

## Reflecting on the 2019 AXYS Family Conference

We promised that our 2019 AXYS Family Conference would be the best one yet and we achieved that goal. Nearly 400 participants from as far away as Brazil and The Netherlands gained knowledge and understanding in Atlanta. You could feel the strong sense of community and watch friendships blossom. Here are some comments from those who attended in their own words:

"The people my daughter and I met were amazing."

"A real sense of community was present at this conference."

"The camaraderie with other parents was invaluable. Seeing our son mingle with the other guys so comfortably. The bowling, pool, and billiards were a hit!"

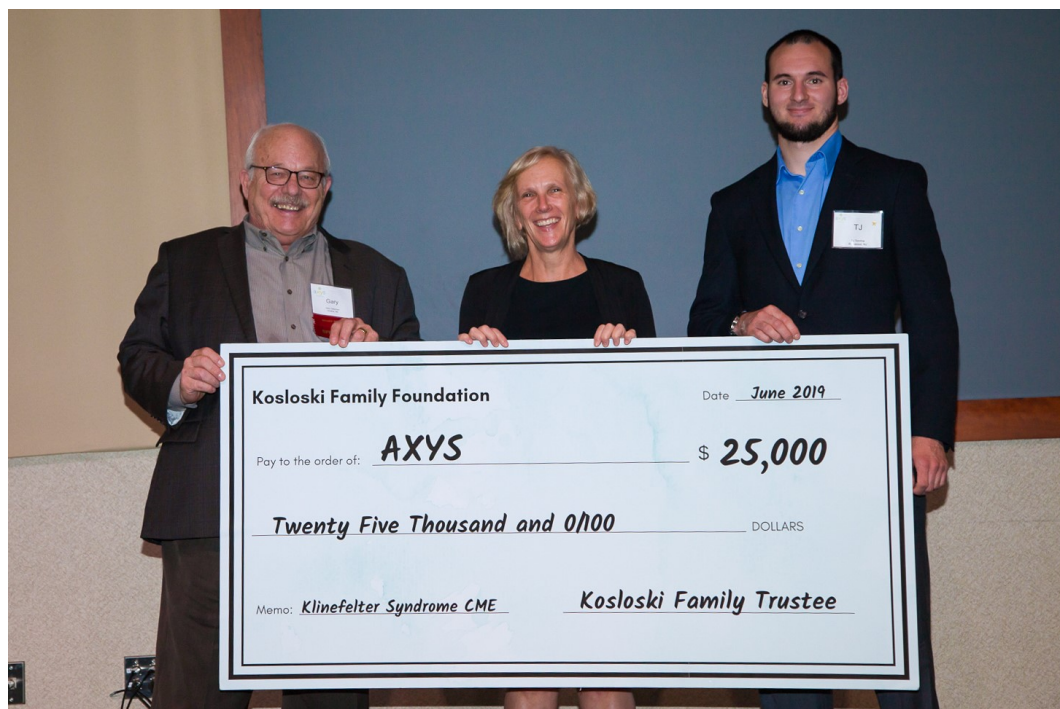
"This conference has changed our lives, and in return our son's...AXYS is a family I am proud to have, my admiration of the doctors is off the charts!!!"

[Read the rest of the article](#)



(Photos by Stuart Hasson Studios)

## AXYS Awarded \$25,000 to Create Continuing Medical Education Course on XXY (Klinefelter Syndrome) in Adults



AXYS Board Chair Gary Glissman and Executive Director Carol Meerschaert accept the Kosloski Family Foundation Grant presented by TJ Torchia (Photo by Stuart Hasson Studios)

June 30, 2019 Atlanta, GA– The Association for X and Y Chromosome Variations (AXYS), dedicated to addressing the needs of those affected by one or more extra X and/or Y chromosomes, was presented a \$25,000 check by TJ Torchia, son of Tony Torchia, CPA, Partner at RotenbergMeril, on behalf of the The Kosloski Family Foundation. The generous \$25,000 grant will be used to develop an online continuing medical education (CME) course to enhance physician knowledge of 47, XXY, also known as Klinefelter Syndrome (KS).

