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# Frequency data for the STR locus ACTBP2 (SE33) in eight populations

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**Abstract** Allele frequency data for the STR system ACTBP2 (SE33) were determined in eight populations by denaturing polyacrylamide gel electrophoresis with automated laser-induced fluorescence detection. No significant deviations from Hardy-Weinberg equilibrium were observed. The power of discrimination and the mean exclusion chance ranged from 96.6% to 98.7% and from 76.3% to 88.9%, respectively. These forensic efficiency values stress the importance of ACTBP2 for individualisation purposes.

**Keywords** Short tandem repeat · Polymorphism · ACTBP2 · Population genetics

## Introduction

The human beta-actin-related pseudogene H-beta-Ac-psi-2 (ACTBP2) locus is one of the most informative tetranucleotide short tandem repeat systems for personal identification and paternity testing (Moos and Gallwitz 1983; Rolf et al. 1997). The aim of this work was the establishment of a database for forensic purposes. In this study we present the allele frequencies and forensic efficiency values for the ACTBP2 locus in eight different populations from four continents.

### **Materials and methods**

The population samples investigated were Caucasoids from Hungary (Budapest area), Moroccans living in Brussels (Belgium) and

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Turks from the Adana region. The Asian population samples were Han Chinese from the Shen Yang area and Japanese from the Shiga area, while the African population sample was comprised of Ovambos belonging to the southwest Bantu group (Namibia). The New Guinean and Australian population samples were Papuans from the eastern highlands and aborigines from the Adelaide area, respectively. The number of unrelated individuals is given in Table 1. Genomic DNA for PCR analysis was extracted from whole blood or oral swabs as described (Walsh et al. 1991). PCR amplification was performed in 30 cycles using 0.3 µM of 5'-Cy5-labelled reverse primer and unlabelled forward primer (Polymeropoulos et al. 1992) according to Wiegand et al. (1993). Typing was carried out using 0.5-3 µl of the amplification product, 10 fmol each of internal size standards 114 bp and 402 bp and 3 µl loading buffer (Amersham Pharmacia Biotech, Uppsala, Sweden). After denaturation, the samples were run on 0.5 mm thick, 6% acrylamide gels in  $0.6 \times TBE$ buffer containing 7 M urea at 700 V, 20 W, 38 mA and 50 °C for 500 min on an ALFexpress DNA sequencer (Amersham Pharmacia Biotech) with the sequenced allelic ladder (Möller et al. 1995) as external size standard in adjacent lanes. Sizing was performed employing the Allele Links software (Amersham Pharmacia Biotech, version 1.0). Allele assignment was made by size comparison with the allelic ladder (range  $\pm 0.5$  bp). Evaluation of Hardy-Weinberg expectations and other forensic statistical parameters was done with the computer programme package HWE-Analysis (version 3.2, C. Puers, University of Münster). The HWE test was performed with 5,000 random shuffles. Alleles observed less than 5 times were pooled with the next highest allele.

The nomenclature used followed the recommendations of GEDNAP (Schneider et al. 1998).

#### **Results and discussion**

In the system ACTBP2, a total of 58 different alleles were observed in the 8 populations with no allele being more frequent than 18%. A typical bimodal distribution at peaks around alleles 18 and 27.2 could be observed in all populations, except for Papuans (Table 1). The apparent lack of the first smaller peak around allele 18 could be due to the rather small sample number or to a founder effect. Interpopulation comparisons using the program  $R \times C$  (M. Miller, available via http://herb.bio.nau.edu/~miller/rxc.htm), that performs Fisher's Exact test on contingency tables through the use of the Metropolis algorithm, revealed a close relationship between the Chinese and the Japanese population **Table 1**Frequency distribu-<br/>tion and statistical parameters<br/>of ACTBP2 alleles in eight<br/>population samples (n number<br/>of unrelated individuals,<br/>H observed heterozygosity,<br/>D discrimination power,<br/>MEC mean exclusion chance,<br/>P-value HWE exact test proba-<br/>bility)

