



## May is National X & Y Chromosome Variation Awareness Month

### Testing Available for Common but Often Undiagnosed Conditions

X & Y Chromosome Variations, also known as sex chromosome aneuploidies (SCA), affect 1 in 500 individuals. More than 600,000 men and women in the United States have an SCA and 4,000 more are born each year. Most go undiagnosed at birth due to limited identifiable physical characteristics that would prompt further testing. Overall, it is estimated that 75% of all individuals with an SCA condition are undiagnosed.

SCA's are randomly occurring genetic conditions, which are not inherited, but caused by chromosomes not separating correctly at conception. Males typically have one X and one Y chromosome. Females typically have two X chromosomes. Those with an SCA condition have one or more extra X or Y chromosomes. The effects of having an SCA condition can range from mild to moderate to profoundly disabling. There are many variations of SCA's, with XXY (Klinefelter Syndrome) being the most common. Other variations in males include XYY, XXYY, XXXY, and XXXXY. Variations in females include X (Turner syndrome), XXX (Trisomy X), XXXX and XXXXX.

A simple blood test called a karyotype can determine if an X and Y chromosome variation is present. Diagnosed individuals benefit enormously from early intervention and a variety of treatments and therapies. Those who remain undiagnosed often struggle through life with incomplete diagnoses such as developmental, behavioral or social delays, ADHD or autism. Boys will often have incomplete puberty. Families should talk to their doctor about ordering a karyotype if they are dealing with any of the above symptoms and still feel there is something missing in their current treatment.

The goal of a National X & Y Chromosome Variation Awareness Month is to spread awareness. Increased awareness will benefit individuals with X & Y variations attain better support from educators, educate medical professionals, establish more specialized clinics, fund research and provide greater acceptance for all those living with an SCA condition.

**About AXYS:** AXYS is the nation's oldest and largest non-profit health-advocacy organization addressing the needs of individuals who were born with X & Y chromosome variations. AXYS provides resources and support for those individuals and their families as well as the clinicians, educators and researchers serving them. The work of AXYS includes a toll-free help line, support groups, webinars, conferences, online and print materials, and the organization of specialized clinics dedicated to X & Y chromosome variations throughout the U.S.