Heterozygote frequencies of common polymorphic markers of factor VIII (*F8*) and factor IX (*F9*) genes in indigenous Nepali population

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OBJECTIVES

To determine heterozygote frequencies of common polymorphic markers of factor VIII (*F8*) and factor IX (*F9*) genes in indigenous Nepali population with an objective of application of the generated data for genetic diagnosis of affected families

METHODS

83 unrelated subjects (46 males and 37 females ; 120 X chromosomes) were included in the present study

Polymorphic markers studied:

***Factor VIII gene* -Bcl1, HindIII, Bsl1**

***Factor IX gene*- Dde1, Hha1, Mse1**

Table 1. Heterozygote frequencies of the polymorphic markers of *f 8* and *f 9* genes in Nepali population

Marker	Allele frequency +	Allele frequency -	Heterozygosity frequency
***F8* gene**			
Bcl1	0.63	0.37	0.38
Hind III	0.34	0.66	0.38
Bsl1	0.28	0.72	0.41
Bcl1 + Hind III	-	-	0.49
Bcl1+HindIII+Bsl1	-	-	0.54
***F9* gene**			
Dde1*	0.09	0.91	0.15
Hha1	0.19	0.81	0.41
Mse1	1.00	0	0
Hha1+Dde1	-	-	0.53
Hha1+Dde1+Mse1	-	-	0.53

RESULTS

Cumulative heterozygotic frequency of Bcl1, Hind III and Bsl1 -54%

Mse1 polymorphism of *F9* gene showed 0% heterozygotic frequency

Ddel and Hhal showed a cumulative heterozygosity of 53%.

CONCLUSIONS

•Nepali population shows considerable deviation from most of the other populations as far as the heterozygosity of some of the markers in *f8* and *f9* genes are concerned.

•MseI marker of *f9* gene may not be used in Nepali population.

• Other RFLP markers like VNTR at intron 7, intron 13 and intron 22 of *F8* gene need to be studied and Taq1, Xmn1, Sal1 , Mnl1

• Using 3 polymorphic markers of *F8* and *F9* genes, more than half of the families can be offered genetic diagnosis in Nepal using the simple PCR-RFLP technique.

Acknowledgements

Authors are grateful to World Federation of Haemophilia (WFH) for providing IHTC fellowship to one of us (BSP). This study is a fruitful culmination of cooperative work between National Institute of Immunohaematology and Haemophilia centre at Nepal

REFERENCES

Furer V, Samuels J. End-stage hemophilic arthropathy in a patient from a developing nation.*J Clin Rheumato*. 2010 ;16:303.

Rijal L, Neogi DS, Ansari MT, Khan SA, Yadav CS. Hemophilic pseudotumor--is there a role of radiotherapy? Literature review and a case report. *Nepal Med Coll J* 2010 ;12:193-7

Shetty S, Ghosh K, Pathare A, Colah R, Badakare S, Mohanty D. Factor VIII and IX gene polymorphisms and carrier analysis in Indian population. *Am J Hematol* 1997 ;54:271-5.

Peake IR, Lillicrap DP, Boulyjenkov V, Briet E, Chan V, Ginter EK, Kraus EM, Ljung R, Mannucci PM, Nicolaides K, et al.Haemophilia: strategies for carrier detection and prenatal diagnosis. *Bull World Health Organ* 1993;71:429-58

Mukundan P, Shetty S, Kulkarni B, Ghosh K. Utility of an exon 14 BsII polymorphism for improved genetic diagnosis of hemophilia A in Indian population. *Prenat Diagn* 2008 ; 28 : 920-2.

DOI: 10.3252/pso.eu.WFH2012.2012