

# Evaluation of 12 single-copy and 2 multi-copy Y-chromosomal STR loci in five German populations

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**Abstract.** The European Minimal Haplotype loci and six further Y-chromosomal loci (one multi-copy) were evaluated with regard to their gene diversity ( $D$ ) in five German population groups. The newly established ranking list showed DYS464 ( $D=0.7733-0.9446$ ), DYS449 ( $D=0.7844-0.8511$ ), and DYS385 ( $D=0.7511-0.8433$ ) on the top. The need for further loci became obvious in one forensic case where the discrimination of two haplotypes was only successfully with DYS446. © 2005 Elsevier B.V. All rights reserved.

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## 1. Introduction

The analysis of Y-chromosomal Short Tandem Repeats is a powerful tool for analysing mixed forensic stains as well as for paternity testing. It was shown that the analysis of a core set of Y-STRs, the European Minimal Haplotype standard (DYS19, DYS385-I and -II, DYS389-I and -II, DYS390, DYS391, DYS392, DYS393), can discriminate between most of the male individuals in a population. The haplotype diversity using the European Minimal Haplotype loci reaches between  $<0.95$  for Albania or Finland and 1.00 for Vienna [1]. This indicates that there is a significant portion of haplotypes in several populations which cannot be resolved. Therefore, the European Minimal Haplotype loci and a set of further Y-chromosomal STR loci [2] were evaluated in five German population groups. Haplotype diversities were calculated for analyzing the European Minimal Haplotype standard and for analyzing at least 12 single-copy and 2 multi-copy Y-chromosomal STR loci.

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## 2. Materials and methods

Blood samples and buccal swab samples from 235 healthy unrelated male German volunteers from Dresden ( $n=89$ ), Rostock ( $n=38$ ), Hamburg ( $n=49$ ), Munich ( $n=30$ ), and the ethnic group Sorbs ( $n=29$ ) were collected for DNA extraction with written informed consent. In a forensic case, DNA was extracted from fingernails (piece of evidence), blood (victim) or buccal swabs (suspect person).

Two multiplex PCR systems were set up and used for the population study:

- Mentype® Argus Y-MH PCR Amplification Kit (Biotype AG, Dresden) — European Minimal Haplotype standard
- Screening multiplex system RAR — DYS446, DYS447, DYS448, DYS449, DYS463, DYS464 (multi-copy locus), DYS385 for overlap.

PCR amplification for the population study was performed in accordance to the manual of Mentype® Argus Y-MH PCR Amplification Kit. DNA of the forensic case was amplified with the PowerPlex® Y System (Promega) and with the screening multiplex system RAR. The PCR products were analyzed by capillary electrophoresis using an ABI PRISM 310 Genetic Analyzer. The alleles were typed automatically using the Genotyper® software and the appropriate allelic ladders. Allele frequency ( $p_i$ ) was estimated by gene counting  $p_i = n_i/N$ , the observed gene diversity ( $D$ ) was computed using the formula  $D = 1 - \sum p_i^2$ . The calculation of the haplotype diversity ( $H$ ) was based on the formula  $H = 1 - \sum f_i^2$  [3].

## 3. Results and discussion

Twelve single-copy and two multi-copy Y-chromosomal STRs were evaluated in the five German populations Dresden, Hamburg, Rostock, Munich, and the Sorbs of Lusatia. The allele frequencies, gene diversities and haplotype diversities were calculated. It was shown that some of the Y-STRs like DYS385, DYS389-II or DYS391 have similar gene diversities in all five populations. Different

Table 1

Gene diversities and ranking of 12 single-copy and 2 multi-copy (MC) Y-STR loci in five German populations

