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47, XYY Karyotype and Borderline Personality Disorder: An Italian Judicial Case and a Review of the Literature

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Abstract

During the 1960s and the 1970s, some studies postulated that Jacobs' Syndrome can lead to aggressive and/or criminal behavior, but the statistical certainty was questioned. In the 1990s new discoveries in neuroscience and genetics brought new attention to a possible biological component of deviance. In this context forensic psychiatrists were required to express opinions about a young Italian man afflicted by 47,XYY karyotype (Jacobs' Syndrome) and Borderline Personality Disorder who had frequent violent behaviors against his relatives, being therefore accused of "abuse against family members or partners" (art. 572 Italian Penal Code). The Court disposed for an evaluation of his mental conditions, his imputability and his possible social dangerousness, analyzing the influences on behavior from both his mental and genetic conditions.

Keywords: Abuse; Criminal behavior; Forensic; Personality disorder; violent

Introduction

The Judge of an Italian Court disposed for an expertise on a young man afflicted by 47,XYY karyotype and Borderline Personality Disorder who had had frequent violent behaviors against his relatives. The goal of the Judge's request was to analyze his mental condition at the time of his crimes and to express an opinion about his imputability (Italian Penal Code, art. 85: 'No one can be punished for an act envisioned bylaw as a crime if, at the time when it was committed, was not imputable. Imputable is who has the ability to understand right from wrong and to determine his volition and his behavior') and his social dangerousness (Italian Penal Code, art. 203: 'Socially dangerous is a person, even if not imputable or not punishable /…/ when it is likely to commit further offenses punishable by laws a criminal offense'). The judge seemed to believe that the genetic condition had determined the behavior of the subject and, since it is a constitutional requirement and cannot be cured, then this karyotype would make the man always socially dangerous.

Forensic psychiatrists made a psychiatric evaluation of the subject, considering the influence on behavior of both his mental and genetic conditions.

In particular, the XYY chromosome abnormality, also called Jacobs' Syndrome, was deeply studied because many researchers in the last fifty years have tried to find a link between this syndrome and violent and/or dysfunctional behavior. Must a man be considered predisposed to commit a crime, just because of its karyotype? How does this genetic abnormality influence his behavior or lead to the presence of a Personality Disorder?

47, XYY syndrome

Before proceeding to the presentation of the case, it seems useful to retrace the most significant evidence related to the Jacobs' Syndrome, an aneuploidy of sex chromosomes in which a human male receives an extra Y-chromosome giving a total of 47 chromosomes instead of the usual 46.

In 1961, Avery Sandberg, internist and cytogeneticist, and colleagues at Roswell Park Memorial Institute in Buffalo, New York, published the first report of a man with a 47, XYY karyotype. It was an incidental finding in an otherwise normal 44-years-old, 183 cm [6 ft.] tall man of average intelligence who was karyotyped because he had a daughter with Down syndrome [1,2].

Four years later, Patricia Jacobs, a British geneticist, made a more detailed study about this chromosome aneuploidy and described several of its characteristics, and since then the presence of an extra Y chromosome is also called Jacobs' Syndrome [3,4].

This syndrome is found in 1 of 1.000 male births [5,6] even if other studies have reported a different prevalence [4,7-12].

The only phenotypic expression that has always been found connected to an extra Y chromosome is an increased growth velocity from early childhood, with an average final height approximately 7 cm (3") above expected final height [13,14]. Nevertheless, in literature varied physical manifestations of Jacobs' syndrome are reported, although none of them statistically related to this chromosome abnormality.

More interesting and studied is the possible influence of the presence of an extra Y chromosome on psychological development and behavior [15]. As a matter of fact, in literature we find this syndrome often associated with low IQ level, cognitive impairment [16-20] delays in speech and language [21-23], poor writing skills [24] and learning

disabilities [23,25] Attention-deficit Hyperactivity Disorder [26,27] social-emotional problems [23,28,29] and sometimes autistic behavior [23,28,30-33].

Other authors described a relation with impairment in impulse control [34] low frustration tolerance, fits of anger from childhood, aggressiveness, tendency to antisocial behavior and even Borderline or Antisocial Personality Disorders [19,35-37]. In fact, since its discovery, the XYY trisomy has been connected with criminal and/or deviant behavior.

