us at info@22q.org. **Thank you**!

Meet our Medical Advisory Board Member

Prof. Peter Scambler, MD, FMedSci

Professor of Molecular Genetics
Great Ormond Street Institute of Child Health
University College London, United Kingdom

To learn more about Prof. Scambler's work, please visit his research website.

Professor Peter Scambler has been working on 22q11.2 deletion syndrome (previously DiGeorge syndrome) since 1992. focusing on the developmental aspects, especially cardiovascular malformations. He has received multiple awards, including the first Angelo DiGeorge Medal in 2010. He was a founder member and first chair of the 22q11.2 Society, where he remains a trustee. In December, 2019, the Society's Post-Doctoral Basic Science Junior Investigator Award was renamed the Scambler Junior Investigator Award.

Prof. Scambler will be retiring in 2022, and we take this opportunity to chat with him.

22q Fdn = International 22q Foundation

PS = Prof. Peter Scambler

22q Fdn: In 1992, you and your colleagues found that patients who were diagnosed with velocardiofacial syndrome had deletions on chromosome 22, just like the patients with DiGeorge syndrome. How did you feel about that discovery?

PS: We (the team) felt wonderful, it was such a fun time. We had been on a high ever since the previous year when we had shown "micro" deletions (i.e. too small to be seen in then standard microscopy of chromosomes) were responsible for most patients diagnosed with DiGeorge syndrome. We had then shown deletions could be inherited in families where affected members had different problems.

The velocardiofacial syndrome advance really hammered home the idea that 22q11.2 deletions could cause a large variety of medical issues. These include behavioural symptoms that, as highlighted by Dr. Shprintzen the same year, develop with time and which might therefore be amenable to future treatments.

22q Fdn: Could you please tell our families briefly about your research?

PS: Back in the 1990's my research could be described as human genetics. I was interested in learning about how genetic variation underlay malformations of development. These include serious ones such as heart defects as well as seemingly trivial ones like extra fingers that still tell us something about how normal babies develop in the womb.

Having found genes important for development the obvious next step was to move into mice, because you can really drill down into how genes work in ways you cannot in humans. Much of this was focussed on 22q11.2 genes, but in recent years I moved into genes one or two steps downstream, so to speak, in cardiovascular developmental pathways. For instance, our most recent paper described how the valves that control direction of blood flow from the heart are sculpted into the correct shape by molecules that communicate between cells.

(This interview continues on the next page)

Meet our Medical Advisory Board Member (Continued)

22q Fdn: You have been working on 22q for almost 30 years. How has basic science research on 22q informed clinical solutions?

PS: The ability to diagnose deletions led to an explosion of work by specialists in other areas. It allowed us to know much more about the life course of the syndrome and how best to treat patients for the plethora of medical issues they may face. It also let us provide support to patients for living a normal life, improving language development or math skills, for example. Newborn screening, to initiate these interventions to all that might need them, is currently being debated (see page 8 of this issue).

At the moment, there is huge interest in exploring basic science underlying the behavioural symptoms, how the immune system works, and in using the latest stem cell technologies to explore new treatments.

22q Fdn: For families who are interested in understanding more about the genes on chromosome region 22q11.2, do you have any advice?

PS: There is no denying this is tough for people without the requisite biological training. Rather than focus on specific genes to start with, look out some basic, instructional videos on Youtube concerning genetics and developmental biology.

You could then progress to something like: https://www.khanacademy.org/science/biology (free, may need to register), which should stand you in good stead for reading scientific review articles talking about how specific genes operate.

Thank you very much to Prof. Scambler for his time and recommendations.

We wish him all the best!

Events in 2022

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

will be held on

May 22, 2022 (Sunday)

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.

The 12th Biennial International 22q11.2 Conference Family Meeting

will be held on

June 26 (Sunday) and 27 (Monday), 2022

This conference will take place **in-person** at the Le Méridien Lav Hotel in Split, **Croatia**.

More information will be available in 2022.
Questions? Please email us at info@22q.org

Research News

Estimate of the contemporary live-birth prevalence of recurrent 22q11.2 deletions: a cross-sectional analysis from population-based newborn screening

Christina Blagojevic, Tracy Heung, Mylene Theriault, Aoy Tomita-Mitchell, Pranesh Chakraborty, Kristin Kernohan, Dennis E. Bulman and Anne S. Bassett
CMAJ Open, 2021, 9 (3) E802-E809; DOI: https://doi.org/10.9778/cmajo.20200294

For individuals with 22q11.2 deletion syndrome (22q), getting the correct genetic diagnosis early in life allows doctors to anticipate and deal with health issues, which helps avoid complications. Therefore, experts in 22q have long advocated for newborn screening for this syndrome. However, to convince policy makers to add 22q to newborn screening programs, researchers must show that 22q is in fact common enough to be worth the effort.

A study published in August 2021 tried to determine: **how often does 22q happen in newborn babies**? The authors found that among 30,074 newborns in Ontario, Canada, 14 had 22q deletions, which means **22q has a prevalence of 1 in 2148 live births**. The authors also found that babies with 22q likely had younger mothers, were smaller compared to babies born after the same length of pregnancy, and had lower counts of a blood test called TREC, which could signal future problems with the immune system.

Based on prevalence data from Ontario, 22q (prevalence 1 in 2148) is more common than cystic fibrosis (CF, 1 in 3600) and severe combined immunodeficiency (SCID, 1 in 50 000 to 1 in 100 000). Both CF and SCID are less prevalent than 22q, but both are genetic conditions that are often screened for in newborn screening programs. Unfortunately, 22q is not known to be in standard newborn screening programs of any countries.

This study confirms that 22q is a relatively common "rare" condition. From a public health stand point, the results support the importance of screening for 22q either prenatally or in the first days of life through newborn screening. An early system of diagnosis would allow individuals with 22q to receive timely management of their health problems, which will decrease adverse outcomes.

The full article is available for free on:
https://www.cmajopen.ca/content/9/3/E802

Eye Check-Up: Don't let eye problems affect your child's learning

Eye problems can happen in children with both 22q deletion and duplication syndromes. Common eye problems in children with 22q deletion include abnormalities of the eyelid, abnormal curving of blood vessels, and strabismus (eyes not aligned). Strabismus is the most frequent eye problem in children with 22q duplications, but many other issues are possible. Some of these conditions can significantly affect vision, making it harder to learn at school. Therefore, children with 22q differences should have regular eye check ups.

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