



The Focus Foundation

research · awareness · treatment

May 2018

The Magnifying Glass: A Newsletter

Our Promise: For all families, The Focus Foundation promises to develop the latest and most innovative care. The Focus Foundation believes in novel and "out of the box" thinking that produces scientific breakthroughs that enable children with neurogenetic disorders to lead better and more fruitful lives.

Join Us as We Celebrate



**X and Y
Chromosomal Variations
Awareness Month
MAY 2018**

VISIT US ON FACEBOOK TO LEARN MORE

A Note from Dr. Sprouse



Dear Friends and Family,

Happy National X & Y Chromosomal Variations Awareness Month! This is such a special time for The Focus Foundation as science, patient care, and awareness are all at the heart of our mission.

Did you know that over 75% of the children who have a chromosome variation will go undiagnosed in their lifetime? Currently, only 25% will receive the proper testing and diagnosis. Imagine. It only takes a simple cheek swab to confirm diagnosis yet some will never receive a diagnosis and most will be misdiagnosed or misled, lacking the ability to properly treat and intervene and allow that child to soar in his or her own future. As a specialist – and a parent – I know we can do better.

We love our community here at The Focus Foundation. Our parents are advocates for their children and together we are creating ripples of change and support for one another's journey. And this month, we are calling you to action:

- **To help change those statistics.**
- **To debunk myths and grossly inaccurate expectations for our children.**
- **To stand up for education and better care.**
- **To help spread the word that there IS hope and there IS treatment.**

You can join us in our Awareness journey this month in several ways.

- **Follow us on Facebook and Twitter** for ideas, inspiration, and innovation daily! Share any and all with people who you think will find it helpful.
- **Show your support** for Chromosome Variation Awareness by featuring one of our custom profile borders on your Facebook picture! Search "The Focus Foundation" in your Facebook profile editor to choose from 3 custom templates! Encourage families members to do the same.
- **Volunteer** for or begin a fundraiser in your community to support research and further testing for all chromosomal variations including 47, XXY, 47, XYY, 47, XXX, 48, XXXY, 48, XXYY, and 49, XXXXY.
- **Advocate for donations** – personal or corporate – to help continue the mission of research and treatment within The Focus Foundation.

I am always moved by your dedication to The Focus Foundation and happy and proud to have you and your children along on our journey. We wish you May flowers, a happy and healthy spring, and of course, an informative and impactful X and Y Chromosomal Variations Awareness Month.

Sincerely,
Dr. Carole Samango Sprouse

Science is Moving Fast and Furiously!

New Publications

Samango-Sprouse, C., Stapleton, E.J., Chea, S., et al. [International Investigation of Neurocognitive and Behavioral Phenotype in 47,XXY\(Klinefelter syndrome\): Predicting Individual Differences.](#) American Journal of Medical Genetics Part A. 2018;1-9.

Receiving Honors

The team was thrilled to attend and present at the American College of Medical Genetics and Genomes (ACMG) Meeting in Charlotte, NC this April with 2 posters, one ribbon for a highly ranked abstract, and a scientific concurrent session!

Posters presented included The Effect of Hormonal Replacement Therapy on Behaviors Related to Executive Function in Boys with 47,XXY (Klinefelter Syndrome) and The Impact and Relationship of Testosterone Replacement Therapy (TRT) on Anxiety Disorders in 47,XXY or Klinefelter Syndrome (KS).



The scientific concurrent session, moderated by our own Dr. Carole Samango-Sprouse and Dr. Cynthia Powell at University of North Carolina, was a multi-institutional effort aimed to educate hundreds of genetic counselors, clinical geneticists and researchers about the latest facts on our disorders. Our experts in X & Y Chromosomal Variations, Non-Invasive Prenatal Testing (NIPT), and endocrinology stressed the importance of comprehensive, coordinated care completed by well-trained professionals. Most importantly, the care that is given needs to be personalized and focused in light of the needs of the child and their families. Hope is powerful, and optimism is self-fulfilling. We demonstrated both the power and impact of both on these children with X and Y

Chromosomal Disorders.

We will continue educating professionals and caregivers around the world as we provide our research results and experience of over 15 years with us to Toronto, Canada in May!

Our Newest Study

The Focus Foundation has been partnering with collaborators throughout the world on a variety of rare disorders for years! We are excited to collaborate on a new study focused on patients with oculo-auriculo-vertebral spectrum (OAVS).

