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KLINEFELTER'S SYNDROME AND PSYCHOPATHOLOGY: A CASE STUDY OF THE COMBINED EFFECTS OF NATURE AND NURTURE

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ABSTRACT

A patient with Klinefelter's syndrome and genotype 47 XXY was abused and ostracized by his family during childhood. Later, this pattern was repeated by a series of homosexual lovers. The separate and interactive effects between genetic and psychosocial risk factors may account for this patient's subsequent psychosexual and personality disorders and reminds scholars of psychopathology of the importance of early detection and intervention. This single case study is presented for heuristic value, illustrative purposes, and serves as a link between mental health clinicians and sexologists.

Klinefelter's syndrome, often seen in combination with the 47 XXY genotype, has been associated with a wide variety of psychiatric disorders including schizophrenia, affective disorders, mild mental retardation, sexual deviance, and personality disorders [1-3]. Whether the incidence of any particular psychiatric syndrome is actually increased among Klinefelter's patients remains questionable because of selection biases inherent in studies of institutionalized or otherwise

clinically identified populations, and because of insufficient characterizations of the patients' psychopathologic signs and symptoms. However, there does appear to be an overall increase in the rates of psychopathology among these persons, with the most consistent association being a form of personality disorder typified by passivity, low social drive, immaturity, paranoid tendencies, apathy, and poor work capacity [4]. It is assumed that such traits are related somehow to the underlying genetic and endocrine abnormalities in these patients, perhaps to low levels of testosterone, but the exact mechanisms for this association remains unclear [4].

Here we report on a case of Klinefelter's syndrome associated with a psychosexual and personality disorder, characterized by cross-dressing, gender dysphoria, confusion, impulsive behavior and repeated psychiatric hospitalizations and suicide attempts. Although similar cases have been reported previously [1, 5], this case is worthy of documentation because: 1) it illustrates the interplay of a known genetic predisposition and early family environment influences in the development of a psychiatric disorder, 2) the patient's psychological and psychiatric status was carefully documented using standardized diagnostic evaluations, and 3) his presentation is somewhat atypical and may have been altered by prior use of exogenous testosterone.

CASE REPORT

Present Illness and Presentation

Mr. H., a nineteen-year-old, single, unemployed Caucasian man with no current address who was traveling cross country, was admitted for treatment following a suicide attempt with a small overdose of Tylenol with codeine. This was his sixth psychiatric hospitalization during the past four years in different areas of the country for suicide attempts related to feelings of frustration, alienation and despair with his homosexual lifestyle, and an inability to develop lasting and satisfying intimate relationships.

Just prior to this most recent admission, while hitchhiking dressed as a female, Mr. H. was assaulted by two truckdrivers who offered him a ride, attempted sexual relations, and later discovered that he was not a woman. He was treated at an emergency room for facial lacerations and contusions. Later the same night he became acutely frightened, disgusted and despondent over his physical appearance and took the overdose with Tylenol. His mental status on admission included suicidal ideation, dysphoric mood, and mild decrease in appetite without weight loss. He denied recent sleep disturbance, feelings of worthlessness, psychomotor retardation, loss of energy, problems with concentration or loss of interest in his usual activities. His behavior was markedly effeminate, and he was misperceived as a woman by unfamiliar staff. No hallucinations, delusions, or other signs of psychosis were noted in the

present or past admissions. He expressed a firm desire to have a sex change because "he felt like a woman."

Family and Personal History

The data presented here are gleaned from extensive review of previous psychiatric records, telephone interviews with family members, and patient report. The patient attributed his sexual identity problems, in part, to his early family environment. His mother required prolonged psychiatric hospitalization prior to and after his birth for an unspecified psychiatric illness. At age two his parents were divorced, and his father, an alcoholic, was killed in an accident two years later. The mother remarried when he was four. Although the family was reportedly unaware of the patient's genetic abnormality, he was identified by them quite early as "being different," was treated like a girl, and was frequently ostracized by his peers [6]. He describes his childhood as one of being "put in the place of a girl" in the family, even though he had half-sisters. This took the typical form of being asked and forced to do housework even though his brothers were not asked to do similar types of household tasks. His stepfather was described as physically abusive toward him when he did not obey. His early relationships with his parents and stepparents were generally characterized by fear and hostility toward males and strong attachment and identification with females. This situation was particularly powerful in that the patient experienced reinforcement for his emotional bonding to females and female images by the support for "opposite" sex role performance expectations by the family.

