THE Y-LINKED LOCUS Y27H39 (DYS19), FREQUENCY DISTRIBUTION IN SOUTH BAVARIAN AND APPLICATION TO PATERNITY TESTING

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Introduction

The short tandem repeat (STR) Y27H39 was discovered by Roewer et al. 1992 on the short arm of the Y-chromosome in 1992. It is based upon a (GATA)n repeat. Due to their heredity along male lines, Y-linked polymorphisms have a considerable potential in paternity expertises, especially in deficiency cases (Santos et al. 1993, Gomolka et al. 1994). We present a genetic population study based upon the analysis of a random sample of non-related men from southern Bavaria. The performance of the Y-linked polymorphism is demonstrated by means of a paternity case in which the two putative fathers were not available.

Materials and Methods

DNA-Preparation: DNA was isolated from whole blood samples using phenol/chloroform extractions (Manniatis et al. 1989). The amount of DNA was determined by comparison of the ultraviolet fluorescence of an aliquot of each sample with known quantities of lambda DNA.
RFLP-Analysis: 3-4 µg DNA were digested with HinfI and separated by electrophoresis in 0,8% garose gels. Single locus probes were provided labeled with the NICE system (ZENECA). Southern transfer and hybridisation were done by standard methods (Manniatis et al. 1989).
PCR-Analysis: PCR reactions were carried out using 1-5 ng of genomic DNA in a 50 µl reaction volume. Reaction mixtures consisted of 10 mM Tris(HCl) pH 8,3; 50 mM KCl; 1,5 mM $MgCl_2$; 200 µM dNTPs; 2,5 U Taq polymerase (Perkin Elmer, USA); 0,20 µM of each Primer. Primer sequences: HumTHO1 (TC11, Edwards et al. 1991), HumVWA31/A (VWA, Kimpton et al. 1992), D21S11 (Sharma et al. 1991), ACTBP2 (SE33, Polymeropoulos et al. 1992), Y27H29 (Roewer et al. 1992). Amplification was carried out a GeneAmp PCR System 9600 from Perkin Elmer Cetus. Temperature cycling conditions were as follows: 30 cycles of 95 °C for 20s, 58 °C (THO1, VWA, SE33, D21S11) or 56 °C (Y27H29) for 30s, 72 °C for 20s followed by a final 7 min incubation at 72°C.

RESULTS AND DISCUSSION

Five different alleles were obtained from a sample of 156 southern Bavarian non-related men. The frequencies of the respective alleles: allele A (186 bp) 16%, allele B (190 bp) 45%, allele C (194 bp) 17%, allele D (198 bp) 19% and allele E (202 bp) 3%. Upon examination of 35 proven father-son-

exclusions, exclusive constellations existed in 28 cases (80%) when employing the Y27H39-polymorphism.

Figure 1:

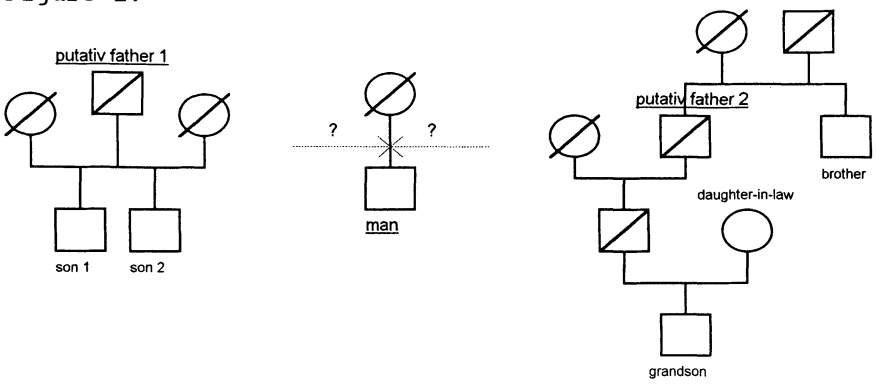

Table 1: Results of a deficienciy case.
Which putative father was the father of the "man"?

Probes	man	son 1	son 2
		putative father 1	putative father 1
MS43a	8,0 / 9,1	5,7 / 9,1	5,1 / 5,7
MS31	6,4	6,4 / 7,0	6,6 / 7,0
MS8	2,3 / 6,8	2,8 / 5,2	2,8 / 4,8
MS1	2,1 / 9,5	4,9 / 8,3	4,6 / 6,7
G3	3,4 / 7,1	7,1 / 2,1	5,0 / 2,1
Y27H39	4	2	2

Probes	man	brother putative father 2	grandson putativ father 2	daughter-in-law putative father 2
MS43a	8,0 / 9,1	9,1 / 5,9	9,6 / 8,2	8,2 / 8,9
MS31	6,4	6,0 / 6,4	8,6 / 7,0	7,0 / 5,8
MS1	2,1 / 9,5	10,2 / 12,4	2,8 / 2,4	2,4 / 5,5
G3	3,4 / 7,0	6,4	7,0 / 9,6	9,6
VWA	16 / 16	17 / 17	18 / 17	17 / 16
THO1	6 / 7	9 / 9,3	9 / 9	9 / 9,3
SE33 [a]	3 / 18	3 / 18	3 / 11	11 / 18
D21S11[a]	9 / 13,2	3 / 13,2	13,2 / 8	8 / 10
Y27H39	4	4	4	

a: Nomenclature like Kratzer A. et al. 1994.

Figure 1 shows the family tree of a deficiency case in which both supposed putative fathers were no longer available for paternity examinations. Two sons, half-brothers of another, were included on behalf of the first deceased putative father. The second putative father was substituted by a brother, a grandson and a daughter-in-law. Results of examinations centring on 4 single locus systems, 4 STR´s and the Y27H39 locus of the persons involved are summarised in table 1.
Constellations of exclusion in regard to putative father 1 could be shown in the Y27H39 and MS1 systems. It was assumed that the men A and B (fig. 1) factually are half brothers.
No exclusive constellation was found in regard to putative father 2. A paternity probability of W = 97,37% was calculated by M.P. Baur, Bonn, Germany.

SUMMARY

The allele distribution in the southern Bavarian population as seen by us as well as the paternity-related data emphasise the potential of the tetranucleotide repeats Y27H39, especially in regard to deficiency cases. In addition, this system lends itself to determination of sex as well as forensic trace analysis.

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