OAVS involves a wide range of congenital malformations that may vary greatly in severity from case to case, resulting in a highly variable phenotypic presentation. Dr. Carole Sprouse collaborated with Cohen et al. in 1995 at Children's National Medical Center to publish an original article on the neurodevelopment of OAVS. They found an increased risk for neurodevelopmental delay, especially those with abnormal muscle tone, bilateral involvement, and cervical vertebral anomalies. Dr. Sprouse and colleagues hope to provide more comprehensive findings on the characteristics of OAVS. Children between the ages of 3-18 years old with OAVS are invited to participate in this study, which will involve a neurodevelopmental evaluation, a neurological functioning assessment, and neuroimaging scans. With this study, Dr. Sprouse and collaborators aim to provide more insight into the phenotype of OAVS and may change the care protocol for these patients. Please feel free to pass on our message to any and all families as we explain these children's profile.

Facebook Live



Next Facebook Live - August 1, 2018

Join Dr. Sprouse as she spreads facts, tips, and general awareness of X and Y Chromosome Variations! She will be LIVE on Facebook for a summer session to answer your questions on **Wednesday, August 1st at 12 pm EST**

Have a topic you want discussed? Email your questions or topic suggestions to cbedard@thefocusfoundation.org.

Missed her other two Facebook Lives? Visit our Facebook page to rewatch her "**General Q&A**" and "**Focus on Developmental Dyspraxia**" sessions from earlier in the year!

FOLLOW US ON FACEBOOK

Conference Announcements

At Home and Abroad

15th Annual 49ers Clinic

Annapolis, Maryland

July 18th – 20th, 2018

We are thrilled to be celebrating our
15th Annual 49ers Clinic!

- **Wednesday, July 18th:** Welcome Reception
- **Thursday, July 19th:** Specialist Rotations/Clinic
- **Friday, July 20th:** Parent Conference



Featuring families from across the globe for a conference and clinic focused on 49, XXXXY.

Registration to open this month via www.thefocusfoundation.org

Accommodations:

Country Inn & Suites, 2600 Housely Rd, Annapolis

Phone: 410- 571-6700

\$99 for Queen/Queen room, \$104 King *Includes indoor pool, continental breakfast and free van shuttle to restaurants and downtown Annapolis

****Mention Dr. Sprouse and The Focus Foundation to receive preferred rate.**

UK Conference

December 1st - 3rd, 2018

Dr. Sprouse and our team of X & Y Variant Specialists are excited to return to the UK this December!

The Focus Foundation will require a minimum of 10 families to register for this annual conference in order to make the jump across the pond. Help us spread the word!



*[Registration](#) will open in July.

[VIEW ONLINE CALENDAR](#)



DeXY's Corner



Bring Out The Tacos This Cinco De Mayo!!

Set up a fun taco bar and allow your little ones to exercise independence and assemble themselves. They will get to practice their critical thinking, planning, coordination, and motor skills as they add toppings themselves! Plus, they're DELICIOUS!

DID YOU KNOW: If you make them with a focus on healthy ingredients, tacos can provide a good balance of protein, fats and carbohydrates and provide ample amounts of fiber, vitamin C, calcium and iron.

Try this easy – and healthy – Mango Salsa recipe:

- 1 Mango, peeled and diced
- ½ cup diced cucumber
- 1 tablespoon finely chopped jalapeno
- 1/3 cup diced red onion
- 1 tablespoon lime juice
- 1/3 cup chopped cilantro leaves
- Salt and pepper

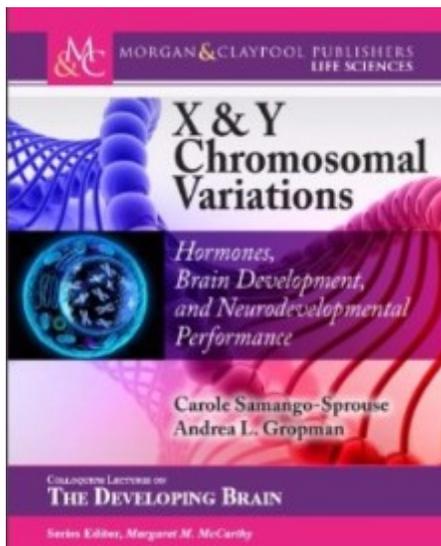
Combine the mango, cucumber, jalapeno, red onion, lime juice and cilantro leaves and mix well. Season with salt and pepper, to taste.



Happy Mother's Day!

To the Mothers, Grandmothers, Aunts, Sisters, and friends who build our day to day family of support and love! Enjoy your day with love from The Focus Foundation!

