

Further evidence of a silent Tf allele

K.Püschel, A.Krüger, R.Söder-Bräunlich

Institut für Rechtsmedizin der Universität Hamburg, Butenfeld 34
D-2000 Hamburg 54

Since the detection of transferrin polymorphism by Smithies in the year 1957 some cases of inherited α transferrinemia have been described in the clinical literature (Čáp et al.1968; Sakata et al.1969; Heilmeyer et al.1981; Goya et al.1972). However, only 3 well documented cases of an apparently "silent" Tf allele have been reported so far (Polesky et al.1983; Weidinger et al.1984; Lukka et al.1985).

Recently, in a criminal case of father-daughter sexual abuse (incest) in which a friend of the mother was originally involved only as a witness we observed a further Tf null allele.

24 blood-group systems (including HLA) were investigated. The paternity of the defendant was excluded in four different systems. An apparent incompatibility in the Tf system was noted between the child and the witness: the child was originally classified as Tf C2, the witness as Tf C1. In 23 other systems the witness could not be excluded as the father of this child.

The calculated plausibility of paternity for the witness (putative father) was $W=99,9965\%$ (Tf system was omitted from the calculation). The high W-value ("paternity practically proved") strongly suggested the existence of a "silent" Tf allele in the child and the putative father.

Quantitative determinations of the serum transferrin were performed employing single radial-immunodiffusion (NOR-Partigen-Transferrin, Behring). In the sera of the child and the putative father less than 50% of the normal transferrin concentration was found.

The child had 150 mg/100 ml, the putative father 160 mg/100 ml transferrin, respectively. We assumed that both are heterozygous for Tf 0 allele (see table 1).

We have been able to investigate sera of some other members of the witness' family and we found that the mother of the witness and her sister are heterozygous for Tf 0 allele, too.

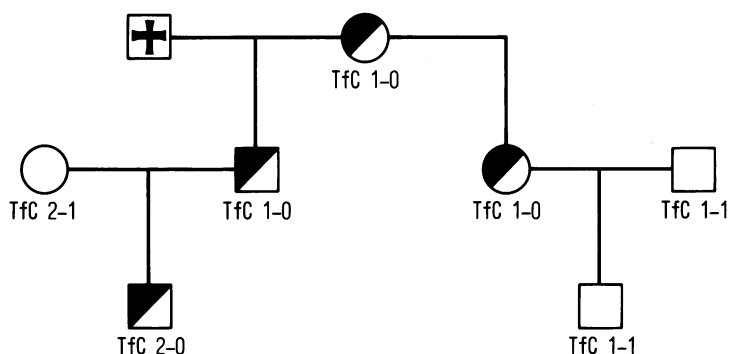
The quantitative determinations of the serum transferrin concentrations of the four examples of Tf 0 allele and the mean value of 3 normal Tf phenotypes (TfC 1-1, 2-1, 2-2) are given in table 1. The pedigree of the family is given in figure 1.

Table 1: Serum transferrin concentrations

Mean value*	573 mg/100 ml	100%
Child, Tf C2-0	150 mg/100 ml	26%
Witness, Tf C1-0	160 mg/100 ml	28%
Witness' mother, Tf C1-0	170 mg/100 ml	30%
Witness' aunt, Tf C1-0	230 mg/100 ml	40%

* N=45, SD=61,4 mg/100 ml; Range: 458-688 mg/100 ml.

Figure 1: Pedigree of the family



Literature:

- Čáp J, Lehotská V, Mayerová A (1968) Kongenitálna atranferrinémia u 11-mesačného dieťaťa (in Czecho-Slovakian). *Česk Pediat* 23: 1020-1021
- Goya N, Miyazaki S, Kodate S, Ushio B (1972) A family of congenital atranferrinemia. *Blood* 40: 239-245
- Heilmeyer L, Keller W, Vivell O, Keiderling W, Betke K, Wöhler F, Schultze HE (1961) Kongenitale Atranferrinämie bei einem sieben Jahre alten Kind. *Dtsch med Wschr* 37: 1745-1751
- Lukka M, Ehnholm C (1985) A silent transferrin allele in a Finnish family. *Hum Hered* 35: 157-160
- Polesky HF, Souhrada JM, Dykes DD (1983) The frequency of "null" genes calculated from trios in disputed parentage cases. *Proceedings: 10th International Congress of the Society for Forensic Haemogenetics, München 11.-15.10.1983: 161-166*
- Sakata T (1969) A case of congenital atranferrinemia, cit. by Goya et al. 1972
- Weidinger S, Cleve H, Schwarzfischer F, Postel W, Weser J, Görg A, (1984) Transferrin subtypes and variants in Germany: Further evidence for a Tf null allele. *Hum Genet* 66: 356-360