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Overview of Klinefelter Syndrome

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Introduction

The clinical and genetic manifestations of Klinefelter Syndrome (KS) are quite heterogeneous. Although the association between clinical phenotype and genetic background has been largely revealed, clinicians are aware that there are still many parts of this subject that are unknown. Patients' interindividual disparities in terms of health state will result in better care of this chromosomal disease if we improve our knowledge of the involvement of specific hereditary components as well as the KS. The additional X chromosome (genotype XXY) in Klinefelter syndrome (KS) was identified instead of the usual male complement (genotype XY). KS is linked to several clinical problems, including hypogonadism and genetic disorders. Tall stature, small testes, late puberty gynecomastia, gynoid aspect of hips (broad hips), sparse body hair, signs of androgen deficiency and low serum testosterone coupled with elevated gonadotropins, and finally azoospermia, oligospermia, hyalinization, and fibrosis of the seminiferous tubules are all symptoms of KS.

Description

Both the supernumerary X chromosome and the effects of hypogonadism influence clinical characteristics. By comparing epidemiological data from prenatal diagnosis with data from men who were diagnosed after birth, it has been calculated that the prevalence of KS is greater than the number of individuals who had a clinical diagnosis. The large prevalence of mild phenotypes explains, at least in part, why most KS patients go undetected, and also justifies our efforts to improve our ability to diagnose patients quickly [1-3].

Because symptoms rarely appear at the same time, the condition is frequently neglected, and diagnosis is missed or delayed. Many instances are projected to go undiagnosed, with just 26% of the expected number of KS adults being appropriately diagnosed late in life, resulting in significant consequences and more difficult clinical management. The age of the patient influences the presentation of signs and symptoms. Furthermore, the phenotypic tends to deteriorate with age, due to the accumulation of characteristics and comorbidities that accompany aging, as well as the worsening of those that are already present. In men with KS, the onset of clinical signs and symptoms is dependent on their age. Despite substantial research, the pathophysiology, or the relationship between the extra X and the phenotypic, remains largely unknown. The parental origin of the X chromosome, gene-dosage effects in combination with (potentially skewed) X chromosome inactivation, and—particularly in spermatogenesis—meiotic failure may all play important roles. In females, one of the X chromosomes is inactivated to accomplish dosage adjustment, and KS is likely to be the same. However, genes from the pseudo autosomal areas and 15% of other genes are unaffected by X inactivation and are candidates for constituting the KS phenotype. The SHOX genes, for example, have been identified as possible causes of the tall height exhibited in KS.

KS is frequently linked to hypergonadotropic hypogonadism and infertility due to azoospermia, however the phenotypic varies greatly. The increased morbidity and mortality in KS highlight the need for a more widespread screening program and early diagnosis of a larger proportion of KS patients. Karyotyping of metaphase spreads from cultured peripheral blood cells remains the gold standard for diagnosing KS. The most significant advantage of karyotyping is the simultaneous examination of chromosome structure for translocations, inversions, and deletions. The KS's genetic foundation is based on chromosome non-disjunction, which results in the existence of an additional X chromosome or chromosomes. Non-disjunction occurs when chromosomes fail to separate at anaphase during meiosis I, II, or mitosis, resulting in cells with an abnormal number of chromosomes. This can happen during oogenesis or spermatogenesis (abnormal chromosomal or chromatid partitioning during maternal or paternal meiosis, respectively) or, less frequently (approximately 3%), during early fertilization division. Although evidence is inconclusive, the genesis of the supernumerary X chromosome has been linked to phenotypic differences. Patients with a paternal origin of the supernumerary X chromosome have a later onset and slower pubertal progression, according to studies. Other research, on the other hand, suggests that the parental origin of the extra X chromosome has no bearing on the patients' phenotype [4,5].

Conclusion

The transcription of one of the two X chromosomes is known to be inactivated at random in female somatic cells to achieve dosage-compensation of the X-encoded genes to that of male cells. Even though certain genes escape inactivation, the Barr body (chromatin) in female cells is evident and reflects the inactivated X chromosome. The coating and silencing of the extra X chromosome in human somatic cells is mediated by the untranslated RNA product of the X-inactive-specific transcript (Xist) gene, which is situated on the long arm of the inactive X chromosome. The existence of the second and any other supernumerary X chromosome in the somatic cell is thus indicated by the expression of Xist.

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