

## Case Report

# Klinefelter syndrome and specific learning disability

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**47XXY aneuploidy is the most common disorder of gender chromosomes with a prevalence of 1: 700 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) were presented with language delay, learning disabilities and behavioral/social problems. Although Klinefelter syndrome (KS) with language based learning disabilities is not uncommon, this case was unusual due to its coexistent condition of attention deficit hyperactivity disorder (ADHD) and specific higher cognitive deficits on psychoeducational profile.**

**Key words:** Klinefelter syndrome, language delay, learning disabilities, behavioral problems, attention deficit hyperactivity disorder (ADHD).

## INTRODUCTION

Klinefelter syndrome (KS) is a group of chromosomal disorders with an extra X chromosome added to a 46 XY male karyotype. 47XXY aneuploidy is the most common disorder of gender chromosomes with a prevalence of 1: 700 males. KS has a varied phenotype with characteristic features of tall height, gynaecomastia, sparse body hair, small testis, decreased muscle mass, feminine distribution of adipose tissue, azoospermia and infertility (Smyth and Bremner, 1998). Majority of KS children have a normal but a characteristic cognitive phenotype (Rovet et al., 1996). 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 was presented to the clinic with academic underachievement for the purpose of certification in order to make available the provisions laid down by the education authorities. He was multilingual from the upper middle social economic status (SES) in

the seventh grade, Central Board of Secondary Education (CBSE), and was presented with academic lags in the areas of reading comprehension in all languages, with poor written expression. He was frequently lost in thoughts and could not express them effectively both verbally as well as on paper. He had difficulty in both copying and writing from memory although oral expression was better than written expression. He made careless mistakes in computation but understood concepts well in both Mathematics and Science however reading comprehension was affected in languages. Occasional spelling errors particularly with unfamiliar words, lengthy words were present.

In mathematics, though analytic reasoning was preserved, simple computation errors occurred due to impulsivity. Additionally, the school reported that he was slow in copying, unable to express verbally, had attention deficits and had considerable emotional immaturity. Concerns of poor concentration and inattention to details were also noted by the parents and teachers alike.

He was reported to have difficulty in sustaining peer interactions and his behaviour was often considered socially inappropriate for his age. He had mild anger management issues and a poor self image and self esteem.

Developmental history was notable for mild motor delays walking (18 months) and language delay (2 years). On examination, he was tall (his height was measured at the 95th centile for his age), generalized mild

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**Table 1.** WISC IQ profile.

Verbal scale	IQ	Performance scale	IQ
Information	15	Picture completion	13
Comprehension	13	Picture arrangement	12
Arithmetic	05	Block design	16
Similarities	13	Object assembly	13
Vocabulary	14	Coding	02
Digit span	14	Mazes	16

Verbal IQ (VIQ) - 116 performance IQ (PIQ) – 115, full scale IQ (FSIQ) – 117.

**Table 2.** Woodcock Johnson III Psychoeducational Tests of Academic Achievement (Standard and Extended Battery).

WJ III achievement clusters	Actual achievement	Predicted achievement	Ability/Achievement SS difference	Discrepancy SD	Significant At -1.50 SD
Math calculation skills	107	110	03	0.25	NO
Math reasoning	96	113	17	1.70	YES
Broad reading	94	112	18	1.72	YES
Reading comprehension	79	112	33	3.29	YES
Written expression	88	109	21	1.68	YES

SS - Scaled scores, SD - standard deviation.

hypotonia, gynaecomastia, flat feet, and soft neurological signs like dysdydokinesia, grapesthesia were also present. He had mild incoordination issues with difficulty in hopping and inability to maintain a steady stance. Chromosomal evaluation done was significant for chromosomal aneuploidy [46 XXY] Klinefelter syndrome karyotype.

