



14 Neurogenetic Basis of Criminal Behaviors in Klinefelter Syndrome: A Case of Uxoricide-Suicide and a Review of the Literature

Isabella Aquila, MD, Viale Europa, località Germaneto, Policlinico Universitario, S. Venuta-Medicina Legale, Catanzaro 88100, ITALY; Ciro Di Nunzio, MFS, PhD, Magna Graecia University, Viale Europa, Germaneto, Legal Medicine, Catanzaro, 88100, ITALY; Miriam Sirianni, MD, Viale Europa, 88100 Germaneto, Catanzaro 88100, ITALY; Carol Pileggi, Magna Graecia University, Dept Legal Medicine, via dei Brutii 16/A, 88046 Lamezia Terme, Catanzaro, ITALY; Pasquale De Fazio, PhD, Viale Europa, 88100, Germaneto, Catanzaro 88100, ITALY; and Pietrantonio Ricci, Viale Europa - Località Germaneto, Catanzaro, ITALY*

After attending this presentation, attendees will be able to better understand cases of Klinefelter Syndrome.

This presentation will impact the forensic science community by demonstrating a correlation between chromosomal abnormalities and criminal behaviors in Klinefelter Syndrome.

Klinefelter Syndrome ("KS," XXY) is the most common sex chromosome abnormality in men. It occurs with a frequency of 1:500 to 1:1000 live male births. The prevalence of Klinefelter Syndrome is around 150 per 100,000 males or one in every 660 males. This chromosomal disorder is characterized by the presence of one extra X chromosome (or more) in a phenotypic male. The most prevalent type of karyotype is 47, XXY.¹ The physical phenotype is characterized by tall stature, small testicles, cryptorchidism, hypogonadism with low levels of testosterone, infertility, gynecomastia, broad hips, narrow shoulders, and sparse body hair.² The abnormalities of the sex chromosome also lead to alterations of brain development. This process may be a genetic basis of the psychiatric disorders described in these subjects. The cognitive phenotype of KS is characterized by language learning problems and mental retardation.³ In these subjects, there is an increased risk of psychiatric disorders, such as symptoms associated with schizophrenia, which are likely to be related with the over-expression of X-linked genes. Patients with KS also show an increased risk of criminal behavior and, in a significant proportion of cases, psychosocial problems have been reported.⁴ The most common offenses reported to be related to KS are sexual abuse, arson, burglary, homicide, and drug-related crimes. KS is often under-diagnosed. The goal of this study is to verify the hypothesis of a correlation between chromosomal abnormalities and criminal behaviors through the analysis of a forensic case of uxoricide-suicide.

Case Report: This study analyzed the case of an older man found dead in his cottage with a gun in his right hand. Judicial inspection revealed a suicidal, single gunshot injury. Social background was reconstructed by psychological autopsy. Data analysis highlighted a personal story of uxoricide, and his life history was surveyed. The investigation demonstrated a medical history of infertility. An analysis of his pending criminal proceedings revealed an incident of uxoricide about 50 years prior (1962). The court's archives were searched for statements made by the man (then-husband) when interrogated. The records revealed that the 25-year-old woman was killed by her husband using a single gunshot injury. She was transported to a nearby hospital, and the doctors found "internal bleeding from gunshot wound penetrating into the abdominal cavity." In the intervening time, her husband vanished but was later arrested.

Autopsy investigation of the man in this case was carried out, and the typical phenotype associated with KS was found. The postmortem clinical diagnosis was confirmed through the histopathological and genetic analysis of his karyotype.

Conclusions: The retrospective analysis of the case showed a correlation between this syndrome, psychiatric disorder, and criminal activity.

References:

1. Hayashi K, Hanaoka Y, Matsumura S, Takagi T, Kajiwara M, Tamaki N, Minaguchi K, and Sato Y. An Autopsy Case of Klinefelter's Syndrome Suspected and its DNA Analysis. *Forensic Sci Int* 2000;113:119-125.
2. Klinefelter H et al: Syndrome Characterized by Gynecomastia, Aspermatogenesis Without A-Leydigism and Increased Secretion of Follicle-Stimulating Hormone. *J Clin Endocrinol Metab* 2: 615-622. 1942.
3. Giedd JN, Clasen LS, Wallace GL, Lenroot RK, Lerch JP, Wells EM, Blumenthal JD, Nelson JE, Tossell JW, Stayer C, Evans AC, Samango-Sprouse CA. XXY (Klinefelter Syndrome): A Pediatric Quantitative Brain Magnetic Resonance Imaging Case-Control Study. *Pediatrics* 2007; 119, E232-E240.
4. DeLisi LE, Maurizio AM, Sventina C, Ardekani B, Szulc K, Nierenberg J, Leonard J, Harvey PD: Klinefelter's Syndrome (XXY) as a Genetic Model for Psychotic Disorders. *American Journal of Medical Genetics. Part. B: Neuropsychiatric Genetics* 2005. 135 B, 15-23.



Psychiatry and Behavioral Sciences Section - 2014

Klinefelter Syndrome, Karyotype, Psychiatric Disorders