



The Focus Foundation

research · awareness · treatment

Fall 2017

The Magnifying Glass: A Newsletter

Our Promise: To develop the latest and most innovative care for all “our children” based on excellent research grounded in the belief that your child’s life should and will improve based on our research findings. We believe 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.



Science is Moving Forward

Publications:

Samango-Sprouse, C. The Benefits and Limitations of cell-free DNA screening for 47, XXY (Klinefelter syndrome) Prenatal Diagnosis, April 2017

Keen, C. Samango-Sprouse, C. Dubbs, H. Zackai, E. 10-year-old Female with Intragenic KANSL1 Mutation, no KANSL1-related Intellectual Disability, and Preserved Verbal Intelligence. American Journal of Medical Genetics Part A. February, 2017

Presentations:

Samango-Sprouse, C. Early childhood development of rare X and Y chromosomal disorders (XYD) 49,XXXXY and 49,XXXYY an oral presentation made at the 20th

SSBP International Research Symposium 14-16 September 2017. Leiden, the Netherlands.

Keen, C., Lasutschinkow, P., Gropman, A. and Samango-Sprouse C. 47, XXY in Two Siblings and Their Varying Neurodevelopmental Presentation. The 20th SSBP International Research Symposium. 14-16 September 2017. Leiden, the Netherlands

Samango-Sprouse, C., Cappello, C., Stapleton, E., Lawson, P., Sadeghin, T., Chea, S, de Sonnevile, Leo, and van Rijn, Sophie. International Investigation of Neurocognitive and Behavioral Phenotype in 47,XXY (Klinefelter Syndrome): Predicting Individual Differences. The 20th SSBP International Research Symposium. 14-16 September 2017. Leiden, the Netherlands

Hensel, Charles; Vanzo, Rena; Serrano, Moises, Wassman, E. Robert; Samango-Sprouse, C. Genetic test results in children under 3 years of age who are at-risk for neurodevelopmental disorders: an update. IMFAR, San Francisco. May 2017.

Keen, C; Cappello, C; Samango-Sprouse, C. Behavior, Social and Executive Function in Children with 47, XXX or 47, XXY. Presented at ACMG Phoenix. March, 2017.

Cappello, C. Gropman.A, Keen, C, Samango-Sprouse, C. Two Unique Family Profiles Involving 16p11.2 Duplication. Presented at ACMG Phoenix. March, 2017.

Samango-Sprouse, C, Cappello, C.; Keen, C.; Sadeghin, T.; Paul M.; Wassman E.; Vanzo,R. Copy Number Variations (CNVs) Associated With XXY: Variability and Impact on Neurodevelopmental Performance. Presented at ACMG Phoenix. March, 2017.

Webinar:

Drs. Gropman and Samango-Sprouse presented to the Prenatal Special Interest Group (SIG) of the National Society of Genetic Counselors on September 11. More than 150 genetic counselors throughout the country learned about 47, XXY: New innovative Treatments and Outcomes.



Our Projects:

The 49, XXXXY and 48, XXXY Project: To develop and implement a standard of care for all boys with these disorders from Medical, Neurodevelopmental, and Endocrinological perspective.

Face2Gene: This is the exciting marriage of clinical care with technology to facilitate early detection of all X and Y Chromosomal Disorders. With a suite of applications and a photograph, this software detects relevant facial and non- facial features and reviews pertinent syndrome matches. It fosters comprehensive and precise genetic evaluations for any patient with neurodevelopmental dysfunction. With our help and the facial features of 47, XXY, 48, XXXY and 49, XXXXY uploaded, early identification and treatment can occur wherever the child lives.

The MIT Study: Genetic Analysis: This study will expand our understanding of the biology of the X&Y chromosomes and their impact on health and well-being of all human beings.

The Johns Hopkins Study: Importance of Methylation: We are looking at methylation characteristics of the genes on X & Y chromosomes. This study will allow for the development of more personalized and precise medicine for “all children” as we begin to understand the relationship between methylation and behavioral outcomes.



The Focus Foundation is using **iMotions** software to analyze eye tracking, facial expression, and galvanic skin response (GSR) to identify ocular, physiological, and emotional biomarkers of anxiety, reading dysfunction, and attentional issues in boys and girls with X and Y Chromosomal Disorders, Dyspraxia and Dyslexia as early as the first year of life. This exciting investigation will lead to improved targeted treatment programs for all our children throughout the world.



