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Naser Kamberi

University for Business and Technology, naser.kamberi@ubt-uni.net

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Genealogical Analysis of Bar's Body Activity in Hemophilia at Two Brothers and a Cousin

Naser Kamberi¹

¹UBT - Higher Education Institution, Lagjja KALABRIA p.n., Prishtinë, Kosovë
10000 Prishtina, Kosovo
naser.kamberi@ubt-uni.net

Abstract: In this study it is included a sample of the Presevo population and villages of 3771 individuals. During family interviews, we encountered cases in women with hemophilia and men with hemophiliacs. Predicting that X-sex chromosome is inactive we did not have the case that females are hemophilic. By doing the genealogical analysis of these families, we will present the thoughts about the role of Bari's body when it comes to hemophilia.

The genealogical method can be used especially in cases of monogenic inheritance. Considering that if the X chromosome is inactive in women then the dose of the chromosome is equal to the male sex. For this reason, likely to get sick of hemophilia would be equal, but in reality this does not stand. So with this study we will give our thoughts about the Bar's body or the gender heterochromatin regarding the dilemma of this case.

Keywords: Kromosom X, Barr body, hamophyli, genealogy.

INTERDUCTION

Bari and Bertram in 1949 observed that X chromosome inactivated at embryo stage at implant time at the end of the first week of pregnancy. This chromosome is accumulated in a cluster in which the genes in it are thought to be inactive and this chromosome is called Bari's body.

With this paper, the effect of "sex inactivated" X chromosome in the cases of individuals with related diseases for this X chromosome is studied. Based on the cases we are going to present; will we clarify that chromosome as the body of Bari has an impact on inheritance or has no influence? We present this phenomenon in the dilemma because only males are sick with hemophilia, while women are not based on data obtained from the four-generation population-generated genealogy study in which hemophilic cases appear.

MATERIAL AND METHOD

For this work as a part of the PhD thesis, the material was taken from the human population in the Presevo municipality where 3771 individuals are involved, including 1727 individuals from Presevo and 2044 individual villages. During the interview method for haemophilists we have had cases of sick boys and women carrying haemophilia but not diseased. To accumulate the material, we combine the method of interview with the method of coding genealogical digit. Using genealogy coding we have been able to convey inherited diseases like hemophilia and other cases. We have assembled the material gathered in the form of a questionnaire in different families without making any selections of them. And all of the cases have been marked in the tables prepared at the excel computer.

Also in the drawings we made genealogy in cases where we encountered hemophilia disease and then genealogy was worked with the genealogy program prepared by S.Vezi-Tirana specializing in genealogical programming. During interviews we can find cases in other villages where they were married by families with hemophilia. But in all cases investigating we did not encounter sick women. Drawing on all these occasions we will give some thoughts about the activity of the burial of Bari

RESULTS AND DISCUSSIONS

The material received from the interviews in three settlements we have managed to include 97 families.

By tracking the visibility of the traits across the generations we have come to observe the cases of families with hemophilia. These cases were present in 7 families but we analyzed in detail three of them. Based on the analysis we found that hemophilia was evident in these families.

From this study we can emphasize that women though based on scientific thoughts have a sexual chromosome X and the other is inactive inactive, are not hemophilic like males and this brings us to a doubt about the activity of the bourgeois Bari. But analyzing the cases we did not have any sick women even though they are on the same dose of X chromosome as men.

From this it is understood that the "inactive" chromosome Heterocyclic or Barr body only in morphological view differs from other sexous chromosome having the form (X), if it is functional and with its own genetic radiation contributes to enzymatic processes and hence inhibits enzyme pathogens in female sex. This is confirmed by the fact that if females find hemophilic occur in both sex chromosomes X women will be sick. Therefore, there is no dilemma that heterochromatin or body barr is partially active but only in shape varies and captures the appearance of a nucleus in the nucleus.