In this regard, the first studies date back to the 1960s and the 1970s, when a lot of research were conducted on offenders, often in prison, trying to find a direct relationship between an extra Y and the deviant behavior [3,38-47].

Afterwards, these studies came to a halt until the 1990s, when new discoveries in neurosciences and genetics brought new attention to a possible biological component of deviance [48-50]. Therefore, some research observed a great number of violent conduct and crimes in subjects suffering from Jacobs' Syndrome [51-53], while others denied a relationship between an extra Y and deviance [24,54-57]. According to other studies, the connection between this chromosome abnormality and dysfunctional behavior was rather considered possibly due to associated intellectual deficits or to underlying aggressiveness [18,53,58]. Contrariwise, other authors hypothesized that these men are more easily recognized as offenders because of their high stature and stocky build, which would make them more identifiable [59-69].

All the aforementioned studies however failed to reach an unequivocal result. The statistical certainty of these studies is questioned and, even today, there are differing opinions about the relevance of Jacobs' Syndrome in predisposing or causing dysfunctional behavior, uncertainty that has to be considered if a forensic psychiatric assessment of imputability and social dangerousness is required.

Mario's history

Mario, a fictitious name, was born of natural birth at term, macrosomic. He was the second child in a middle-class family without apparent problems. His psychosomatic development was normal except for an early and impressive height growth. Since kindergarten, he manifested learning, relational and behavioral problems, which became even more evident after he started primary school.

When he was 7 years old, his father died and he developed a Prolonged Grief Disorder that had been treated for two years with neuroleptics (Neuleptil, Haldol, and Depakin).

Mario's problems continued however to get worse, and, because of the learning difficulties, on two occasions at school he failed to advance to the next grade, thus finding himself placed in a class with younger companions. Moreover, he looked very tall and strong and, because of this particular phenotype, his classmates frequently mocked him, so he often reacted aggressively. During childhood and adolescence, Mario was unable to establish real friendships or romantic relationships.

Because of both his physical appearance and his aggressive behavior, he was soon labelled as being different and isolated from peers, starting therefore to congregate with other problematic or deviant subjects and to develop a drug addiction, with drugs (synthetic drugs, marijuana, heroin, cocaine) and even alcohol abuse. Because of his behavioral problems and his drug addiction, at the age of 15 he was taken into care by psychiatric services. He stopped studying at the end of compulsory schooling, he refused to take any further school training and he showed no interest in any work activity.

At the age of 17, the Juvenile Criminal Court decided his placement into a therapeutic community, where he was diagnosed afflicted by Borderline Personality Disorder with difficulties in anger management and frequent recourse to aggressiveness and because of his tallness and strong build, a chromosome analysis was also made, showing a 47,XYY karyotype. One year later, Mario left the community and returned to his family. Almost immediately he stopped taking his medications as prescribed and instead he began again the abuse of alcohol and drugs, so that, having reached the age of 18, soon squandered a big part of the inheritance left to him by his father to buy drugs.

The situation became even more critical as he began to steal valuable objects from home and continually asked money from his mother and sister, reacting with aggressive behavior and temper tantrums if they refused. His family, troubled about his continuous anger's outburst and worried of being attacked, repetitively turned to law enforcement.

In this context, in accordance with the Italian Penal Code, he was accused of 'abuse against family members or partners' (Italian Penal Code, art. 572: 'whoever, /…/ mistreats a person of his family or other cohabitant, /.../ is punished with imprisonment from two to six years'). Because of the possibility of repeating the same crime, the Court decided his removal from the family and his hospitalization for treatment in a Forensic psychiatric hospital. At the same time, the judge, considering the diagnoses of Borderline Personality Disorder and especially of immutable Jacobs' Syndrome, decided to apply for forensic psychiatric assessment in order to understand his mental condition at the time of his crime commission.

Mental examination

The mental examination was performed in the Forensic psychiatric hospital in which he was hospitalized.

