

June 2016
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We're six months into our 10th
Anniversary

**LOOK WHAT YOU HELPED US
ACCOMPLISH -it is only JUNE !**



The Focus Foundation has been spreading the word on XY disorders around the world. We're going Global ! In beautiful Waikiki, Hawaii the **17th International Conference on Autism, Intellectual Disability & Developmental Disabilities** brought together a unique blend of researchers and clinicians centered around the continued advancement of education and life outcomes for individuals with autism, neurodevelopmental disorders and intellectual disability. Here, The Focus Foundation had a presentation entitled "ASD or XY Disorder?" which discussed the prevalence, characteristic features and therapeutic interventions for XY disorders and the overlap of features with ASD. We want professionals throughout the world to think about XY disorders whenever they see atypical development, delays or dysfunction. The earlier diagnosis---the better the outcome !

Dr. Samango-Sprouse was an invited speaker at the **2nd International Workshop on Klinefelter Syndrome in Muenster, Germany**. This was a fascinating conference centered around innovative findings by the most renowned experts in the field. Topics for presentation included X chromosome inactivation, metabolic and cardiovascular risks, transitional care, neuropsychology and fertility preservation in 47,XXY. This was a wonderful opportunity for The Focus Foundation to receive exposure in a prestigious academic environment specializing in the care of individuals with 47,XXY. Our presentation focused on our comprehensive study of more than 170 prenatal diagnosed boys with XXY and how to personalize care to optimize outcome. Reported by **Colleen Keen, BA**

Dr. Samango-Sprouse and her two research assistants recently attended IMFAR (**The International Meeting for Autism Research**) and PAS (**Pediatric Academic Societies**) conferences. These conferences are large gatherings for clinical researchers, scientists and practitioners to learn the latest and most innovative findings on neurodevelopmental disorders as well as state of the art pediatric care for common disorders like asthma and diabetes among others.

At IMFAR, the two presentations were given one focusing on CASS-i, an assessment developed by Dr. Samango-Sprouse to identify the biomarkers for infants at risk for Developmental Language Disorders (DLD) and ASD prior to the 12 months of age. This is an exciting and easy assessment to identify our babies at risk for XY disorders who are not diagnosed prenatally. It is quick, efficient and easy for pediatricians to identify our boys and girls prior to 12 months of age. The second presentation was on the Phelan-McDermid Syndrome, a rare genetic disorder which expanded the phenotypic profile to include childhood apraxia of speech (CAS). We posited that CAS may play a larger role in Phelan-McDermid syndrome expressive language delays than has been previously appreciated. CAS is common in many children with X and Y chromosomal variations. Its significant impact on neurodevelopmental progression is often not appreciated by clinicians and researchers.

At PAS, presentations were completed on the neurodevelopmental differences in two populations of boys with 47,XXY: one from the Netherlands, and one from the US. This is the first of many papers which lays the foundation for an "outcome algorithm". It identifies those boys with XXY who are most vulnerable for neurodevelopmental disruption as early as possible. Then, targeted intervention may be prescribed based on specific features rather than "generic or one size fits all" therapeutic services.

The second presentation was focused on another biomarker from the CASS-i which is severe stranger anxiety. It assists us in the identification of these very vulnerable children with XY disorders who have overlapping features with ASD but do NOT have ASD.

We are globe trotting to spread the news that there is great hope for our boys and girls to do well and succeed with personalized care, targeted treatment and early diagnosis !

Reported by **Patrick Lawson, BA**

***Our Publications and Our New Adventures
The Focus Foundation promises excellent
research with novel and creative approaches
that will help your children be the best! We
promise that we will bring our findings to all
families.***

Dr. Sprouse's book **X & Y Chromosomal Variations: Hormones, Brain Development, and Neurodevelopmental Performance** will be available for purchase by the fall of 2016. We have lots of great information for you, your doctor and your therapists. It is being circulated to more than 50 medical schools throughout the USA so we can educate the



next generation of the "Real Facts" about XY disorders with good science and thoughtful statistics !

Expanding the Phenotypic Profile of Kleeftstra Syndrome: a Female with Low-Average Intelligence and Childhood Apraxia of Speech. American Journal of Medical Genetics. in press

Samango-Sprouse C, Keen C, Mitchell F, Sadeghin T, Gropman A. 2015. Neurodevelopmental variability in three young girls with a rare chromosomal disorder, 48, XXXX. Am J Med Genet Part A 9999A:1-9.

