

# 木村資生記念進化学セミナー 第1回

## Human Evolution I

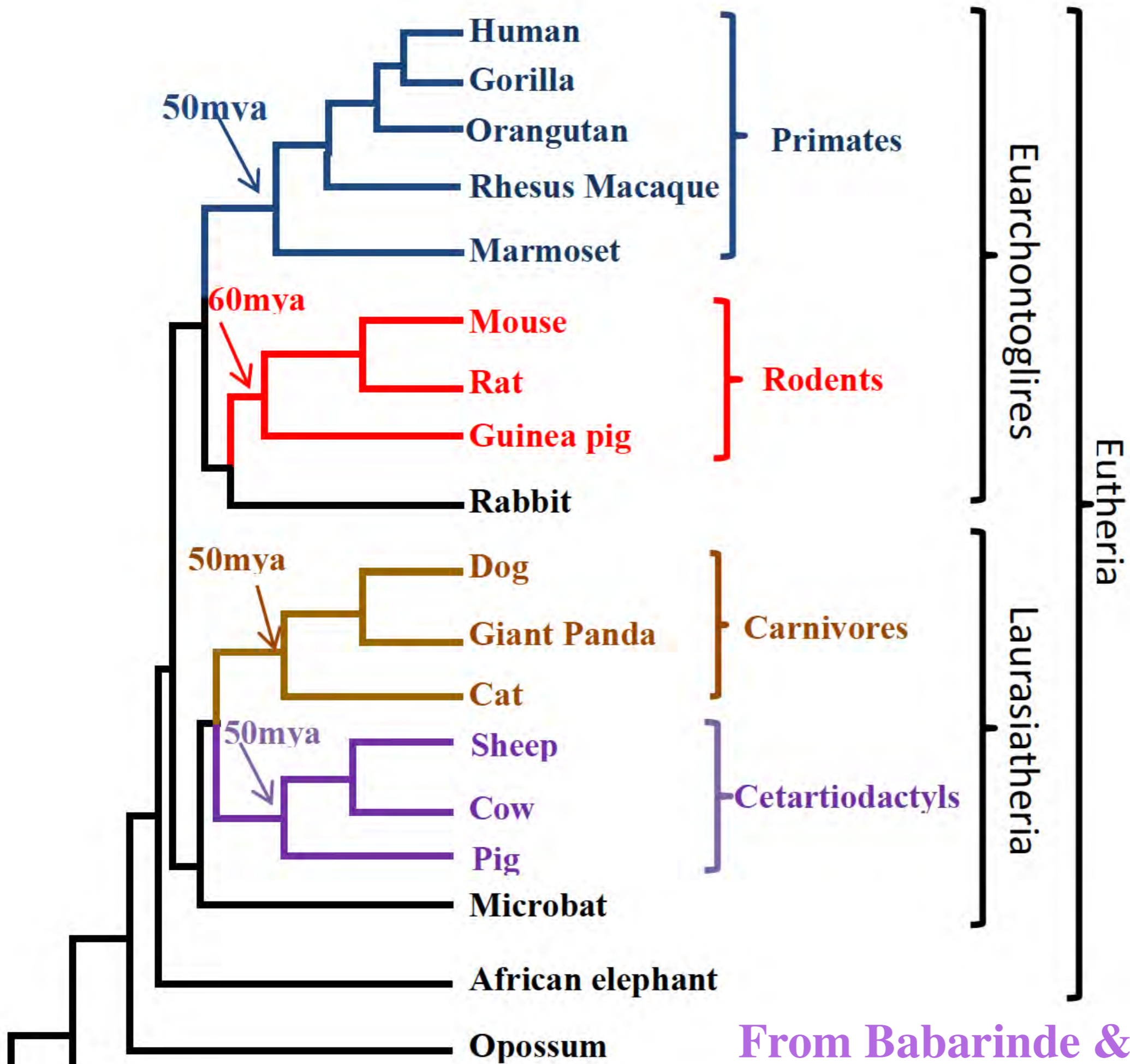
**SAITOU Naruya**

**National Institute of Genetics**

**Mishima, Japan**

**August 4th, A.S. 0017**

**NIG Auditorium, Mishima**



From Babarinde & Saitou (2013)

**Table S2: Setting of whole coding divergence threshold for each lineage**

	<b>Synonymous (S)</b>	<b>Nonsynonymous (N)</b>	<b>Genomic noncoding</b>	<b>Whole coding divergence (C)</b>	<b>Mean divergence proportion (P)</b>
Primates	0.2224 (1.81)	0.0441 (0.20)	0.1218	0.0575 (0.03)	0.0603
Carnivores	0.3868 (2.06)	0.0572 (0.15)	0.1861	0.0817 (0.04)	0.0633
Cetartiodactyls	0.5201 (2.87)	0.0687 (0.24)	0.2143	0.0979 (0.05)	0.0561
Rodents	0.9601 (2.99)	0.1101 (0.25)	0.239	0.1656 (0.06)	0.0578

The mean values of the divergences are given while the values in parentheses are the standard deviations. Synonymous substitution, genomic noncoding divergence and coding divergence are normally distributed ( $P\text{-value} < 10^{-8}$ ). The normality test was performed based on the combination of skew test and kurtosis test using scipy (Jones et al. 2011). Whole coding divergence, which is significantly lower than synonymous and average genomic noncoding divergences, but higher than nonsynonymous divergence, was used as threshold. Mean divergence proportion (P) is given by  $(C-N)/S$ .

# Topics covered in this talk

- PCA (Principal Component Analysis)
- Individual-based admixture analyses
- Population-based phylogenetic analyses
- Search of genes existing in genomic regions with large population difference
- Haplotype estimation (phasing)
- Introgression from archaic humans

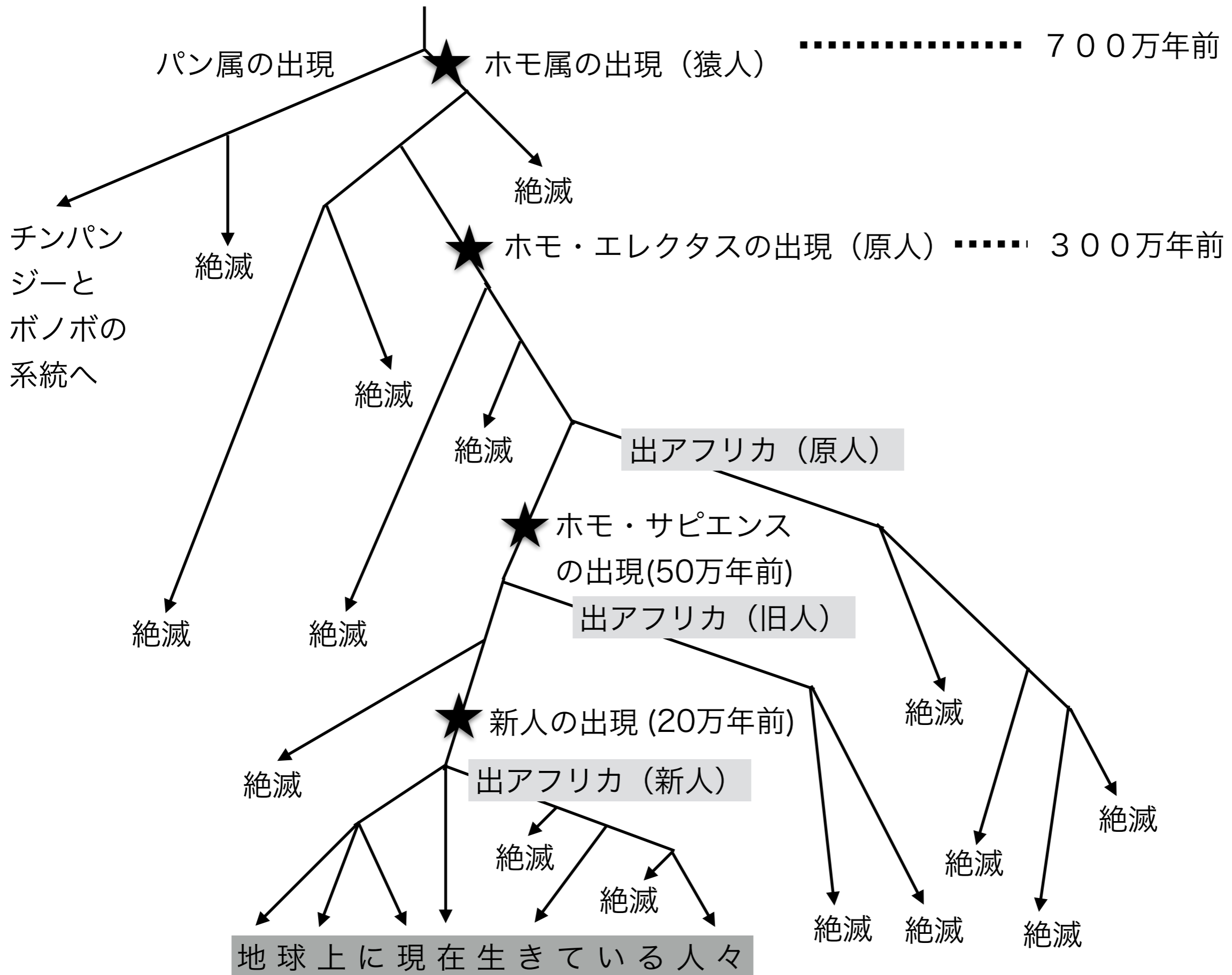


図 1

**Table 2**Estimated Relative Ratios of the Mutation Rates to  $\mu_H$ 

Relative Ratio to $\mu_H$	$\mu_H$	$\mu_C$	$\mu_G$	$\mu_O$	$\mu_{HC}$	$\mu_{HCG}$	$\mu_{HCGO}$
Whole genome	1	1.004	1.034	1.091	1.005	1.025	1.091
X chromosome	1	0.9965	1.073	1.159	1.001	1.070	1.159

mammals. In contrast, the cause of the variation in mutation rates across autosomes remains unclear, though such variation is clearly observed between the human and chimpanzee genomes (Hodgkinson and Eyre-Walker 2011). We did not find statistically significant correlations between mutation rates and genomic characteristics such as GC contents, CpG proportions, chromosomal sizes, SNP densities, or

**Table 3**

Estimated Speciation Times and Ancestral Population Sizes

$\mu_H$ (/Year·Site)	$T_{HC}$ (Ma)	$T_{HCG}$ (Ma)	$T_{HCGO}$ (Ma)	$N_{HC}$	$N_{HCG}$	$N_{HCGO}$	$N_{HC(X)}$	$N_{HCG(X)}$	$N_{HCGO(X)}$
$0.436 \times 10^{-9}$	7.57	9.70	18.8	75,600	65,500	203,000	43,800	49,200	180,000
$0.556 \times 10^{-9}$	5.94	7.61	14.7	59,300	51,400	159,000	34,300	38,500	141,000
$1.00 \times 10^{-9a}$	3.30	4.23	8.19	33,000	28,600	88,600	19,100	21,400	78,400

<sup>a</sup>The value traditionally used. This value was not used for the conclusive estimation.

# ヒトゲノム突然変異率の推定

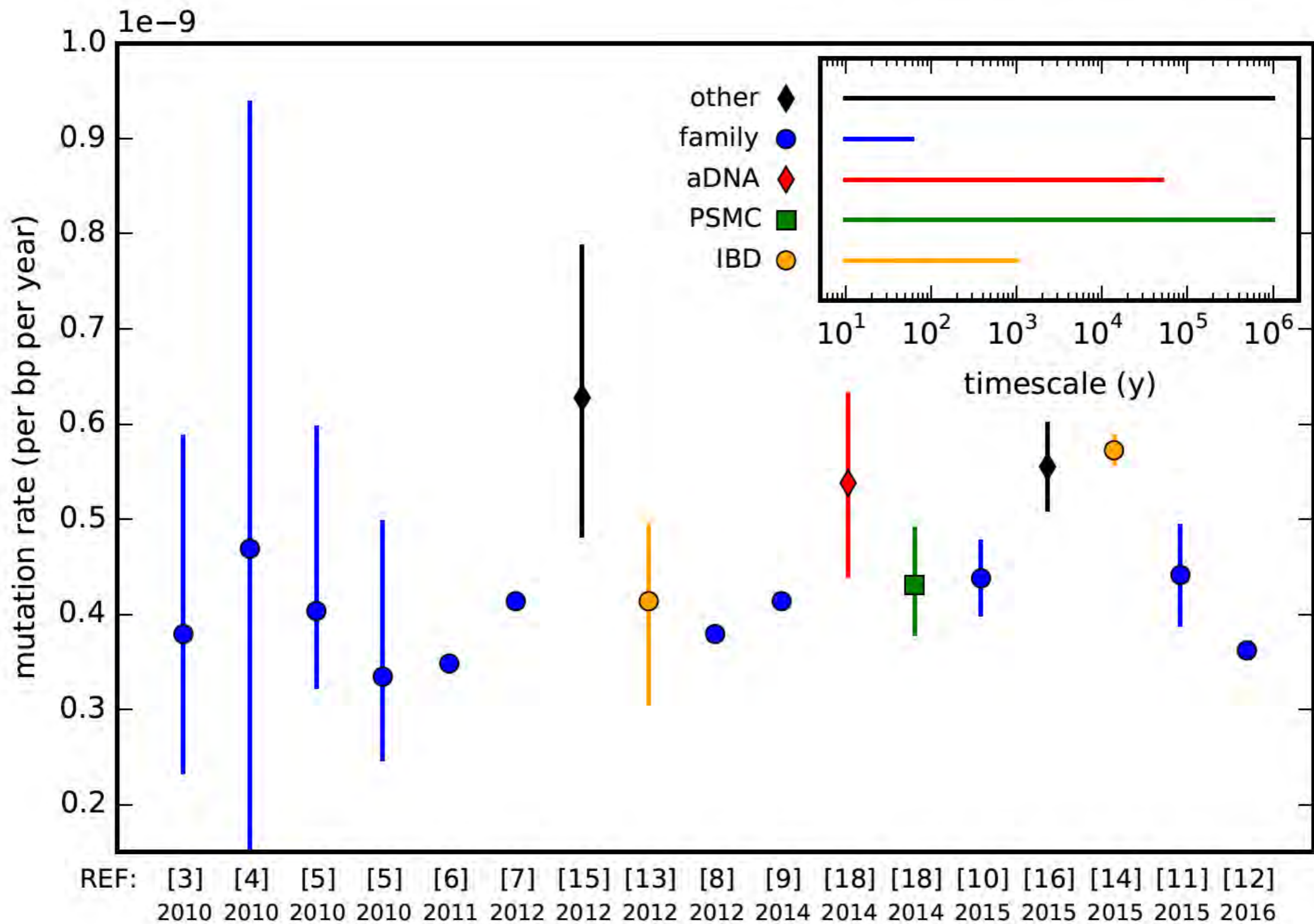


Figure 1: **Recent estimates of the human genome-wide mutation rate.** Estimates are