Join Us On Facebook Live!

Exciting News:

January 2018 Dr. Sprouse gives tips and takes your questions live on our Facebook page in a new quarterly segment based on your interests!

What would you like to hear about?

- Strategies for addressing anxiety
- The best exercises for building strength and endurance
- Managing difficult behaviors
- Increasing compliance with Testosterone application
- Optimizing neurodevelopmental outcomes

Let us know – we want to hear from YOU!

Be sure to follow us on Facebook for regular updates.

RSVP for Dr. Sprouse's 1st Facebook Live!



Conference In Great Britain Our Specialists on the Road November 6 – 8, 2017

**Educational Conference for X & Y Chromosomal Variations
including 47, XXY, 48, XXXY and 49, XXXXY**

Specialist Interviews Sessions and Conference

FEE: 148 Pounds (\$175 USD) per family.

50% discounted rates on accommodations at Thornton Manor in Wirral, Cheshire in the UK. Registrations will be accepted until October 6, 2017

Register Online



Colton's XXtraordinary Cause

A RARE AFFAIR - Raising Funds and Awareness for X&Y Variations

When: September 23, 2017 from 5 – 10 pm

Where: Globe Hill at Ronnybrook Farm in the Hudson Valley, Pine Plains, NY

What's Included: Dinner, Open Bar, Live Music, Dancing

Silent Auction includes VIP tickets to the taping of Live! with Kelly & Ryan as well as Harry!

WE ARE HAPPY TO ANNOUNCE THIS EVENT IS SOLD OUT!

THANK YOU TO THE HUTCHINSON FAMILY FOR ALL OF YOUR HARD WORK AND SUPPORT!



The 7th Annual Oktoberfest

“Here’s looking at you, kid!”

When: October 19, 2017

Where: The Ballroom at the Ben; 834 Chestnut Street, Philadelphia, PA 19107

Cost: \$90 per person

Philadelphia Fundraising Committee for The Focus Foundation invites you to our 7th annual event. The efforts of this committee continue to change the lives of children around the world as they have been enabled to reach their full potential.

Event underwritten by Melissa & George Stratts

Buy Tickets

Falling into the Holidays!

We are just around the corner from the start of the holiday season and shopping!



Don't forget to choose The Focus Foundation as your charity of choice through [amazonsmile](https://www.amazon.com/amazonsmile)!



Donations and Fundraising



AIG celebrated Global Volunteer Month with an Air Hockey Tournament to benefit The Focus Foundation.

AIG continues to support The Focus Foundation finding fun, creative ways to raise funds and awareness for the children living with X & Y Chromosomal Disorders. Participants included AIG staff members along with visitors from JLT Re.

Many thanks to the Family of Richard R and friends who contributed donations made in his memory to the Focus Foundation.

It was amazing to receive monies **in honor of Kayla for Gregg's 40th birthday.** What a great idea and wonderful celebration for **Gregg's next 40 years!**





Thanks to Crofton Papa John's and Wegman's in Waugh Chapel for saying "we care" with donations for our Family Affair at the Annapolis Conference.

THANK YOU ALL for your donations and fundraising. Your generosity is directly helping children with X and Y Chromosomal Disorders throughout the world!

DeXY's Corner



A Closer Look at Executive Function

Note from a parent:

My son is in Kindergarten and will be 7 next March. He has the many issues which I understand is linked to executive functioning disorder caused by the extra X. Does the extra X cause "delayed growth" in the Frontal lobes of the brain?

What is "executive function"? Dr. Samango-Sprouse has written two chapters on the frontal lobe of the brain and its relationship to executive function. She describes that "Executive function is a commonly used term to describe planning, initiation, selective and appropriate attention and finally execution of everyday tasks in our life. The Frontal Lobe is "the governor of the brain" and the largest region of the human brain. It allows us to complete simple as well as complex tasks by identifying, planning, organizing and executing them.

Individuals with executive dysfunction have deficits with initiating, selective planning, organizing and managing time and space. They also show weakness with "working memory" or visualizing the necessary steps in an organized fashion to finalize the goal in a timely and orderly manner. Executive dysfunction may impact on every aspect of a child or adult's life regardless of how intelligent that individual may be. Executive dysfunction is commonly associated with learning disorders both verbal and nonverbal types.