Books We Love:



X & Y Chromosomal Variations: Hormones, Brain Development, and Neurodevelopmental Performance

by Dr. Carole Samango-Sprouse and Dr. Andrea L. Gropman

This popular book is [now available](#) in hardback! We suggest considering offering a copy of this book to your team of physicians so they can more clearly understand your child's diagnosis and today's treatment options. **Our XY families receive a 25% discount with the coupon code XYPARENT.** (Also available on all major book sites, but discount code only valid through publisher.)

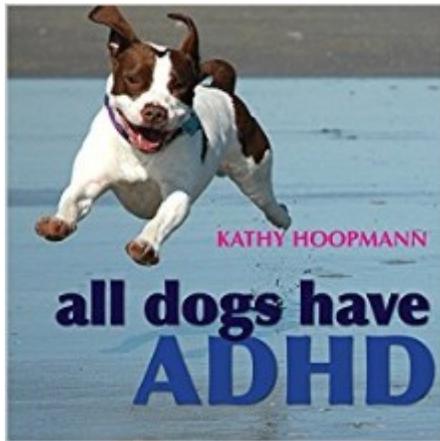


Hello, Universe

by Erin Entrada Kelly

A 2018 Newbury Medal Winner

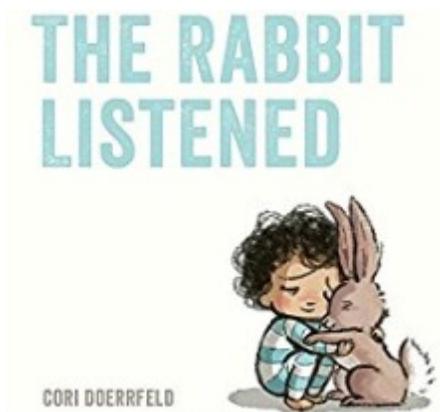
Filipino folklore and real life converge at the bottom of a well. Even while following signs and portents, the characters are the definition of creative agency. Masterfully told through shifting points of view, this modern quest tale shimmers with humor and authentic emotion.



All dogs have ADHD

by Kathy Hoopmann

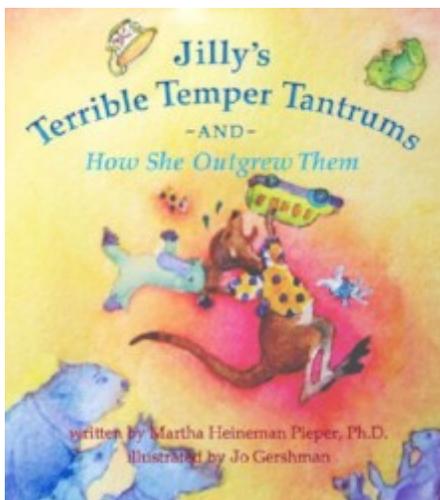
Takes an inspiring and affectionate look at Attention Deficit Hyperactivity Disorder (ADHD), using images and ideas from the canine world to explore a variety of traits that will be instantly recognizable to those who are familiar with ADHD.



The Rabbit Listened

by Cori Doerrfeld

A universal, deeply moving exploration of grief and empathy. With its spare, poignant text and irresistibly sweet illustrations, *The Rabbit Listened* is a tender meditation on loss.



Jilly's Terrible Temper Tantrums and How She Outgrew Them

by Martha Heinemen Piper, Phd

The Winner of a Gold Mom's Choice Award

When Jilly, a happy little kangaroo, has a series of Terrible Temper Tantrums, her parents lovingly help her through them. Children will be very familiar with the frustrations that trigger Jilly's upset feelings, and will applaud her dawning understanding that seeking help and a hug is far superior to the misery of a temper tantrum.

Help Wanted

**" IF EVERYONE IS
MOVING FORWARD
TOGETHER, THEN
SUCCESS TAKES
CARE OF ITSELF. "
~ HENRY FORD**

We want to celebrate the accomplishments of our children. If you have any success stories to share (great or small) please email them along with any photos. Names will not be shared. Please email: cbedard@thefocusfoundation.org

How to Get Involved:



DONATE TO THE FOCUS FOUNDATION WITH A CORPORATE OR PERSONAL GIFT

You can give more without spending more. **Many companies will match your donation to The Focus Foundation.** Check with your personnel or human resources office to see if your company participates in a matching program.

THANKS to all who are helping support the mission of The Focus Foundation through your continued support spreading awareness and through your varied contributions. Your efforts are helping ensure a brighter future for all of our children.

Visit thefocusfoundation.org or click the button below to donate.

DONATE ONLINE TODAY

We welcome suggestions and comments for future newsletter or Facebook content. Email dexy@thefocusfoundation.org