We promise that we will be bring our care to families everywhere

Through the generosity of a European family The Focus Foundation will be traveling to Great Britain in the Fall to have a conference for boys with 49, XXXXY. We will be at the **Thornton Manor (www.thorntonmanor.com)** in Thornton Hough **December 1 – 4**. Details for the families will be sent out soon Thank you to the Hamilton Family for their wonderful generosity!

Keep watching --we are coming to Italy in 2017 !



**Announcing the
Thirteenth Annual 49ers Conference**

For children with 49 XY and their families

Ringo Starr: When I was 13 I wanted to be a drummer.

We are 13 and drumming globally!

Michael Azerrad: (drumming) involves all four limbs, and you're hearing stuff and you're converting your ideas into physical motions, getting physical feedback from things you are touching - it's pretty cool. **We are proud drumming about 49, XXXXY!**

When: July 20-22, 2016

Where: Annapolis, MD



I have been volunteering with the Focus Foundation since 2008 and have been involved in 49ers conferences since then. I have been helping with the family reception to the meet and greet. I assisted with child care and parent education conference where families learn from Dr. Sprouse and other specialists. Then as I grew older, I developed the child care plans and became the supervisor of the child care ! Then I began helping with 12 member multidisciplinary clinic where all children and families receive expert advice on their children's disorder. I have learned so much from the wealth of information presented in both the conference and the clinic. I really enjoy watching the families talk and share their experiences with each other so they know they are not alone. Over the years, I enjoy seeing the families and children who return and see how much they grow, change and improve ! " Our

frequent flyer families" have been coming to numerous conferences and the progress of their children is amazing! The added benefit is these experienced families provide reassurance to the families of the young children. I am honored to be a part of an organization that makes a difference in these families' lives. Over the years I have gotten some of my friends, family, and previous Girl Scouts involved in the process by having them help me with the child care.

Reported by **Katie Perez**

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Calling all 49ers!

We are so excited to be beginning a study on **growth and development** for the boys with 49, XXXXY. This information will help us understand the boys with 49, XXXXY growth and determine how the additional X affects their growth. Please send Teresa your growth charts on your son from the time he was born until current.

We are working with a world renowned pediatric endocrinologist, Dr. Alan Rogol on the boys with 49, XXXXY and their growth. Dr Sprouse and he are both very interested in writing about **Growth Hormone Deficiency** in 49, XXXXY. If you are interested in participating, contact us – teresa@thefocusfoundation.org

PATH TO DIAGNOSIS, A Survey for Postnatally Diagnosed Children

The Focus Foundation created this survey because we are seeking to better understand the factors that lead to a formal postnatal diagnosis of X and Y

chromosome abnormalities. Your participation will provide insight that could lead to more children receiving proper diagnosis and care for these disorders at an earlier age. To get started, here are some important points about this survey:

Intended for parents/primary caregivers of children with:

47,XXY, 47, XYY and 47, XXX

Child must have been postnatally diagnosed and born in 1980 or later

The survey is less than 30 minutes

No personal identification information is asked for

We are very excited to receive your insight and feedback. We truly believe it will make a difference for families across the U.S. and around the world. Thank you again for your time and support of our efforts. It is greatly appreciated.

Please pass on the information for this survey to other families. Thanks!

TO PARTICIPATE GO TO

<http://pathtodiagnosis.questionpro.com/>



Dexy's Corner – ***Social Language:***

From: Jillyan StLaurent, MA CCC-SLP

Social language deficits (pragmatics) are mostly thought of with children who have autism spectrum disorder (ASD) however children with any type of speech and language dysfunction can demonstrate these weaknesses. Their ability to communicate is often hindered, which then makes social situations more difficult for the child. As parents and professionals, we need to assist children with X and Y disorders to understand how to observe social clues in their environment. These observations help them to cope better with the different "roads" in their lives, i.e, school, family life, friends' homes, etc. Lack of social skills may damage self-esteem, peer interactions and social emotional development.

Both positive and negative behaviors are forms of communication and each has different impact on social engagement. When negative behaviors increase, there are many reasons why social language deficits can be the cause. The inability to express one's feelings and thoughts can be extremely frustrating for a child and can often result in negative behaviors. It is important that social communication be developed and supported in children with language leaning disorders since this is an area of significant vulnerability. With the development of these skills, negative behaviors can be shifted to positive outcomes from a weakness to a strength!