As with other manifestations of Learning disorders , executive function may occur in multiple family members. Deficits may be seen at any age but tend to be increasingly apparent as children move through the early elementary grades. The demands of completing schoolwork independently may often signal signs that there are executive dysfunction. Is present.

So, how can you help?

Here are activities and games that require players to follow directions, take turns, and plan strategies. Why does music matter? It offers concentration, sequencing, planning and attention. Evidence shows that music enhances areas of cognition, working memory and processing speed. Pathways can be built to executive function skills beginning very early in life. So, let's have fun in encouraging our creative thinkers to set and accomplish their own goals. And at the same time, we may build positive relationships and interactions with our children.

Ages 6- to 18-months

Simple games engage babies in your lap. Repetition helps infants build motor plans and practice execute them. Play Peekaboo and Pat-a-Cake. Hide a toy under a cloth and ask your child to find it. Teach the infant that signs that are meaningful such a the sign for eating or drinking so he can learn that planning and executing an action changes your behavior.

Ages 18- to 36-months

Toddlers become more physical at this age. Provide many materials and opportunities to try new skills. Throw and catch large soft balls, walk on a different surfaces whether are soft, hard, sandy or wet. Play follow the leader and fingerplays or songs and rhymes that have hand gestures to match because this requires planning, execution and integration of singing and gestures. Play 'Simon Says' since it requires listening, shifting and planning quickly

Ages 3 – 5

Movement challenges: songs and games support executive function because children must move to a specific rhythm and synchronize words to actions and the music. These tasks contribute to regulate, develop control and working memory.

Provide many opportunities for children to test themselves physically through access to materials such as climbing structures, balance beams and seesaws.

- Make simple obstacle courses
- Play skipping games like snake in the grass, jumping rope with a song
- Balance a beanbag on the head or an stuffed animal while hopping

Encourage attention control through quieter activities to reduce stimulation and focused listening—such as using a balance beam or yoga poses that include holding for 30 seconds, then one minute and upward. Encourage the effort and do not worry about the accuracy of the poses!

Play some music and have children dance fast, then slowly then in circles, diagonals. Each type of movement demands different and more sophisticated planning and execution.

Sing songs that repeat and add onto earlier sections (either through words or motions) are a great challenge to working memory, such as

- She'll be coming 'round the mountain
- The ants go marching one by one
- Ten in the bed... roll over, roll over!

Ages 5 – 7

At this age, children begin to enjoy games with rules. Games can exercise children's

executive function and self-regulation skills, and allow them to practice these skills in different ways.

Play games:

- For memory: Concentration, uncover cards and remember the location of matches. Play Go Fish, and Old Maid.
- Matching games: Crazy Eights and Uno
- Games that involve strategy such as Sorry! Parcheesi, checkers, mancala, and Chinese checkers.
- Physical activities and quick response games help children practice attention. They include freeze dance, musical chairs, Red Light, Green Light, Mother May I?

Ages 7 – 12

We begin to provide challenges and increase the complexity of games and activities. Playing cards and games exercise working memory in planning and strategy

- Play hearts, spades, bridge, and Rummy games require matching
- Poker and mahjong challenge older children
- Physical activities at this age organized sports become popular. They have to follow complicated rules and strategies, make quick decisions and respond to play.
- Children can become very skilled at jump rope, double Dutch, Chinese jump rope, and other such challenges. They must recall words of the chants while attending to the motor movements
- Learning to play an instrument requires attention and coordination as well as working memory.
- Crossword puzzles are available for all skill levels.

Activities for Adolescents

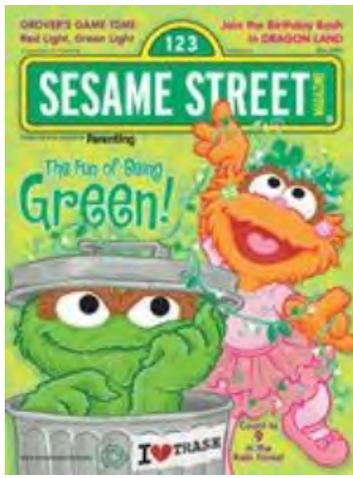
In school, adolescents are expected to be greater degree of independence and organization. This can be difficult. To help, break a project down into manageable pieces. Identify timelines to complete each piece. Keep a calendar of project deadlines and steps along the way. Create an excel sheet that provides the weight of the project and the deadlines. Calendar reminders can be put on cell phones so that organization becomes more automatic for the child prior to college.

