American Journal Of Biomedical Science & Pharmaceutical Innovation

VOLUME Vol.05 Issue04 2025
PAGE NO. 19-20
DOI 10.37547/ajbspi/Volume05Issue04-04

Sexual Chromatin: Structure, Functions and Significance in Medical Practice

Qurbonov A. Q.
Department of Histology and Medical Biology, Tashkent Medical Academy, Uzbekistan

Yuldasheva Z.Z.
Department of Histology and Medical Biology, Tashkent Medical Academy, Uzbekistan

Received: 17 February 2025; **Accepted:** 15 March 2025; **Published:** 16 April 2025

Abstract: The concept of sex chromatin plays an important role in genetics, cytology and medical practice. Sex chromatin is a condensed inactive X chromosome in the nucleus of somatic cells in female mammals. This phenomenon is explained by a dosage compensation mechanism that ensures equal amounts of X-sex chromosome gene products in males and females. Sex chromatin is an important object of cytogenetic studies, allowing the diagnosis of disturbances in the number of X-sex chromosomes and the differential diagnosis of hereditary chromosomal diseases.

Keywords: Sex chromatin, Barr bodies, X-sex chromosome, genotype, trisomy, Klinefelter syndrome, Turner syndrome, amniotic fluid.

Introduction: Sexual chromatin was first described in 1949 by Canadian scientist Murray Barr and his colleague E. Bertram while studying the cells of the nervous tissue of cats. They found a dense colored body near the nuclear membrane, which was present only in females. Later, it was found that this formation corresponds to the inactivated X-sex chromosome. Further studies confirmed the presence of Barr bodies in other mammals and explained the mechanism of X-sexual inactivation.

Sex chromatin, a dense, staining body found in the non-dividing nuclei of cells in heterogeneous (having X and Y sex chromosomes) animals and humans. Sexual chromatin is divided into X-chromatin, or Barr's body, and Y-chromatin (discovered in 1970 by Swedish scientists T. Kaspersky and L. Tsekh). X-chromatin is a body that is intensely stained with basic dyes (0.7—1.2 microns), more often adjacent to the nuclear envelope and having a triangular semilunar or rounded shape. Y-chromatin is much smaller in size and is detected when the nucleus is stained with fluorochromes (akrikhin, akrikhiniprit) and examined in ultraviolet light. In female individuals (type XX), one of the X chromosomes is inactive, which is manifested in its stronger spiralization and densification. In the interphase nucleus, this spiralized X chromosome is visible as X-chromatin. Y-chromatin in humans and some primates has a large heterochromatin region that produces intense fluorescence. Thus, a technically simple study of the interphase nucleus makes it possible to judge the state of the sex chromosome system. X-chromatin is more or less often found in women in the nuclei of cells of all tissues (for example, in the epithelial cells of the oral mucosa in 15-60% of the nuclei). The number of nuclei with X-chromatin depends on the intensity of cell reproduction in a given tissue and on the hormonal state of the body. A change in the amount of sex chromatin indicates a change in the number of sex chromosomes, which is detected by the cytogenetic method of human genetics.

The nature and formation of sex chromatin was first explained by the English geneticist Mary Lyon in 1961. When comparing the female (XX) and male (XY) sex chromosomes, it was found that the genes in the X chromosome can show all the signs on their own without forming XX. For example, men (XY) have one X chromosome, but it alone carries all the information.

Based on this, Lyon explained the formation of sex chromatin. In women, one of the X-sex chromosomes is inactive, it is formed at the stage of the interphase of DNA division from an inactive strongly twisted and spiralized chromosome, and it also forms a small body clearly visible under the microscope. And the second X chromosome is active, it carries genes that manifest their functions, and the DNA is in an unspiralized state, in the form of a thin long strand. For this reason, this chromosome is not visible under a microscope.

The transition to the inactive state of one of the X chromosomes in the female body is observed randomly in the early embryonic period. However, the property of DNA division in an inactive chromosome remains. In the female body, one X chromosome is derived from the father, the other from the mother. The manifestation of symptoms depends on the transition of one of the sex chromosomes to an inactive state. If the chromosome received from the mother contains dominant genes, but becomes inactive, then the chromosome received from the father with recessive features will manifest itself.

Since the inactivation of the X-sex chromosome occurs randomly, women are mosaics of X-linked genes. This leads to unique biological features. The coat color of tortoiseshell cats is a good example of mosaic gene expression. Mosaicism in the case of X-linked diseases – some cells may be healthy, while others may be mutant, which mitigates the manifestations of diseases such as hemophilia or Duchenne syndrome in humans.

A method for determining sex chromatin in medicine:

1. In the diagnosis of hereditary chromosomal diseases. If no sex chromatin is detected in a woman's body, it is revealed that she has one X-sex chromosome. This genotype is observed in women suffering from Shereshevsky-Turner disease. If there is one extra sex chromatin, then the woman has an excess of the X chromosome, i.e. a trisomy in the genotype. Usually, men do not have sex chromatin, but people suffering from Klinefelter's disease have excess sex chromatin.

2. In obstetric and gynecological practice, this method is used to determine the sex of the fetus, where the test material is amniotic fluid, into which fetal cellular substances are secreted. Also, this research method makes it possible to find out the probability of having a hemophiliac if the family already has a son suffering from hemophilia. If there is sexual chromatin in the amniotic fluid, the sex of the fetus is a girl and she will be born healthy. If sexual chromatin is not observed, then the sex of the child is a boy and the probability of hemophilia is 50%.

3. In pediatric practice, in some cases children are born bisexual, i.e. hermaphrodites. According to the presence or absence of sexual chromatin, the child's proper sex is determined and various measures are taken to form a natural medical gender.

4. In transplantation.

5. In the forensic medical examination. Using the method of determining sex chromatin, it is possible to determine the sex of a person.

CONCLUSION

Sex chromatin is an important object of study in the field of human genetics and medicine, since the method of determining sex chromatin allows us to obtain valuable data on sex chromosomes and their behavior in cells. The use of sex chromatin research methods is widely used in the diagnosis of various human chromosomal diseases, as well as in various areas of medical practice.

REFERENCES

К.Н. Нишанбаев, П. Р. Алимходжаева, Д. Х. Хамидов, «Медицинская биология и генетика», 2008г., Государственное Научное издательство «Узбекистон миллий энциклопедияси» Ташкент;

К.Н. Нишанбаев, П.Р. Алимходжаева, Л.И. Шамансурова, «Медицинская биология и генетика», 2009г., Государственное Научное издательство «Узбекистон миллий энциклопедияси» Ташкент;

Гинтер, Е. К., «Медицинская генетика», 2009г., СПб.: Издательство СПбГУ;

Жуков, Б. Н., & Иванова, Л. Н., «Основы цитогенетики человека», 2010г., М.: Наука;

В. Н. Ярыгин, «Биология» учебник, под редакцией академика РАМН профессора, 2018г, том 1,2; издательство ГЭОТАР – Медиа, Москва;

Халиков П.Х., Курбанов А.К., Даминов А.О., «Медицинская биология и генетика», 2023г.