Based on the DSM IV-TR (Diagnostic Statistical Manual) clinical criteria, a diagnosis of the Attention based hyperactivity disorder, which predominantly consists of an inattentive subtype, was made. Psychological tests as the WISC-R (Weschler's Intelligence Scale for Children- Indian adaptation by Mahindrika Bhatt) provides a broad assessment of general cognitive skills and describes the typical functioning in specific areas of cognition broadly divided into the verbal intelligence quotient (VIQ), performance intelligence quotient (PIQ) and full scale intelligence quotient (FSIQ). The average above his cognitive profile done as per the WISC-R (Table 1) revealed a full scale IQ - 117 with a VIQ - 116 and PIQ – 115, that is, IQ was above average without significant differences in verbal conceptual and non verbal reasoning. But on further analysis of the WISC report, the scaled score of Arithmetic subset in the verbal scale was very low (05) suggesting that severe deficits in arithmetic reasoning were present. The coding subset in the performance scale had low scaled scores (02) implicating fine motor issues, poor processing speed and inattention. Psycho educational assessment for specific learning disability was done using Woodcock Johnson III (WJ III) test of

Academic Achievement (Standard and extended battery) (Table2). The WJ III measures broad reading, reading comprehension, written expression, mathematic reasoning and calculation skills among other clusters. Significant discrepancies in the scaled scores of predicted ability versus actual achievement were seen in areas of broad reading (94), written expression (88). High order cognitive skills such as reading comprehension were much lower (79) when compared to broad reading (94) skills, the mathematics reasoning cluster (96) was also low. 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. The significant discrepancies established in the areas of broad reading, reading comprehension, broad written expression suggested a language based learning disability. In spite of the child's innate intelligence skills of word decoding, spelling, fluencies were affected due to the poor processing speed.

## DISCUSSION

Cytogenetic surveys of neonates have found that approximately one boy in 500 is born with an extra sex chromosome. Only about 10% children with KS are diagnosed prenatally, while another 25% are diagnosed during childhood or adulthood and alarmingly about two thirds of affected individuals tragically remain unidentified forever in their lifetime (Bogesen et al., 2003). Specific

learning disability especially in reading is seen in 50-75% of KS cases (Verri et al., 2010). Since children with KS have a predominantly language based learning disability, language difficulties are known to occur in almost 70-80% of them. Therefore, speech and language problems especially during early age of life, acts as a red flag sign in predicting and may actually predict the later academic achievement deficits (Boada et al., 2009). A recent study on the description of the cognitive phenotype of KS in a sample of 50 children concluded that specific language, academic, attention and motor abilities tend to be impaired and are a major cause of concern (Ross et al., 2008) Although the cognitive profile in KS is characterized typically by a low VIQ and high PIQ due to poor verbal skills versus the non verbal performance based skills in older children this discrepancy diminishes with advancing age (Verri et al., 2010). Many features in our assessment such as the language based learning disability, ADHD, social immaturity, poor motor coordination are seen to be in synch with that of the literature reviewed.

Due to the oral and written language problems of boys between 5 to 12 years of age, anticipatory guidance is recommended for these boys to prevent secondary maladaptive behaviors (Graham et al., 1988). Boys with KS do poorly on word decoding, written language skills and these deficits result in language based learning disability, for example, reading and spelling (Graham et al., 1988). The poor reading comprehension severely compromises their capacity to derive information from print. Clinicians should be aware of the possibility of KS in a child with physical features and behavioral and academic problems. The inability of these children to convey their specific needs to the teacher leads to behavioral problems in them and possible psychiatric complications later. KS boys are shy, immature, sensitive, anxious and prone to psychiatric ailments later in life. Difficulties in fine motor and coordination issues results in limited participation of sports related activities which are essential in developing social relationship and peer approval during the schooling period. These may further impact peer relations thereby limiting the child's social milieu. Awareness of this particular psycho educational and neurological profile can help in planning multidisciplinary intervention goals such as remedial education, occupational therapy, speech therapy, social skills training, behavior modification and medications Psychological and emotional support to parents while communicating the diagnosis is vital and of equal importance (Graham et al., 1988). Although hormonal treatment at puberty improves motor skills, testis development it has no known impact on improving the child's cognitive ability. A language based learning disability with features of KS should justify karyotyping in a child given the high prevalence of the syndrome especially if behavioral issues are also present. Physicians should have a low threshold for obtaining a

karyotype in children with ADHD and learning disabilities, particularly in boys who are tall. Early detection could improve the prognosis of the learning and behavioral problems through medications and other non pharmacological interventions. Hence the diagnosis of KS should be considered in tall boys with characteristic physical features, language, academic problems as this disorder has a significant impact on learning and academic success.

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