Self monitor by setting a timer for increasing attention, time on task, understanding the task and finally completion of the task. Identify ways to reduce distractions, find a room that works for the individual sometimes quiet and lower light but for some it is music in background on a comfortable chair. Be creative to identify what makes your child the most productive and attentive. Multitasking will come, but first we need to tackle one task at a time. Build success then self confidence comes along with more mature skill development.

Use memory supports to remember information. Writing notes or learning mnemonic devices are powerful tools. Use the cell phone to place reminders on the calendar with alerts or recording notes from the many APPS for recording or obtain the notes ahead time and add to them as the teacher talks.

Gifts that Give

Thinking about those Holiday or Birthday presents? How about the gift of a magazine that keeps on coming throughout the year?! There's something for every age ! And you can find more on the internet!



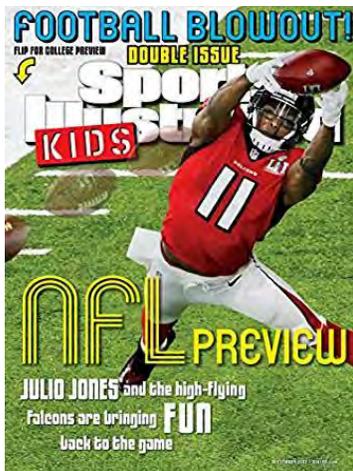
Sesame Street: Join Elmo, Cookie Monster, Big Bird and all of the other Muppets in the new Sesame Street magazine! Each issue of Sesame Street is full of stories, activities, games, science fun, a workbook, collectible poster, plus much more. For boys and girls ages 3 to 6.



ChickaDEE is a lively “hands-on” magazine that combines fun and learning to help kids ages 6-9 explore their world. Each issue includes puzzles, mazes, games, crafts, animal facts, simple experiments and more.



National Wildlife for Kids Ranger Rick is packed with awesome animal facts, stories, and photos; comic adventures; outdoor activities; wildlife puzzles, riddles, contests, and crafts; plus amazing nature discoveries to share with friends and use in school assignments. For kids 7 & up!



Sports Illustrated Kids is a sports magazine for kids ages 8-15 from the publishers of Sports Illustrated magazine.

Sharing is Caring

Thanks to Maddox's Gram for sharing this poem:

The **C**olor of Love
(for Maddox)

What color is this one? I ask.

He frowns.

Blue, he says firmly, then grins as

he peeks up at me, waiting

for my reaction.

Blue? I say in mock astonishment. Really?

He giggles.

Red, he laughs.

What color is this one? I ask.

Yellow! he cries.

Yellow is his favorite color, or at least his favorite word for a color. He searches the room for similar shades . . .

Yellow! Yellow! Yellow! . . .

grinning with delight as he shares

his 2 yr old view of the world.

Donations - we need your help for two special projects!

CASS-i

We are on the home stretch of finishing our seminal study focused on identifying ASD and language delayed (our undiagnosed X and Y infants) in babies prior to 12 months of age. We have tested a remarkable 800 babies and are looking to reach a final sample size of 1000. We now need to replicate our CASS-i (Comprehensive Autism Screening Scale for Infants) in additional medical settings. Our scale affords the opportunity for medical practitioners to identify infants at risk for ASD and LD by 9 months of age. The typical age of diagnosis for ASD is 3 years in an urban area and can be as late as 6 years of age in rural settings and even later for those with language learning disorders. We know that infancy and early childhood is a critical time for brain development.

The CASS-i allows early detection during infancy, a remarkable period of brain growth, then resulting in earlier treatment and greater recovery rates. This is a formidable goal and we need your help! The benefits of early detection cannot be overstated! 50% of children with ASD recover if identified by 24 months of age — one year earlier could lead to every more children recovering sooner! We are going global and bringing our vision of hope and help to every child who is affected by one of these disorders! We need your help to finalize the CASS-i and to investigate the ocular biomarkers in children at risk for social anxiety, reading dysfunction and motor planning deficits. The impact of your support could be immense for thousands of children throughout the world!



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. Many thanks to the Families employed at companies who have doubled their donations through community partnership programs!

[Donate Online Today](#)



Copyright © 2017 The Focus Foundation, All rights reserved.

Want to change how you receive these emails?
You can [update your preferences](#) or [unsubscribe from this list](#).

MailChimp