



# The Even Exchange

For more news, upcoming events and information visit the AXYS website at [www.genetic.org](http://www.genetic.org)

## Present and Future Enhancements at AXYS

### A Message from the Chair of the Board of Directors

by Myra Byrd

AXYS would like to welcome Robby Miller as our Interim Executive Director. Robby previously served as the Executive Director of the National Fragile X Foundation and has worked with many other patient advocacy organizations as a consultant. He will be assisting us in our search for a permanent Executive Director over the next few months as he continues in his role developing the AXYS Clinic and Research Consortium.



This summer, AXYS will host a meeting of professionals in the AXYS Clinic and Research Consortium (ACRC). The meeting will be held in conjunction with a Scientific Conference on X and Y Chromosome Variations at Emory University in Atlanta, Georgia. We are excited about bringing together the professionals who are committed to providing care for individuals with one or more X and/or Y chromosomes. These ACRC meetings are the foundation for the sharing of knowledge that is so vital to the success of our clinics and fostering research for patients and families.

I am also pleased to tell you that AXYS has engaged a web design firm to completely overhaul our website. Once completed, we are confident that it will be a site that you will want to regularly visit for the latest news and knowledge, all presented in an easy to find and family-friendly format.

There are many exciting changes ongoing and planned at AXYS. Please bear with us during this time of transition. We are, more than ever, committed to our mission to help individuals with an X or Y chromosome variation lead, healthier, fuller and more productive lives.



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## The AXYS Clinic & Research Consortium (ACRC)

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by Robert Miller, AXYS Interim Executive Director



In 2015, the AXYS Board of Directors voted to approve the development of a consortium of clinics, organized, coordinated and funded, in part, by AXYS. Though each clinic operates independently, as members of a consortium they collaborate with one another, share informational resources, and have the opportunity to participate in joint research projects. In addition, AXYS organizes annual meetings of the consortium at which members meet to discuss topics important to the SCA community. The goal of AXYS is to ensure that all families impacted by any of the chromosomal variations have access to the best available evaluation and treatment or treatment recommendations. The AXYS Clinic & Research Consortium is one of the important ways we are achieving that goal.

Read more about the clinics [here](#)

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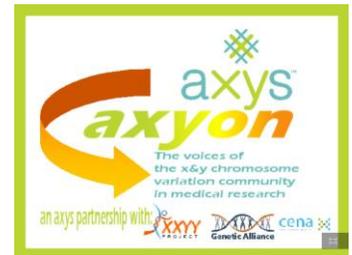
## Advance research by participating in the AXYON Registry

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by Susan Howell, MS, CGC, MBA and member of the AXYS Board of Directors

*We need your help to advance research by participating in the AXYON Registry!*

In December of 2015, AXYS was proud to launch the AXYON Registry, a grant-funded, web-based, self-reported registry specific to X and Y chromosome variations. This registry allows people like you to contribute information specific to your experience with an X and Y chromosome variation. Your contribution to the registry is CRITICAL to helping advance research in X and Y chromosome variations!



To date, 206 people have entered their data into the registry, but many more are needed. The more people who enter information into the registry, the more powerful and useful the information is to researchers, and the more likely they will use our data and publish scientific papers about X and Y variations. This will lead to: More awareness; Better educated professionals; More funding to help promote further research; development of new treatments.

Common Questions:

1. If I participate in the registry, do I have to disclose my identity?

Answer: No! While some people choose to disclose their identity to allow researchers to possibly contact them for additional studies, the registry is designed to be anonymous if you prefer, so please do not let this deter you from participating!

2. What types of questions is the registry going to ask me?

Answer: All sorts of questions, which is why it's so powerful for research! The registry collects information about how a person was diagnosed, what types of medical and developmental problems they may have, lifestyle questions, quality of life questions and much more! The more information we provide for researchers to use; the more researchers we can attract to take an interest in X and Y chromosome variations!

3. What if I don't want to answer all of the questions? What if I don't have time to answer them all?

Answer: No problem! You can skip questions you would rather not answer and if you run out of time, save your answers and come back to the registry later. Just remember, the more information you enter into the registry, the more information researchers could possibly use to help our cause!

4. Does the registry collect information about my rare type of X&Y chromosome variation?

Answer: Yes! The registry collects information about ALL X&Y chromosome variations! Even Mosaic variations with multiple types.

5. Who can access my information?

Answer: Only researchers who apply and are approved by AXYS to use the registry data can be given a data export for their research project and publications. AXYS wants to insure that only top-notch researchers who want to help our cause are able to use our data.

Click [here](#) to access the AXYON registry and enter your information today.

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## May is National X and Y Chromosome Variation Awareness Month!

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by Sandy Schindler, AXYS Board of Directors

Awareness is crucial. With awareness comes greater knowledge of X and Y chromosome variations, making it easier for those in need to find help and support. But awareness is only the first step. It takes resources, services, and support to let individuals and families know they are not alone.