Mario was a young 20-years old man, over 2 meters tall and weight over 100 kg. He showed himself polite and cooperative, trying to appear as a person aware of the incongruity of his precedent conducts in the light of the alleged actual achievement of a stable mental and behavioural balance. He claimed that now he had understood the importance of a correct and regular drug treatment and that he was now willing and able to do even some jobs that require responsibilities.

In telling his life history, he justified his aggressiveness and his deviant conduct with the early loss of his father who was supposed to contain his impulses and to train and educate him. On the other side, he tended to find explanation and responsibility for his own dysfunctional behaving in attendance bad company or false friends, at the same time minimizing the importance and significance of past aggressive conducts.

At the time of mental examination, Mario appeared in a discrete psycho-behavioural status, but he still had a great intra-psychic tension. His affectivity was strictly controlled and censored in order to contain the impulses and instincts; his mood was dysphoric. He showed deficits in cognition abilities, with a response to certain stimuli from emotional reactivity rather than from conscious cognition. His capacity for critical analysis and judgment was inadequate in case of unexpected or stressful events. He showed tendency to manipulation,

to projective conception, to minimization and irresponsibility, he appeared unable to learn from punitive experiences and was lacking in insight of disease.

The MMPI-2 showed a person generally characterized by antisocial behavior and a poorly developed conscience, which led frequently to implement socially unacceptable conducts (alcoholism and substance use, aggressive acting out and violent outbursts of anger), properties also difficult to control because of the lack of ability to learn from experiences of punishment and rejection of responsibility for his behavior. Such a framework brought Mario to have problems in all major areas of life (emotions, work, and social life) and to have trouble with the law. There was a resistance to treatment; he started to take medicine only because forced by environmental pressures. The configuration so far delineated was enriched with delusions of persecution and grandeur. Because of the presence of a hypertrophic confidence suffocating reality testing, he seemed unable to make an efficient analysis of the context, which is necessary to assess the real difficulties that it may suggest.

In summary, relying on psychiatric examination, Mario presented a pervasive pattern of instability of interpersonal relationships, of self-esteem and mood, and marked impulsivity began since childhood. He presented frequent and unexplained mood swings, persistent instability in self-image, chronic sense of emptiness, fear of abandonment, which frequently led to manipulative attitudes, impulsive behaviours in risk areas (e.g.: substance abuse), often marked dysphoria and emotional reactivity, inability to control anger, and/or hetero-directed aggressive behaviour, dichotomy, idealization and devaluation (in relationships with others). All these characteristics indicated the diagnosis of a Borderline Personality Disorder, diagnosis already previously done by doctors who had treated him before.

Discussion

As described above Mario's life history is differing from a normal life experience since his early childhood; he had learning disabilities, he was taller and had a stronger build compared to other children who isolated him, he showed aggressiveness.

At age of seven, his father died, loss that resulted in feelings of abandonment and affective instability, and his problems became more serious. In adolescence, he began using drugs and alcohol and he made deviant and violent conducts, especially against his mother and sister, in order to obtain money for buy drugs. He was soon reported to the Juvenile Criminal Court, which decided to place him in a therapeutic community, where was diagnosed a Borderline Personality Disorder and Jacobs' Syndrome. He has never done any professional training course or worked continuously. Because of numerous aggressive acting out, Mario was finally placed in Forensic psychiatric hospital. Some psychiatrists who had treated Mario in the previous year thought that his mental status could not improve because of his genetic condition, which is immutable and so cannot be treated.

The mental examination showed his severe difficulties in the management of instinctual instances, a pathologically asthenic volition and some cognitive deficits, which deprive him of additional resources that may be mobilized in order to contain and process adaptively negative stimulation. Mario has developed a working model in which the non-fulfillment of immediate impulses gives rise to a nearly constant stress experience. In this condition, even very small quantities of exogenous or endogenous insults engage an excessive stressful stimulus. The co-presence of some deficit of intellectual faculties, which affects his ability to analyze them functionally as stressors, the insulting content of them are inevitably transferred on the extratensive side with the implement of uncontrolled aggressive behavior. He had difficulty in managing feelings of anger and he frequently recurred to aggression shake. In other words, he seemed to be unable to control his impulses and to learn from the negative consequences of his actions.

