

The latest information about conferences, education, research opportunities, clinics and more.



## THE EVEN EXCHANGE AXYS Newsletter - Fall 2017

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[For more news, upcoming events and information visit the AXYS website](#)

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### Upcoming Events

Click [here](#) to visit the support group page where you will find more detail about each of the following U.S. events. International XXYY events can be found [here](#).

**October 26:** Online Webinar, 8:30pm Eastern / 7:30 Central / 6:30 Mountain / 5:30 Pacific. Dr. Nicole Tartaglia: X&Y Chromosome Variations from Birth to Age 5 - Developmental and Medical Care

**November 4:** NEXXYS (the New England XXY Support Group) 1:00-4:00pm, Braintree, MA

**November 5:** 12:00-2:00pm, AXYS San Francisco Bay Area Support Group, Mountain View, CA

**November 5:** 2:30-5:00pm, NY Tristate Support Group, Montclair, NJ

**November 28:** Online Webinar (see below)

**January 25:** Online Webinar (see below)

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## AXYS Webinars

AXYS wishes to thank Endo Pharmaceuticals and Roche Diagnostics Corporation for their support of the 2017-2018 AXYS Webinar Series

**October 26, 2017**

**8:30pm Eastern / 7:30 Central / 6:30 Mountain / 5:30 Pacific**

**Dr. Nicole Tartaglia**

**X&Y Chromosome Variations from Birth to Age 5 -  
Developmental and Medical Care**

Everything you wanted to know about early child development in X&Y chromosome variations! We will discuss monitoring of developmental milestones, common developmental concerns in X&Y chromosome variations, when and how to evaluate for and access early intervention therapies. We will also discuss medical screenings and procedures important in this age group. I will share information about the exciting new research opportunities for infants and young children with X&Y chromosome variations including the eXtraordinary Babies Study, the TRIXY study, and the TESTO study. [REGISTER](#)

**November 28, 2017**

**8:30pm Eastern / 7:30 Central / 6:30 Mountain / 5:30 Pacific**

**Cory Nourie**

**Transition to Adulthood**

**January 25, 2018**

**8:30pm Eastern / 7:30 Central / 6:30 Mountain / 5:30 Pacific**

**Dr. Adrian Dobs**

**Promoting Good Health in Adults with XXY**

*Note: Registration for each webinar will be made available, on the AXYS website and through an email reminder, approximately one month prior to the scheduled date.*

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## AXYS Launches Professional Directory

[View](#)

**View a list of medical and other types of professionals who are known to provide services to those with an X or Y variation.**

[Add](#)

**Submit your recommendation of someone to be listed in the directory.**

*(Submissions will not appear until reviewed by staff.)*

Please also be sure to always check the AXYS Clinic & Research Consortium (ACRC) page to see if there is a [clinic](#) within a reasonable distance of you.

Please inform AXYS via [info@genetic.org](mailto:info@genetic.org) if you become aware of any changes to the provider's service.

**Disclaimer:** AXYS maintains a directory of healthcare providers and education advocates as a service to the X and Y Variation Community. These names are provided by members of the community. AXYS

makes no representations or warranties regarding their clinical competencies or specialized knowledge regarding sex chromosome aneuploidy. AXYS reserves the right to edit the submitter's comments.

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## New research opportunities

### **The eXtraordinaryY Babies Study: Researching the Natural History of Health and Neurodevelopment in Infants and Young Children with Sex Chromosome Trisomy**

**X and Y variations:** XXY, XYY, XXX, XXYY, and other sex chromosome variations are eligible.

**Genders being studied:** All genders **Age Range:** 6 weeks to 12 months old at the time of enrollment.

### **TESTO Study: Testosterone Effects on Short-Term Outcomes in Infants with XXY X and Y variations**

**Genders being studied:** Male **Age Range:** Birth to 12 weeks of age at enrollment

### **Growing up with X and Y Chromosome Trisomy: The TriXY Study**

**X and Y variations being studied:** XXY, XYY, and XXX

**Genders being studied:** All genders **Age Range:** 12 months to 5 years

**Other eligibility requirements:** Non-mosaic diagnosis, born at 37+ weeks gestation

### **Accessible Genetic Counseling Information for Parents Undergoing Non-Invasive Prenatal Screening (NIPS)**

**X and Y variations being studied:**

Applicable to parents of children with any X and/or Y chromosome variation

**Genders being studied:** Males and Females **Age Range:** Adults (18+)

**Other eligibility requirements:** Mothers and fathers who have had non-invasive prenatal screening

[More details about these studies and others](#)

**Participate in Research:** AXYS maintains a webpage called "Research Opportunities for Families." There are many ways your participation can make a difference. Not all opportunities involve travel. In fact, some simply require the completion of an online survey. Take a look [here](#).

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**XXY**

## Criminal Justice Support Program

by Gary Glissman, AXYS Board Chair

I wanted to let everyone know that AXYS is in the process of working on several new major initiatives related to assisting individuals and families that request help with legal problems. This is a topic we seem to get consistent requests for information and support and some of the stories we hear about people being imprisoned or facing lengthy prison sentences instead of receiving treatment are simply heart-wrenching.

A large percentage of the SCA population *do not* experience legal problems but research indicates they can be more at-risk compared to non-SCA populations, particularly when a person has more risk factors in their life such as late diagnosis, lack of effective medical treatment or support, reduced family involvement, learning difficulties and low income. One of the important goals we are working on involves helping legal professionals and health care providers understand that certain individuals with x and y chromosome variations can be more at risk for criminal charges but this risk can be significantly reduced if they are provided effective medical, psychological and educational support from childhood through young adult ages.

