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Opinion Article

The Role of X-Chromosome Inactivation and its Implications for Disease Prevention

Dipak Bodhi*

Department of Genetic Medicine, JSS Medical College, Mysuru, Karnataka

DESCRIPTION

In mammalian female cells, X-Chromosome Inactivation (XCI) is a type of dosage compensation used to balance X-linked gene expression levels between the sexes. Because of inactivation escape and skewing, many diseases are linked to XCI, and the symptoms and severity of many diseases are heavily influenced by XCI status. There are three types of X-chromosome aneuploidy: X-linked disorders, diseases influenced by XCI escape, and X-chromosome aneuploidy. We examine representative disorders in terms of their definitions, symptoms, and the involvement of XCI in their development.

The XCI process has been linked to a variety of disorders. They can be divided into three groups: (1) X-linked gene diseases, whose severity is influenced considerably by the direction and degree of X-inactivation skewing; we review FD that falls under this category here. (2) X-Chromosome Dosage Effect: Systemic Lupus Erythematosus (SLE) has a higher incidence in females due to the existence of an extra pair of X chromosomes and the possibility of escaping gene expression from the Xi: Turner syndrome (TS, 45, X), triple X syndrome (47, XXX), and Klinefelter syndrome (47, XXY) are examples of X-chromosome aneuploidy. These diseases are discussed below briefly.

X-linked diseases

Clinical manifestations of practically all X-linked disorders are more severe in males than in females. In comparison to male carriers, female carriers are either asymptomatic or have milder characteristics. XCI is responsible for the gender disparities in certain X-linked disorders. Because most X-linked genes are hemizygous in males, a male carrier of a mutant allele is frequently affected with all of the disease's clinical characteristics. In terms of XCI status, females have two sorts of cells: those with active maternal X and those with active paternal X. Because a percentage of the female cells have the mutant X inactivated, even if a female carries one copy of the mutant allele, she is unlikely to have full clinical manifestation. According to OMIM data, there are over 500 X-linked disorders that are more severe in men. This includes X-linked retinitis pigmentosa, Duchenne muscular dystrophy, FD, and fragile X syndrome.

X-chromosome dosage effect diseases

X-inactivation escape is another form of illness that is influenced by XCI. Disease presentation and sex bias in such disorders may be linked to the escape and consequently overexpression of particular genes in female cells. Autoimmune diseases (SLE and autoimmune thyroid diseases, for example) and several psychiatric problems are among them (bipolar disorder and major depression). SLE is a chronic autoimmune illness that manifests differently depending on which primary organ is afflicted. The rash that patients commonly get on their skin is referred to as erythematosus. Globally, the incidence of SLE ranges from 2.2 to 23.1/100,000 person-years, with North America having the highest estimated incidence. SLE is more common in women, and African-Americans have a higher incidence and prevalence than Caucasians.

Aneuploidy of X-chromosome

Human disease phenotypes caused by X-chromosome aneuploidy include TS (45, X), Klinefelter syndrome (47, XXY), and triple X syndrome (47, XXX). Only one X chromosome remains active in both Klinefelter syndrome and triple X syndrome, and all extra pairs of X chromosomes are inactivated. Overexpression of escape genes is thought to cause the phenotypic abnormalities found in such disorders. Due to haploinsufficiency, the expression would be decreased in TS. Indeed, the XCI-resistant SHOX gene has been linked to tall stature in Klinefelter syndrome and triple X syndrome, as well as low stature in TS. However, with decreasing X-chromosome dosage, some X-linked genes show increased expression, suggesting a compensating mechanism in the complex interaction between X-chromosome dosage and X-linked gene expression level.

CONCLUSION

Inactivation of the X-chromosome is a physiological mechanism that balances the effects of gene dosage on the sex chromosomes. The phenotype observed in females bearing X-linked mutant genes or chromosome aberrations is affected by the presence of this natural process. As revealed in research in rodent models and cell lines, targeted reactivation of the normal allele on the Xi gene should be examined further to enable the development of further therapy options.

Correspondence to: Dipak Bodhi, Department of Genetic Medicine, JSS Medical College, Mysuru, Karnataka, E-mail: dranils3452@yahoo.com

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