



## **The Magnifying Glass – A Newsletter**

**October 2014**

**Vol. 4, Issue 2**

Dear Focus Foundation Friends and Families,

We are preparing to mark the upcoming 10th anniversary of The Focus Foundation. In just a decade, each of you has helped to change the lives of children around the world, helping them realize the beautiful potential within each of us and giving parents and families hope and help.

Our research has made The Focus Foundation the go-to source on X and Y chromosomal disorders here in North America, Europe, and as far away as Australia. Leading scientists are requesting our slides to present at worldwide meetings to “enlighten medical doctors” on the benefits of Early Hormonal replacement and making the case for prenatal diagnosis. Our many published papers on early detection, treatment and recovery are changing the medical landscape.

Parallel with our research is our work, child by child, family by family, year by year, to help obtain services, advise and assist with medication, and provide educational and interventional strategies before symptoms manifest. The Focus Foundation works with the largest cohort of children with X and Y Chromosomal Variations in the world. Our work and our results are making dramatic differences for each child and each family.

Together, we are telling the story that chromosomal disorders are common, that there is treatment, and that it works if diagnosed early, when brains change the fastest.

There are no words to thank you all for your unwavering belief in our work and your generosity that funds its continuing progress. These successes are yours, and the future is amazingly exciting. We are close to our goals of universal testing and helping to create a different future for the next generation of children with these disorders and it is because of YOUR help, YOUR work and YOUR support. Thank YOU!



Our Greater Philadelphia Fundraising Committee has become a critical part of our fundraising team and their efforts shine through this incredible event. Oktoberfest “Under the Big Top!” was a great success! We were happy to see many families from the region, with more than 400 people in attendance. More than \$200,000 was raised for The Focus Foundation, and the event was featured on FOX Philly news by reporter Jennaphr Frederick (visit <http://www.myfoxphilly.com/story/26784062/boys> to check out the story). It was a great night, and we hope to see you there next year! The Stratts family and the entire Oktoberfest committee has been wonderful--thank you!

Sincerely,

*Dr. Carole*

## **What's New? Upcoming Fundraisers and Events!**

-Dr. Samango-Sprouse will be speaking at a Natera webinar (**Early**

**Detection Means Early Treatment for Children with Developmental Delays due to X and Y Chromosome Aneuploidies)** regarding her work on studying X and Y chromosome aneuploidies on November 5th. This is part of our continued outreach to the medical community at large and worldwide.

-HOW CAN YOU HELP? Parents and children have been staging **grassroots fundraising campaigns** across the United States, hosting TFF awareness and fundraising events in lieu of birthday parties and gathering groups to run in local races! Contact us for ideas on how you can raise funds for The Focus Foundation.

## **CONFERENCES & RESEARCH**

The Focus Foundation has made significant progress in working towards a better life for children.

**We presented at the following venues:**

- o Pediatric Academic Societies' (PAS) for Pediatric Research,
- o American College for Medical Genetics Clinical Genetics Meeting.
- o CME Satellite Dinner Symposium
- o Dartmouth Pediatric Endocrinology Education Day
- o The Eastern Society for Pediatric Research,

- In October, Dr. Samango-Sprouse and Corley Gibbs, one of our two new research assistants, presented at the **17th Society for the Study of Behavioral Phenotypes International Research Symposium and Educational Day**. The poster presentation discussed our girls with 48, XXXX and their neurodevelopmental profiles. Dr. Sprouse presented research on the benefits of androgen on brain function in 47, XXY to an international audience. This symposium highlighted a wide variety of research and clinical work in neurodevelopmental and neurobehavioral

aspects of genetic disorders from all over the world.



Dr. Samango-Sprouse, Corley Gibbs, and Diana Sisson, our newest research assistant, presented at the **64th annual meeting of the American Society of Human Genetics** - the largest human genetics meeting and exposition in the world. This meeting provides a forum for the presentation and discussion of cutting edge science in all areas of human genetics. Dr. Sprouse highlighted our

continuing research on the benefits of androgen on brain function in XXY. One poster demonstrated the variability and strengths of our girls with 48, XXXX. A second poster highlighted the impact of childhood apraxia of speech on Kleefstra syndrome, a very rare disorder.

Next month, Dr. Samango-Sprouse will be presenting at the **American Speech-Language-Hearing Association's** conference in Orlando, Florida.

