CASE REPORT

PSYCHOEDUCATIONAL PROFILE OF KLINEFELTER SYNDROME

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Abstract

Forty-seven XXY aneuploidy is the most common disorder of sex chromosomes with a prevalence of 1:500 males. However only 1/3 cases are diagnosed due to phenotypic variability and insufficient professional awareness of the syndrome. School children with Klinefelter syndrome (KS) present with language delay, learning disabilities and behavioral / social problems. We present a case of Klinefelter syndrome (KS) with language based learning disabilities.

Keywords: Klinefelter syndrome, language delay, learning disabilities, behavioral problems

Introduction

Klinefelter syndrome (KS) are a group of chromosomal disorders with an extra X chromosome added to a 46 XY male karyotype. (1) KS has a varied phenotype leading to gross underdiagnosis. (2) The characteristic features of KS are tall height, gynecomastia, sparse body hair, small testis, decreased muscle mass, feminine distribution of adipose tissue, azoospermia and infertility. (3) Majority of KS children have a normal but a characteristic cognitive phenotype. (4) They have a deficit in verbal processing affecting comprehension and learning leading to significant under achievement and a generalized learning disability. Identification of this is of immense importance in planning early psychoeducational interventions.

Case Report

An eleven year old boy presented to the clinic with academic underachievement for purpose of certification to avail of provisions laid down by the education authorities. He knew multi-languages, was from upper middle socio-economic class in the seventh grade of a central board of secondary education (CBSE) school. He had academic lags in the areas of reading, comprehension in all languages and poor written expression. In mathematics, analytic reasoning was preserved but computation errors occurred due to impulsivity. The school report said additionally he was slow in copying, unable to express verbally, had attention deficits and emotional immaturity. The parents expressed concern regarding their child?s inability to remember names of commonly used words and to develop a sustained coherent narrative. Developmental history was notable for mild motor delay with walking achieved at 18 months language delay (2 years). On examination, height was 155cms (95th centile for age), he had hypotonia, gynecomastia and flat feet. Soft neurological signs like dysdydokinesia, graphesthesia, difficulty in hopping and maintaining balance were elicited. Chromosomal evaluation showed 46 XXY Klinefelter syndrome karyotype. Based on the Diagnostic and Statistical Manual of Mental Disorders, 4th edition text revision (DSM IV-TR) a diagnosis of attention based hyperactivity disorder, predominantly inattentive subtype was made. His cognitive assessment revealed a full scale intelligence quotient (FSIQ) of 117 with a verbal intelligence quotient (VIQ) of 116 and performance intelligence quotient (PIQ) of 115. Intelligence quotient (IQ) was above average without significant differences in verbal conceptual and non verbal reasoning. But deficits in arithmetic reasoning, visual memory were seen due to poor processing speed and inattention. Psychoeducational assessment was done using Woodcock Johnson tests of achievement (Standard and extended). The IQ scores and the educational assessment scores were compared to calculate discrepancy between potential and academic achievement for fulfilling the essential criteria in diagnosis of a learning disability. Significant discrepancies were established in the areas of broad reading, reading comprehension, broad written expression suggesting a language based learning disability. Skills of word decoding, spelling, fluencies were affected due to poor processing speed. On testing for auditory processing deficits he had poor auditory word discrimination, auditory sequencing deficit with an impaired auditory memory.

Discussion

Cytogenetic surveys of neonates have found that approximately one boy in 500 is born with an extra sex chromosome. Only 10 percent with KS are diagnosed prenatally with another 25% diagnosed during childhood or adulthood and alarmingly two thirds of affected individuals remain unidentified. (5) Specific learning disability especially in reading is seen in 50-75% of KS cases. (6) Language difficulties have been identified in 70-80% in them from an early age hence speech and language problems precede the academic achievement deficits. (7). Auditory processing and verbal memory are the core deficits that underline linguistic deficits in these children. Boys with KS do poorly on word decoding, reading comprehension, spelling, written language skills and these deficits result in language based learning disability e.g. reading and spelling which was seen in our patient. (8) The poor reading comprehension which was also present in our child severely compromised his capacity to derive information from print. Patients with KS have inability to convey their specific needs to the teacher which leads to behavioral problems. Cognitive profile in KS is characterized by a low VIQ and high PIQ but in older children this discrepancy diminishes with advancing age as seen in our child in whom the V-P split was not appreciable. KS boys are shy, immature, sensitive, anxious and prone to psychiatric ailments later in life. (7) Difficulties in motor domains result in limited participation of sports activities. Treatment of a child with language based learning disability includes early speech ? language therapy, appropriate educational interventions, behavioral, physical therapy. Psychological and emotional support to parents while communicating the diagnosis is vital. Hormonal treatment at puberty improves motor skills but has no impact on cognition.

Conclusion

Diagnosis of KS should be considered in all tall boys with language, learning problems as this disorder has a significant impact on school learning and success.

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