"For 30 years AXYS has worked to increase access to the multidisciplinary care that those with X and Y variations need throughout their lives," said Carol Meerschaert, AXYS Executive Director. "This generous

## ACRC Clinic Spotlight: MassGeneral Hospital Klinefelter Syndrome Clinic

The MassGeneral Hospital Klinefelter Syndrome Clinic is the most recent addition to the [ACRC \(AXYS Clinic and Research Consortium\)](#). They offer care throughout the lifespan, from caring for those with a prenatal KS diagnosis to adults of all ages. While the clinic is named for KS, they specialize in all male X and Y chromosome variations, including 47,XXX, 48,XXYY, and 48,XXXY. Individuals with 47,XXX can also receive care at MGH through a separate team within the Medical Genetics department.

This clinic was created in part due to the efforts of the NEXXYS Support Group. Several members of this group saw the need for a clinic in New England and worked with MassGeneral Hospital to establish the clinic.

"Our multidisciplinary clinic was inspired by patient feedback, and it is our hope that those with Klinefelter syndrome and other sex chromosome variations can consider our clinic a "medical home," says Emma Snyder, the Clinic Coordinator. "I am the first point of contact for new patients and an ongoing resource to patients with further questions about our services."

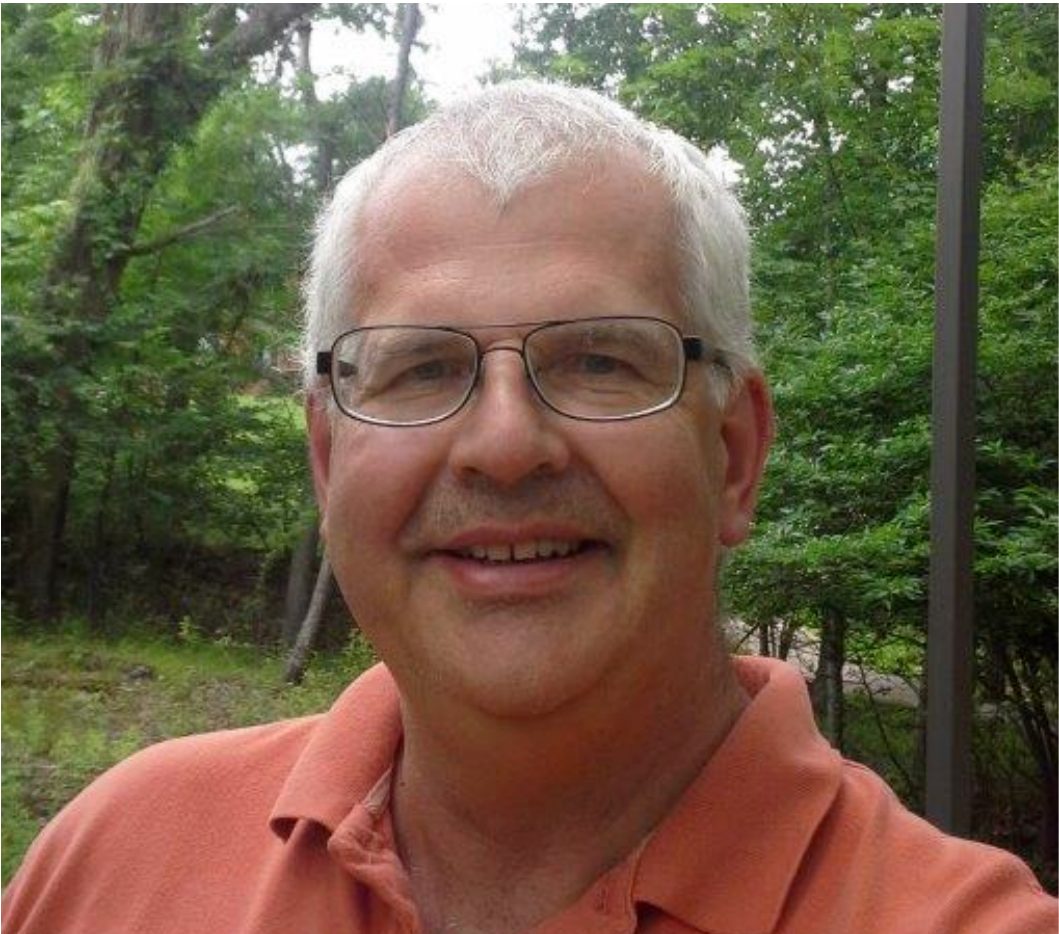
Led by co-directors [Frances A. High, MD PhD, specializing in Medical Genetics](#) and [Frances J. Hayes, MBBCh BAO, who specializes in Reproductive Endocrinology](#), the MassGeneral Clinic takes a multidisciplinary approach to providing coordinated care. They offer a comprehensive evaluation and work with your primary care providers to identify



grant from the Kosloski Family Foundation will support AXYS’ efforts to enhance physician knowledge of the most common X and/or Y chromosome variation, with the ultimate goal of improving the quality and accessibility of medical care for adults with Klinefelter Syndrome.”