In the last year, Dr. Sprouse published two articles:

- **Expanding the phenotypic profile of boys with 47, XXY: the impact of familial learning disabilities** in June
- **Identification of infants at risk for autism spectrum disorder and developmental language delay prior to 12 months** on ePub in April, and will be in print in December.
- We had our **Eleventh Annual 49er Conference** in Annapolis with 19 families including 8 children under 6. Our keynote speakers, Robert Bullock, Esq., CELA, CAP, and Abigail Wolf, Esq. from The Elder & Disability Law Center, shared **Planning the Future for a Child with**

Disabilities. We initiated two exciting studies during the conference:

- o Our senior PT, Francie Mitchell, has received her Doctorate of Physical Therapy (DPT) with a Capstone project on the 49ers. She used the PEDI-CAT to evaluate function in daily activities. This will help us evaluate the functional skills of the boys and identify goals.

- o The second study is looking at methylation on the extra X. We will be collecting saliva (spit) samples to collect DNA that will provide useful genetic information in conjunction with our neurodevelopmental assessments.

As always, we would like to thank all of the families that participate in our studies, allowing us to expand understanding of these rare disorders. Please contact our office if you would like to receive a copy of either of these articles.

## **WHY OUR RESEARCH MATTERS:**

### **Making a Difference One Family at a Time**

I have a son who is 5 yrs 4 months old. He is recently diagnosed with Karyotype XXY on his chromosome analysis. He has been facing problems like Receptive and Expressive Speech delay, attention problems (major), ADHD, behavioral issues (like temper tantrums, frequent meltdowns, etc.) He has major issues with speech like comprehension problems, fluency, decreased vocabulary and he struggles with sentence structure (developmental dyspraxia).

He has been having all of the issues which kids with XXY face right from birth; it was just none of the doctors were able to diagnose him including his pediatrician until just now when we insisted that we want every possible test to be done to pinpoint his problems. We have done quite

some research in the last few days and are so happy to have found Dr. Samango-Sprouse, an expert in Sex Chromosome Disorders.



## **Dexy's Corner – Succeeding in School**

Reading is sometimes challenging, but DeXY has put together some strategies to make learning easier! There are apps, organizers, and exercises to help you do your best!

### **Reading - Passage Comprehension**

Reading skills involving comprehension and recall of facts starts with the parts of speech, and extend through language growth, recognition, memorization, fluency, and story recall. Here we recommend apps, activities, and strategies for succeeding in reading homework! Reading should be fun, so we recommend these games:



[Compare A Twist](#) uses compare and contrast practice to improve fluency and comprehension, and general academic progress. (K-12)

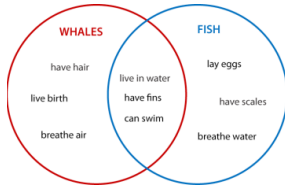


The Smarty Ears App, [Reading Comprehension Camp](#) is designed to encourage language growth and reading comprehension. It has multiple levels, 50 stories, and the ability to create personal stories. (grades 2 - 7)

More than just games, these activities and organizers can help you with your reading skills by letting you act out the big scenes, or write down facts in order to help them make sense!

- Act out a book to better understand the story.

- Draw out the main scenes
- Use a graphic organizer when working on comprehension - like these!

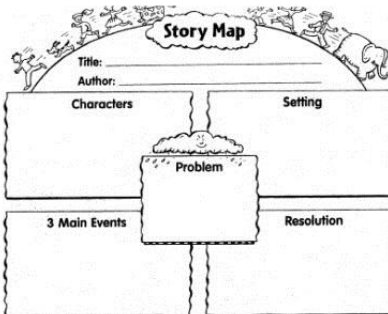
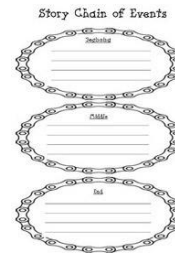


### Venn-Diagrams

- Used to compare or contrast information from two sources. For example, comparing two Dr. Seuss books.

### Storyboard/Chain of Events

- Used to order or sequence events within a text. For example, listing the steps for brushing your teeth.



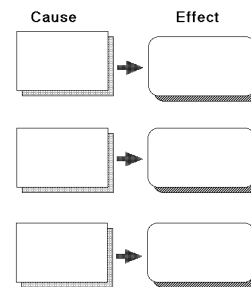
### Story Map

- Used to chart the story structure. These can be organized into fiction and nonfiction text structures. For example, defining characters, setting, events, problem, resolution in a fiction story; however in a nonfiction story, main idea and details would be

identified.