Aware of Mario's life history, the forensic psychiatrists had to determine if Mario was or was not insane at the time of his crimes ('abuse against family members or partners') and if at the time of the examination, he presented a substantial risk of repeating new offenses. The Italian Penal Code provides that there is an abolished or diminished imputability for crime if a correlation between the mental disease and the crime can be demonstrated, if the disease was in course at the time of the crime, and if there is a link between the motives behind the crime and the disease. In other words, somebody is considered to have a mental impairment if the crime can be shown to be a symptom of his disease. Although until 2005 only different forms of psychosis were considered suitable to influence the imputability, according to the pronouncement of the Italian Court of Cassation, also different forms of 'severe' Personality Disorder can be considered able to affect the mental state at the time of crime commission.

The examinee, bearer of 47, XYY chromosome abnormality, was also diagnosed affected by Borderline Personality Disorder.

The particular question in this case is how did Mario's two diseases condition and influence his violent behaviour? The Judge wanted especially to know if his crime was due to his genetic condition or to his mental disorder.

First of all, the analysis of the literature shows that about the 75% of cases of Jacobs' Syndrome are not detected prenatally or postnatally [61], because these subjects often have an adequate mental and psychological development [70,71] and therefore often the diagnosis is accidental, delayed or even absent [5,61].

Moreover, studies on the Jacobs' Syndrome have never reached unique results; the only certain correlation with an extra Y seems to be an excessive height and a strong build, and often the anomaly is associated with a low IQ level, with learning disabilities. While some authors underlined the great number of violent behavior and different forms of crime or deviance in subjects suffering from this syndrome, they did not achieve statistically significant results. Even the recent researches in neuroscience on genetic anomalies can only suppose a major risk to commit deviant conduct, especially if other external factors are present. Some authors have written about a possible correlation between an extra Y and mental illness, such as Personality Disorders, but this link is neither automatic nor certain, and therefore many authors suggested that also other factors, such as environmental and social ones must be considered.

As regards the Borderline Personality Disorder, the most recent studies in the aetiology of BPD are added together and intersect several factors: a genetic and constitutionally component, a mother figure too overwhelming, a history of family's abuse (psychological, physical and / or sexual) or some trauma or abandonment during childhood. Obviously, the relative importance of each of these factors remains to be determined case-by-case [62]. Once again, a bio-psycho-social model of interpretation thus prevails.

Taking into account the findings of the literature, we can say that Mario has the characteristics described in the literature both about the Jacobs' syndrome (such as high robustness and stature, learning

deficits, mild cognitive impairment, aggressive behavior), and about BPD (unstable interpersonal relationships, impulsivity, emotional instability, chronic feelings of emptiness, intense anger, difficulty controlling anger). Briefly, both the diagnoses seem to be characterized by impulsivity and aggressiveness.

In this context, it may be interesting to mention a new approach to the study of aggressive behavior. Recently, neurosciences have developed new bio-social theories, which empathize how criminal behavior cannot be interpreted in a 'deterministic' way in which the central role is performed only by constitutional factor, but rather on the basis of evidence that biology and environment interact continuously in the characterization of the human being, from the creation of the embryo and throughout the whole existence. It is a growing interdisciplinary approach to the study of crime in which the interaction between biology and environment is the keystone [63]. Basic assumptions on which this discipline moves are evidence that the biological predisposition to antisocial behavior has a greater effect in those who live in crime-genetic environments and that the social causes of crime affect more significantly more biologically vulnerable individuals.

Over the past two decades, theoretical models have strongly influenced research on aggression: 'social learning' and the 'disease outset'. According to these theories, the children would learn aggressive behavior by the environment in which they grow and the development of destructive behavior ('illnesses) would be triggered by excessive exposure to harsh environmental models, such as family violence and the media [63].

In line to more recent studies, however, aggressive behavior is not learned, but the child will become a violent teenager and a violent adult if he has not adequately learned to control his behavior. The research is therefore directed to the study of the factors that make learning self-control difficult, especially from early childhood [64,65].

According to the current of the so-called bio-social criminology, the various factors involved (genetic, neuropsychological, environmental and evolutionary factors) should be analyzed and studied in the actual of their interaction to form a unitary and coherent interpretative perspective in the study of crime [66].