[Read the full article here.](#)

## What is XYY? Meet Uldis and Find Out



*AXYS shares articles about our community members to help you gain understanding of X and Y chromosome variations on a personal level. Please enjoy this article and learn about [XYY](#).*

You’d notice Uldis Silgailis. A stocky guy, 6’6” tall, is hard to miss. But like most men with an X or Y variation, medically known as a sex chromosome aneuploidy (SCA), everyone missed his condition.

As a child, Uldis liked to sit quietly in the back of the class and daydream. He was a smart kid from an academically accomplished family. His teachers and family thought he was just not applying himself. “It was tough to hear the comments from ‘why are you acting so immature?’ to ‘what’s wrong with you?’ over and over.”

He knew he was different and that school was harder for him, but nobody could explain why. In middle school his family finally got him tested.

[See the full article](#)

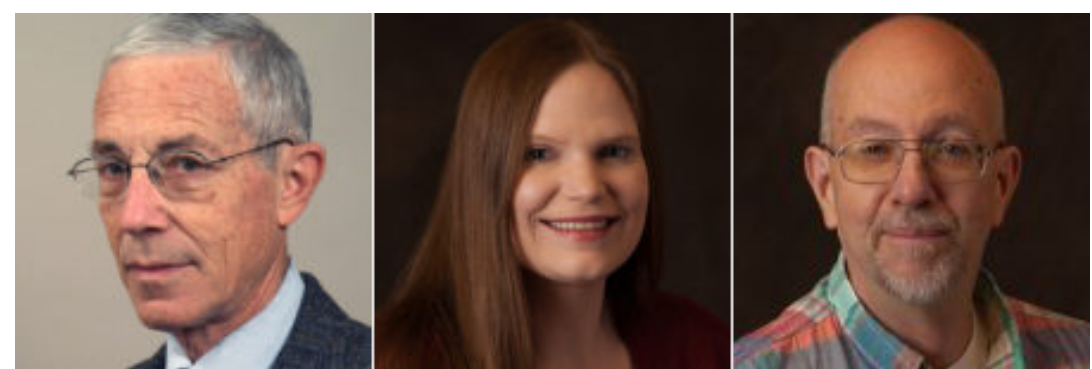
specialty needs, coordinate care, and improve outcomes.

The clinic offers a multidisciplinary team of medical, surgical, and neuropsychological specialists, and can refer to many other subspecialties at MassGeneral as needed. “We want to grow the ease of transition from pediatric to adult care,” says Emma. “As you reach your 20s, you often lose access to services in the education system but may continue to need support, including mental healthcare.” This clinic was designed to meet these needs.

New to the team is genetic counselor, Ashley Wong, MS. In her role in the KS clinic, Ashley focuses on the psychosocial counseling aspect of genetic counseling. She is a resource for patients and their families as they navigate various aspects of a KS diagnosis, particularly the neurodevelopmental components.

To learn more about the clinic or make an appointment, call Emma at 617-726-5521 or send her an email at [esnyder2@mgh.harvard.edu](mailto:esnyder2@mgh.harvard.edu). For MGH Trisomy X care, call Medical Genetics at 617-726-1561.

## AXYS Welcomes New Board Members



(Photos by Stuart Hasson Studios)

AXYS is thrilled to announce that three members have joined our board in June 2019: Stuart Howards, Andrea Millett and Bill Mulkern.


Stuart S. Howards, MD will serve as the AXYS treasurer. He is a professor emeritus at the University of Virginia Medical School and an active professor of Urology at Wake Forest School of Medicine. Dr. Howards has vast experience having served as a board member of the American Board of Urology and was their CEO for 15 years. He served as the treasurer for the American Society of Reproductive Medicine for 15 years. “I’m happy to serve as the AXYS treasurer, says Dr. Howards, “I want to assure our financial health and grow our funds so we can serve the X and Y community.”

