

ASSOCIATIONS OF PSYCHIATRIC DISORDERS WITH SEX CHROMOSOME ANEUPLOIDIES IN THE DANISH IPSYCH2015 DATASET: A CASE-COHORT STUDY

Sánchez XC, Montalbano S, Vaez M et al. Lancet Psychiatry. 2023 PMID: 36697121.

Most studies of sex chromosome aneuploidies have focused on children and adolescents, and risk estimates for most disorders with typically adult onset have therefore been scarce.

This study ([Calle Sánchez et al.¹](#), Lancet) relies on the iPSYCH2015 case-cohort dataset, based on a source population of individuals born in Denmark between 1981 and 2008 with “case” comprising individuals with a diagnosis of schizophrenia spectrum disorder, bipolar disorder, major depressive disorder, autism spectrum disorder or ADHD and “cohort” consisting of individuals randomly selected from the source population.

Analysis of microarray data from bio-banked blood samples of 119 481 individuals (78 726 in the case sample and 43 326 in the cohort) allowed the identification of 387 (0.3%) individuals as carriers of sex chromosome aneuploidies. Each sex chromosome aneuploidy karyotype was associated with an increased risk of at least one index psychiatric disorder and all karyotypes were associated with an increased risk of ADHD, autism spectrum disorder and schizophrenia spectrum disorder. The proportion of sex chromosome aneuploidy carriers who had been clinically diagnosed was 93% for 45,X but lower for 47,XXY (22%), 47,XXX (15%), and 47,XYY (15%).

Increased risks of psychiatric disorders associated with sex chromosome aneuploidies, combined with low rates of clinical diagnosis of sex chromosome aneuploidies, compromise the adequate provision of necessary health care and counselling to affected individuals and their families.

1- [https://www.thelancet.com/journals/lanpsy/article/PIIS2215-0366\(23\)00004-4/fulltext](https://www.thelancet.com/journals/lanpsy/article/PIIS2215-0366(23)00004-4/fulltext)