Scally (2016)より

Figure 17-1: A schematic gene genealogy of two populations which differentiated long time ago

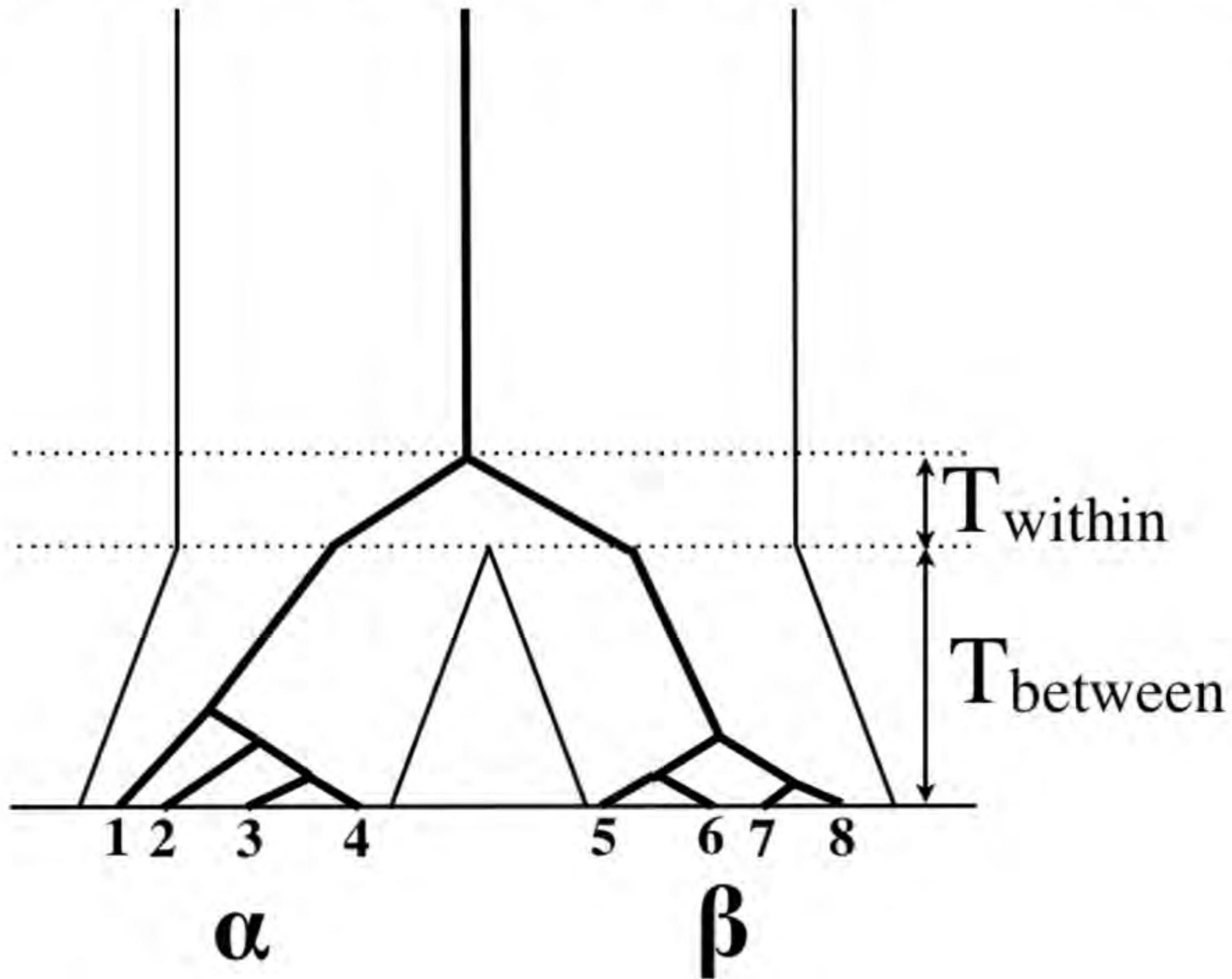
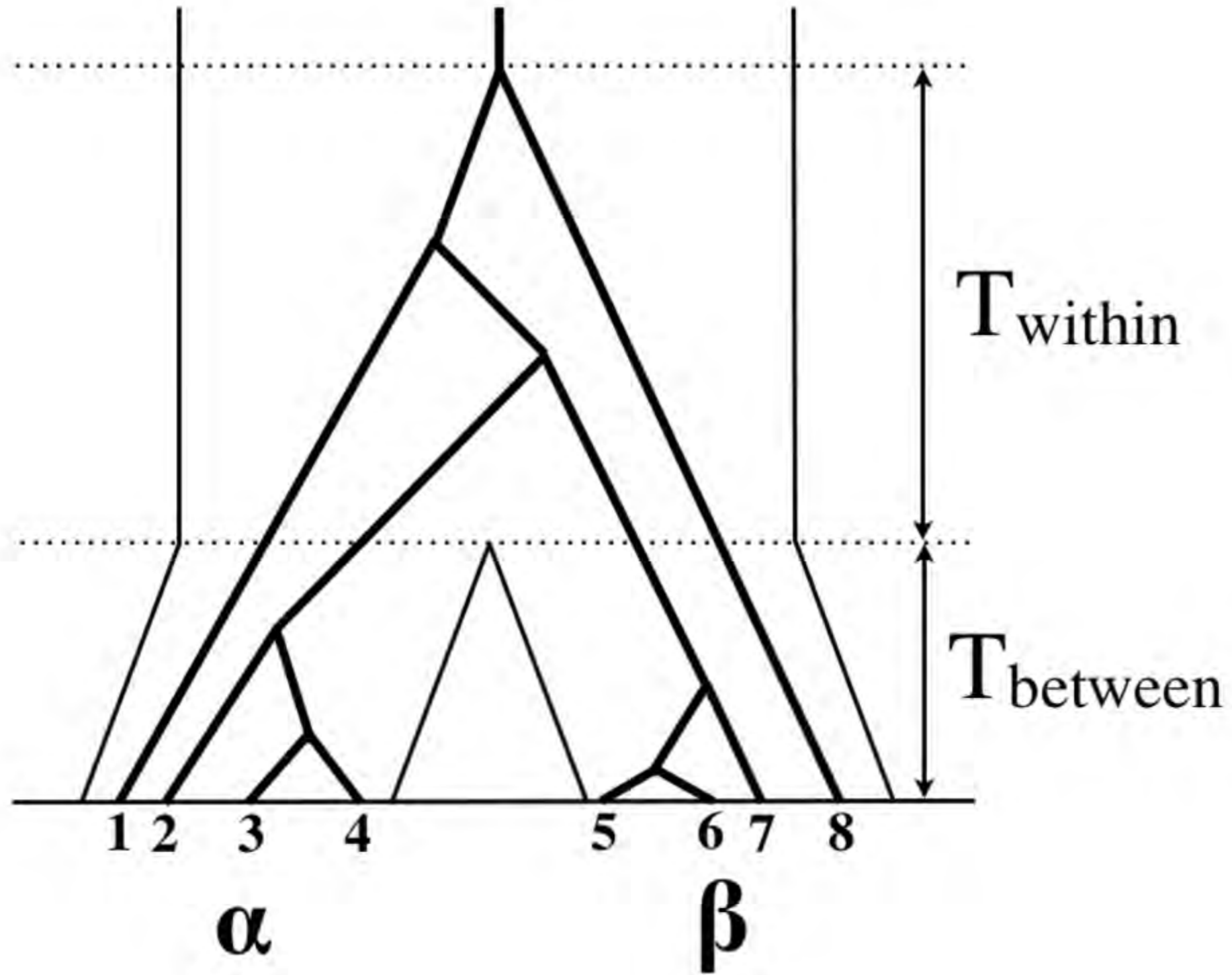
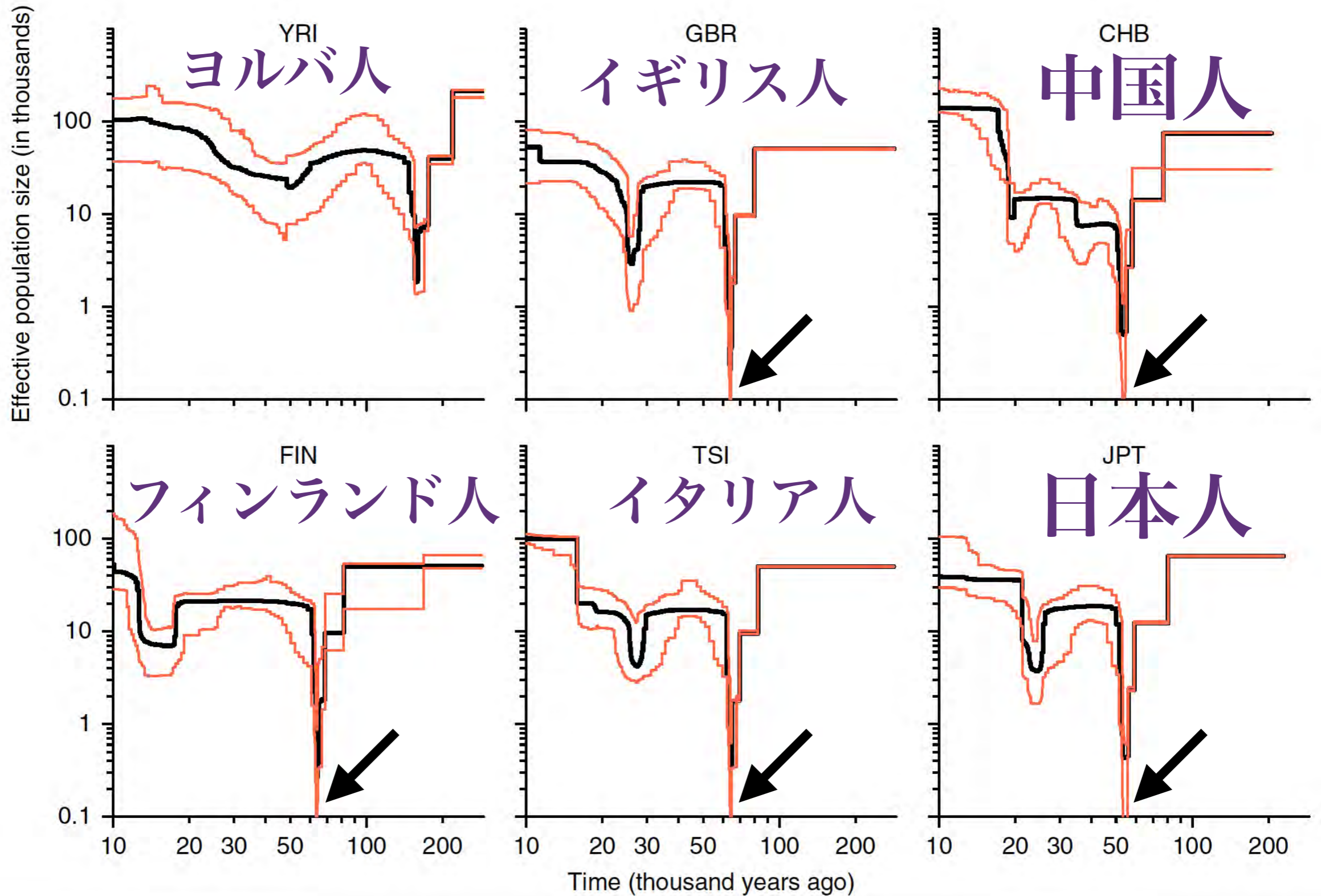




Figure 17-2: A schematic gene genealogy of two populations which differentiated recently



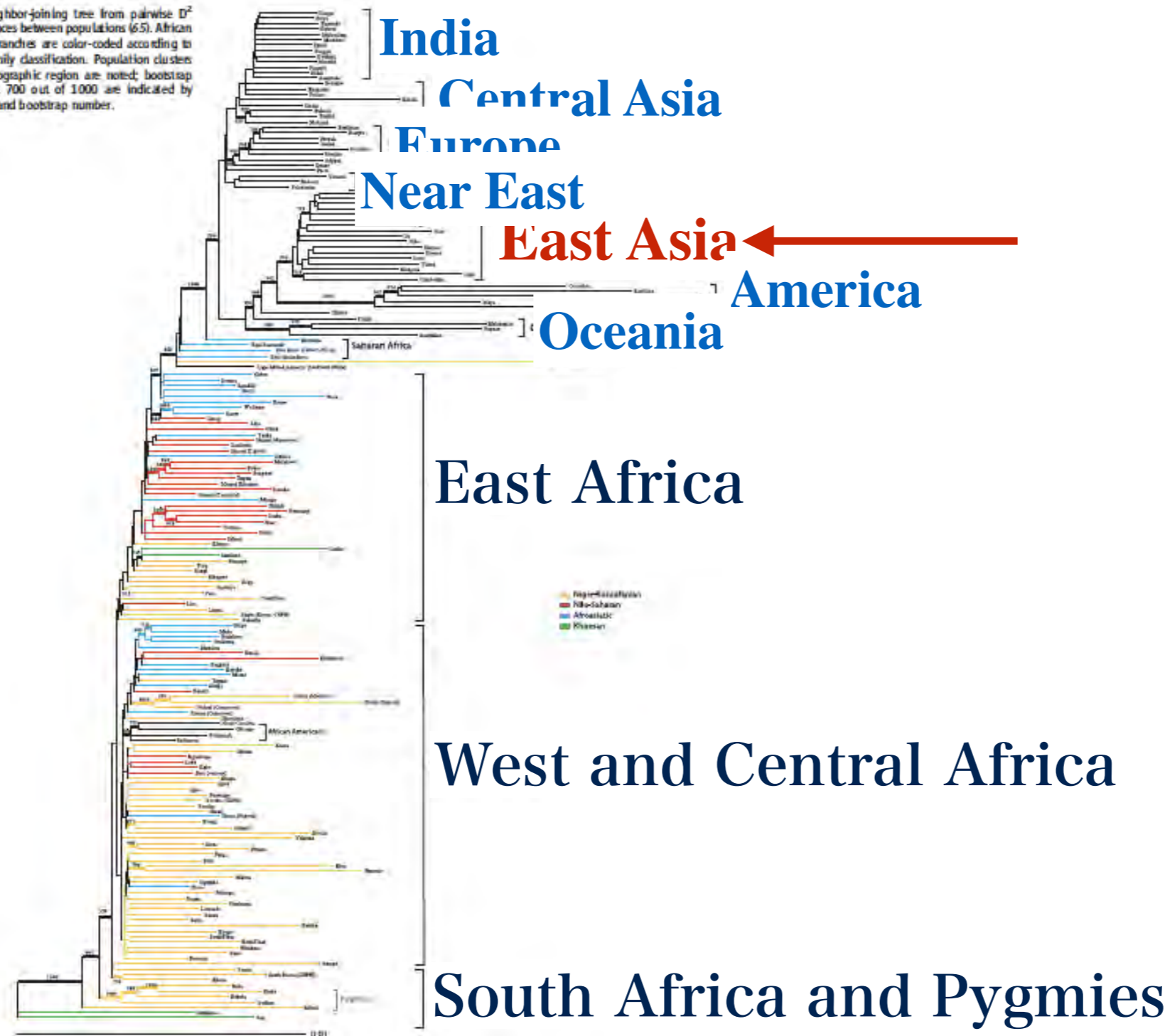
# ゲノムデータを用いた人口変化の推定



“stairway” From Liu and Fu (2015)

