Case Report

A Case Report of Klinefelter Syndrome with Schizophrenia-Like Psychosis and Seizure Disorder

Anu Rita Jayaraman, Maithreyi Poguri, Nambi Siva

ABSTRACT

Klinefelter syndrome is a disorder of variation of sex chromosome, the most common karyotype being 47XXY. Multiple case reports and articles have been published linking the increased prevalence of psychiatric disorders like Schizophrenia, Schizophreniform psychosis, Attention deficit hyperkinetic disorder, Learning disorder, etc. and seizure disorder in Klinefelter syndrome than in general population, attributing to the extra X chromosome. Here is a case of a 45-year-old gentleman with Klinefelter syndrome with schizophrenia-like psychosis and seizure disorder. He was diagnosed as Klinefelter syndrome 15 years back by genetic testing (47XXY) when he was investigated for infertility. His luteinizing hormone (LH) (32.04 mIU/ml) and follicle-stimulating hormone (FSH) (50.70 mIU/ml) levels were high and his testosterone level was low (1.76 ng/ml). He had four episodes of seizures in 2004 for which he was started on phenytoin and sodium valproate, and was seizure-free for past 10 years. He was brought to our hospital in July 2014 with complaints of talking and laughing to self, suspicion, hearing voices and aggressive behaviour, which were persistent mildly for past 15 years and aggravated for past 6 months. He was not going for work for past 15 years, does not mingle with relatives or friends.

Key words: *Klinefelter syndrome, schizophrenia-like psychosis, seizure disorder*

INTRODUCTION

Klinefelter syndrome is a disorder of variation of sex chromosome, the most common karyotype being 47XXY. It is characterised by hypogonadism, gynecomastia and azoospermia. There is increased prevalence of psychiatric disorders like schizophrenia,[1] schizophreniform psychosis, attention deficit hyperkinetic disorder, learning disorder, etc. and seizure disorder[2] in Klinefelter syndrome than in general population, attributing to the extra X chromosome. Many with Klinefelter syndrome are not aware of their genetic impairment and diagnosis is significantly delayed in 75% of subjects.[3]

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CASE REPORT

A 45-year-old gentleman was brought to the outpatient department by his wife. He was already diagnosed as Klinefelter syndrome 15 years back by genetic testing (47XXY) when he was investigated for infertility. His luteinizing hormone (LH) and follicle-stimulating hormone (FSH) levels were high and his testosterone level was low at that time in 2000, when he was 30 years of age. He had four episodes of seizures in 2004 for which he was started on phenytoin and sodium valproate, and was seizure-free for past 10 years. He was brought to our hospital in July 2014 with complaints of talking and laughing to self, suspicion, hearing voices and aggressive behaviour, which were persistent mildly for past 15 years and aggravated for past 6 months.

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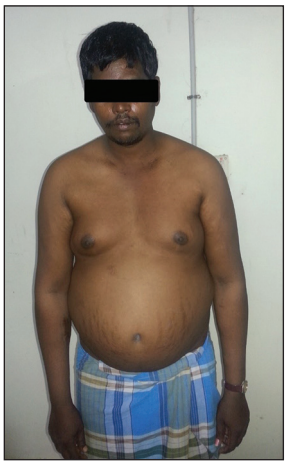

Figure 1: Photograph of the patient

He was not going for work for past 15 years, does not mingle with relatives or friends.

On physical examination, bilateral atrophic testes, gynecomastia were present and waist circumference was 110 cm. On mental state examination, delusion of infidelity, delusion of reference, 2nd person auditory hallucinations were noted. Poor motivation to work, to take care of family and self were present. He was diagnosed to be suffering from schizophrenia-like psychosis and started on antipsychotics. Positive symptoms improved with medications [Figure 1].

Investigation

Genetic testing: 47XXY
LH: 32.04 mIU/ml
FSH: 50.70 mIU/ml
Testosterone: 1.76 ng/ml
Echocardiography (ECHO): Normal
Computed tomography (CT) brain: Normal study
Electroencephalography (EEG): Epileptiform activity

DISCUSSION

This rare case of Klinefelter syndrome is presented here to emphasise both the significance of genetic model[4] of psychotic disorders and the need for assessment of psychiatric comorbidities in such patients. The association between the presence of extra X chromosome in males and the increased prevalence of schizophrenia, schizophreniform psychosis, attention deficit hyperkinetic disorder, learning disorder and criminality[5] are to be addressed in future research.

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