Allele	Hungar- ians $(n = 240)$	Moroc- cans (n = 141)	Turks ( <i>n</i> = 198)	Japanese $(n = 136)$	Han Chinese (n = 95)	Ovambos $(n = 187)$	Aborigines $(n = 79)$	Papuans $(n = 107)$
7	_	_	0.0025	-	_	_	_	_
8.1	_	_	0.0051	_	-	_	_	-
9.2	-	-	-	-	-	_	_	0.0047
10	_	0.0071	_	_	_	_	_	_
11	0.0021	0.0071	0.0025	_	_	_	0.0063	_
11.2	0.0021	-	-	-	-	0.0053	0.0253	-
12	0.0021	_	0.0025	_	_	0.0027	_	_
12.2	_	_	_	_	_	0.0027	_	_
13	0.0063	0.0319	0.0152	_	_	_	_	_
13.2	_	_	_	_	_	0.0080	0.0127	0.0093
14	0.0250	0.0390	0.0278	_	0.0053	0.0214	_	_
14.2	_	0.0035	0.0025	_	_	0.0134	0.0443	_
15	0.0208	0.0780	0.0253	0.0147	0.0158	0.0107	0.0063	_
15.2	0.0042	_	_	_	_	0.0027	_	0.0047
16	0.0438	0.0603	0.0328	0.0294	0.0211	0.1016	_	_
16.2	0.0021	_	_	_	_	_	0.0127	_
17	0.0604	0.0603	0.0808	0.0404	0.0421	0.0829	0.0316	0.0047
17.2	0.0021	_	_	_	_	0.0053	_	_
17.3	_	_	0.0025	_	_	_	_	_
18	0.0667	0.1206	0.096	0.0846	0.0526	0.0963	0.0127	0.0047
18.2	0.0021	_	0.0076	0.0074	_	0.0080	_	_
19	0.0771	0.08528	0.0833	0.0809	0.0632	0.1096	0.0127	_
19.2	0.0146	0.0035	_	_	0.0053	0.0027	_	_
20	0.0667	0.0390	0.0657	0.0515	0.0842	0.1257	0.0063	_
20.2	0.0042	0.0035	0.0051	0.0074	0.0012	_	-	_
20.2	0.0188	0.0035	0.0303	0.0588	0.0055	0.0561	_	_
21 21 21 21 21 21 21 21 21 21 21 21 21 2	0.0167	-	0.0076	0.0110	0.0158	0.0027	0.0063	_
21.2 22	0.0063	_	0.0126	0.0110	0.0105	0.0187	-	_
22 77 7	0.0005	0.0355	0.0120	0.0257	0.0263	0.0027	0.0063	0.0234
22.2	0.0230	-	0.0120	0.0237	0.0203	0.0027	-	-
23 73 7	0.0500	0.0390	0.0328	0.0441	0.0053	0.0053	0.0570	0.0327
23.2 74	0.0021	0.0370	0.0320	0.0441	0.0520	-	-	-
24 24 2	0.0021	0.0709	0.0379	0.0441	0.0632	0.0134	0.0443	0.0280
24.2 25	0.0004	0.0709	0.0379	0.0441	0.0052	0.0154	0.0445	0.0280
25 25 2	0.0021	0.0248	0.0253	-	0.0055	-	-	-
25.2 26	0.0438	0.0240	0.0255	0.0000	0.1156	0.0401	0.0090	0.1495
20	0.0021	-	-	0.0037	-	0.0615	-	-
20.2	0.0313	0.0390	0.0379	0.0919	0.0421	0.0013	0.0090	0.1729
21 07 0	-	-	-	0.0037	-	0.0080	-	0.0047
21.2	0.1000	0.0215	0.0303	0.1000	0.1211	0.1125	0.1013	0.1028
20 28 2	-	-	0.0023	-	-	-	0.0003	-
28.2	0.0040	0.0490	0.0808	0.0809	0.0421	0.0374	0.1772	0.1402
29	-	-	-	-	-	-	-	0.0047
29.2	0.0365	0.0461	0.055	0.0755	0.0379	0.0154	0.0825	0.1508
29.3 20	-	_	_	0.0037	_	-	-	_
30 20 2	0.0021	-	-	-	-	0.0027	0.0127	-
30.2 21	0.0500	0.0248	0.0657	0.0294	0.0632	0.0107	0.1013	0.1121
31 21 2	0.0021	-	0.0025	-	-	-	0.0063	-
31.2 22	0.0375	0.0248	0.0354	0.0037	0.0211	0.0055	0.0380	0.0421
32	0.0021	-	0.0025	-	-	_	-	-
32.2	0.0188	0.0142	0.0202	0.0147	0.0105	_	0.0253	0.0280
33	0.0063	-	-	-	-	_	-	_
33.2	0.0167	0.0142	_	_	0.0053	_	0.0063	-
34	-	-	0.0051	-	-	-	-	-
34.2	0.0063	0.0035	0.0051	_	_	-	0.0063	_
35	_	0.0107	0.0126	_	_	-	-	_
36	-	-	0.0025	-	-	-	-	-
36.2	-	-	-	-	-	-	0.0127	-
39.2	-	-	-	0.0037	-	-	-	-