Social Language Activities:

Ideas found by **Katie**

Ways to work with your child –

Role playing: One of the biggest activities for learning social norms for a child is role-playing activities with same aged peers, or learning appropriate role playing type behaviors for varying situations.

Exercise increases endorphins. Following through on a commitment to walk will build confidence thereby countering anxiety.

Have a routine. This helps steady the mind, but also gives confidence to emotions. In other words the child will feel in control.

Practice affirmations. Often anxiety puts pictures in our mind about how we are going to freak out in a certain situation. Help the child change the picture. See it differently. See yourself calm and happy. Do this on purpose 2-3 times a day, especially when the child is calm. Don't wait until they are panicked to do this! You are retraining their mind to have new associations. Literally changing their brain and equally important developing new habits to old behaviors. Be patient. Keep it up. The human brain loves habits since it takes less energy, is easily available and predictable. You will notice a big difference !

<http://www.healthyplace.com/parenting/the-parent-coach/teaching-children-social-skills-emotional-skills/>

Kid activities:

Blowing Bubbles

Children can learn deep breathing skills by using bubbles. Deep breathing is an excellent way to manage anxiety. Demonstrate and have children use bubbles while focusing on how they breathe to create a big bubble.

[See Therese J. Borchard's article "[3 Deep Breathing Exercises to Reduce Anxiety](#)" for more information about deep breathing.]

Worry Can

To help kids identify and discuss their worries, they can write down their worries and place them inside a can (with a lid). This helps kids to identify and express their worries instead of keeping them bottled up inside which can lead to even more difficulties. This activity can be modified to fit for the child's particular situation and personality.

[This activity was adapted from [Hall, Kaduson, and Shafer, 2002.](#)]

Calm-Down Box

Fill a box with things that will help the child calm down when they feel anxious. Helping children to become more aware of their own functioning including what triggers them to become anxious (or feel any sort of emotion), what helps them to relax, and to have the knowledge and belief that they can master control over their inner experience can greatly help them in managing their emotions and overcome their challenges.

Use a box and help the child come up with objects to put in the box. Some examples to include in the box are: stress balls, bubbles, or a book.

<http://pro.psychcentral.com/child-therapist/2014/08/anxiety-relief-3-activities-to-help-kids-manage-their-anxiety/>

Summer Treats

Gathered by Teresa

Now that I'm an adult, I still have seasonal food memories. One of my neighbors made popsicles the minute the weather got hot. All the neighbor kids flocked to her home for the treat! I thought I'd share something the kids could easily make with you... and then, since I got nostalgic, I decided to make the popsicle a root beer float – one of my Dad's favorite treats with me and then with my kids. I hope you enjoy:

Root Beer Float Popsicles (you will need popsicle molds)

1 can root beer, cooled down in the freezer about 50 minutes (so ice cream doesn't melt)

Soften the ice cream by setting on the counter about 30 minutes.

Add $\frac{3}{4}$ cup vanilla ice cream to the cold root beer (keep in chunks)

Spoon about 1 tablespoon ice cream into bottom of the mold. Add the root beer/ice cream mixture. Continue to pour until the mold is full.

Top off with a tablespoon of ice cream.

Freeze about 4 hours.

One bite and you'll be a kid again and your kids will have a new favorite treat!





This looks like a fun snack! We call my daughter, Farideh Flutterfly, so this matches her personality!

Apple Butterfly (makes 4)

½ cup peanut butter
4 fresh baby carrots
1 large red apple
8 pieces celery, thin strips(2-inches long)

Spoon 2 tablespoons peanut butter onto center of each of 4 individual plates. Shape peanut butter on each plate into a mound about 2 inches long and 1 inch wide. Place

1 carrot down center of each mound of peanut butter.

Cut apple into quarters; remove core. Cut each quarter into 7 thin slices; cut 1 slice in half crosswise. Fan 3 apple slices on each side near top of each carrot to form upper wings. Place 1 half-slice on each side near bottom of each carrot to form lower wings.

Place 2 strips of celery at top of each carrot to form antennae. Sprinkle apple slices with decorator sugar (optional).

Decorate wings with raisins, craisins, peas, fruit loops or Cheerios.

from food.com