



# KLINFELTER SYNDROME

IMPORTANCE OF COMMUNITY ENGAGEMENT

HEIGHTENED AWARENESS

RECOGNITION OF POSITIVE ATTRIBUTES



School of Rural Medicine

## A RURAL MEDICAL STUDENT PROJECT

EXPLORING THE RURAL EXPERIENCE OF INDIVIDUALS WITH KLINFELTER SYNDROME (XXY AND MOSAIC PLUS OTHER VARIANTS SUCH AS XXXY ETC) AND PARENTS OF CHILDREN WITH KS

### RESEARCH TEAM

Dr Catherine Keniry - CSU, School of Rural Medicine  
Dr Himanshu Goel - Clinical Geneticist Hunter New England Genetics Service  
Dr Komal Vora - Paediatric Endocrinologist - John Hunter Children's Hospital  
Maddison Symons - Medical Student, CSU SRM  
Mr Bruce Hopper - Senior Genetic Counsellor, Forster Genetics

### WHAT YOUR PARTICIPATION WILL INVOLVE

An online interview (approx. 40-60mins) to discuss your journey, challenges, and support systems related to Klinefelter Syndrome.

### BACKGROUND

Klinefelter Syndrome (KS) is an underdiagnosed sex chromosomal aneuploidy and affects about one in 550 Australian men, however only half of those affected are ever diagnosed. The diagnosis of KS in children often occurs during evaluations for developmental delays, learning disabilities, or atypical physical development. In adults, KS is frequently diagnosed during investigations for infertility, as many men with KS present with oligospermia (low sperm count) or azoospermia (no sperm). The diagnosis of KS is increasing following the introduction of Non-invasive Prenatal Testing (NIPT) in 2012. NIPT is increasingly undertaken as a routine test during pregnancy.

### WHY PARTICIPATE

Your insights are invaluable in helping us understand the unique experiences of families and individuals affected by Klinefelter Syndrome, especially in rural areas.

### WHO IS IT FOR

Parents of Klinefelter Syndrome children and individuals with Klinefelter Syndrome

### AFFILIATIONS

Charles Sturt University, SRM  
Hunter New England Local Health District

### HOW TO PARTICIPATE

Please contact: [srm-research@csu.edu.au](mailto:srm-research@csu.edu.au) to arrange an interview.

### FURTHER DETAILS

<https://science-health.csu.edu.au/schools/medicine/research/student-md-projects>



TOGETHER, WE CAN IMPROVE AWARENESS AND ACCESS TO SUPPORT RURAL AREAS