It is also worthwhile to note that the patient experienced inconsistent and contradictory parenting as he had to deal with a series of stepfathers each with a different set of ideas and expectations. This "push and pull" along the lines of sexual roles and behavior, family expectations, and shaping of identity may have sown the seeds for the patient's later sexual identity disturbance and psychological difficulties. Since age nine, the patient himself reported feeling "different" from other boys and was attracted more to boys than girls. He dropped out of high school in the 10th grade and worked intermittently on assembly line type jobs to support himself. He assumed an active homosexual lifestyle at age seventeen and began cross-dressing as a dancer in a gay bar at age eighteen. Since age seventeen, the patient has lived an itinerant life-style, traveling throughout the country, and typically moving on or requiring hospitalization when rejected by homosexual companions. At times he has consumed up to one fifth of whiskey daily for extended periods, but denied all other signs and symptoms of alcohol abuse.

Conceptualization, Assessment, and Treatment

Our initial impression was that the patient suffered from a psychosexual disorder, either transsexualism or transvestism, and from other personality

problems associated with his traumatic childhood. A genetic disorder was not considered likely because of his slightly sparse but otherwise normal distribution of male pubic hair, apparently normal intelligence, average height, testes within the normal range of size, and penis on the low side of average. The patient also reported that he was able to achieve an erection and sporadically achieve an orgasm (see Raboch, Mellan and Starka's [7] report on the sexual development and activity of a large sample of Klinefelter's syndrome patients), but that this was managed mainly when he assumed a receptive position with a gay partner via anal intercourse. However, laboratory testing revealed a serum testosterone of 49 ng/dl (normal male: 300-1000) and a 47 XXY karyotype. Medical records from a prior hospitalization subsequently also revealed that the diagnosis of Klinefelter's syndrome had been made earlier when the patient was sixteen years old and that he had been treated for a year with testosterone. The patient discontinued this treatment because he disliked the subsequent development of secondary male sex characteristics. Other laboratory studies, including T_3, T_4, TSH, LH, FSH, VDRL and CBC were within normal limits.

On evaluation, the patient failed to meet criteria for an affective or psychotic disorder based upon the Schedule for Affective Disorders and Schizophrenia [8], but did meet the criteria for borderline personality disorder using the Borderline Personality Inventory [9]. The patient also met DSM III criteria for transsexualism except for presence of genetic abnormality. Neuropsychological testing revealed specific defects in reading, verbal production, and ability to change cognitive sets rapidly, suggesting a mild learning disability. His IQ based upon the WAIS-R was 88–verbal, 89–performance, and 88–full scale. This result is slightly different from that reported by Ratcliffe, Bancroft, Axworthy, and McLaren [10] which reported on twelve boys with Klinefelter's syndrome matched with twelve controls. Ratcliffe and colleagues found slightly higher IQs (verbal M = 96.5; performance M = 97.0; full scale M = 96.3) for those cases. The patient's MMPI profile showed elevations on scales associated with depression and schizophrenia. Projective test assessment revealed themes of low self-esteem, sexual identity confusion, and family conflict, but no psychotic features. The Hamilton Rating Scale for Depression showed no significant elevation over the course of hospitalization. Brief Psychiatric Rating Scales (BPRS), collected by staff nurses as a repeated measure, showed no symptoms of a psychotic nature, but transient intense anger. On the gender dysphoria subscale (a 31-item subscale taken from the MMPI), which has accurately discriminated between gender identity patients and matched controls [11], the patient scored a 23 out of a possible 31 suggesting the presence of gender dysphoria. The Rotter Incomplete Sentences Task 1950 also supported the findings of gender dysphoria (e.g., item #7: *Boys* are cute; item #2: *The happiest time* I ever had was when I was dressing as a female; item #34: *I wish* I could be a girl; item #36: *I secretly* played with dolls). Assessment using the Gender Identity Scale for Males also revealed patterns similar to those seen in homosexual patients on the feminine gender identity scale [12].

The patient's treatment during this four-month hospitalization emphasized insight and supportive psychotherapy, a structured milieu to management intermittent suicidal behavior and sexual provocations, and attempts at development of longer term discharge plans to help the patient stabilize his life-style once out of the hospital. He adjusted quickly to the hospital, proved himself adept at both complying with ward routines as well as such tactics as manipulation and splitting of staff. His inpatient time was marked by highly charged emotional interactions with staff. Upon learning of his discharge date, the patient barricaded himself in his room and set fire to his bed, but sustained no serious injury. After this incident, he engaged actively with his therapist in dealing with termination, displayed considerable insight over this incident and his hospitalization, and was discharged in good condition. He returned to live with a sister elsewhere in the country, but as of last report had once again taken to hitchhiking across the country. He was not treated with any psychotropic medications during the hospitalization. Discharge diagnoses were: Axis I–Dysthymic disorder; Axis II–Borderline personality disorder; Axis III–Klinefelter's syndrome; Axis IV–stressors, moderate; Axis V–highest level of function, fair. A gender identity disorder was not diagnosed on Axis I because of the patient's chromosomal abnormality but was present according to psychological examination.

