Prevalence of Sex Chromosome Aneuploidy (SCA) Stratified by Genetic Ancestry The bars and error bars represent the calculated prevalence and 95% confidence interval (CI) for each genetic ancestry group. The horizontal line and shaded area represent the calculated prevalence and 95% CI overall. The lighter shaded areas within the bars represent individuals with a clinical diagnosis in their electronic medical record. Credit: *JAMA Network Open* (2024). DOI: 10.1001/jamanetworkopen.2024.4113
Researchers at the University of Colorado Anschutz Medical Campus and collaborators across the country have conducted the largest and most diverse study of men with extra X or Y chromosomes in the US using a large dataset of military veterans. The results could lead to better diagnoses of these underrecognized conditions and earlier treatment of associated diseases.

The study was published in JAMA Network Open.

"One in 400 males have an additional X or Y chromosome, how. However, 86 percent of these individuals are not diagnosed," said the study's first author Shanlee Davis, MD, Ph.D., associate professor of pediatrics at the University of Colorado School of Medicine. "This study supports that men with X and Y variations successfully serve in the US military but experience many medical and psychiatric comorbidities that could potentially be prevented with appropriate diagnosis and care."

Men with an extra X chromosome, known as Klinefelter syndrome, or Y chromosome, also known as Jacobs syndrome, have an increased risk of delayed development, learning disabilities, type 2 diabetes, and cardiovascular disorders. Men with Klinefelter syndrome also have impaired testicular function associated with low testosterone and infertility. All of the research we have on these conditions is from the