### Cause/Effect

- Used to illustrate the cause and effects told within a text. For example, staying in the sun too long may lead to a painful sunburn.



For free graphic organizers, follow this link!

- [http://www.educationoasis.com/curriculum/graphic\\_organizers.htm](http://www.educationoasis.com/curriculum/graphic_organizers.htm)



## **“I need help with my reading homework!”**

When working on your reading assignments, there are four different types of questions you may be asked about your reading assignments.

- **"Right There"**

- o These questions ask you to look in the text to find the one answer, such as a word or a sentence.

- o *Example: Who is Frog's friend? Answer: Toad*

- **"Think and Search"**

- o You will have to think about facts in the text, usually in more than one place - you will have to "think" and "search" through the passage to find the answer.

- o *Example: Why was Frog sad? Answer: His friend was leaving.*

- **"Author and You"**

- o Use what you already know, with what you have learned from reading the text.

- o *Example: How do you think Frog felt when he found Toad? Answer: I think that Frog felt happy because he had not seen Toad in a long time. I feel happy when I get to see my friend who lives far away.*

- **"On Your Own"**

- o Questions are answered based on your prior knowledge and experiences. Reading the text may not be helpful to you when answering this type of question.

- o *Example: How would you feel if your best friend moved away? Answer: I would feel very sad if my best friend moved away because I would miss her.*

- Want to try it out? Go to these links to practice!



- <http://www.englishmaven.org/Pages/Paragraph%20Correction.htm>
- <http://mrnussbaum.com/readingpassageindex/>
- <http://englishforeveryone.org/Topics/Reading-Comprehension.htm>
- <http://www.k12reader.com/reading-comprehension-guide/>



## DeXY's New Recipe for Fall

### Fall Leaves Sugar Cookies

#### Ingredients

- 1 cup Land O Lakes Butter, softened
- 1 cup sugar
- 1 egg

- 2 tablespoons milk
- 1 ½ tablespoons almond extract
- 2 1.2 cups all-purpose flour
- 1 teaspoon baking powder
- Red, orange, and yellow gel food colors
- Coarse white, orange, and/or yellow decorator sugars

#### Directions

1. Combine butter and sugar in bowl; beat at medium speed until creamy. Add egg, milk and almond extract; continue beating until well mixed. Add flour and baking powder; beat at low speed until well mixed.
2. Divide dough into thirds; place each portion into separate bowls. Add very small amount of red gel food color to 1 portion; mix well. Add additional red gel for desired color, if necessary. Repeat with remaining dough portions and gel food colors.
3. Shape each dough into ball; flatten each ball to 1/2-inch thickness. Wrap each in plastic food wrap; refrigerate 2-3 hours or until firm.
4. Heat oven to 400° F. Working with half of each dough color (keeping remaining dough refrigerated), drop tablespoon-size pieces of dough from each color onto a well-floured surface in a random pattern with dough pieces touching. Roll out dough to 1/4- to 1/8-inch thickness,

forming a marbled design. Cut with 3-inch leaf-shaped cookie cutters.  
5. Place cookies, 1 inch apart, onto ungreased cookie sheets. Sprinkle with decorator sugar. Bake 7-9 minutes or until cookies are lightly browned. Cool 1 minute on cookie sheet; remove to cooling rack.

from: <http://www.landolakes.com/recipe/1761/fall-leaves-sugar-cookies>

## **Remember The Focus Foundation This Year!**

Did you read the letter featured above in this newsletter from a relieved and happy parent? We at The Focus Foundation know how important an early diagnosis can be in the development of a child. We also know that getting the right information at the right time is crucial for parents, especially when receiving a diagnosis. We think every parent, healthcare provider and educator should be informed about early testing and treatment for X and Y Chromosome Variations and other related disorders. We can't do it alone though. Your tax-deductible donation to The Focus Foundation supports our efforts to disseminate information about and continue researching X and Y Chromosome Variations and its possible treatments. Consider The Focus Foundation in your year-end giving this year, so fewer parents have to deal with a scary, unknown diagnosis. Help us focus on research that will foster a change in the lives of children and families affected by this and related disorders. As we get closer to the end of the year, consider giving to The Focus Foundation in support of X and Y Chromosomal Variation research, awareness, and advocacy.

*Donating online is easier than ever, just click on our page below. Your generosity will improve the lives of many individuals!*

<http://www.thefocusfoundation.org/FF/supportingthefoundation.php>



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