# Neighbor-Joining Tree of Modern Human Populations

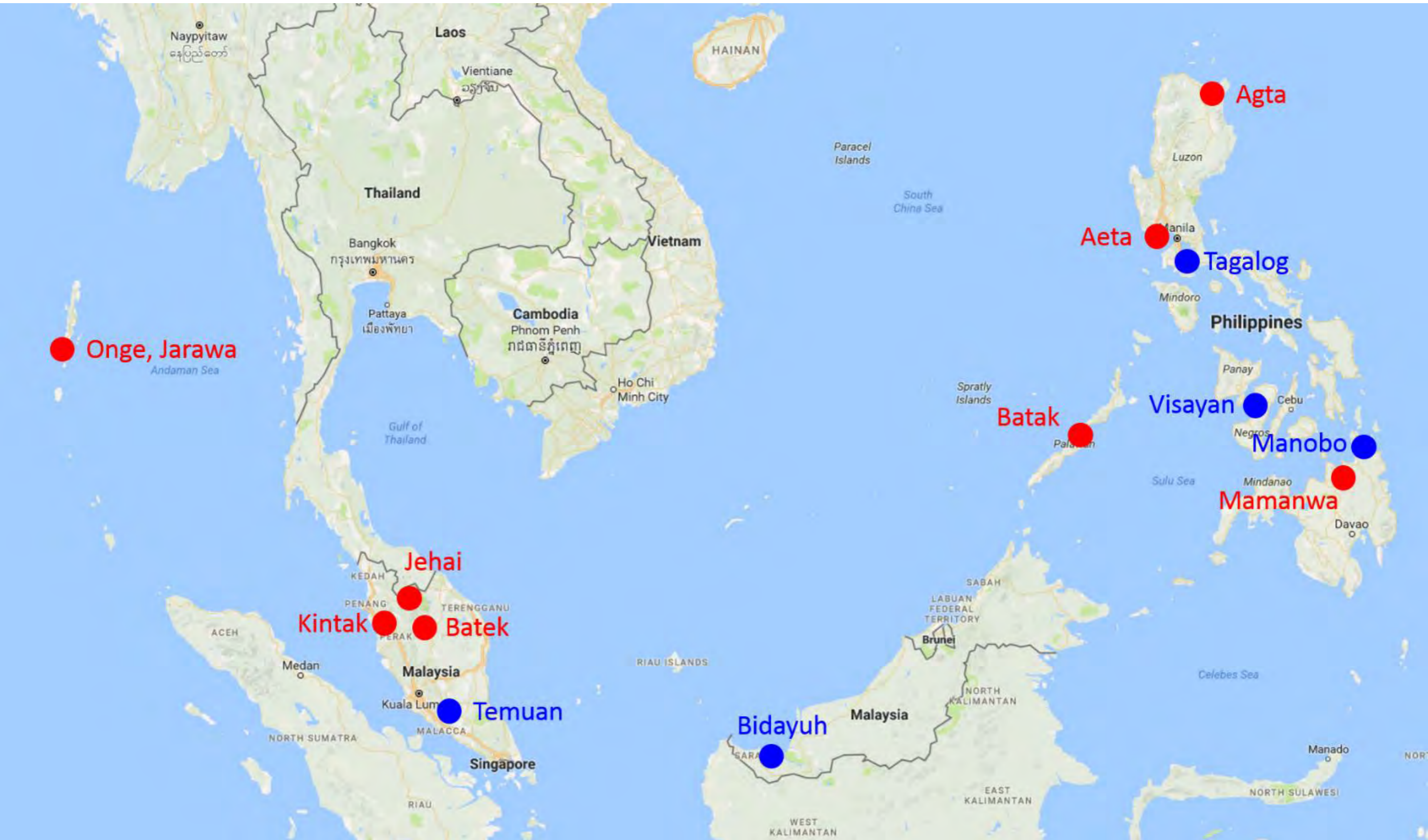
Fig. 1. Neighbor-joining tree from pairwise  $D^2$  genetic distances between populations (65). African population branches are color-coded according to language-family classification. Population clusters by major geographic region are noted; bootstrap values above 700 out of 1000 are indicated by thicker lines and bootstrap number.



From Tishkoff et al. (2009)

● Negritos

● Non-Negritos





**January, 1982; Palawan Island**



February, 1982; Tappa, Pamplona





February, 1982; Palaui Island



# Evolutionary History of Continental Southeast Asians: “Early Train” Hypothesis Based on Genetic Analysis of Mitochondrial and Autosomal DNA Data

Timothy A. Jinam,<sup>1,2</sup> Lih-Chun Hong,<sup>3</sup> Maude E. Phipps,<sup>4</sup> Mark Stoneking,<sup>5</sup> Mahmood Ameen,<sup>3</sup> Juli Edo,<sup>6</sup> HUGO Pan-Asian SNP Consortium,<sup>7</sup> and Naruya Saitou<sup>\*,1,2</sup>

<sup>1</sup>Department of Genetics, The Graduate University for Advanced Studies (SOKENDAI), Mishima, Japan

<sup>2</sup>Division of Population Genetics, National Institute of Genetics, Mishima, Japan

<sup>3</sup>Department of Molecular Medicine, Faculty of Medicine, University of Malaya, Kuala Lumpur, Malaysia

<sup>4</sup>Jeffrey Cheah School of Medicine and Health Sciences, Monash University (Sunway Campus), Selangor, Malaysia

<sup>5</sup>Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany

<sup>6</sup>Department of Anthropology, Faculty of Arts and Social Sciences, University of Malaya, Kuala Lumpur, Malaysia

<sup>7</sup>Human Genome Organisation, Singapore

\*Corresponding author: E-mail: [saitounr@lab.nig.ac.jp](mailto:saitounr@lab.nig.ac.jp).

Mol. Biol. Evol. 29(11):3513–3527  
(2012)



# Individual-based PCA using SNP genotype data

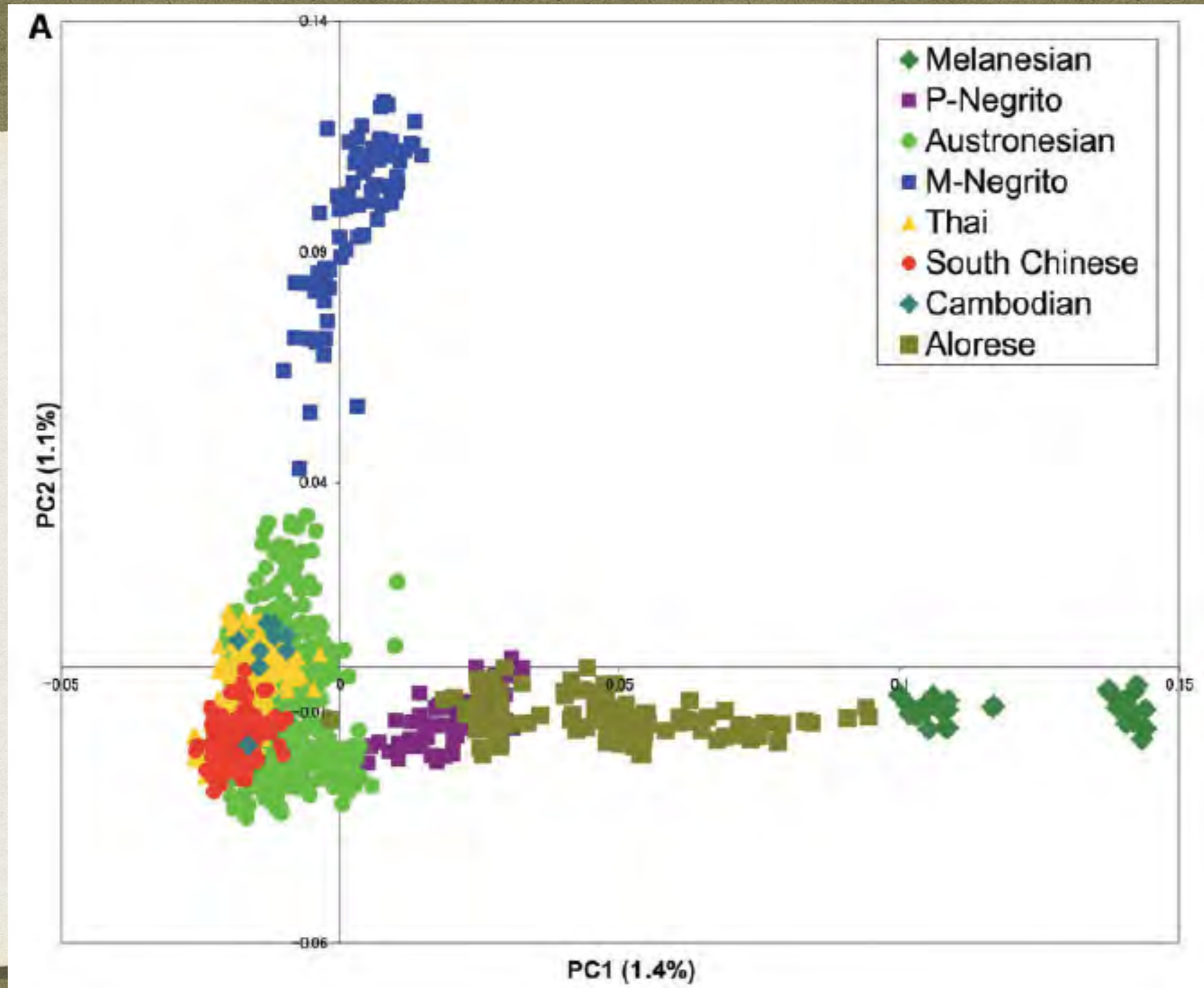
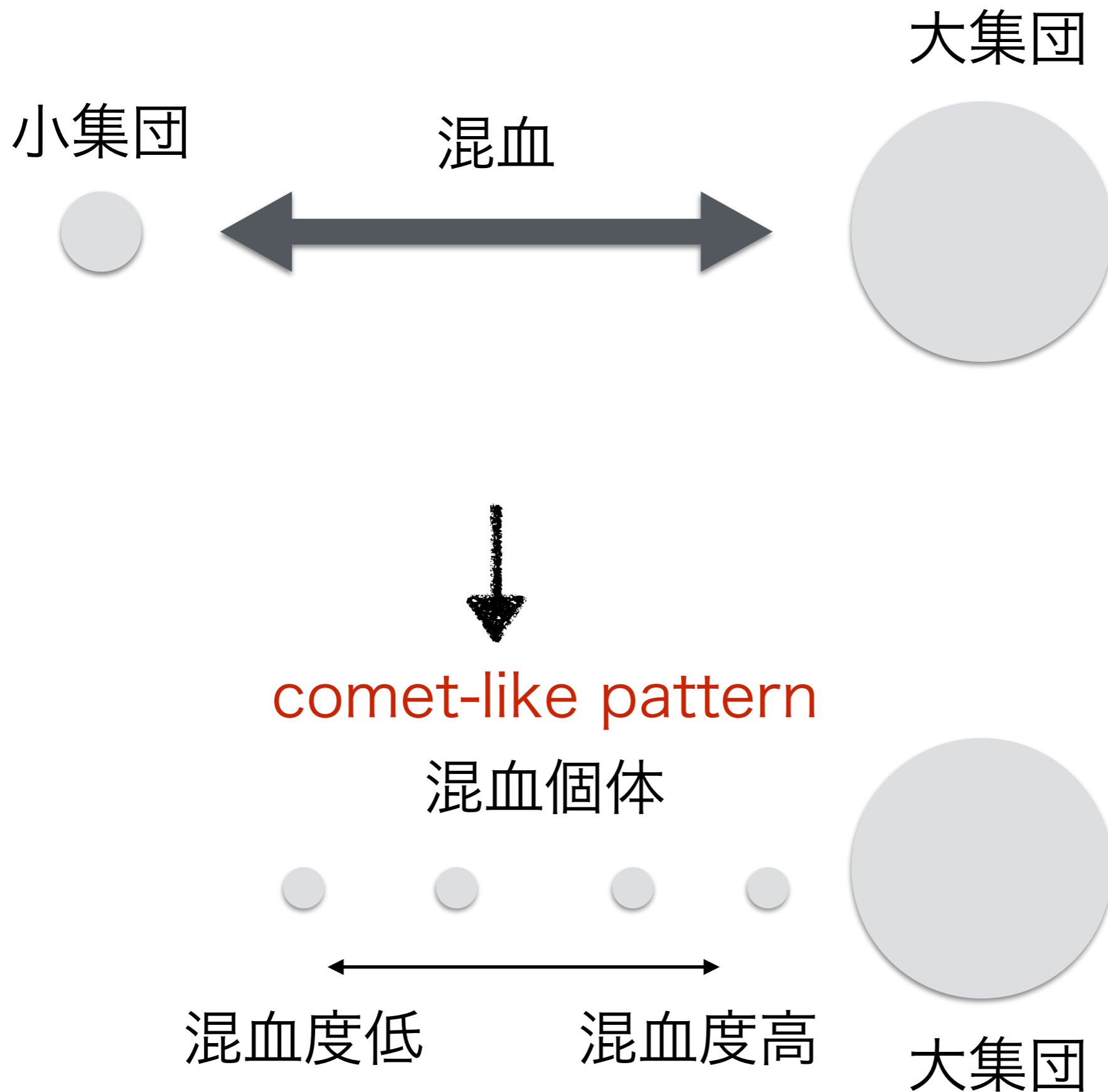
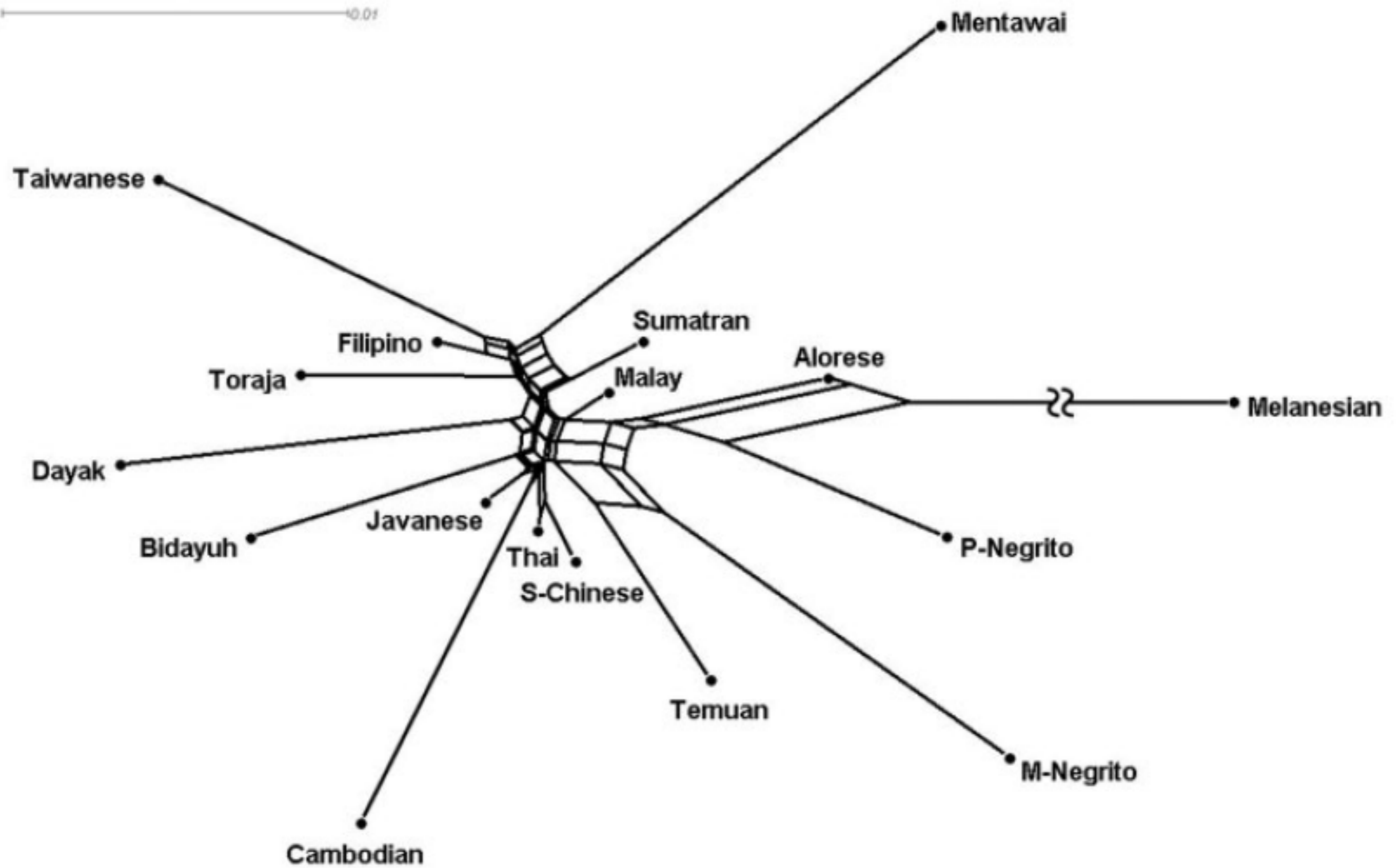


