

Advocating for the Health and Well-being of all Individuals with a 45,X Cell Line July 2022

The Turner Syndrome Society of the United States' Scientific Advisory Board is reaching out to the members of the Turner Syndrome Society to advocate for the health and well-being of all individuals with a 45,X cell line. Everyone diagnosed with Turner syndrome has a deficiency of the second sex chromosome, which in genetic terms (karyotype) is called 45,X. Studies have shown that all those with a 45,X cell line are at risk of congenital heart disease, short stature, infertility, and other associated health concerns. Throughout the years, females with a 45,X cell line have been actively recruited to participate in many studies, and their participation has led to great advances in our understanding of the health of females with these chromosomal findings.

Through the strong participation of our community in Turner syndrome research, clinical guidelines were developed that helped to improve the overall health and well-being of our members and the greater TS community. These Turner syndrome clinical practice guidelines serve to educate physicians and other practitioners on the care of our community and serve as a standard of quality care. However, there is an increasing number of males who are being diagnosed with Turner syndrome. Unfortunately, there is no clear guidance on how males, those who are intersex and those across the gender spectrum with a 45,X cell line should be cared for. This group includes people who have 46,XY (male) genetics. How will they access needed routine cardiac, kidney, reproductive, autoimmune, or endocrine screening unless their healthcare providers have clear guidelines to follow for their care? The concern is that they may go without necessary treatments or not qualify for insurance coverage of recommended testing or medications. Because these individuals may suffer from a lack of appropriate health surveillance and treatment, they need our community's support.

It is common for individuals with 45,X cell lines to learn of their chromosomal differences when undergoing evaluations for infertility or short stature. However, more and more of these differences are being identified through maternal prenatal testing at a time when biological sex is not established or remains fluid. Individuals with 45,X cell lines and their parents are often frightened and uncertain as to what the future may hold and many reach out to TSSUS for guidance and support.

TSSUS can be a strong and effective advocate for the wellbeing of all those with 45,X cell lines. Together we can advocate for everyone to receive quality and comprehensive care. We welcome your thoughts, comments, and ideas on how to move forward to offer our support at info@turnersyndrome.org.