Andrea Millett has a degree in psychology and has worked as a retail manager and preschool teacher. Andrea says “I have been involved in the AXYS community for about 10 years, watching it grow has been incredible. From being part of the ambassador team, to gaining knowledge from doctors and peers from the PCORI scientific conference, we’ve brought together similar minded people who continue to thrive for change. I want to assist in creating and achieving new goals for our future.”

Bill Mulkern is the current leader of the New England XXYS Support Group (NEXXYS), and a soon-to-be-retiree from the retirement plan law and administration field. Bill says “I want to take this opportunity to again thank Stefan Schwarz, who was generous and gracious in his support of me when I was diagnosed in early 2000. A few months after diagnosis, I attended my first meeting of the New England group he had founded, and it was then that I volunteered to become co-chair when he announced that he was leaving. And here I still am, 19 years later.”


Welcome Stuart, Andrea and Bill!

## Adult Clinic Update: Developing a Model for the Transition from Pediatric to Adult Care






**Developing a Model for the Transition from Pediatric to Adult Care**

Sharon Close, PhD, MS, CPNP-PC, FAAN; Susan Howell, MS, CGC; Nicole Tartaglia, MD; Amy Talbot, MD; Virginia Cover, MSW; Robert Miller; Carol Meerschaert, MBA, RD



### Adult Specialty Clinics for Sex Chromosome Aneuploidy

WITH Grant Progress Report

Introduction	Plan	Challenges
<p>Adults with X and Y variations experience difficulty in finding adult specialists familiar with the range of medical issues that affect them into adulthood.</p> <p>A range of endocrine disorders, neurological complications, and autoimmune disorders affect the health of these adults. Many have continuing problems with psychiatric complications, autism spectrum disorders, and learning disabilities.</p> <p>Nearly 20% of the 500 annual requests to the AXYS Help Line request assistance finding medical professionals who can provide specialty consultation and treatment for adults with an X or Y variation.</p> <p>Some adults with an X or Y variation may not be completely independent of their parents, and therefore the approach must be family centered.</p> <p>Pediatric programs have experience with providing multi-disciplinary care; adult clinics find more difficulty in coordinating care. Multi-specialty evaluations can document medical and neuro-developmental barriers to successful employment, enabling workplace accommodations and supports. In addition, the evaluations allow this population to access public programs that promote their ability to live independently.</p>	<p>Thanks to the generous support of the <b>WITH Foundation</b>, AXYS has partnered with the <b>eXtraordinaryY Kids Clinic at Children's Hospital Colorado</b> and the <b>eXtraordinaryY Clinic at Emory University</b> in Atlanta to pilot two efforts to better serve individuals with an X or Y variation transition from pediatric care to adult care.</p> <div></div> <ul style="list-style-type: none"><li>AXYS executed cooperative research agreements with our partnering clinics.</li><li>Leads from each clinic identified potential leads for an adult clinic at their institution or an affiliated institution serving adults.</li><li>Pilot programs for adult clinics are being formed using the protocols developed by the multidisciplinary clinics for children</li><li>Additional services protocols will be developed for services needed by adults such as vocational services and assistance with public support programs.</li></ul> 	<p>Because adult medical services are often provided by different departments, in separate clinical buildings, creating a program successfully requires a clinic coordinator who can devote dedicated time to evaluating the needs of each patient, coordinating appointments for them, particularly if the patient is traveling from a distance, and compiling visit and lab records into a report for the patient's primary care physician.</p> <p>Providing clinic coordinator startup costs will allow these specialty clinics to begin operating at least one day per month, with the goal of five adult patients per month per clinic during the initial year of operation.</p> <p>Ideally, AXYS will extend its project to two years, with a goal to expand the number of adults in interdisciplinary care to 150 patients annually.</p> <p>Creating pilot adult clinics can pave the way for interdisciplinary care for adults with X and Y variations.</p>

At the 2019 AXYS Family Conference, a progress report on the effort to create adult specialty clinics for sex chromosome variations was presented via [this poster](#).