図13：大集団と小集団の混血により、彗星状パターンが生じる様子



# Neighbor-Net network constructed from Nei's standard genetic distance matrix



11-27-2013

# Admixture Patterns and Genetic Differentiation in Negrito Groups from West Malaysia Estimated from Genome-wide SNP Data

Timothy A. Jinam

*Department of Genetics, School of Life Science, Graduate University for Advanced Studies (SOKENDAI), Mishima, Japan*

Maude E. Phipps

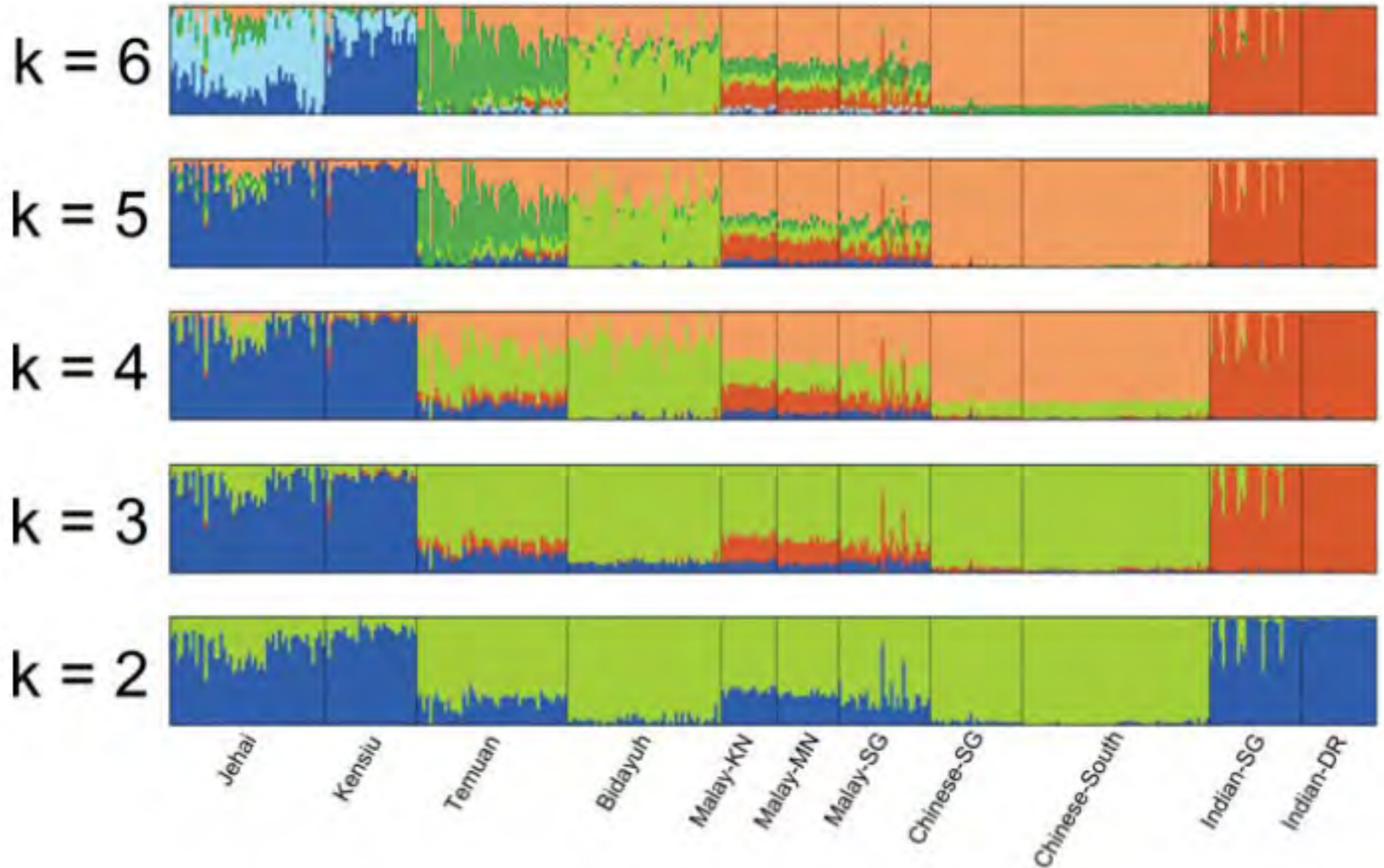
*School of Medicine and Health Sciences, Monash University (Sunway Campus), Selangor, Malaysia*

Naruya Saitou

*Division of Population Genetics, National Institute of Genetics, Mishima, Japan, [saitounr@lab.nig.ac.jp](mailto:saitounr@lab.nig.ac.jp)*

The Hugo Pan-Asian SNP Consortium

# Structure analysis (k = 2~6)



PC2 (2.3%)



0.14      -0.08      -0.04      0      0.06



PC1 (2.6%)

# **Discerning the origins of the Negritos, First Sundaland Peoples: deep divergence and archaic admixture**

**Genome Biology and Evolution (advance pub. July 11, 2017)**

**Timothy A. Jinam,<sup>1</sup> Maude E. Phipps,<sup>2</sup> Farhang Aghakhanian,<sup>2</sup> Partha P. Majumder,<sup>3</sup> Francisco Datar,<sup>4</sup> Mark Stoneking,<sup>5</sup> Hiromi Sawai,<sup>6</sup> Nao Nishida,<sup>6,7</sup> Katsushi Tokunaga,<sup>6</sup> Shoji Kawamura,<sup>8</sup> Keiichi Omoto,<sup>9</sup> Naruya Saitou<sup>1</sup>**

**1) Division of Population Genetics, National Institute of Genetics, Mishima, 411-8540, Japan**

**2) Jeffrey Cheah School of Medicine and Health Sciences, Monash University Malaysia, Sunway City, 46150 Selangor, Malaysia**

**3) National Institute of Biomedical Genomics, Kalyani 741251, West-Bengal, India**

**4) Department of Anthropology, University of the Philippines, Diliman, Quezon City 1101, the Philippines**

**5) Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology, Deutscher**

**6) Department of Human Genetics, Graduate School of Medicine, The University of Tokyo, Tokyo 113-865**

**7) Department of Hepatic Disease, Research Center for Hepatitis and Immunology, National Center for Global Health and Medicine, Chiba 272-8516, Japan**

**8) Department of Integrated Biosciences, Graduate School of Frontier Sciences, The University of Tokyo,**

**9) Department of Anthropology, Faculty of Science, The University of Tokyo, Tokyo 113-0033, Japan**



**Dr. Timothy A. Jinam  
at Kanazawa in 2015**



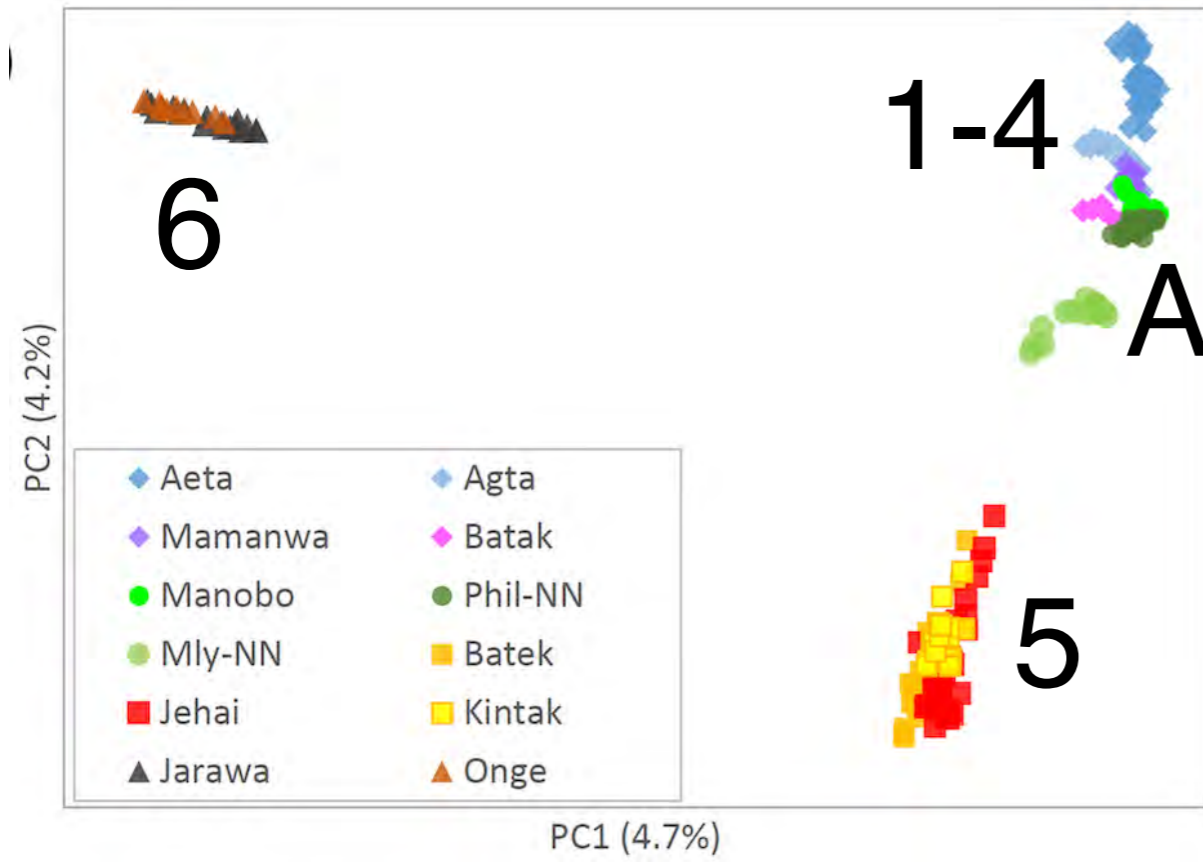
**Table S2:** Types of analyses performed using merged datasets

