Forensic evaluation and haplotypes of 19 Y-chromosomal STR loci in Koreans

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Y-chromosomal STRs

- Applications
  - forensic investigations
  - genealogical purposes
  - evolutionary studies

- Advantages to Human Identity Testing
  - male component isolated without differential extraction
  - paternal lineages

- Needs
  - population studies to evaluate diversity of haplotypes
  - robust assay for accurate characterization of Y markers
**Minimal haplotype loci**

Y-STR Haplotype Reference Database
http://www.ystr.org/asia

9 best studied Y STRs

- DYS19
- DYS389I
- DYS389II
- DYS390
- DYS391
- DYS392
- DYS393
- DYS385

**Percentage of unique haplotypes**

* n = 2576 7-locus Asian haplotypes, DYS385 excluded, logged in the database
** n = 2576 minimal Asian haplotypes logged in the database
*** n = 850 European mt-DNA D-Loop sequences (from Legal Medicine Magdeburg, Germany)
**** n = 551 extended Asian haplotypes logged in the database

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**Previously study of Y-STRs**


*Y-chromosome multiplexes and their potential for the DNA profiling of Koreans.*


*Polymorphism and Haplotypes of 10 Y-STR Loci in Koreans*
Zhe-Jia Zheng, Yong-Ji Zhang, Jin-Chool Park, Hai-Yu Lin, S.—Young Kim, Young-Tae Choi, Jung Bin Lee


*Allele frequencies and haplotypes of six new Y-specific STR loci in Koreans*
Hwan Young Lee, Ji-Eun Oh, Gil-Ro Han, Kyoung-Jin Shin
Selection of new Y-STRs

Forensic Science International 130 (2002) 97-111

DYS464 : four-copy locus

DYS464 a/b/c/d

He human Y chromosome

Object

- Usefulness of DYS464, DYS449, DYS446
- Gene/haplotype diversity and discriminatory power of 19 STR loci containing minimal haplotype loci, DYS388, DYS434, DYS435, DYS436, DYS437, DYS438 and DYS439
- Assess of contribution of respective Y-STRs to the increase of haplotype diversity
Materials and Methods

- **DNA samples**
  - 301 buccal swab samples from unrelated Korean population
  - DNA was extracted using QIAamp DNA Mini Kit
- **Multiplex PCR**
  - Total 10 µl PCR reaction: 1-2 ng of DNA, 1.4 -1.6 µl of Gold STR buffer, 2.0 U of AmpliTaq Gold polymerase and primers
  - Cycling condition: 95°C 1'; 94°C 1' 55-59°C 1' 72°C 1' x 30; 60°C 45'
- **Detection Systems**
  - ABI prism 310 genetic analyzer, Gene Scan software 3.1, and Genotyper 2.5 software (PE Applied Biosystems, CA, USA)

Statistical analysis

- Gene, haplotype diversity and linkage disequilibrium values were calculated using Arlequin version 2.0 software.
### Allelic Structure

**DYS449**

- Repeat structure: (CCTT)\textsuperscript{14}CTT(CCTT)\textsuperscript{2}N\textsuperscript{3}3(CCTT)\textsuperscript{4}N\textsuperscript{4}(CCTT)\textsuperscript{2}N\textsuperscript{5}(CCTT)\textsuperscript{2}
- Microvariant allele
- Mutant allele

**DYS464**

- Repeat structure: (TTTC)\textsuperscript{x}N\textsuperscript{y}[(TTTC)\textsuperscript{2}N\textsuperscript{4}(TTTC)\textsuperscript{2}N\textsuperscript{16}]\textsuperscript{2}(TTTC)\textsuperscript{16}N\textsuperscript{88}
- Repeat

### Single Locus Analysis

<table>
<thead>
<tr>
<th>Marker</th>
<th>Motif</th>
<th># Allele seen</th>
<th># Repeat unit</th>
<th>Gene Diversity</th>
</tr>
</thead>
<tbody>
<tr>
<td>DYS19</td>
<td>TAGA</td>
<td>6</td>
<td>13–18</td>
<td>0.7167</td>
</tr>
<tr>
<td>DYS389I</td>
<td>(TCTG)(TCTA)</td>
<td>6</td>
<td>11–16</td>
<td>0.6659</td>
</tr>
<tr>
<td>DYS389II</td>
<td>(TCTG)(TCTA)</td>
<td>7</td>
<td>26–32</td>
<td>0.7257</td>
</tr>
<tr>
<td>DYS390</td>
<td>(TCTA)(TCTG)</td>
<td>7</td>
<td>21–27</td>
<td>0.6689</td>
</tr>
<tr>
<td>DYS391</td>
<td>TCTA</td>
<td>4</td>
<td>7, 9–11</td>
<td>0.2924</td>
</tr>
<tr>
<td>DYS392</td>
<td>TAT</td>
<td>7</td>
<td>10–16</td>
<td>0.6931</td>
</tr>
<tr>
<td>DYS393</td>
<td>AGAT</td>
<td>6</td>
<td>10, 12–16</td>
<td>0.6338</td>
</tr>
<tr>
<td>DYS385</td>
<td>GAAA</td>
<td>51</td>
<td>9–18–19–20</td>
<td>0.9590</td>
</tr>
<tr>
<td>DYS388</td>
<td>ATT</td>
<td>5</td>
<td>10, 12–15</td>
<td>0.5083</td>
</tr>
<tr>
<td>DYS446</td>
<td>TCTCT</td>
<td>9</td>
<td>11–19</td>
<td>0.7873</td>
</tr>
<tr>
<td>DYS449</td>
<td>TTTC</td>
<td>11</td>
<td>26–30, 30.2, 31–35</td>
<td>0.8433</td>
</tr>
<tr>
<td>DYS464</td>
<td>CCTT</td>
<td>67</td>
<td>12–14–15–16–17</td>
<td>0.9668</td>
</tr>
</tbody>
</table>
# Haplotyping analysis

