# Null and Rare Alleles in Paternity Testing

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## INTRODUCTION

The iron-binding protein of human plasma transferrin (Tf) was discovered in 1946 by Schade and Caroline and the polymorphism of human red esterase D was first demonstrated by Hopkinson et al. (1973). But only recently was an extensive genetic polymorphism revealed due to the high resolving power of protein separation by isoelectric focusing (IEF).

The purpose of this work is to present the evidence of rare alleles in Tf and EsD systems in the population of North of Portugal.
An apparent maternity exclusion found in the Duffy system due to a silent allele will also be present. The frequency of this allele in a large number of trios tested in Medico Legal Institute of Oporto because of disputed paternity was determined.

## MATERIALS and METHODS

Blood samples were obtained by venopuncture from healthy individuals involved in paternity cases in the North of Portugal.
Phenotyping of Tf and EsD was performed by IEF using polyacrilamide gels with pH 5-8 Ampholines and with Pharmalyte carrier ampholytes pH 4.5-5.4, respectively.
Duffy typing was done using indirect Coombs test.

## RESULTS and CONCLUSION

The analyse of 873 blood samples of unrelated persons typed for Tf showed the common C subtypes C1, C2, and C3 as well as the rare C6 and B variants. The frequencies of those alleles are: C1 = 0.7697, C2 = 0.1723, C3 = 0.0532, C6 = 0.0011, B = 0.0034.

EsD was typed in 502 blood samples. The following frequencies were observed: EsD1 = 0.8256, EsD2 = 0.1623, EsD5 = 0.0079, EsD7 = 0.0039.

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Of interest was the observation of a mother child pair with apparent maternity exclusion in the Duffy system. However, the analysis of 20 markers indicated a very high probability for maternity, so that the presence of a "null" allele was assumed in the child and mother. The allele frequencies obtained after typing 992 blood samples are: Fya = 0.3462, Fyb = 0.6486, Fy = 0.0050.

The results of this study underline the usefulness of these systems for population studies and paternity testing. IEF with immobilized pH gradients is a supplementary method for identification and delineation of newly observed Tf and EsD subtypes.

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