AXYS is working to raise awareness for X & Y chromosome variations. With the help of many of you, we have been successful in getting proclamations Colorado, Delaware, Georgia, Illinois, Mississippi, New Hampshire, New Jersey, West Virginia, and we hope to receive and Pennsylvania. As you can see, we



signed by the Governors of Arizona, Kansas, Massachusetts, Maryland, Oklahoma, Vermont, Wisconsin and proclamations from Florida, Texas still have a lot of work to do!

It is our hope to have all 50 states supporting a *national* awareness month. The easiest way to get this accomplished is for each and every one of you to go to [www.whitehouse.gov/contact](http://www.whitehouse.gov/contact) and fill out the online form to President Obama. Simply select administration in the subject box and type in "Proclaim May as National X & Y Chromosome Variation Month" and include why this is so important to you in the "What would you like to say?" box. Some of the benefits of having a National Awareness Month are:

- a platform to continually increase awareness

- earlier diagnosis and early interventions
- medical professionals who are educated on X and Y chromosome variations
- eliminating stigmas associated with X and Y chromosome variations
- greater fund raising opportunities
- easier to get funding for much needed research

We need to let our voices be heard! We are so close in getting a Presidential proclamation but we need YOUR help. Without the support of everyone who is affected by these conditions, it makes it harder for us to accomplish all of the above. If you are tired of having to educate medical professionals, tired of keeping your or your child's condition a secret, tired of teachers not understanding the challenges you are dealing with, then you need to let your voice be heard and advocate for change!

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## **XXY** Specialty Clinics

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by Gary Glissman, Vice-Chair, AXYS Board of Directors

As you will read in other articles in this newsletter edition, there are many exciting developments taking place in research and treatment for all SCA conditions and especially for XXY. The continued growth of the eXtraordinaryY Kids clinic at Children's Hospital of Colorado in Denver has been equal to their name in recent years (extraordinary) and their interdisciplinary treatment model has been published in journal articles and now is being replicated by other well-known health systems in the U.S. You can read more about their model program and how successful it has been [here](#) and the full text version [here](#).

There is no way to adequately describe the importance of these specialty clinics. The ability for parents and individuals dealing with various XXY challenges to obtain the best and most knowledgeable care and support is so critical for getting positive results. For years it has been difficult at best to find experienced health professionals that understand XXY. Often it was impossible. The results of inaccurate diagnoses and ineffective treatment interventions have been devastating and impacted tens of thousands of individuals. But we're seeing positive changes in professional awareness, a willingness to learn, and interest in more targeted care. This will really accelerate with new clinics opening around the country.

Read the article, share it with your local physicians and educational professionals. Ask them if you can send them other research articles or help them connect with one of the specialty clinics. Help them understand that XXY can be a unique condition with unique characteristics that will benefit from knowledgeable treatment or support. If you're in a larger urban area, talk with your hospitals and see if they would be interested in starting a specialty clinic. Show them the locations that are taking action. Think about getting involved with a support group, either an existing one or maybe start one. AXYS can help with this. Use the support group to raise awareness.

This is an exciting time and we're seeing changes that have taken decades to achieve. The time is now and it is important that you take action and get involved. Please join us in making this a whole new world for the current XXY generation and future ones to come. Remember, there are 500,000 XXY people counting on you and 4,000 new ones every year.

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## **XXY NIH Research Study**

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by Myra Byrd, Chair, AXYS Board of Directors

The biggest news for our 47, XYY community is the study underway at the National Institutes of Health. There is so little good information on XYY compared to other variations and the more who participate the better! Everyone who has been a part of this study has reported how wonderful the experience was.

The research study at the NIH in Bethesda, Maryland (outside Washington, DC) involves a 2-day visit that includes a physical, MRI scan of the brain, a visit with psychologists experienced with autism, an appointment with the craniofacial and dental clinic, a thorough diagnostic mental health appointment, and cognitive testing.

During the visit, parents have the opportunity to ask the doctors questions and will receive immediate feedback. After the visit, an in-depth report will be written and provided to the families. The goal is to have as many as 100 families participate before January 2017 so the full range of experiences can be observed and then published in a journal that pediatricians can read.

The age range is 5 to 25 years of age but parents of 4 year olds can contact the study now for participation when their son turns 5. Round trip air travel within the US and 3 nights of hotel lodging are provided for participating family members, along with \$300 participant payment for the son and \$100 for each parent, in addition to a meal allowance of \$20/person/day. Families can extend the visit by paying for additional nights at the hotel to explore Washington DC (like the Air & Space Museum or National Zoo).

Jonathan Blumenthal is glad to answer any questions you have and you are welcome to send an email to [jb364e@nih.gov](mailto:jb364e@nih.gov) or call him at 301-435-4516.

Please be sure to read the announcements in this newsletter regarding regional support group meetings. If you are interested in starting a local support group, contact us at [info@genetic.org](mailto:info@genetic.org) for assistance.