Dataset	Typing platform	Populations included	No. of SNP	Type of analysis
1	<del>Affymetrix</del> 6.0 <del>genechip</del>	Agta, Acta, <del>Mamanwa</del> , Batak, Tagalog, Visayan, Manobo, <del>Jehai</del> , <del>Temuan</del> , <del>Bidayuh</del>	519,832	PCA (Fig. 1B, Fig. S4B); Neighbor-Net network (Fig. S11)
2	<del>Affymetrix</del> + Illumina SNP chip	Dataset 1 + Jarawa, Onge, <del>Batek</del> , <del>Kintak</del>	112,845	PCA (Fig. 1A); <del>Est</del> calculation; Neighbor-Net network (Fig. S7); Divergence time estimate (Table S4)
3	<del>Affymetrix</del> + Illumina + HGDP Human Origins SNP chip	Dataset 2 + Papuan, Melanesian, Cambodian, French, Han Chinese, <del>Biaka</del> , <del>Yoruban</del> , <del>Denisovan</del> , Neanderthal	44,960	ADMIXTURE (Fig. 2); NJ & <del>Treemix</del> (Fig. 3, Fig. S8); f4 ratio test (Fig. 4); PCA (Fig. S4A); D-statistics (Fig.S6); $R_D(X)$ statistic (Fig. S9); Neighbor-Net network (Fig. S12)
4	Variants from whole genome sequences	<del>Denisovan</del> , Neanderthal, Papuan, Australian Aborigine, Acta, Han Chinese	763,275	Allele sharing analysis (Fig. 5)
5	<del>Affymetrix</del> + whole genome sequences	Dataset 1 + Acta, Agta, Batak	513,136	PCA (Fig. S3)
6	<del>Affymetrix</del> 6.0	Dataset 1 + CEU, CHB, YRI	483,197	Divergence time estimate (Fig. S10, Table S4)
7	Illumina SNP chip	Jarawa, Onge, <del>Batek</del> , <del>Kintak</del> , <del>Jehai</del> , <del>Temuan</del> + HGDP French, Papuan, <del>Yoruban</del> , Han, Cambodian	292,599	Divergence time estimate (Table S4)

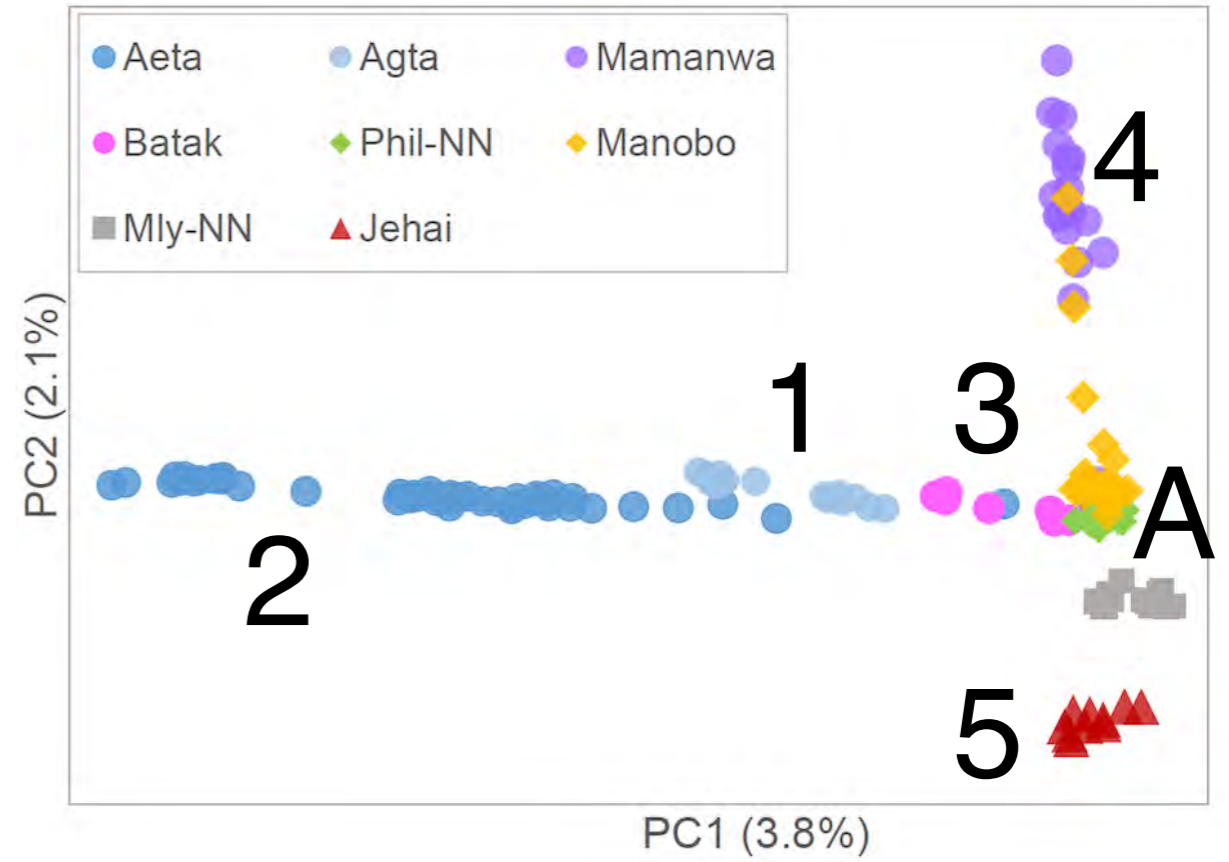
図12：東南アジアのネグリト人のゲノム多様性

(グレイスケールにする)