<table>
<thead>
<tr>
<th>Y-STR marker</th>
<th>Number of haplotypes</th>
<th>Haplotype diversity</th>
<th>Discriminatory power</th>
</tr>
</thead>
<tbody>
<tr>
<td>Multiplex I (minimal haplotype)</td>
<td>230</td>
<td>0.9963</td>
<td>0.7641</td>
</tr>
<tr>
<td>Multiplex II</td>
<td>90</td>
<td>0.9509</td>
<td>0.2990</td>
</tr>
<tr>
<td>Multiplex III</td>
<td>269</td>
<td>0.9989</td>
<td>0.8937</td>
</tr>
<tr>
<td>DYS385+DYS464</td>
<td>200</td>
<td>0.9946</td>
<td>0.6645</td>
</tr>
<tr>
<td>19 Y-STRs haplotype</td>
<td>297</td>
<td>0.9999</td>
<td>0.9867</td>
</tr>
</tbody>
</table>

# Contribution of Y-STR loci to haplotype diversity

<table>
<thead>
<tr>
<th>Y-STR marker</th>
<th># of LD loci&lt;sup&gt;a&lt;/sup&gt;</th>
<th>Gene diversity</th>
<th># of haplotype</th>
<th>Haplotype diversity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Minimal haplotype</td>
<td>-</td>
<td></td>
<td>230</td>
<td>0.9963</td>
</tr>
<tr>
<td>Minimal haplotype + DYS388</td>
<td>7</td>
<td>0.508</td>
<td>239</td>
<td>0.9968</td>
</tr>
<tr>
<td>Minimal haplotype + DYS434</td>
<td>7</td>
<td>0.361</td>
<td>233</td>
<td>0.9967</td>
</tr>
<tr>
<td>Minimal haplotype + DYS435</td>
<td>6</td>
<td>0.259</td>
<td>234</td>
<td>0.9966</td>
</tr>
<tr>
<td>Minimal haplotype + DYS436</td>
<td>5</td>
<td>0.102</td>
<td>233</td>
<td>0.9967</td>
</tr>
<tr>
<td>Minimal haplotype + DYS437</td>
<td>7</td>
<td>0.413</td>
<td>236</td>
<td>0.9968</td>
</tr>
<tr>
<td>Minimal haplotype + DYS438</td>
<td>8</td>
<td>0.672</td>
<td>232</td>
<td>0.9964</td>
</tr>
<tr>
<td>Minimal haplotype + DYS439</td>
<td>6</td>
<td>0.610</td>
<td>255</td>
<td>0.9980</td>
</tr>
<tr>
<td>Minimal haplotype + DYS446</td>
<td>7</td>
<td>0.787</td>
<td>250</td>
<td>0.9980</td>
</tr>
<tr>
<td>Minimal haplotype + DYS449</td>
<td>8</td>
<td>0.843</td>
<td>258</td>
<td>0.9985</td>
</tr>
<tr>
<td>Minimal haplotype + DYS464</td>
<td>8</td>
<td>0.967</td>
<td>276</td>
<td>0.9989</td>
</tr>
</tbody>
</table>

<sup>a</sup> Number of minimal haplotype core loci in significant linkage disequilibrium with each added Y-STR locus.
Contribution of Y-STR loci to haplotype diversity

Conclusions

- Haplotype diversity of 19 Y-STR loci was 0.9999 and 297 different haplotypes was identified.
- DYS464 (0.9668), DYS385 (0.9590), DYS449 (0.8433) and DYS446 (0.7873), in order of those, are highly diverse in a Korean population.
- DYS464, DYS449 and DYS446 are significantly increase the haplotype diversity of minimal haplotype.
- DYS434, DYS435, DYS436, DYS437, DYS438 did not significantly increase the number of different haplotypes and the haplotype diversity.