
Genetic characteristics of 22 Y-STRs in Koreans:

Intermediate, null, and duplicated alleles and deletions in the DYS385 flanking region

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Establishment of a Korean Y-STR database for 22 Y-STRs

❖ 22 Y-STRs

- DYS19, DYS385, DYS388, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS446, DYS447, DYS448, DYS449, DYS456, DYS458, DYS464, DYS635 and GATA H4.1

❖ 3 multiplex PCR systems

❖ 708 unrelated Korean males

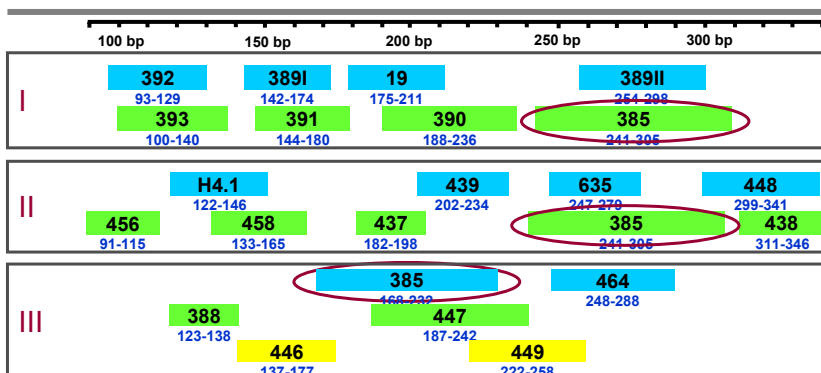
❖ Haplotype analysis and characterization of atypical alleles



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Schematic of 3 multiplex PCRs for 22 Y-STRs



- ❖ DYS385 was included in each multiplex to detect sample switching
- ❖ Small amplicon was designed for **DYS385** in multiplex III
(168-232 bp vs. 241-305 bp)



Y-STR analysis in 708 Koreans

| Y-STR haplotype | No. of haplotype | Haplotype diversity | Discriminatory power |
|---------------------------|------------------|---------------------|----------------------|
| Minimal Haplotype loci | 485 | 0.9966 | 0.6850 |
| 17 Y-filer loci haplotype | 657 | 0.9995 | 0.9280 |
| 22 Y-STRs haplotype | 693 | 0.9999 | 0.9788 |

- ❖ Atypical alleles
 - **Intermediate alleles** at DYS447, DYS449, DYS458 and DYS464
 - **Duplicated alleles** at DYS19, DYS390 and DYS447
 - **Null allele** at DYS448
 - **Different allele designation at DYS385** depending on the primer binding site

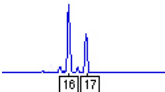
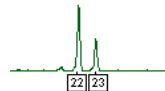
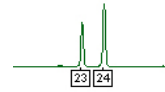


Microvariant and intermediate alleles in DYS447, DYS449, DYS458 and DYS464

| Locus | Allele | Sequence structure | N |
|--------|------------|---|----|
| DYS447 | 18 | (TAATA) ₇ TAAAA ₁ (TAATA) ₁₀ TAAAA(TAATA)_n | 2 |
| | 19 | (TAATA) ₇ TAAAA ₁ (TAATA) ₁₁ TAAAA(TAATA)_n | 4 |
| DYS449 | 26 (25)* | (TTTC) ₁₂ - tctc -N10-N32-cttc-(TTTC) ₁₄ | 1 |
| | 30 (30.1)* | (TTTC) ₁₆ -tctc-N10-N32-cttc-(TTTC) ₁₄ | 1 |
| | 33 (42)* | (TTTC) ₁₅ -tctc-N10- [N32-cttc]₂ -(TTTC) ₁₈ | 1 |
| | 27.2 | (TTTC) ₃ TC (TTTC) ₁₀ -tctc-N10-N32-cttc-(TTTC) ₁₄ | 1 |
| | 28.2 | (TTTC) ₃ TT (TTTC) ₁₁ -tctc-N10-N32-cttc-(TTTC) ₁₄ | 1 |
| | 29.2 | (TTTC) ₃ TC (TTTC) ₁₀ -tctc-N10-N32-cttc-(TTTC) ₁₄ | 1 |
| | 30.2 | (TTTC) ₁₆ -tctc-N10-N32-cttc- (TTTC) ₁₀ TT (TTTC) ₄ | 12 |
| DYS458 | 14.1 | (GAAA) ₁₄ G | 1 |
| | 17.2 | (GAAA) ₁₅ AA (GAAA) ₂ | 1 |
| DYS464 | 12.3 | (CCTT) ₇ CTT (CCTT) ₅ | 1 |
| | 14.3 | (CCTT) ₃ CTT (CCTT) ₁₁ | 5 |

* Ostensible alleles at DYS449

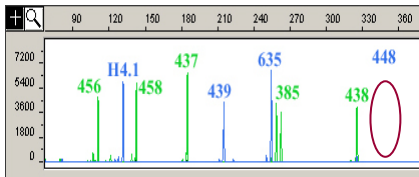
Duplicated alleles at DYS19, DYS390 and DYS447

| Locus | Allele | Sequence structure |
|--------|---|--|
| DYS19 |  16,17 | (TAGA) ₃ tagg (TAGA)_{13,14} |
| DYS390 |  22,23 | (TCTG) ₈ (TCTA)_{9,10} (TCTG) ₁ (TCTA) ₄ |
| DYS447 |  23,24 | (TAATA) ₇ TAAAA (TAATA)_{7,8} TAAAA(TAATA) ₇ |