Discussion

Case studies, such as this one, are useful for the description of new syndromes, for the testing of new treatment techniques, or as illustrative of especially interesting cases. It is clear they are never sufficient to test complex etiological theories. As true single case experimental designs they also have some special strengths and some inherent limitations [13]. Generalizations based on this case must be cautious since the patient's presentation was quite atypical. Associations between genetic abnormalities and psychopathology can be fraught with difficulties in interpretation, as this case illustrates. The patient's clinical assessments revealed a complex picture. In the literature Klinefelter's syndrome has been reported in association with schizophrenia, neurologic syndromes, mental retardation, personality disorders, sexual deviance, criminality, and alcoholism [5]. Other personality traits sometimes associated with the illness include passivity, lack of sexual drive, immaturity, touchiness, paranoid sensitivity, insecurity, apathy, and poor capacity for work. Some authors have also pointed toward a connection between Klinefelter's syndrome and bipolar affective illness, often characterized by meglomania and short periods of manic-like disorders [15]. This patient was found to have Dysthymic disorder, personality disturbance (borderline personality disorder and personality features of immaturity, touchiness, paranoid sensitivity, and insecurity), sexually bizarre behavior, mild neuropsychological deficits, and significant gender dysphoria.

Before knowing that the patient had Klinefelter's syndrome, it was quite plausible to attribute his confusion about his sexual identity, his difficulties in establishing close interpersonal relationships, and his fragile self-esteem to childhood developmental traumas and disturbed family dynamics. Similarly, after the diagnosis of Klinefelter's syndrome was made, it was tempting to explain these psychological problems on the basis of his chromosomal and endocrine abnormalities. But it was most probably nature and nurture at work in concert that accounted for his sexual identity confusion and interpersonal difficulties. His development seemed to provide an excellent example of the transactional model of early social relations in which the interplay between constitutional and environment acting through parent-child interactions exerts at least as much influence on development as either nature and nurture alone [16]. In this case, the patient, who was at increased risk for a variety of psychopathologic conditions because of his XXYgenotype and his family history of mental illness, grew up in a chaotic, rejecting family environment. The patient's exclusion from male peers appeared to have been related to his temperament which was moody and unpredictable, and his dislike of traditional male activities. At home, he was expected to do household chores with his sisters rather than outdoor chores with his brothers, and was physically abused by his stepfather when he protested this. The patient's early rejection by his family appeared to be a function of his mother's poor parenting and her transient relationships with masochistic men and a high degree of family discord. Even without the genetic abnormality, he was at increased risk for major psychological difficulties later in life. His genetic and associated physical abnormalities probably influenced the form of his family rejection (i.e., humiliation by treating him like a girl, and marked him as the scapegoat for various family conflicts). It is not typical for persons with Klinefelter's syndrome to show this patient's level or form of psychopathology. His specific psychological problems can best be explained through the transactional influences of nature and nurture.

There are many clinical, ethical, and legal issues to be considered in treatment of an individual with sex chromosome abnormality [17]. Our interventions with the patient in the hospital were successful in resolving his immediate crisis, but probably had little effect long-term. He has resumed an itinerant life-style and will undoubtedly require similar inpatient care again. An optimistic outpatient treatment plan for him would include long-term psychotherapy, a stable living environment, and involvement in meaningful employment, all with the goal of reducing the severity of his life crises and improving his self-esteem. Transsexual surgery was considered as a high risk option because of the patient's borderline personality disorder and the low degree of life satisfaction associated with these procedures and was, therefore, not recommended. Much more might have been accomplished with earlier intervention. Early treatment with this family, in which both parents exhibited psychopathology, may have reduced their maladaptive parenting that focused so much conflict and rejection on this patient with a genetic disorder. In addition there is some evidence that aggressive

treatment of Klinefelter's patients with exogenous testosterone at the time of puberty may reduce subsequent psychopathology, presumably by producing both a physiologically and psychologically more normal adolescence [18-21]. Such testosterone treatments for this patient were begun late in adolescence, seemed to have only added to his internal emotional turmoil and sexual identity confusion, were probably less effective than if they had been initated earlier [19], and were ultimately rejected by him. In summary, primary and secondary preventive efforts of a biological and psychosocial nature, including early genetic detection, enlightened pediatric care, and sensitivity to family dynamics may significantly reduce the risk of dysfunctional outcomes for such persons.

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