## It started with a letter to Ann Landers

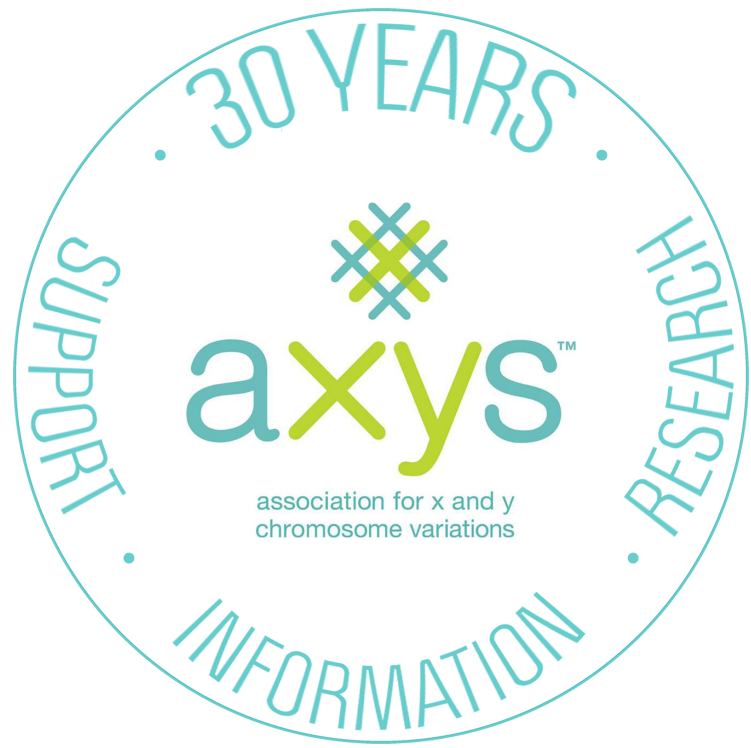
Back in 1989 Melissa Aylstock, a mother of an 8-year-old with Klinefelter syndrome, wrote a letter to Ann Landers. She offered to connect families who had a member with KS. She received over 1,000 letters. That was the beginning of AXYS.

For the next year we will be celebrating 30 years of information, support, and research. Our first celebration event will be a No Show Gala and Online Silent Auction in September. Throughout the month of August we will be collecting auction items. High value items that have nationwide appeal or are easy to ship are most needed.

Possible items to donate:

- weekend at a vacation home
- gift cards to a national chain store or restaurant
- tickets to a play or concert
- tickets to a popular sporting event
- hand crafted quilt

Please send an [email to our Executive Director Carol Meerschaert](#) describing the item you wish to donate or to ask any questions.



## Support Groups

AXYS Support Groups are facilitated by volunteers who schedule the programs and often arrange for speakers on topics of interest to individuals and families affected by X and Y variations. These groups offer the opportunity to share experiences and resources that they have found valuable. Some of our groups are affiliated with AXYS Clinic and Research Consortium hospitals, and they meet in those facilities. Other groups meet in libraries, medical centers, or churches. Here are some upcoming meetings. See our [Support Group page](#) for a full listing of all groups.

### Upcoming Support Group Meetings:

[September 29, 2019: San Francisco Bay Area Support Group Family Picnic, Mountain View, CA](#)

October 5, 2019: [Florida Support Group](#) Meeting, Orlando, FL



## New Addresses

Our mailing address has changed to:

**AXYS**  
**PO Box 659**  
**Paoli, PA 19301**

And you can now reach out to our Helpline volunteers for support by sending an email to [helpline@genetic.org](mailto:helpline@genetic.org).

## Library

When you seek information on X and Y conditions, please visit the [Library](#) tab on our website. The [Research Articles List](#) page makes it easy to search for a specific article in our library. You can search by name, author names, or date of publication.

## Videos

Did you know past webinars and conference presentations are available on demand on the [AXYS YouTube channel](#)? Take some time to view one of the 100+ videos in our library. Do you have a suggestion for a topic or speaker? [Write to Carol](#) with your suggestions.

## Volunteer

Do you want to get more involved? Visit our [committee webpage](#) to learn about the committees and let us know if you'd like to be involved. We have opportunities of all sizes from a one-time task to ongoing planning. Send an email to our [Executive Director Carol Meerschaert](#) and let her know how you can help.

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