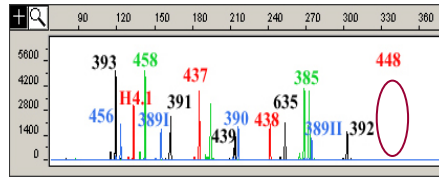
Null allele at DYS448

- ❖ In 6 samples, null allele at DYS448 were commonly observed using multiplex II and Y-filer

Multiplex II



AmpFISTR® Yfiler™ kit

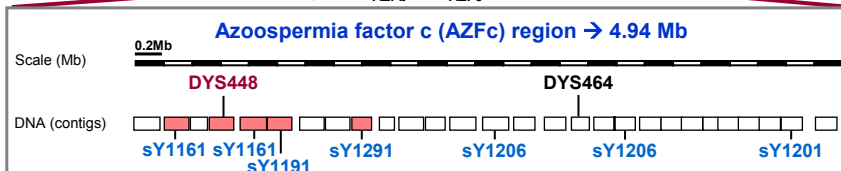
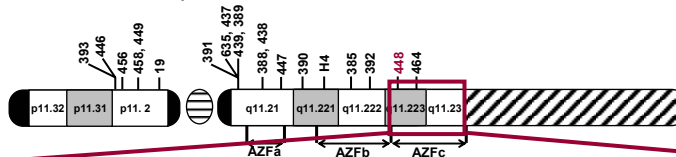


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Characterization of the null allele at DYS448

- ❖ Schematic representation of Y-STRs and STS markers



3 of 708 samples

3 of 708 samples

Null alleles at DYS448 were occurred with partial AZFc deletion.

Frequencies of null allele at DYS448 in various ethnic groups

| | | | |
|---|-----------|--------------|---|
| Korean Present study | 6 in 708 | 0.85% | } Total 14 in 2787 Asians 0.50% |
| Nepalese ParkIn et al. (2006) FSI | 3 in 769 | 0.40% | |
| Malay Chang et al. (2006) FSI | 1 in 334 | 0.30% | |
| Indian Chang et al. (2006) FSI | 1 in 315 | 0.32% | |
| Chinese Chang et al. (2006) FSI | 1 in 331 | 0.30% | |
| Asian in Yfiler database | 2 in 330 | 0.61% | } Null allele at DYS448 is observed much more frequently in Asians compared with African Americans and Caucasians. |
| African American in Yfiler database | 2 in 985 | 0.20% | |
| Caucasian (USA) in Yfiler database | 2 in 1276 | 0.16% | |
| German Rodig et al. (2006) IJLM | 0 in 234 | 0.00% | |
| Austrian Berger et al. (2005) IJLM | 0 in 135 | 0.00% | |



Different allele designation at DYS385 in multiplex I/II and multiplex III



Design of DYS385 for small PCR products in multiplex III

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Forward primer in the multiplex I and II
agcatgggtg acagagctag acaccatgcc aaacaacaac aaagaaaaga
aatgaaattc agaaaggaag gaaggaagga gaaagaaagt aaaaaagaaa
gaaagagaaa aagagaaaaa gaaagaaaga gaagaaagag aaagaggaaa
gagaaaagaaa ggaaggForward primer in the multiplex III
aagg aaggaaggaa gggaaaagaaa gaaagaaaga
aagaaaagaaa gaaagaaaga aagagaaaaa Reverse primer in the multiplex I and II
gaaaggagga ctatgtaatt
gg aatagata gattatTTTT taaaatattt ttattacctt tacagttttt
ttaa atctg gtcagcagcc ctta ccagct
Reverse primer in the multiplex III

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- ❖ Allele size ranges
 - Multiplex I and II: 241-305 bp
 - Multiplex III: 168-232 bp



Characterization of allele designation difference at DYS385

- ❖ Due to the deletion mutation in 8 samples, DYS385 allele designation in multiplex I and II was differently observed from that in multiplex III.

8 bp deletion: (gagaaaa)₂→₁-N38-(aagg)₆(GAAA)_n

4 bp deletion: (gagaaaa)₂-N38- (aagg)₆→₅(GAAA)_n

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F primer in multiplex I/II
agcatgggtg acagagctag acaccatgcc aaacaacaac aaagaaaaga
aatgaaattc agaaaggaag gaaggaagga gaaagaaagt aaaaaagaaa
gaa8 bp deletion gagaaa aagagaaaaa gaaagaaaga gaagaaagag aaagaggaaa
gagaaaagaaa ggaaggForward primer in multiplex III
aagg aaggaaggaa gggaaaagaaa gaaagaaaga
aagaaaagaaa gaaagaaaga aagagaaaaa R primer in multiplex I/II
gaaaggagga ctatgtaatt
gg aatagata gattatTTTT taaaatattt ttattacctt tacagttttt
Reverse primer in multiplex III
ttaa atctg gtcagcagcc ctta ccagct

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Concluding remarks

1. 18 microvariant alleles at DYS449, 6 intermediate alleles at DYS464, 2 intermediate alleles at DYS458, and 1 duplicated allele at DYS19, DYS390 and DYS447 have been discovered and characterized.
2. The use of DYS448 needs to be considered for genotyping of Asians due to the relatively high frequency of null allele in Asians.
3. By applying small amplicon of DYS385 in multiplex III, we could efficiently avoid or prevent the allele mis-designation at DYS385 which occurs as a result of deletion mutation.
4. This work will provide a well-established Korean Y-STR database which can be used as a reference for the interpretation of Y-STR genotyping results in Koreans.



Acknowledgement

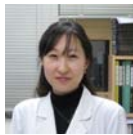
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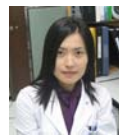
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