Table 1 (continued)

Allele	Hungar- ians $(n = 240)$	Moroc-cans(n = 141)	Turks ( <i>n</i> = 198)	Japanese $(n = 136)$	Han Chinese (n = 95)	Ovambos $(n = 187)$	Aborigines $(n = 79)$	Papuans $(n = 107)$
Н	0.9458	0.8652	0.8535	0.9265	0.9263	0.9198	0.8608	0.8785
D	0.9876	0.9851	0.9773	0.9867	0.9822	0.9830	0.9771	0.9659
MEC	0.8892	0.8832	0.7991	0.8708	0.8667	0.8409	0.8395	0.7633
P-value	0.0640	0.0520	0.058	0.129	0.737	0.517	0.119	0.175

samples (p = 0.2), while all other populations were found to be more distantly related (p < 0.05). These results clearly underline the importance of establishing ACTBP2 databases for particular populations (e.g. Pestoni et al. 1999). Comparison of the data for the Japanese and Chinese population samples with published Japanese and Chinese data (Liu et al. 1997) revealed no significant differences (p = 0.47and p = 0.28, respectively). The observed genotype frequency distributions do not deviate from Hardy-Weinberg expectations based on the exact test (Table 1). The observed heterozygosity ranged from 85% to 95%, while the power of discrimination and the mean exclusion chance ranged from 96.6% to 98.8% and from 76.3% to 88.9%, respectively. According to these statistical parameters the highly polymorphic system ACTBP2 is a powerful tool for forensic casework and paternity testing in all investigated populations.

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## References

Liu C, Harashima N, Katsuyama Y, Ota M, Arakura A, Fukushima H (1997) ACTBP2 gene frequency distribution and sequencing of the allelic ladder and variants in the Japanese and Chinese populations. Int J Legal Med 110:208–212

- Möller A, Schürenkamp M, Brinkmann B (1995) Evaluation of an ACTBP2 ladder composed of 26 sequenced alleles. Int J Legal Med 108:75–78
- Moos M, Gallwitz D (1983) Structure of two human  $\beta$ -actin-related processed genes one of which is located next to a simple repetitive sequence. EMBO J 2:757–761
- Pestoni C, Laren MV, López-Gómez J, Carracedo A (1999) Genetic data on three complex STRs (ACTBP2, D21S11, Hum FIBRA/FGA) in the Galician population (NW Spain). Int J Legal Med 112:337–339
- Polymeropoulos MH, Rath DS, Xiao H, Merril CR (1992) Tetranucleotide repeat polymorphism at the human beta-actin related pseudogene H-beta-AC-psi-2 (ACTBP2). Nucleic Acids Res 20: 1432
- Rolf B, Schürenkamp M, Junge A, Brinkmann B (1997) Sequence polymorphism at the tetranucleotide repeat of the human betaactin related pseudogene H-beta-Ac-psi-2 (ACTBP2) locus. Int J Legal Med 110:69–72
- Schneider HR, Rand S, Schmitter H, Weichhold G (1998) ACTBP2nomenclature recommendation of GEDNAP. Int J Legal Med 111:97–100
- Walsh PS, Metzger DA, Higuchi R (1991) Chelex-100 as a medium for simple extraction of DNA for PCR-based typing from forensic material. Biotechniques 10:506–513
- Wiegand P, Budowle B, Rand S, Brinkmann B (1993) Forensic validation of the STR systems SE33 and TC11. Int J Legal Med 105:315–320