

Transferrin Subtypes in Haryana Determined by Agarose Gel Isoelectric Focusing

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ABSTRACT Distribution of transferrin subtypes in Haryana, a north-west Indian state, based on isoelectric focusing analysis has been reported. In the present investigation all the common subtypes were detected excepting TFC3. The allele frequencies ($TF^*C1 = 0.757$, $TF^*C2 = 0.155$, $TF^*C3 = 0.088$) are in genetic equilibrium and within the frequency ranges reported from other parts of the country.

INTRODUCTION

The transferrin (TF), an iron-binding β -globulin fraction in human serum was discovered by Schade and Caroline (1946). Electrophoretic studies of human transferrin categorised it into three main types based on the mobility of the bands. The most common type was designated as TFC, while the other two were called TFB and TFD (Smithies, 1957; Giblett et al., 1959 and Sutton et al., 1960). The allelic frequency of TFC has been found greater than 98% in most human populations (Dykes et al., 1982). With isoelectric focusing technique TFC has been resolved into six common subtypes : TFC1, C2, C2-1, C3, C3-1 and C3-2 controlled by three common alleles TF^*C1 , TF^*C2 and TF^*C3 (Kuhnl and Spielman, 1978, 1979 and Kuhnl et al., 1979). In addition, several subtypes due to rare alleles TF^*C4 , TF^*C5 , TF^*C6 , TF^*C7 , TF^*C8 , TF^*C9 and TF^*C10 have also been reported in some populations (Constans et al., 1980; Kuhnl et al., 1981; Dykes et al., 1982 and Weidinger et al., 1984).

Although, the TF subtypes have been extensively studied in various human populations of the world, from India only limited studies have been reported on some tribal and non-tribal

populations from Jammu and Kashmir, Himachal Pradesh, Delhi, Gujarat, Assam, Manipur, Sikkim, West Bengal, Karnataka (Bhasin et al., 1992, 1994), and infact no such data are available from Haryana, a north-west Indian state located between 27.5° and 31° northern altitudes and between 74.5° and 77.5° eastern longitude. Hence, as part of our ongoing serogenetic studies in Haryana aimed at generating allele frequencies of various genetic markers for use in forensic analysis, this report presents the baseline data on the polymorphism of TF subtypes and the corresponding allele frequencies in the people of Haryana.

MATERIAL AND METHODS

Plasma samples from 74 apparently healthy and unrelated individuals belonging to various Hindu caste groups of Haryana state were analysed for TF subtypes on 0.5mm agarose gel using IEF technique as described by Dykes (1985). Serum samples of known phenotype TFC1, C2, C2-1, C3-1 and C3-2 were used as controls.

RESULTS AND DISCUSSION

The observed and expected number of TF subtypes and the corresponding allele frequencies are presented in table 1. The observed number of phenotypes are in close agreement with those expected assuming Hardy-Weinberg equilibrium $\chi^2_3=5.43$; $0.20 > p > 0.10$). No rare variant was found and only three TFC alleles occur in the following order : $TF^*C1 > TF^*C2 > TF^*C3$. The allele frequencies ($TF^*C1 = 0.757$, $TF^*C2 = 0.155$ and $TF^*C3 = 0.088$) es-

Table 1: Transferrin subtypes and allele frequencies in Haryana

| Phenotype | Observed number | Expected number | Allele frequencies |
|-----------|-----------------|-----------------|--------------------|
| C 1 | 45 (60.81%) | 42.41 | $TF^*C1=0.757$ |
| C 2 | 4 (5.41%) | 1.78 | $TF^*C2=0.155$ |
| C 2-1 | 12 (16.22%) | 17.37 | $TF^*C3=0.088$ |
| C 3 | 0 (0.00%) | 0.57 | |
| C 3-1 | 10 (13.51%) | 9.86 | |
| C 3-2 | 3 (4.05%) | 2.02 | |

timated from present sample from Haryana are well within the allele frequency ranges ($TF^*C1 = 53 - 81\%$, $TF^*C2 = 17 - 40\%$ and $TF^*C3=0.0 - 3.3\%$) reported from other regions of Indian subcontinent (Bhasin et al., 1992, 1994). TF subtyping has enhanced the discrimination potential and probability of excluding a man falsely accused of paternity to 40% and 60%, respectively.

REFERENCES

- Bhasin, M.K., Walter, H. and Danker-Hopfe, H. : *The Distribution of Genetical Morphological and Behavioural Traits Among the People of the Indian Region*. Kamla-Raj Enterprises, Delhi (1992).
- Bhasin, M.K., Walter, H. and Danker-Hopfe, H. : *People of India - An Investigation of Biological Variability in Ecological, Ethno-Economic and Linguistic Groups*. Kamla-Raj Enterprises, Delhi (1994).
- Constans, J., Kuhl, P., Viau, M. and Spielmann, W. : A new procedure for the determination of transferrin C(TfC) subtypes by isoelectric focusing. Existence of two additional allele, *TfC4* and *TfC5*. *Hum. Genet.*, **55** : 111-114 (1980).
- Dykes, D.D. : Detection of red cell enzymes and serum protein polymorphism in bloodstains. In : *Serological Methods in Forensic Science*. Susan D. Rolih and W. John Judd (Eds.). AABB Arlington (1985).
- Dykes, D.D., DE Furio, C.M. and Polesky, H.F. : Transferrin (Tf) subtypes in US Amerindians, Whites and Blacks using thin-layer agarose gels : Report on a new variant TfB. *Electrophoresis*, **3** : 162 (1982).
- Giblett, E.R., Hickman, C.G. and Smithies, O. : Serum transferrin. *Nature*, **183** : 1589-1590 (1959).
- Kuhl, P. and Spielmann, W. : Transferrin : evidence for two common subtypes of TfC allele. *Hum. Genet.*, **43** : 91-95 (1978).
- Kuhl, P. and Spielmann, W. : A third common allele in the transferrin system, TfC3 detected by isoelectric focusing. *Hum. Genet.*, **50** : 193-98 (1979).
- Kuhl, P., Spielmann, W. and Constans, J. : Identification of serum transferrin c allele subtypes by isoelectric focusing. *9 Int. Congr. Soc. Forens. Haemogenet. Bern* (1981).
- Kuhl, P., Spielmann, W. and Weber, W. : Isoelectric focusing of the rare transferrin (Tf) variants and common TfC subtypes. *Hum. Genet.*, **46** : 83-87 (1979).
- Schade, A.L. and Caroline, L. : An iron binding component in human blood plasma. *Science*, **104** : 340-341 (1946).
- Smithies, O. : Variations in human serum β -globins. *Nature*, **189** : 1482-1483 (1957).
- Sutton, H.E., Matson, G.A. Robinson, A.R. Koucky, R.W. : Distribution of haptoglobin, transferrin and haemoglobin types among Indians of southern Mexico and Guatemala. *Am. J. Hum. Genet.*, **12** : 338-348 (1960).
- Thymann, M. : Identification of a new serum protein polymorphism as transferrin. *Hum. Genet.*, **43** : 225-229 (1978).
- Weidinger, S., Clave, H., Schwarzfischer, Postal, W., Weser, J. and Gorg, A. : Transferrin subtypes and variants in Germany; further evidence for a Tf null allele. *Hum. Genet.*, **66** : 356 (1984).