According to that, the aggressive behavior cannot be considered just the consequence of the 47, XYY syndrome.

Mario's psycho-social experience has certainly played a role in the development of his mental illness. His father's early and unexpected death, with the consequent lack of a person appointed to direct the control of his impulses, his cognitive deficit, his being physically diverse from other children of his age, have certainly isolated him from classmates, he was shunned by them and so he felt different from others. In this sense, this psycho-social experiences can be considered also a consequence of the phenotypic expression of the 47, XYY karyotype, of course with other life experiences.

As aforementioned, according with the Italian Penal Code, the answer given to the question of the Court was that the behaviour of Mario, in his nature of impulsive aggressive and violent acting against his family, was expression of the symptoms of the disease affecting the subject. Mario had no possibility to control or contain his impulse and his violent behaviour. Consequently, the results of the forensic psychiatric evaluation led to consider Mario not imputable at the time of the crime commission, in accordance with the requirements of the Italian Penal Code, because he was suffering from a 'severe' Personality Disorder that caused an impairment of his . On the other side, his habitual drug abuse was not considered influencing his imputability in accordance to the Italian Penal Code (Article 94. 'When the offense is committed in a drunken state, and this is habitual, the penalty is increased. /…/ also apply where the offense is committed under the influence of drugs by those who are addicted to the use of these substances').

Mario was also considered still mental ill at the time of the evaluation, so forensic psychiatrists prescribed a regimen of psychiatric and psychological care and treatments inside a suitable facility.

Conclusion

More than 50 years of studies on the Jacobs' Syndrome, new knowledge in the field of neurosciences and the recent bio-social approach do not seem to have yet given definite answers about the problem of deviant behaviour, although it appears to emerge a prevailing multifactorial interpretation.

The presence of an extra Y seems just to be a genetic substrate, which cannot be considered the only cause of a deviant behaviour. Even although the results of recent researches in neuroscience claim that one or more genetic abnormalities may be associated with a statistically higher risk of manifesting a deviant behaviour, at the same time show that this anomaly is not a condition nor necessary nor sufficient for the development of a dysfunctional conduct. In fact, many studies have underlined the importance of environmental, social and family background in the developing of a criminal conduct [24,47,54-56]. In addition, the BPD cannot be considered just as a direct and certain consequence of the presence of an extra Y, because also social and environmental factors may contribute to the establishment of such a mental disorder. So antisocial and deviant behaviours are thought to have multifactorial aetiology, in which the genetic component may play some role, but should not be considered the sole cause.

Consequently even the psychiatric assessment of subjects with this chromosome abnormality has to be approached in the usual way: only a depth analysis of subject's life history, considering environmental, social, familiar, cultural and genetic factors, and a clinical examination of the offender, eventually supported by some psychological tests, can point out psychopathological signs and symptoms which could lead the physician to a diagnosis and to the evaluation of the mental competency at the time of crimes [67,68]. However, in a psycho-pathological examination of such subjects it is necessary to consider also their genotype and the influence that it can produce on the social context.

In this case report, the authors can suppose that the 47, XYY syndrome has perhaps had an indirect influence in Mario's life history and mental disease, because the connected phenotypic characteristics, such as high stature, the large body size, aggressiveness, may have fostered the development of an unfavourable socio-environmental context, on which also other factors worked, such as his father's death, his being isolated by peers, to determine his dysfunctional conduct. Mario was considered insane at the time of his crime because of Borderline Personality Disorder, which affected him but not predisposed or 'forced' to violent behaviour in direct correlation with Jacob's Syndrome. On the other side, a possible conclusion of a direct correlation between the chromosomal abnormality and deviant behaviour would moreover make a therapeutic treatment impossible

and Mario would never have the possibility to come out of the judicial circuit.

The old question, if a human being is free to decide or if his behaviour dependents from several internal and/or external factors and if he is forced to be deviant or if he can decide to become deviant, however remains open. Mario seemed to have found the answer and so once he said: 'An extra-Y can influence, but I'm the only one who can decide who I am and how to behave, as well as I'm the only one who decided by himself to take drugs and to be a delinquent'.

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