The XXYY Project Joins the AXYS Family

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The Association for X and Y Chromosome Variations (AXYS) and The XXYY Project

Introduction

X and Y chromosome variations occur in approximately 1:500 people. They are common genetic conditions, but often go undiagnosed. The most common variations involve a trisomy, which means three sex chromosomes instead of the typical two. This includes 47, XXY (Klinefelter syndrome), 47, XYY (Jacobs Syndrome) and 47, XXX (Trisomy X).

Approximately 1 in 18,000 males are born with XXYY. World-wide, 400 individuals with XXYY Syndrome are known to us at this time. While once thought of as a variation of Klinefelter Syndrome, XXYY Syndrome has more numerous and severe symptoms and may require special treatment and management.

AXYS Mission

The mission of AXYS is to help individuals with one or more extra X and/or Y chromosomes and their families to lead fuller, more productive lives.

XXYY Project Vision Statement

Our vision is a world where men and boys with XXYY Syndrome lead purposeful, productive lives.

Goal of Partnership

The XXYY Project partnered under the umbrella of AXYS to amplify our presence and bring more awareness to XXYY by reaching more people.

Shared Characteristics of Klinefelter and XXYY Syndromes

Many of the most common symptoms and characteristics of Klinefelter Syndrome are also seen in XXYY Syndrome, however they may be more severe in those with XXYY.

- Developmental delays
- Speech impairment or delay
- Tall, considering family history
- Behavior outbursts, mood swings
- Learning disabilities
- Intellectual impairment
- ADD or ADHD symptoms
- Autism, autism spectrum, PDD-NOS
- Scoliosis
- Clinodactyly (Curved-in pinky fingers)
- Low muscle tone
- Flat feet/club feet
- Sterility
- Delayed sexual development
- Undescended testes
- Low or no testosterone
- Dental problems
- Leg ulcers
- Heart defects (i.e. VSD)

History of the XXYY Project

1998 XXYY Parent Network created, an informal association of parents and caregivers. Utilize listserv for private communication.

2003 XXYY Project formed by founding parents of the XXYY Parent Network;
XXYY Project becomes a project of the Colorado Nonprofit Development Center (CNDC), a 501(c)(3) and nonprofit incubator, which provides fiduciary and legal support.

2007 Assisted Dr. Nicole Tartaglia in the development of the eXtraordinarY Kids Clinic at Children's Hospital Colorado through fundraising and research support. Developed a clinic grant program to assist families in need of financial support to visit the Denver clinic.

2008 Recruited study subjects and helped fund Dr. Tartaglia's study: "A New Look at XXYY Syndrome: Medical and Psychological Features." Hired part-time social worker to work with families. Hired part-time Executive Director.

2012 Updated organizational mission and vision statements. Created new XXYY Project logo and selected organizational colors.

2015 Merged with AXYS, the umbrella patient advocacy organizations for individuals and families of all sex chromosome aneuploidies. Relationship with CNDC ends. XXYY Project Advisory Board member joins AXYS Board of Directors.

2017 Transferred $10,000 from the XXYY Project general fund to the AXYS Clinic and Research Consortium fund to support development of the clinic network. Create closed Facebook Group, ***XXYY Parents and Caregivers***, to provide online, real-time support for XXYY families. Second XXYY Project volunteer joins AXYS Board of Directors.

2018 Migrated the content of the XXYY Project website xxyysyndrome.org to the AXYS website genetic.org

History of AXYS

1989 Klinefelter Syndrome and Associates (KS&A) founded by Melissa Aylstock, the mother of an 8 year old with Klinefelter syndrome when her letter to Ann Landers was published.

1991 KS&A incorporated in California as a 501(c)(3). Began to include other SCA conditions including 47, XYY (Jacobs Syndrome) and 47, XXX (Trisomy X). Renamed *Knowledge, Support & Action*.

1994 First KS&A Family Conference

2014 Rebrand with new name AXYS

2018 Hire first full time Executive Director

The XXYY Project: Part of the AXYS Family

Operations

- AXYS staff Executive Director and Communications Manager support XXYY programs.
- XXYY sends own newsletters and eblasts. XXYY Project members receive AXYS communications.
- Callers to Help Line with XXYY related questions referred to XXYY volunteers.

Governance

- XXYY Project volunteers serve on AXYS Board of Directors.

Fundraising

- Both logos are on Annual Fund appeal mailings.
- Donors can designate donations to XXYY-specific fund.

Research

- AXYS Clinical and Research Consortium supports XXYY as well as all X and Y variations.
- XXYY Project also funds condition specific projects.

Website

- Specific XXYY content within genetic.org.

Conference

- AXYS Family Conference has condition specific sessions as well as sessions common to all conditions.
- XXYY hosts separate family gathering after the conference.

Research

AXYS has a long history of supporting scientific research on all X and Y variations and sharing sound information based on peer reviewed science. AXYS helps recruit study subjects, fund research projects and disseminate findings via a webinar series and a communication titled *Research Paper of the Month*.

The XXYY Project funds research on XXYY Syndrome

2008 Recruited study subjects and helped fund Dr. Tartaglia's et al study: "A New Look at XXYY Syndrome: Medical and Psychological Features."

2009 Recruited study subjects and helped fund Dr. Tartaglia's study: "Tremor in 48 XXYY Syndrome."

2011 Recruited study subjects and helped fund Dr. Tartaglia's study: "48,XXYY, 48,XXXY and 49,XXXXY Syndromes: Not Just Variants of Klinefelter Syndrome."

2016 Funded the eXtraordinarY Kids Clinic project: "XXYY Sibling Video."
Helped fund the Dr. Sharron Close, Emory University study: "Voices of XXYY."

2018 Helped fund the Amy Blumling, PhD candidate dissertation: "The Relationship of Physical Function and Psychosocial Health on Quality of Life in Individuals with 48,XXYY."

Contact

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