KLINEFELTER SYNDROME PRESENTING WITH LEARNING DISABILITIES: 2 CASE REPORTS

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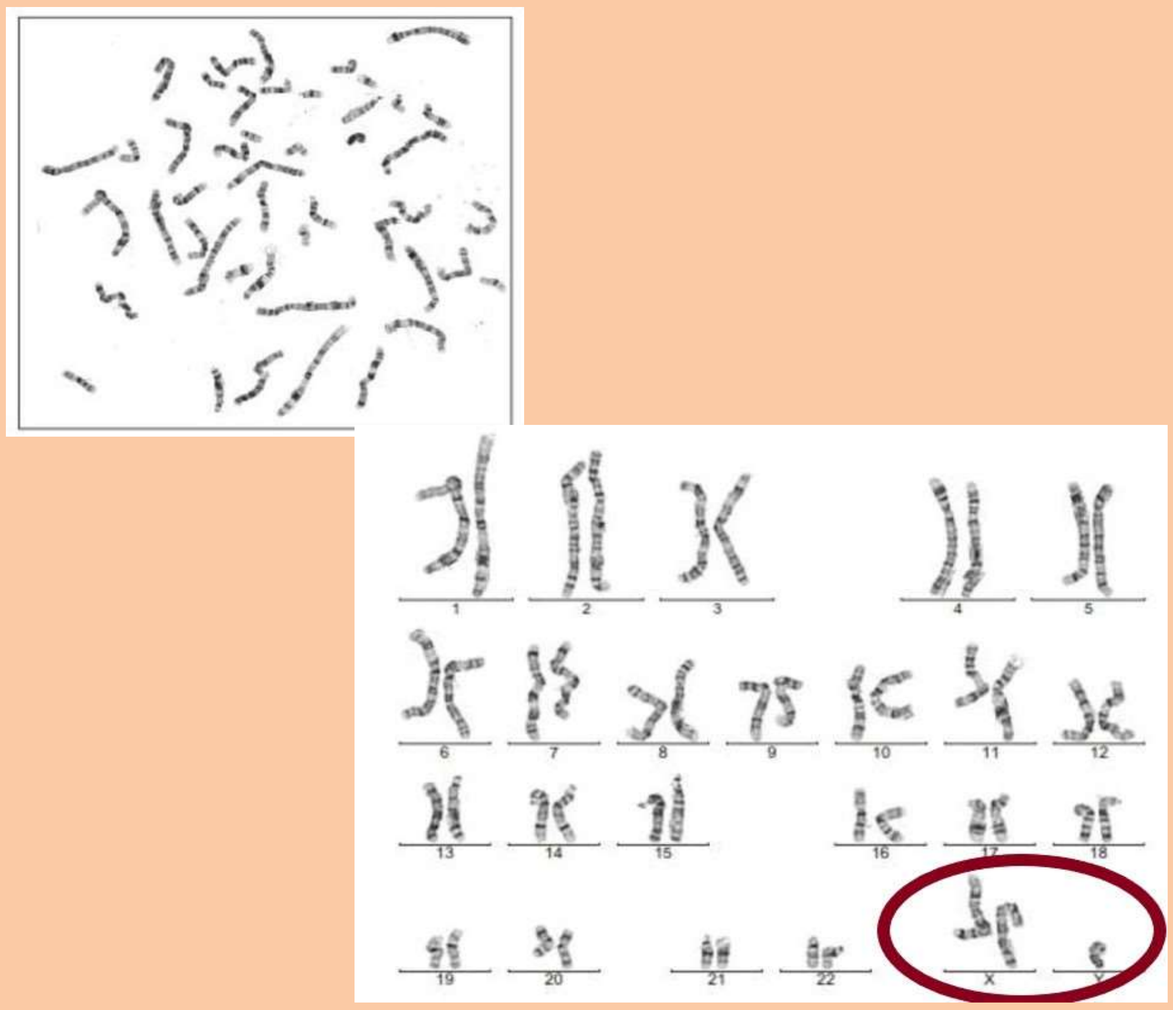
INTRODUCTION

Klinefelter syndrome (KS) occurs in approximately 1 in 650 males, representing the most common sex chromosome disorder. However, it is estimated that only 25% of KS patients are ever diagnosed, and 90% of them are not identified until 15 years of age. The disease is caused by congenital aneuploidy of the sex chromosomes; the most usual karyotype being 47, XXY. Typical phenotype includes tall stature, hypergonadotropic hypogonadism, small testicular volume, and fertility issues. Characteristic neurocognitive, behavioral, and language deficits of varying severity have been reported in patients with KS, including verbal disabilities, learning disabilities, executive function impairments, psychiatric disorders, autism spectrum disorders, and attention-deficit/ hyperactivity disorders.

CASE 1

An 11-year-old boy was referred to the Pediatric Endocrinology Outpatients Unit of our Hospital due to obesity. The parents reported nothing unusual in their son's medical history, except from learning disabilities and behavioral issues. The boy primarily exhibited social and emotional developmental delays, as well as school difficulties, including deficits in speech and language skills. The child's height was 155cm (95th percentile) and he weighed 60kg (>95th percentile), with a BMI of 24.97 (>95th percentile). On physical examination, sexual maturity rating of his gonads, pubic hair and axillary hair were at stage 1. Additionally, gynecomastia was noted. Laboratory testing revealed prepubertal FSH, LH and testosterone levels. Chromosome analysis detected a 47,XXY karyotype.



Chromosome analysis revealing a 47, XXY karyotype in case 1 patient.

CASE 2

We also report the case of another 11-year-old boy with KS who initially presented 5 years ago with prominent learning disabilities and speech difficulties. Developmental milestones were slightly delayed. Physical examination and past medical history were unremarkable. Cytogenetic analysis eventually revealed the diagnosis. The child has been receiving speech, occupational and behavioral therapies for the last 5 years. These interventions have greatly improved behavioral and learning skills.

CONCLUSION

Although many individuals with KS become highly successful in their academic pursuits and social lives, it is clear that there is a distinct behavioral and neurocognitive phenotype associated with the extra X chromosome. As the learning disabilities, speech delays, and behavioral difficulties usually develop in childhood they present a unique opportunity for early detection of KS and timely intervention. Clinicians should therefore maintain a high level of vigilance for KS in boys with learning difficulties.

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Sex differentiation, gonads and gynaecology or sex endocrinology

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