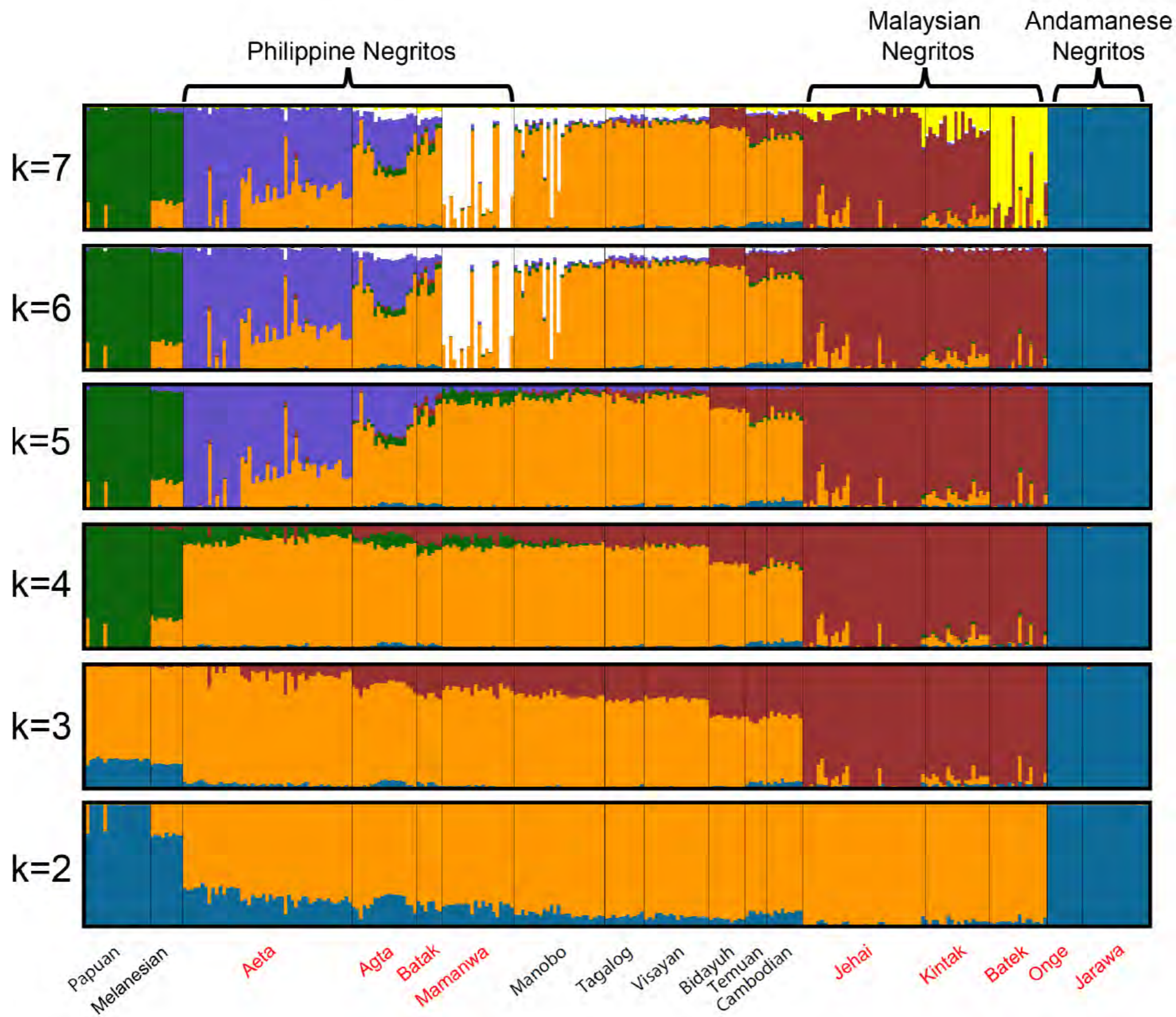
(A)ネグリト集団全体の比較



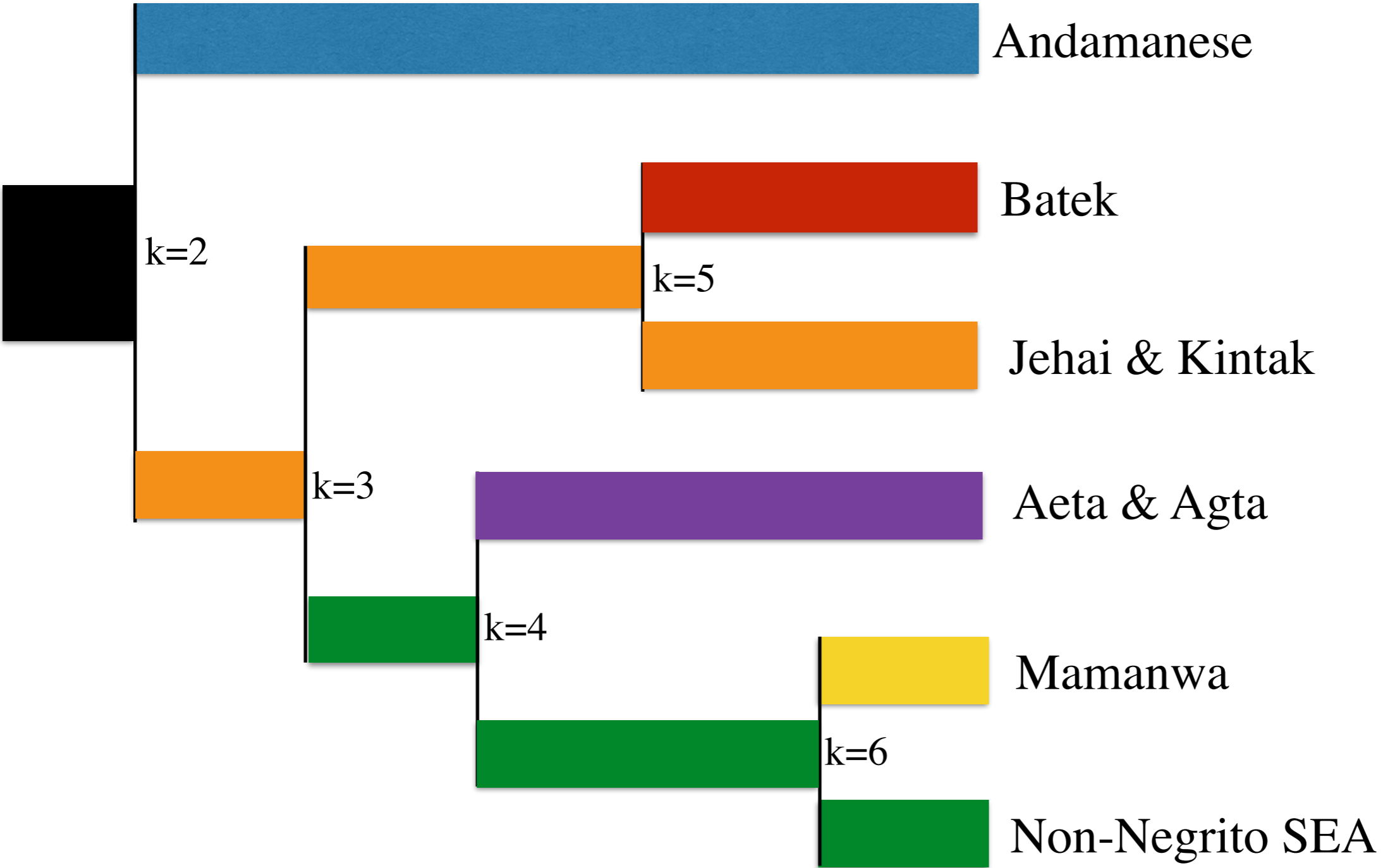
(B)アンダマン諸島人を除いた比較



# Admixture Analysis

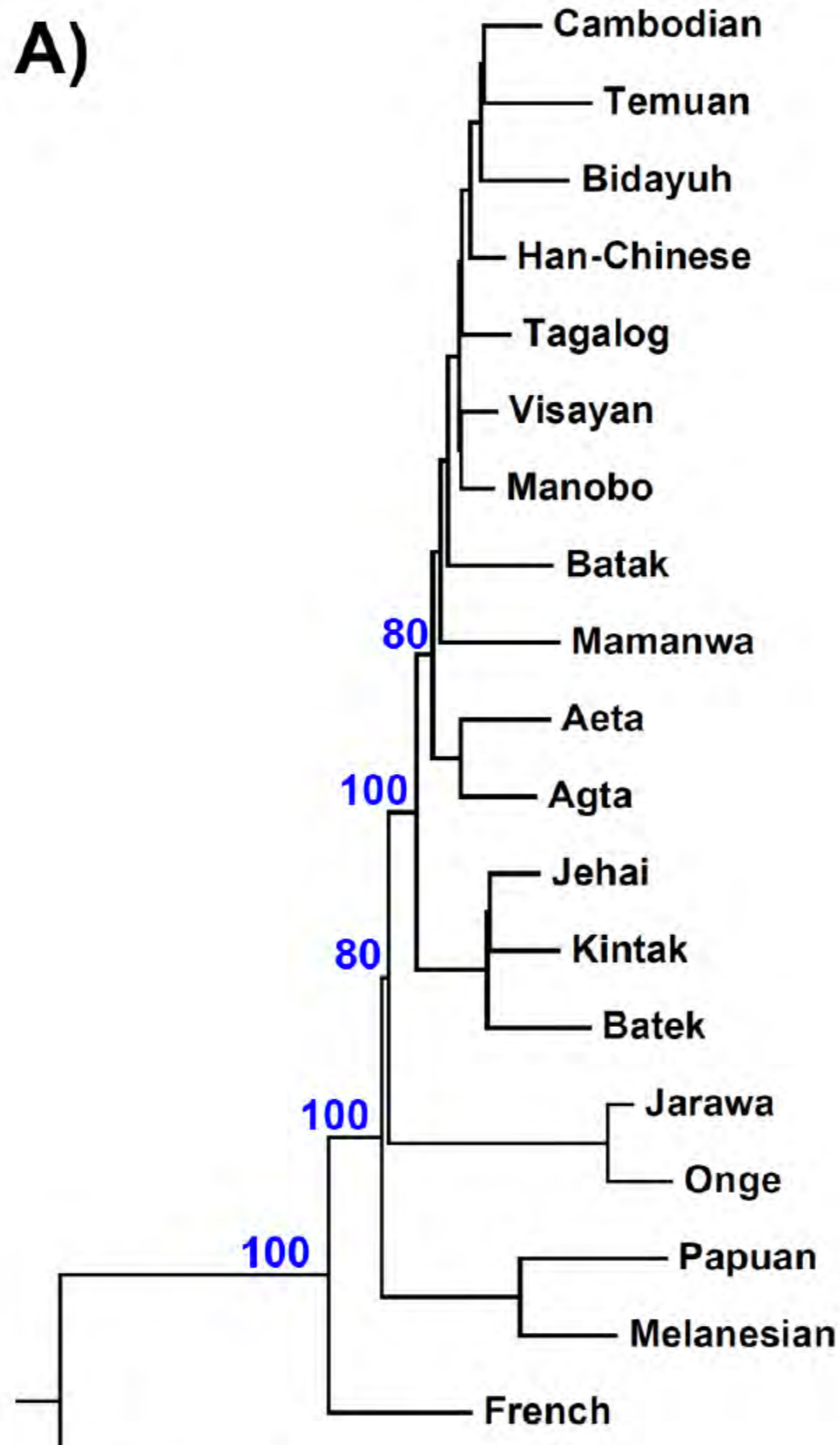


# Phylogenetic interpretation of admixture analysis results

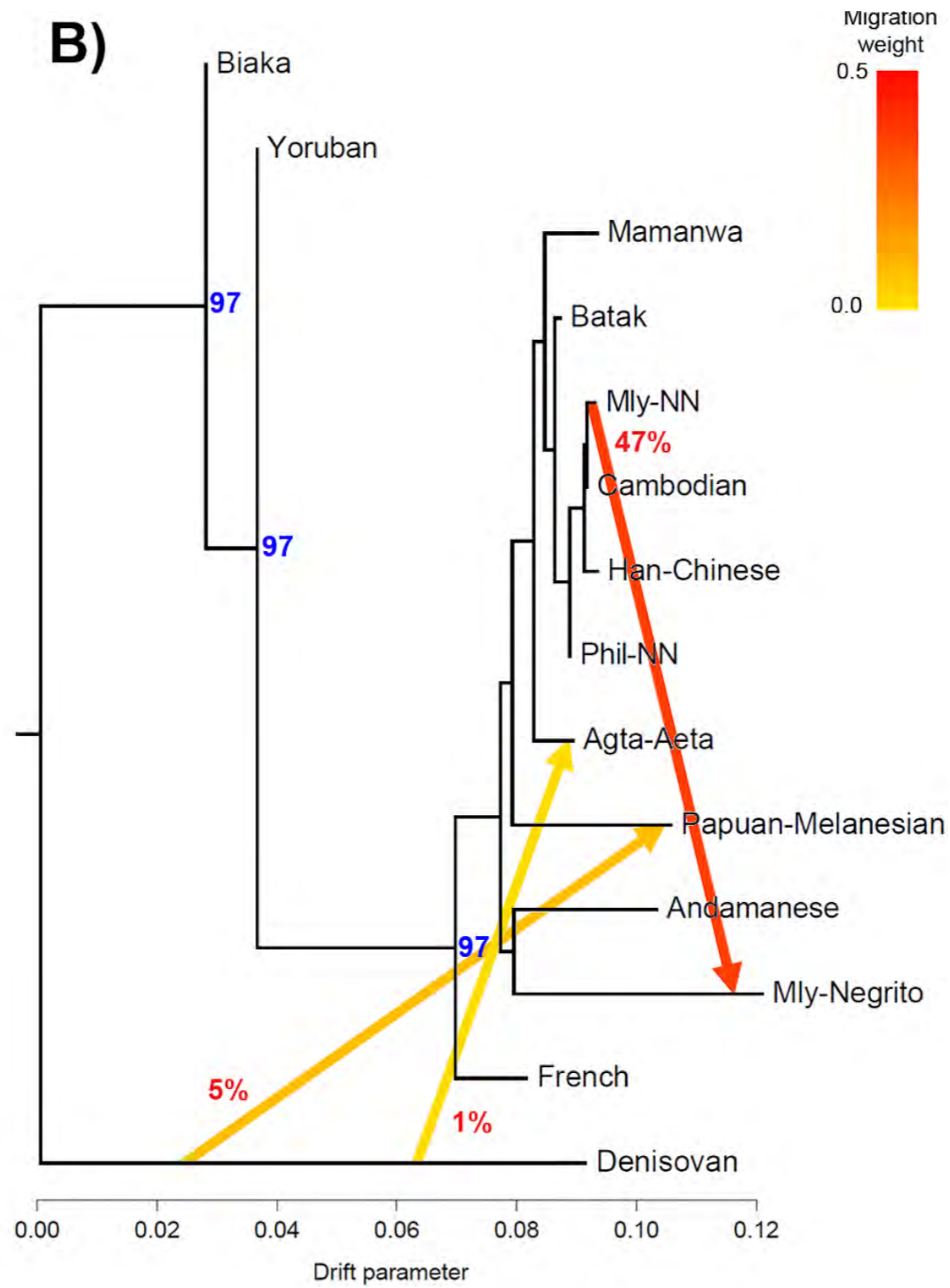


# Phylogenetic tree using Neighbor-Joining method

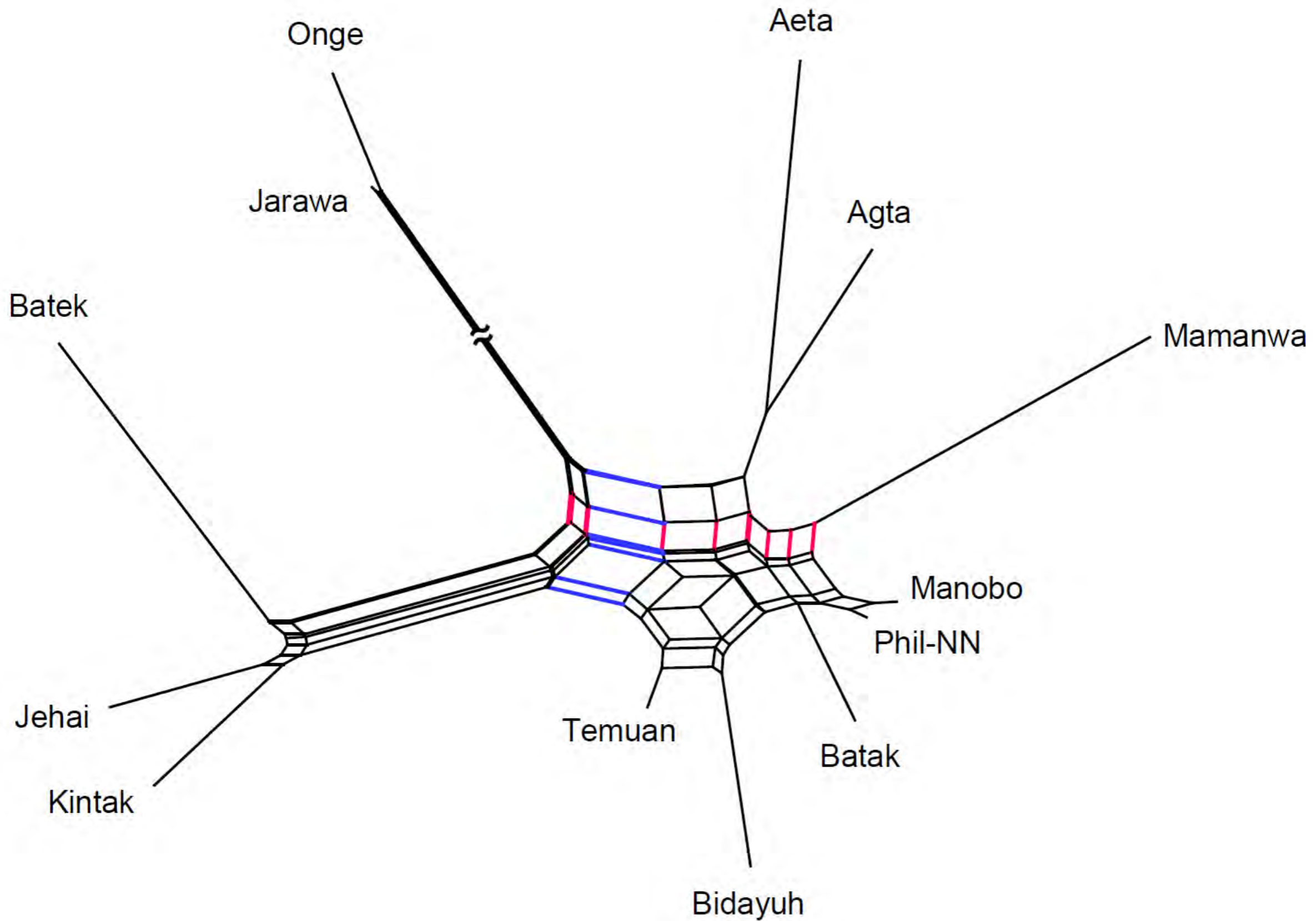
**A)**



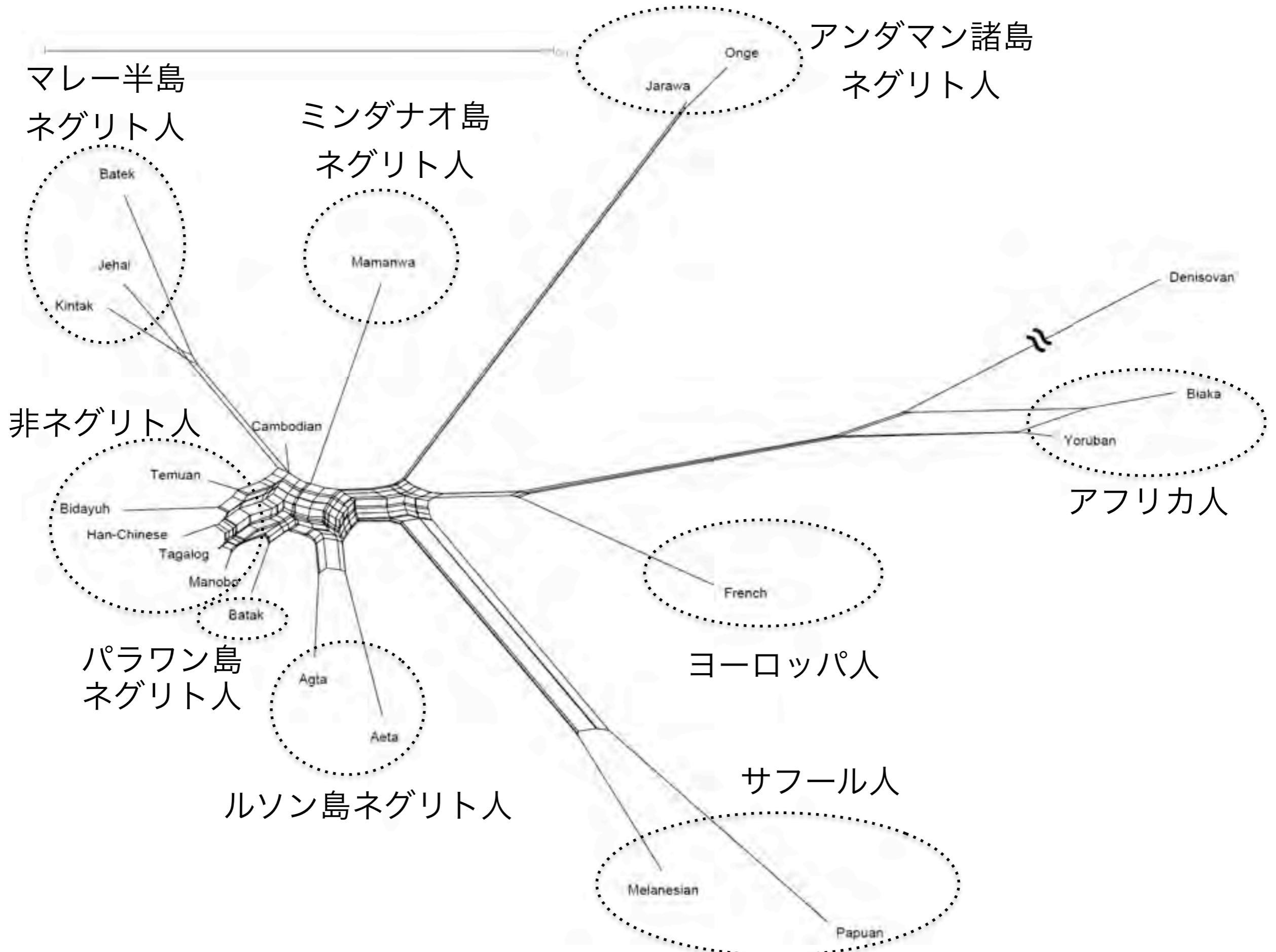
# Phylogenetic tree using TreeMix method