	Dresden		Hamburg		Rostock		Munich		Sorbs		Cumulative	
DYS464 (MC)	0.9317	1	0.9271	1	0.9452	1	0.9303	1	0.7658	3	0.9446	1
DYS385 (MC)	0.8433	2	0.8813	2	0.8313	3	0.7638	3	0.8086	1	0.8638	2
DYS449	0.8249	3	0.8247	3	0.8400	2	0.8491	2	0.7943	2	0.8523	3
DYS447	0.7629	4	0.7480	4	0.7889	4	0.7638	4	0.6730	6	0.7634	4
DYS390	0.7560	5	0.7239	5	0.6910	7	0.7622	5	0.5779	8	0.7555	5
DYS389-II	0.7330	6	0.7039	6	0.7012	6	0.7156	7	0.7253	4	0.7524	6
DYS19	0.7183	7	0.5389	10	0.7553	5	0.6467	9	0.7111	5	0.7159	7
DYS446	0.7156	8	0.6031	7	0.6910	8	0.6576	8	0.6373	7	0.6983	8
DYS463	0.6056	9	0.6022	8	0.5833	9	0.7409	6	0.4090	10	0.6275	9
DYS448	0.5705	10	0.5364	12	0.5493	11	0.5682	10	0.3805	12	0.5494	10
DYS389-I	0.5184	12	0.5864	9	0.4894	13	0.5578	11	0.4352	9	0.5408	11
DYS392	0.5381	11	0.5381	11	0.5668	10	0.5511	12	0.3472	13	0.5294	12
DYS391	0.4757	13	0.4665	13	0.5245	12	0.4444	14	0.3995	11	0.4748	13
DYS393	0.4214	14	0.3807	14	0.4587	14	0.5511	13	0.2497	14	0.4221	14

Cumulative gene diversity means diversity over all 235 DNA samples.

Table 2

Haplotype diversities for analyzing the European Minimal Haplotype standard or 12 single-copy (SC) and 2 multi-copy (MC) Y-STR loci in five German populations

	Dresden	Hamburg	Rostock	Munich	Sorbs	Cumulative
Minimal Haplotype	0.9847	0.9779	0.9709	0.9622	0.9512	0.9921
12 SC+2 MC	1.0000	1.0000	1.0000	0.9644	0.9560	0.9955

Table 3

Alleles of 5 single-copy and 1 multi-copy Y-STRs from the piece of evidence and the suspicious person

	DYS446	DYS447	DYS448	DYS449	DYS463	DYS464
Piece of evidence	19	23	19	32	21	15, 15, 16, 17
Suspicious person	20	23	19	32	21	15, 15, 16, 17

Only difference: DYS446.

diversities were determined for DYS464, especially for the ethnic group Sorbs ( $D=0.7658$ ) in comparison to the four other populations ( $D=0.9271$ – $0.9452$ ). In addition, cumulative values were calculated for allele frequencies and gene diversities. The loci with the highest cumulative gene diversity are DYS464 ( $D=0.9446$ ), followed by DYS385 ( $D=0.8638$ ) and DYS449 ( $D=0.8523$ ). Several Minimal Haplotype loci like DYS391, DYS392, and DYS393 were displaced to the end of the ranking list which is in concordance to the data published [4] (Table 1).

The calculated haplotype diversities for the European Minimal Haplotype standard are between 0.9560 for the ethnic group Sorbs and 0.9847 for Dresden. The additional use of the markers DYS446, DYS447, DYS448, DYS449, DYS463, and DYS464 increased the haplotype diversities for Dresden, Hamburg, and Rostock significantly to 1.0000. Diversities of Munich and Sorbs were increased to 0.9644 and 0.9560; no complete discrimination of all of the non-related male individuals was possible for these two population groups (Table 2).

The need for further Y-STRs became obvious in a case of homicide. The victim was choked and murdered. The Minimal Haplotype loci as well as DYS437, DYS438, DYS439 showed the same alleles for the suspicious person and the piece of evidence. The analysis of further Y-chromosomal STRs on difference between the samples was revealed only at the locus DYS446. The small difference in the typed alleles for DYS446 indicates that the suspicious person was not the murderer but probably his/her close relative (Table 3).

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