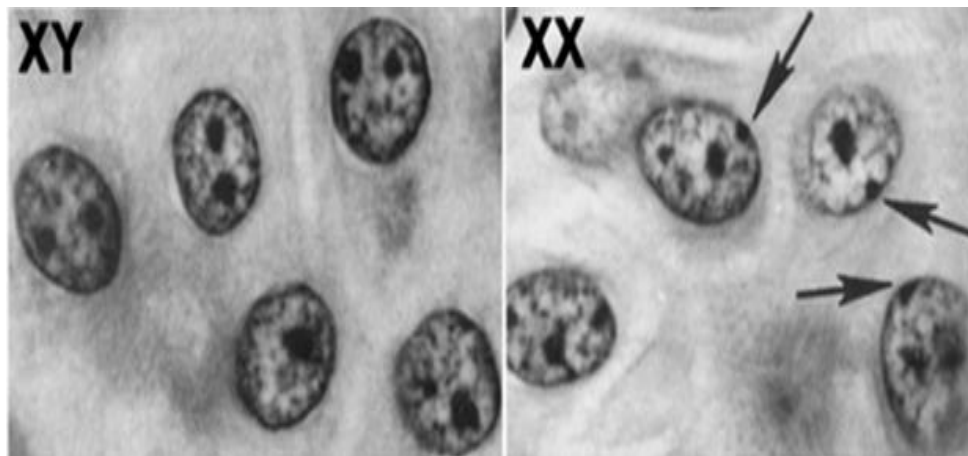


Fig.1. Heterochromatina

In this family we had a case of hemophilia where the individual number III8 of the female gender is the carrier of hemophilia, but not sick. So it is not possible to say that the mutation of this disease has occurred in the body of Bari and not in the sex X chromosome. Also the same case is in the IV10 generation where we have sick hemophilic (dead) son and two sisters in generation IV14 and IV15 as haemophilic carriers, the subconscious that finds mutation is found in the body of Bari or in the X chromosome. From female IV14 we have a boy with hemophilia

Three cases with hemophilia Biochemical and genetic results	The first individual (first brother)	The second person (second brother)	The third person, the cousin of the two brothers
Type of hemophilia	Hemophilia type A	Hemophilia type A	Hemophilia type A
Blood group	A, Rh negative	A, Rh negative	A, Rh negative
Factor VIII level	1.7% (Preferably 50-150%)	1.5% (Preferably 50-150%)	2% (Preferably 50-150%)
Factor I level	338,5%	338%	339%
Factor IX level	91,5%	91%	91%
Factor XI level	82.8%	83%	83%
Fibrinogen	4.01%	4%	4%
Genotype	ccddee	ccddee	ccddee
Intron 22	Region Xq28	Region Xq28	Region Xq28

Therefore, we can conclude that the inheritance of these factors is done with a high and independent genetic stability without any factor VIII interference resulting from mutation in intron 22 in Xq28 region of chromosome X. So we conclude that the other factors analyzed in this study have not changed from the mutation of the gene that induces hemophilia.

In these cases, we can say that the mutation of the hemophilic gene does not represent any correlation with factors I, IX, XI and fibrinogen, while there are signs with the blood group and the rhesus factor, therefore a genealogical and molecular research should be performed.

Conclusions

1. By analyzing the genealogy tree or the genetic tree it is clearly seen that only masculine is affected by hemophilia, while the female gender is a carrier, but heterozygous is not ill. By this we certify that the so-called inactive chromosome Heterochromatin or the body of Bari is not completely inactive, but in these cases it is self-evident that it is functional forming blood clotting enzymes.
2. Regardless of who and what happens X inactivation, however, activity in the formation of the encoding enzyme is active. So it just seems to be ineffective but in reality the genius works when it comes to the disease. The hemophilic source is female with a number 8 strain of generation III that has undergone intron mutation 22 in Xq28 region of chromosome X.
3. Analyzing genealogy of families, we can conclude that it comes to a new mutation for hemophilia.
4. Factor VIII level of action in three cases is very low: 1.7%; 1.5% and 2% compared to the preferred value, (50-150%).
5. Also Bari's body may be partially inactive, but it is completely safe.
6. In the early stage, at the end of the first week of pregnancy, before implantation of the embryo in the uterus occurs the morphological change, the inactivation of the X chromosome originating from the sperm, and this proves that their father was not hemophilic and the body of Bari is non-carrier of hemophilia in these three cases, but the mother was carriers but not sick.
7. As a solution to this problem, evolution has determined that women always modify an X chromosome, because the space in the nucleus is not inactive completely. And this is accomplished precisely by DNA methylation in histone on a X chromosome of the father who is subject to the egg cell's egg-like factor because it does not find fit in another cell that has a completely different opposite to where it came from. And due to the action of the cytoplasm

and cellular enzymes of the chromosome X egg cell from the father undergoes a condensation in the body of Bari.

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