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## **XXYY Going About the Business of Solving Problems Associated with XXYY**

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by Dr. Sharron Close, Chair, AXYS Professional Advisory Committee

Individuals with XXYY and families who care for them encounter a multitude of problems associated with physical symptoms, medical care, psychosocial health and educational/vocational issues. From a patient's or a family member's point of view, the compounding of problems can become overwhelming and seemingly unmanageable. At times it may seem that nobody sees, hears or understands what life is like for those who have XXYY.

Useable solutions come from truly gaining an in-depth understanding of problems. It has long been a criticism of research that results are mostly "descriptive" without offering help or treatment alternatives that would benefit patients and families. While there is truthfulness to this, the descriptive parts of research provide the foundation for how scientists begin to understand the scope of problems. Advances in science

move systematically with descriptions and evidence for treatments all building together. This requires the attention and a lot of work from different types of researchers, different institutions and, of course, different types of funders. Some things can be solved with research and some things can be solved with advocacy and policy. The “voices” of those affected by XYY will help direct what and how problems can be solved.

This year, Emory University is launching a study entitled, “Living with XYY: Voices of Patients and Caregivers” as a step in the process of deeply probing problems so that solutions can be worked upon. The Emory Team is led by myself and Dr. Kristy Martyn with assistance from 2<sup>nd</sup> year Doctoral student, Amy Blumling. The study has been funded by the Ryan Scovell Memorial fund and will soon be seeking 10 adult males with XYY and 10 adult caregivers of XYY males to participate in interviews. Results from this study will help the research team as well as other researchers to form new ideas about possible treatments and other interventions in the future. The Emory Research Team is deeply invested in bringing solutions that matter back to patients' especially those affected by X & Y Variations.

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## **XXX** Research opportunities available to women and children with Trisomy X

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by Erin Frith, AXYS Board of Directors

There are multiple current research opportunities available to women and children with Trisomy X. Please consider participating in one of the following to help researchers learn more about this condition. Here is your chance to make a difference!

All ages: XYY registry: <http://www.genetic.org/Action/AXYSXYYONRegistry.aspx> An AXYS backed project to provide valuable data to researchers interested in Trisomy X and other SCAs

Ages 7 and up: <http://geisingeradmi.org/care-innovation/studies/sex-chromosomes/> As part of Geisinger's MAP study, we are interested in understanding the extent to which family background influences learning and behavioral symptoms in individuals with Turner syndrome, Klinefelter syndrome, and Trisomy X.

Ages 5 - 25: <http://www.nimh.nih.gov/labs-at-nimh/research-areas/clinics-and-labs/chp/sex-chromosome-aneuploidy.shtml> The Child Psychiatry Branch of the National Institute of Mental Health is conducting a study of the effects of sex chromosomes on human brain development.

Ages 18 - 55: [http://clinicalstudies.info.nih.gov/cgi/wais/bold032001.pl?A\\_12-HG-0181.html@chromosome](http://clinicalstudies.info.nih.gov/cgi/wais/bold032001.pl?A_12-HG-0181.html@chromosome) This study is seeking to learn more about the genetic and clinical characteristics of disorders related to the X and Y chromosomes.

Please visit <http://www.genetic.org/Action/CurrentResearch.aspx> for other current research opportunities.

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## **EVENTS**

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**May 14, 2016:** Staten Island **Bowl-a-thon Fundraiser for XYY Project.** 2:30 - 5:00pm, Rab's Country Lanes, 1600 Hylan Blvd., Staten Island, NY 10305. Cost is \$25 includes bowling, shoes, pizza & soda. Sponsored by the "All Island Kiwanis Club." Raffle baskets as well as a 50/50. The bowling alley is donating 10% of their proceeds.

**June 5, 2016: AXYS Midwest Support Group** meeting 1:00-3:00pm. This meeting is for **all X & Y chromosome variations**. Childcare is not available at this meeting. Light refreshments will be provided. 330 Arkansas, Lawrence, Kansas 66044: Conf. Rm. A - lower level. Please RSVP Julie Urban ~ 785-766-4801 ~ [jurbanslp@yahoo.com](mailto:jurbanslp@yahoo.com)

**June 5, 2016: AXYS Southeastern Support Group** meeting 2:00 – 6:00pm at Jones Bridge Park, 4901 East Jones Bridge Road, Norcross, GA 30092. (Picnic starts around 4pm) Park located on the Chattahoochee River. Please bring food for a potluck. Contact Dorothy Boothe at [theboothefamily@yahoo.com](mailto:theboothefamily@yahoo.com) or 706-224-4437.

**October 13, 2016:** Guest bartender night at [BBC Tavern and Grill](#) located at 4019 Kennett Pike, Greenville, DE 19807 from 6:00 - 9:00 pm to **benefit the Nemours eXtraordinarY Kids Clinic**.

**October 23, 2016:** Mid Atlantic Support Group meeting 1:00 – 4:00pm at Nemours Alfred I. duPont Hospital for Children, 1600 Rockland Road, Wilmington, DE 19803 in Classroom 1.

**XXYY Project Events:** The XXYY Project tracks and promotes XXYY-related events throughout the world. Please visit <http://xyysyndrome.org/main/events/> for a complete listing.