Social adaptation along with general adherence to cultural norms and legal compliance are complicated, learned behaviors that are developed over time and involve coordination between

many areas in the brain. People are not born with these skills, they have to learn them. Research has demonstrated that some individuals with SCA conditions can have an impaired ability to learn from positive and negative experiences that many non-SCA individuals simply take for granted. This learning difficulty can lead to a marked lack of insight, poor judgement and behavioral indiscretion that can result in criminal charges. The problem is often magnified because the individual is charged as an adult due to their chronological age. We are working with staff from Emory Law School on publishing a research paper with case studies that illustrate the problems that we hope will motivate courts to consider treatment vs punishment.

In the past AXYS developed a "white paper" document for attorneys and courts to improve their understanding about KS and the developmental challenges that can be involved with some individuals. While this has been helpful for many families, we need more detailed and compelling documents to convince courts and health professionals that are not experienced with SCA conditions. We need clinical and legal experts to assist us with drafting documents that explain the specific kinds of neuropsychological challenges some KS individuals have and treatment interventions that could help them develop more effective skills. And more published research papers that explain how KS and other SCA conditions can influence people being unfairly punished because of a genetic condition. All of this will require time, expense and other resources to not only develop these documents but also find ways to get them into the hands of people that can make a difference. We would greatly appreciate any assistance you can provide to fund or support our efforts so that we can help people before they are facing serious, life-changing consequences.

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**XYX**

## **Mental Health: Issues and Challenges in XYX**

A presentation by Erin Torres, Nurse Practitioner

**From the AXYS Family Conference at the Children's Hospital in Aurora, Colorado which was held on June 24-25, 2017.**

[Click here to view this YouTube video](#)

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**XXX**

## **Trisomy X Panel and Q&A**

with Rebecca Wilson, PsyD and others

**From the AXYS Family Conference at the Children's Hospital in Aurora, Colorado which was held on June 24-25, 2017.**

[Click here to view this YouTube video](#)

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**XXYY**

## **The XXYY Project: XXYY Sibling Project Reaching New Audiences**

by Mika Nagamoto and Jenna Nagamoto

As research assistants in the eXtraordinarY Kids Clinic, Children's Hospital Colorado, we helped to manage and organize research data. We also provided support for research patients and our work included frequent interactions with families. Often brothers and sisters accompanied families during appointments to the clinic. Our initial interactions with siblings of boys with XXYY were limited to brief conversations in waiting rooms as their brothers completed visits with Dr. Tartaglia and the rest of the clinic team. [Read more...](#)

[Click here to view this YouTube video](#)

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## Capital Cup Fundraiser

Some months ago, Michael Kvarme reached out to AXYS and told us that, earlier in the year, his family had lost their youngest son, Jimmy Kvarme, to cancer. He also informed us that he had been invited to participate in this year's Capital Cup golfing tournament and had decided to make AXYS one of two recipients of his fundraising efforts. On his promotional materials, he shared his reasoning to his supporters as follows:

"AXYS is an organization that provides a range of direct services as well as research to help those with X and Y chromosomal variations. Jimmy was born with one of those, called Klinefelter syndrome. No one would have ever known this about Jimmy in large part because of the resources provided by AXYS. Julie previously served on the board and I provided legal assistance to the organization a few years ago. AXYS has a detailed website at genetic.org, organizes national conferences and provides telephone support by trained volunteers."

As a result of the Kvarme family's kindness and generosity, AXYS has so far received over \$6500 in donations. Thank you!

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## All Variations

### Coming soon... New Educational Content

AXYS is working on an addition to its website that will provide **more and better content regarding educational options, recommendations, services** and more. We recognize and appreciate that parents should not have to struggle in the dark or reinvent the wheel when so many others have already successfully tackled the numerous educational challenges facing those with an X and Y variation. You can expect to see the section no later than this coming January.

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## AXYS Annual Fund

### Please consider AXYS when planning your 2017 charitable giving

A message from Robert Miller, AXYS Executive Director

We hope you've seen the results of our renewed commitment to ensuring that the X and Y variation community receives the latest and best consensus and evidence-based information. Even if you were unable to join last summer's conference, we hope you've seen the **tremendous amount of content we added to our website** from that event. Even if you've been unable to visit one of the AXYS Clinic & Research Consortium (ACRC) clinics, we hope you're aware of our beginning success in **bringing together all of the leading medical institutions in the U.S.** and creating an environment where dedicated clinicians and researchers are working together. And even if you've, personally, never had the need to

access our telephone or email support system, we hope you understand that it is there for you - should you ever need it - and that it has, and will continue to, **help countless numbers of families.**

Add to that:

- our ever-improving website
- the brand new professional directory
- the new webinar series featuring X and Y variation experts
- the AXYS support groups
- regular postings on social media
- consensus based treatment recommendations being developed by the ACRC

and other efforts too numerous to mention, and you'll understand why I feel so comfortable asking you to keep us high on your list for your charitable giving.

You can expect to see our online campaign in the second half of November and our direct-mail campaign arriving in your mailbox sometime around Thanksgiving. Of course, you could always check that off your to-do list now by clicking [here!](#)

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AXYS | (888) 999-9428 | Email | Website

STAY CONNECTED:



AXYS, P.O. Box 861, Mendenhall, PA 19357