# Phylogenetic Network using NeighborNet

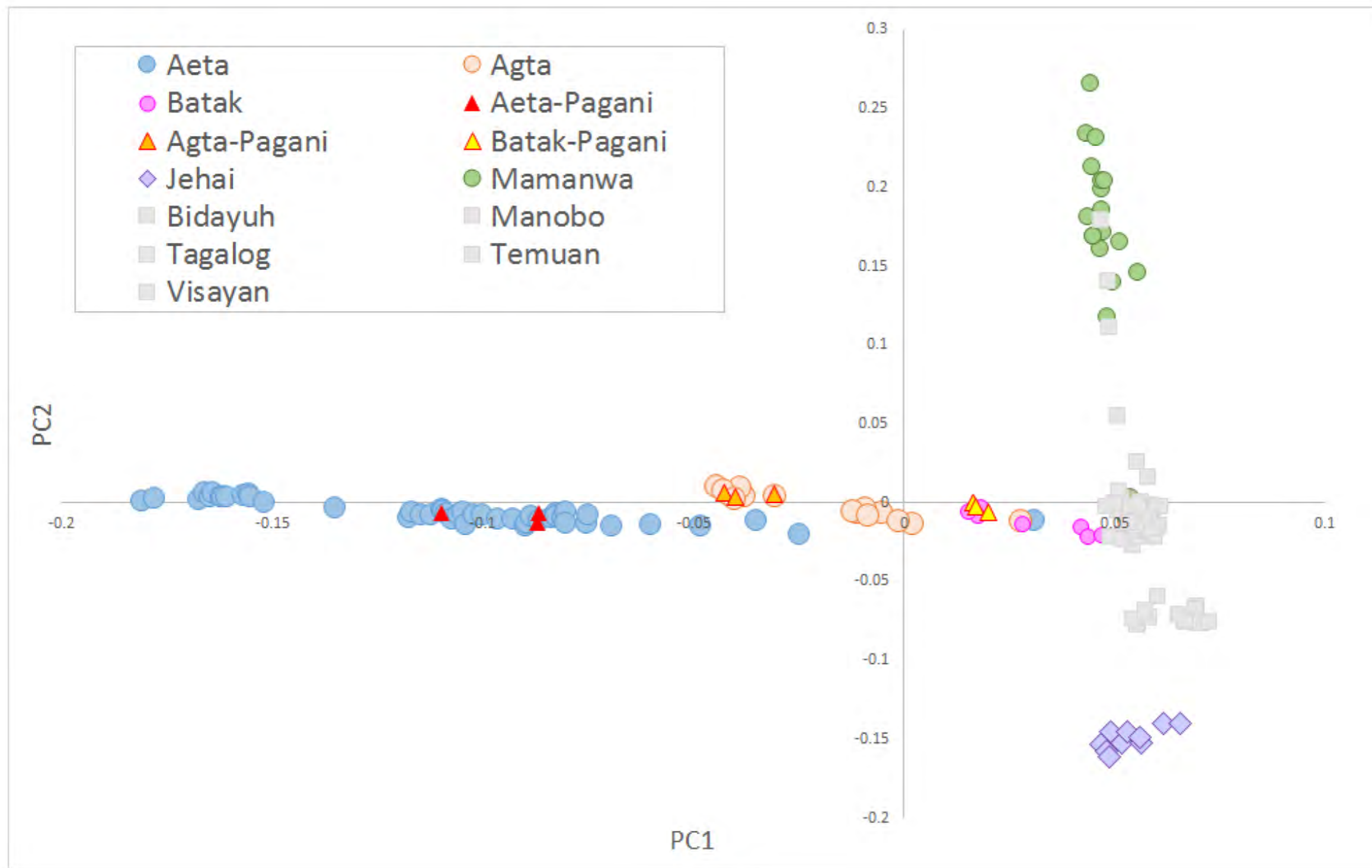


# 新人20集団とデニソワ人の系統ネットワーク



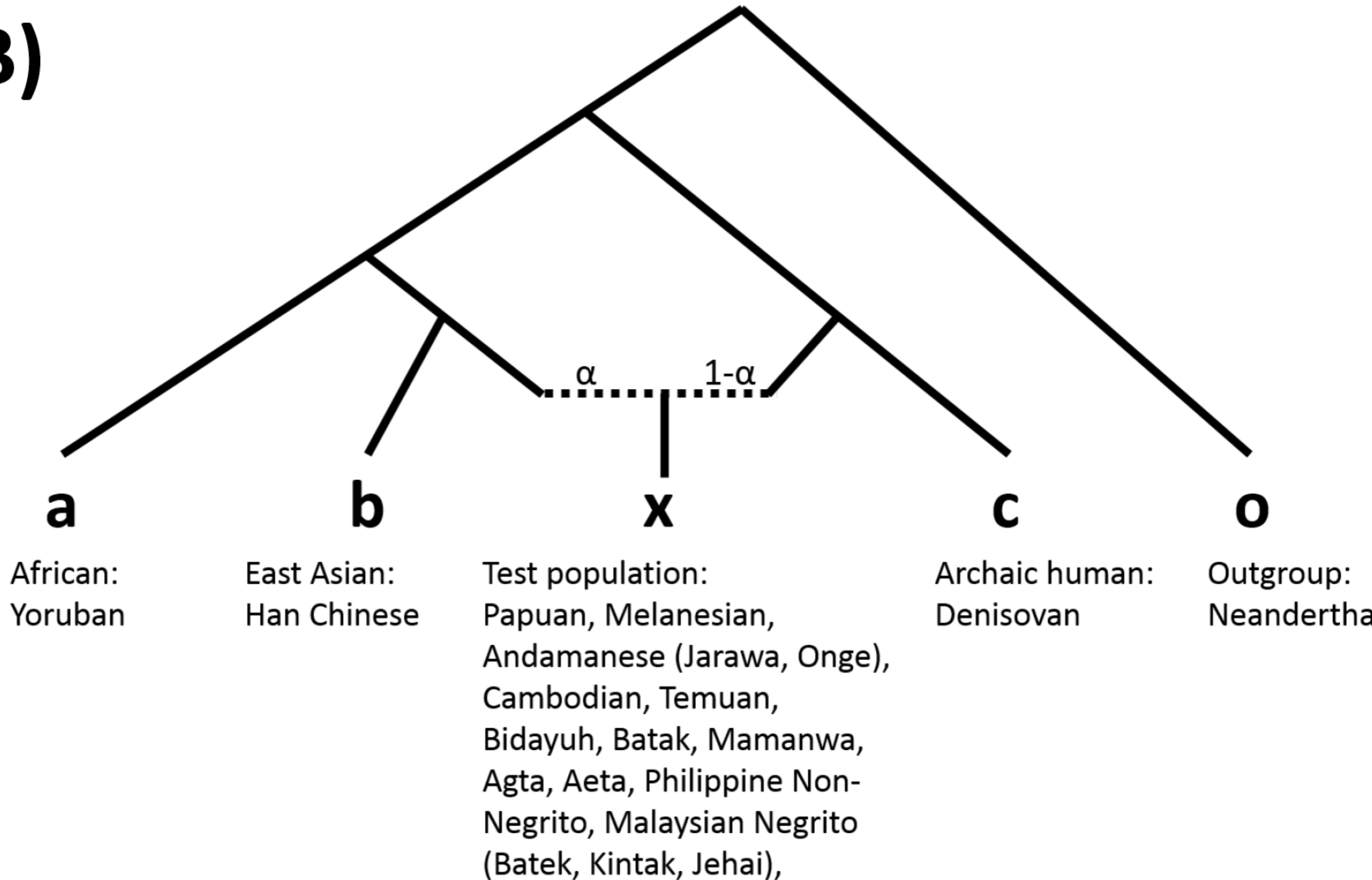


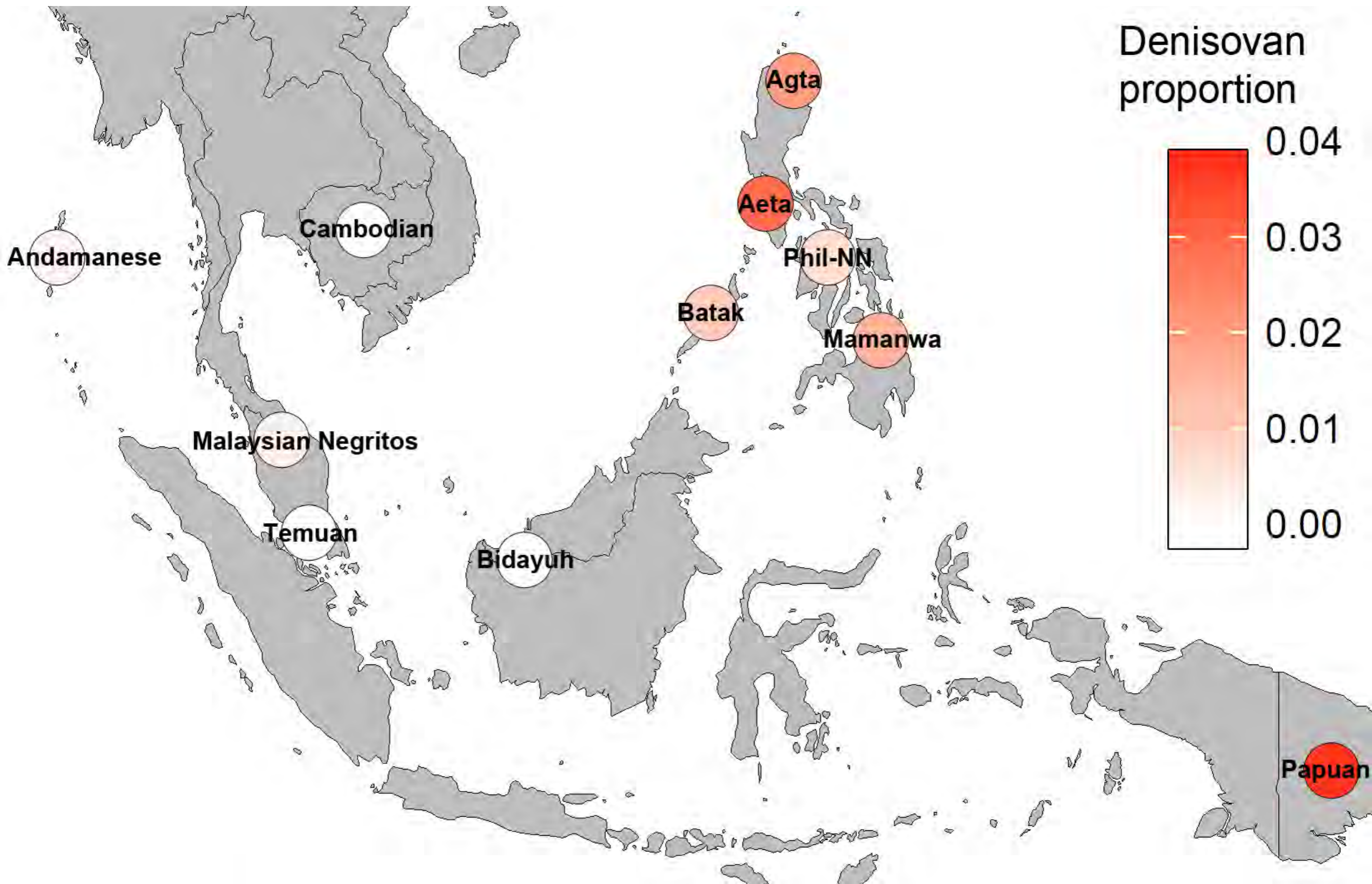
# Introgression of Denisovan DNAs to Papuans and Aeta Negritos

