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Romanian Prader-Willi Association

ASOCIACIÓN MADRILEÑA  
PARA EL SÍNDROME DE  
PRADER-WILLI



## A BOY WITH KLINEFELTER SYNDROME AND PRADER-WILLI SYNDROME: A CLINICAL REPORT.

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**INTRODUCTION:** A number of X and Y chromosome abnormalities have been reported in children with Prader-Willi syndrome (PWS). This report describes a 16-year-old boy with 47, XXY, UPD 15, karyotype and a Prader-Willi syndrome (PWS) phenotype. He was admitted to the outpatient service of our multidisciplinary clinic for children with mental retardation and psychiatric and behavioural problems with worsening behavioural problems. We compared the phenotype of our patient to that of other patients in the literature for whom detailed phenotypic information was available and against the clinical criteria for PWS and Klinefelter syndrome (KS).

**METHODS:** In this descriptive and mainly retrospective case study, we describe a 16-year-old boy with Klinefelter syndrome and Prader-Willi syndrome. The patient underwent psychiatric assessment by a psychiatrist who was skilled in assessment of children with intellectual disabilities. Symptomatology was collected on basis of clinical interview, clinical observation, family and carer informants, medical records and psychiatric reports.

**RESULTS:** This boy was born after 36 weeks gestation. Klinefelter syndrome was diagnosed on prenatal amniocentesis. As an infant, he was hypotonic and demonstrated a poor suck. He had delayed milestones. By parental report, he had a history of eating behaviors typical for PWS. He had been diagnosed as suffering from Prader-Willi syndrome at the age of three years and genetic studies confirmed a maternal uniparental disomy. At 13 years, there were behavioral problems, with episodes of temper tantrums, obsessions and compulsions. A variety of behavioural disturbances that has been reported (stubbornness, temper-tantrums, skin picking, obsessive-compulsive behaviour and internalising emotional problems), conformed to the criteria of the behavioural phenotype of PWS.

**DISCUSSION:** We reported a patient with PWS and KS and describe the difficulties with the clinical diagnosis of these conditions when they coexist. For the clinical point of view, the affected individual is expected to have a PWS phenotype. The chromosome X and 15 events commonly occur in different parents and pre- and post-conception, thus the mechanism are likely distinct and coincidental. These two conditions would be expected to occur together, by chance alone, in 1 in 20 million live births. While some speculate the frequency of these reports alone suggests that the events are not coincidental, we are hesitant to attribute this specific combination to a concordant etiology. On the other hand, most of those cases include non-disjunction events that are associated with advanced maternal age; thus it may not be unexpected to see the concordance of UPD 15 with sex chromosome aneusomy.