Confirmation of Y haplogroup tree topologies with newly suggested Y-SNPs for the C2, O2b and O3a subhaplogroups

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Y SNP and Y haplogroup

- Specific and distinct geographic distribution
  → Investigation of the history of human migration.
- The inference of the biographic ancestry through haplogroup identification.
- Forensic fields, anthropology, genetic genealogy and human evolution.
Y-chromosome phylogenetic Tree

New binary polymorphisms reshape and increase resolution of the human Y chromosomal haplogroup tree

REPORT

A Revised Root for the Human Y Chromosomal Phylogenetic Tree: The Origin of Patrilineal Diversity in Africa

Fulvio Cruciani,1,* Beniamino Trombetta,1 Andrea Massia,1 Giovanni Destro-Bisol,2,3 Daniele Sellitto,4 and Rosaria Scozzari1,*

OPEN ACCESS Fully available online

Molecular Dissection of the Basal Clades in the Human Y Chromosome Phylogenetic Tree

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Y haplogroup in East Asia

Y-Chromosome Evidence of Southern Origin of the East Asian–Specific Haplogroup O3-M122

Hong Shi,1,2,4 Yong-ji Dong,1 Bo Wier,9 Chuan-He Xiao,9 Peter A. Underhill,1 Pei-dong Shen,1 Ranajit Chakraborty,1 Li Jin,1,5 and Bing Su,1,2,4

SHORT REPORT

An updated tree of Y-chromosome Haplogroup O and revised phylogenetic positions of mutations P164 and PK4

Shi Yan1,2, Chuan-He Wang1.

ORIGINAL ARTICLE

Assignment of Y-chromosomal SNPs found in Japanese population to Y-chromosomal haplogroup tree

Sue Naitoh1,3, Isao Kando-Numata1, Hideki Shigemitsu3, and Shinobu Harasawa3,2
Goals

Re-analysis of recently suggested or positioned in different phylogenetic topologies Y-SNPs with Korean male samples belonging to predominant Y haplogroup in Korean.
Predominant Y haplogroup in Korean

<table>
<thead>
<tr>
<th>Haplogroup</th>
<th>N</th>
<th>Freq</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>16</td>
<td>0.005</td>
</tr>
<tr>
<td>B (C1b)</td>
<td>95</td>
<td>0.033</td>
</tr>
<tr>
<td>C1f</td>
<td>1</td>
<td>0.0004</td>
</tr>
<tr>
<td>C1f (N1c)</td>
<td>1</td>
<td>0.0014</td>
</tr>
<tr>
<td>C</td>
<td>13</td>
<td>0.0054</td>
</tr>
<tr>
<td>N</td>
<td>1</td>
<td>0.0014</td>
</tr>
<tr>
<td>N1c</td>
<td>27</td>
<td>0.0082</td>
</tr>
<tr>
<td>O1</td>
<td>77.5%</td>
<td></td>
</tr>
<tr>
<td>O1a1</td>
<td>10.2%</td>
<td></td>
</tr>
<tr>
<td>O1a1a</td>
<td>32.0%</td>
<td></td>
</tr>
<tr>
<td>O1a1b</td>
<td>22.4%</td>
<td></td>
</tr>
</tbody>
</table>


Materials

► 545 unrelated Korean males previously classified as one of the C, O2b and O3a haplogroups.

(This study was approved by the Institutional Review Board of Severance Hospital, Yonsei University in Seoul, Korea.)

► The DNA quantification using a NanoDrop 1000 spectrophotometer (Thermo Fisher Scientific).

► Dilution to 1.0 ng/μl for PCR amplification.
Methods

C2 (L1373, Z1338, JST002613-27, Z1300, Z8440, F845), O2b(F3356, L682), O3a1c(F11, F238, F449) haplogroup.

- Multiplex PCR and SBE reaction.

O3a2c1 haplogroup (M134, M117, F444)

- Multiplex PCR and analyzing by length differentiation.

Results
C2 haplogroup – 89 samples

O2b haplogroup – 226 samples

M312 marker was known to define O2b1 haplogroup

→ Mutation was not observed!
O2b1 haplogroup is defined by F3356 not M312!

O2b haplogroup – 226 samples

O2b1a (59.3%)
- **O3a1c haplogroup – 72 samples**

  - O3a1c*
  - O3a1c1
  - O3a1c2
  - O3a1c* (78.2%)

- **O3a2c1 haplogroup – 158 samples**

  - O3a2c1*
  - O3a2c1a
  - O3a2c1b
  - O3a2c1c

  - O3a2c1c (57.0%)
Y haplogroup in 706 Korean males

Summary

- C2[L1373(C2b), Z1338/JST002613-27(C2e), Z1300(C2e1), Z8440(C2e1b), F845(C2e2)], O2b[F3356(O2b1), L682(O2b1b)], O3a1c[F11(O3a1c1), F238/F449(O3a1c2)] and O3a2c1[F444(O3a2c1c)].

- JST002613-27 marker was relocated to the appropriate position to C2e haplogroup.

- F3356 marker was newly confirmed to define the O2b1 haplogroup.

- O2b1b haplogroup had the highest frequency among Korean males (19.0%).
Funding

◆ Bio & Medical Technology Development Program of the National Research Foundation funded (NRF) by the Ministry of Science, ICT & Future Planning (No. 2013-057192).

Thank You For Your Attention!