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

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

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 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, 48, XXY, 48, XXXY. Individuals with 47, XXX can also receive care at MGH through a separate team within the Medical Genetics department.

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 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 NEXXS 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. Hioh, 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
A 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

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

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. For MGH Trisomy X care, call Medical Genetics at 617-726-1561.

AXYS Welcomes New Board Members

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 (NEXXXYS), 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!
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.