

## **Psychiatric manifestations and psychosocial impairment in a case of delayed diagnosis of Klinefelter Syndrome**

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**TOPIC/TARGET AUDIENCE:** Adult and child mental health advocates, early detection and treatment programs, public assistance programs for people with genetic disorders, psychiatrists, psychologists, psychiatric nurses, Mental Health NP, school counselors

**ABSTRACT:** The chromosomal aneuploidy causing Klinefelter Syndrome (KS) has a wide variety of developmental implications; however, many cases go undiagnosed until adulthood which can result in children with Klinefelter Syndrome not having access to essential community resources. Men with KS are phenotypically male, but carry the chromosomal makeup of 47, XXY. The random genetic error that leads to the presence of the extra chromosome results in physical, psychiatric and psychosocial manifestations. Males with KS present with low testosterone levels, a tall, thin body habitus, decreased body hair and muscle mass, as well as hypo fertility or infertility. Alterations in intellectual development as well as behavioral and social difficulties are seen in this population.

This case study will examine the effects of undiagnosed KS in a young adult male who presented to an inpatient psychiatric facility with symptoms of psychosis, anxiety, depression and substance use. The process of formulating the diagnosis of KS, challenges of the co-existing psychiatric diagnoses, and availability and accessibility of community health resources will be discussed. KS is a lifelong condition with a myriad of symptoms. Improved understanding of the clinical picture of KS and associated comorbidities may help to improve recognition, diagnosis, and understanding of the condition.

### **OBJECTIVE(S):**

- Explain the diagnosis and clinical picture of Klinefelter Syndrome in terms of its well-known pathophysiology in addition to the less known impact on psychosocial development.
- Discuss a case of delayed diagnosis of Klinefelter Syndrome in a young adult male in an inpatient psychiatric setting.
- Identify the important role of early diagnosis with accompanying community resources in children with Klinefelter Syndrome.

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