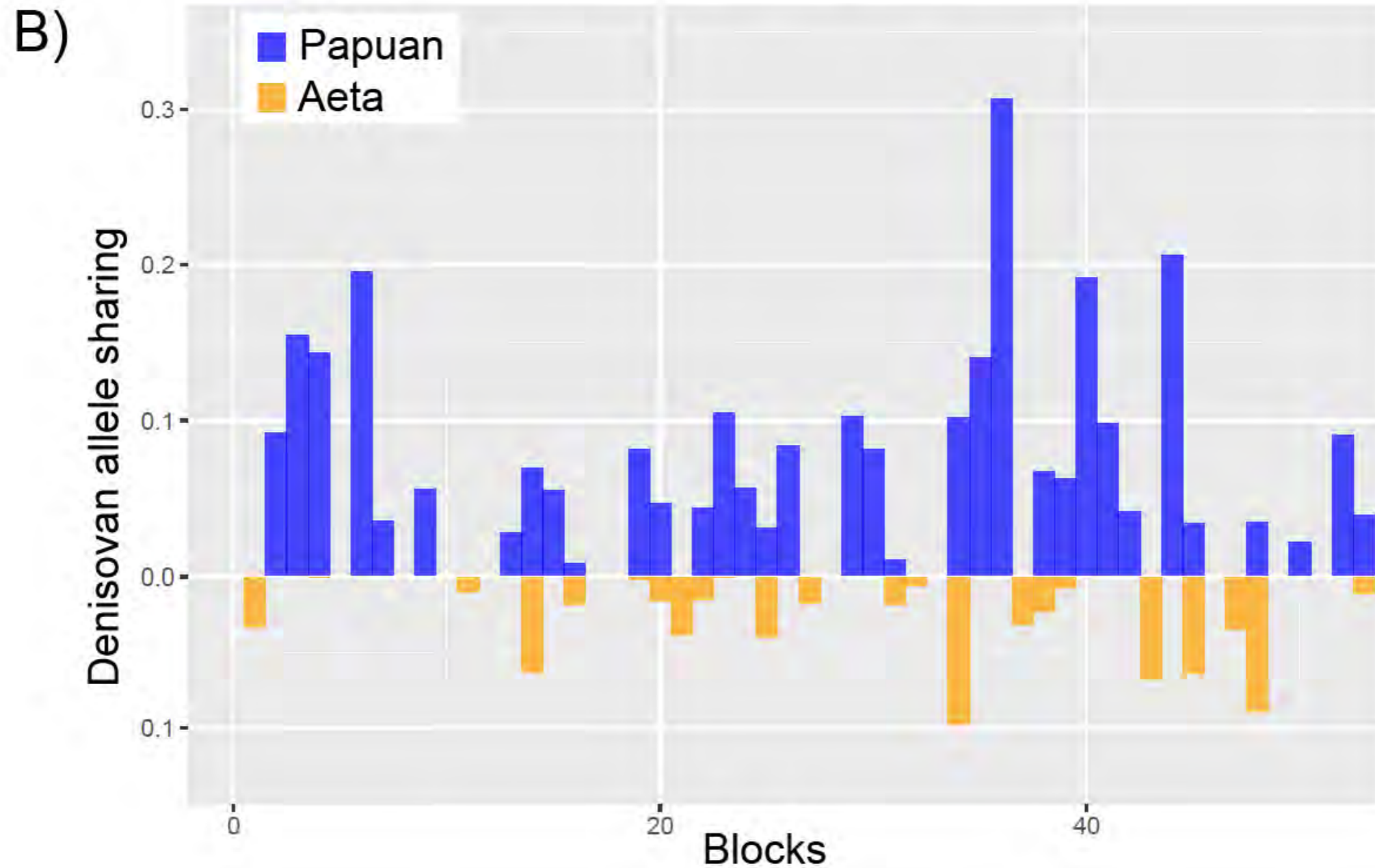
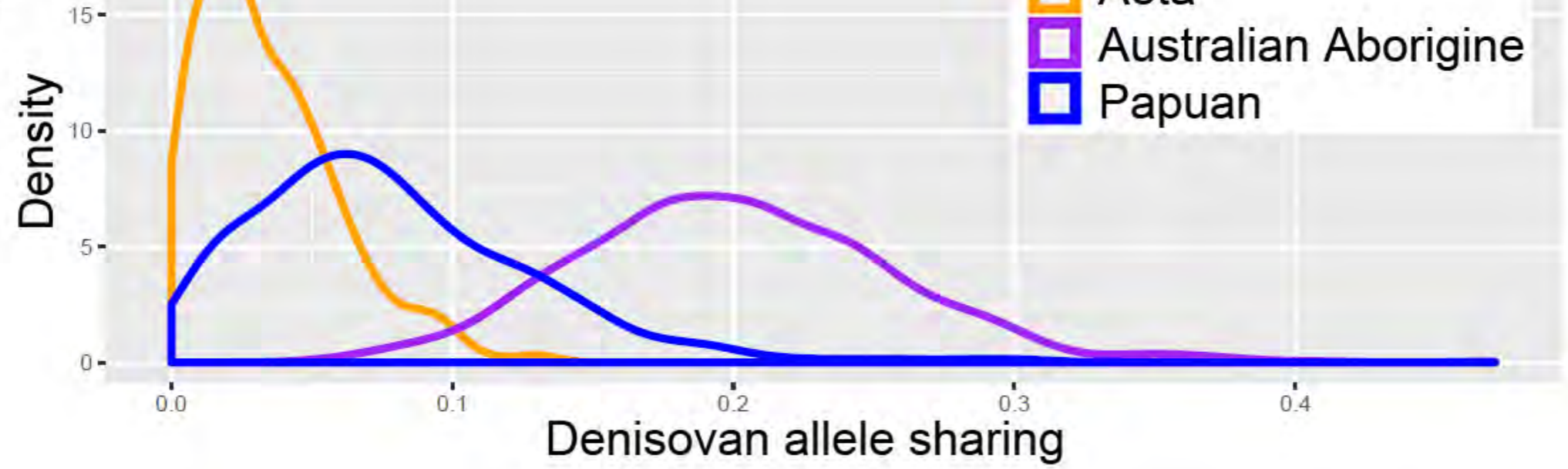


# Introgression test of Denisovan DNAs to Negritos

**B)**







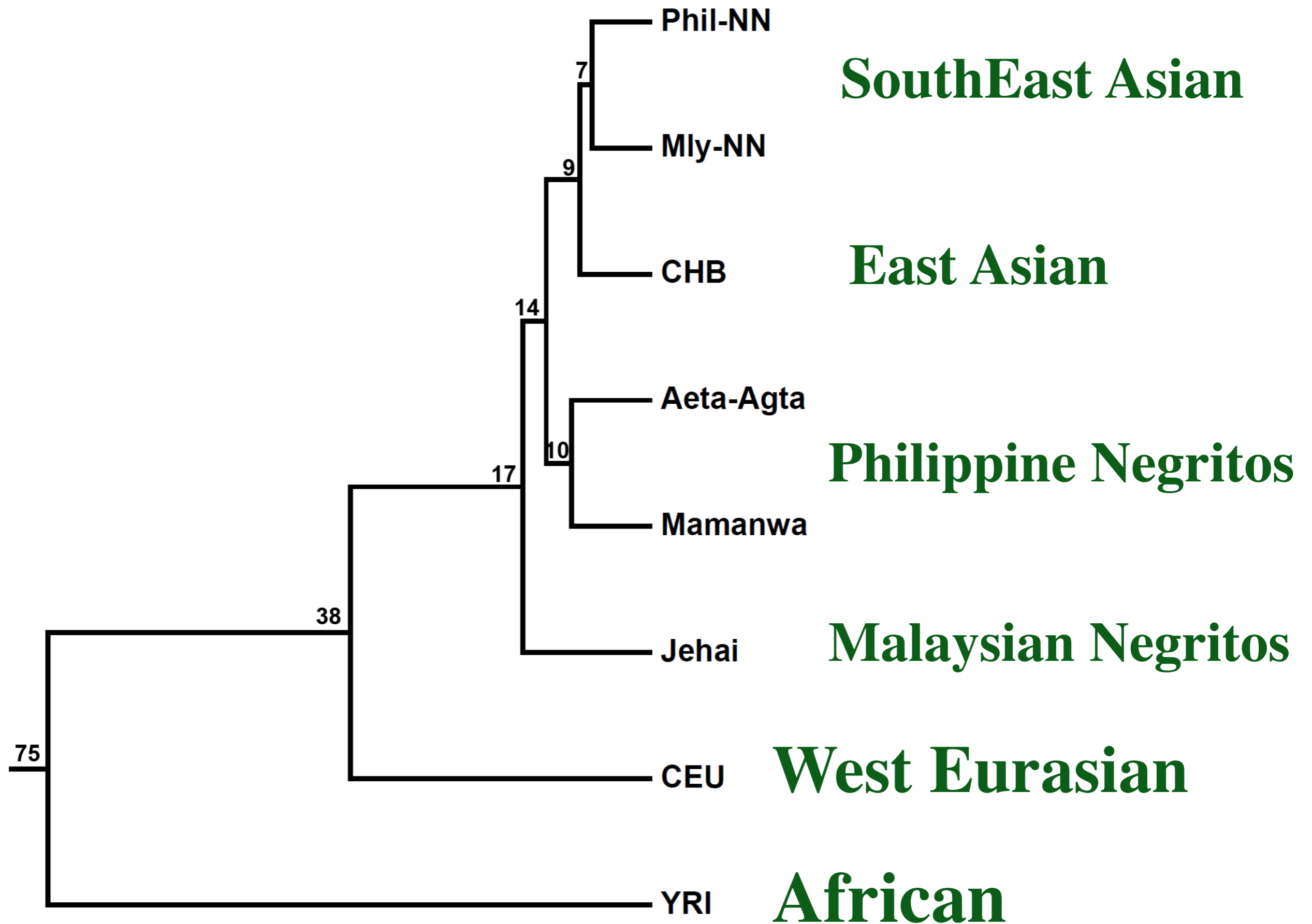
**Table S5:** Gene Ontology terms that are overrepresented in SNP loci that are shared among Negrito groups

<u>Biological process</u>	Fold enrichment	p-value
Cell communication (GO:0007154)	1.48	8.47E-11
Cellular process (GO:0009987)	1.26	7.42E-10
System development (GO:0048731)	1.75	1.83E-09
Single-multicellular organism process (GO:0044707)	1.63	8.18E-09
Multicellular organismal process (GO:0032501)	1.62	9.98E-09
Developmental process (GO:0032502)	1.48	1.89E-08
System process (GO:0003008)	1.7	2.53E-08
Nervous system development (GO:0007399)	1.88	9.10E-08
localization (GO:0051179)	1.42	9.11E-07
Regulation of molecular function (GO:0065009)	1.69	1.12E-06
Cell adhesion (GO:0007155)	2	1.17E-06
Ectoderm development (GO:0007398)	1.91	1.63E-06
Regulation of catalytic activity (GO:0050790)	1.69	1.85E-06
Transport (GO:0006810)	1.42	1.95E-06
Biological adhesion (GO:0022610)	1.96	2.00E-06
Neurological system process (GO:0050877)	1.66	6.33E-06
Cell-cell adhesion (GO:0016337)	2.17	7.50E-06

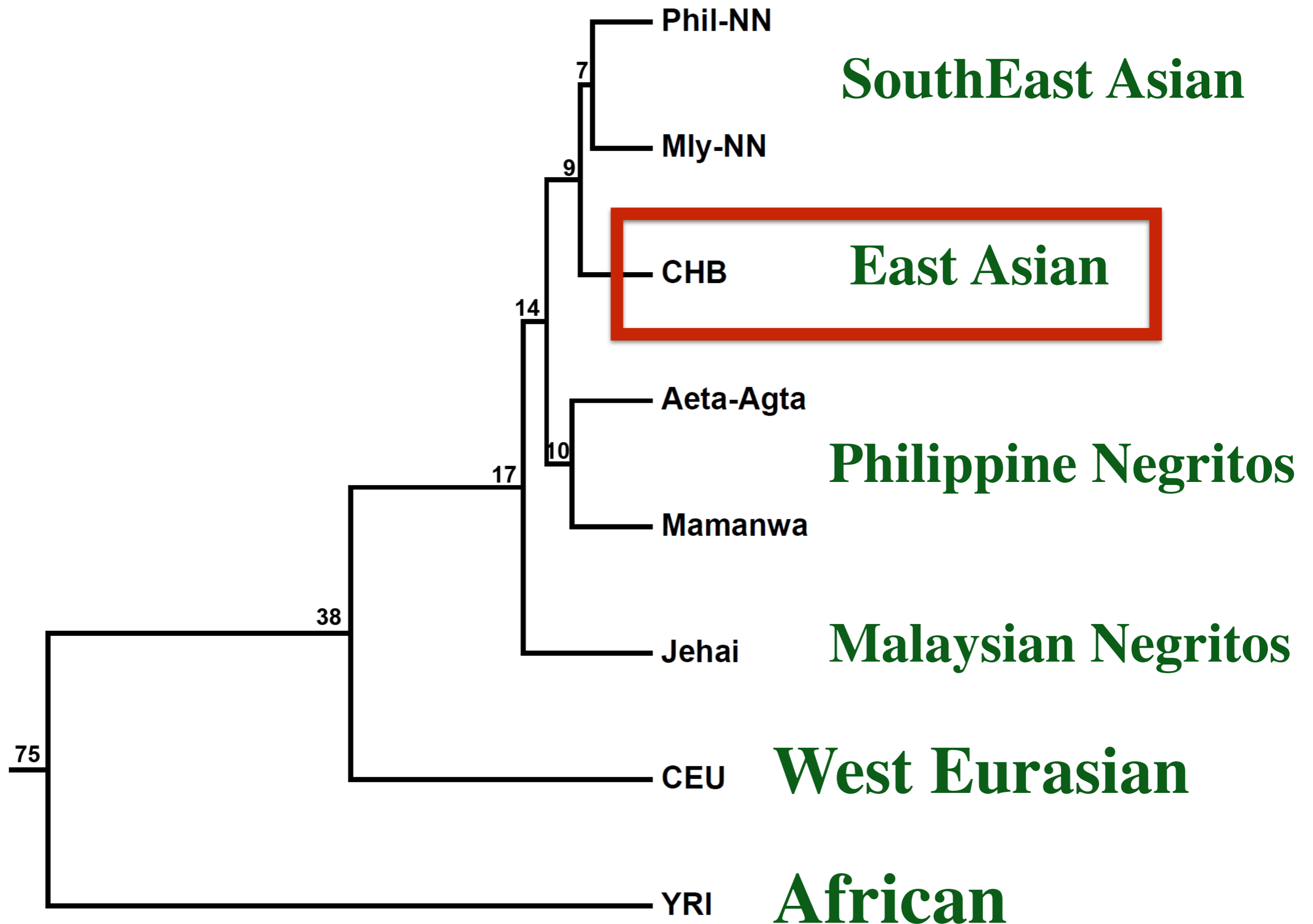
**Table S6:** Annotation of shared SNP loci among Negrito groups. SNPs associated with physical features and malarial resistance are listed.

Associated phenotype	rsID	Chr.	Among-Negrito mean Est	Negrito-CHB mean Est	Associated gene	SNP type
Skin pigmentation (Stokowaski et al. 2007)	rs1800414	15	0.0409	0.5884	OCA2	Synonymous
	rs6451050	5	0.0013	0.0714	SLC45A2	Intronic
Height (Wood et al. 2014)	rs939587	15	0.0133	0.3280	ACAN	Intronic
	rs1042631	15	0.0060	0.1663	ACAN	Synonymous
	rs11631646	15	0.0002	0.0181	ADAMTS17	Intronic
	rs4246299	15	0.0000	0.2473	ADAMTS17	Intronic
Facial and scalp features (Adhikari et al. 2016)	rs2276630	2	0.0011	0.0244	PAX3	Intronic
	rs1617445	6	0.0079	0.1122	PREP	Intronic
	rs3011704	10	0.0000	0.1692	GRID1	Intronic
	rs2140661	10	0.0000	0.1743	GRID1	Intronic
	rs1106789	10	0.0006	0.0249	GRID1	Intronic
	rs1880385	10	0.0013	0.1000	GRID1	Intronic
	rs11201909	10	0.0022	0.1754	GRID1	Intronic
	rs2607839	10	0.0026	0.3160	GRID1	Intronic
Malarial resistance (Liu et al. 2015)	rs2243270	5	0.0049	0.0818	IL4	Intronic
	rs1564029	16	0.0047	0.0742	CDH13	Intronic
	rs4783293	16	0.0000	0.2995	CDH13	Intronic
	rs4508407	16	0.0005	0.1495	CDH13	Intronic
	rs6565153	16	0.0013	0.1109	CDH13	Intronic
	rs1125244	16	0.0094	0.2059	CDH13	Intronic
	rs734888	16	0.0146	0.1919	CDH13	Intronic
	rs1364309	16	0.0100	0.3120	CDH13	Intronic
	rs9935086	16	0.0010	0.0683	CDH13	Intronic
	rs735674	16	0.0057	0.0764	CDH13	Intronic
rs2326024	16	0.0053	0.1545	CDH13	Intronic	

# Divergence time estimates based on SNP data

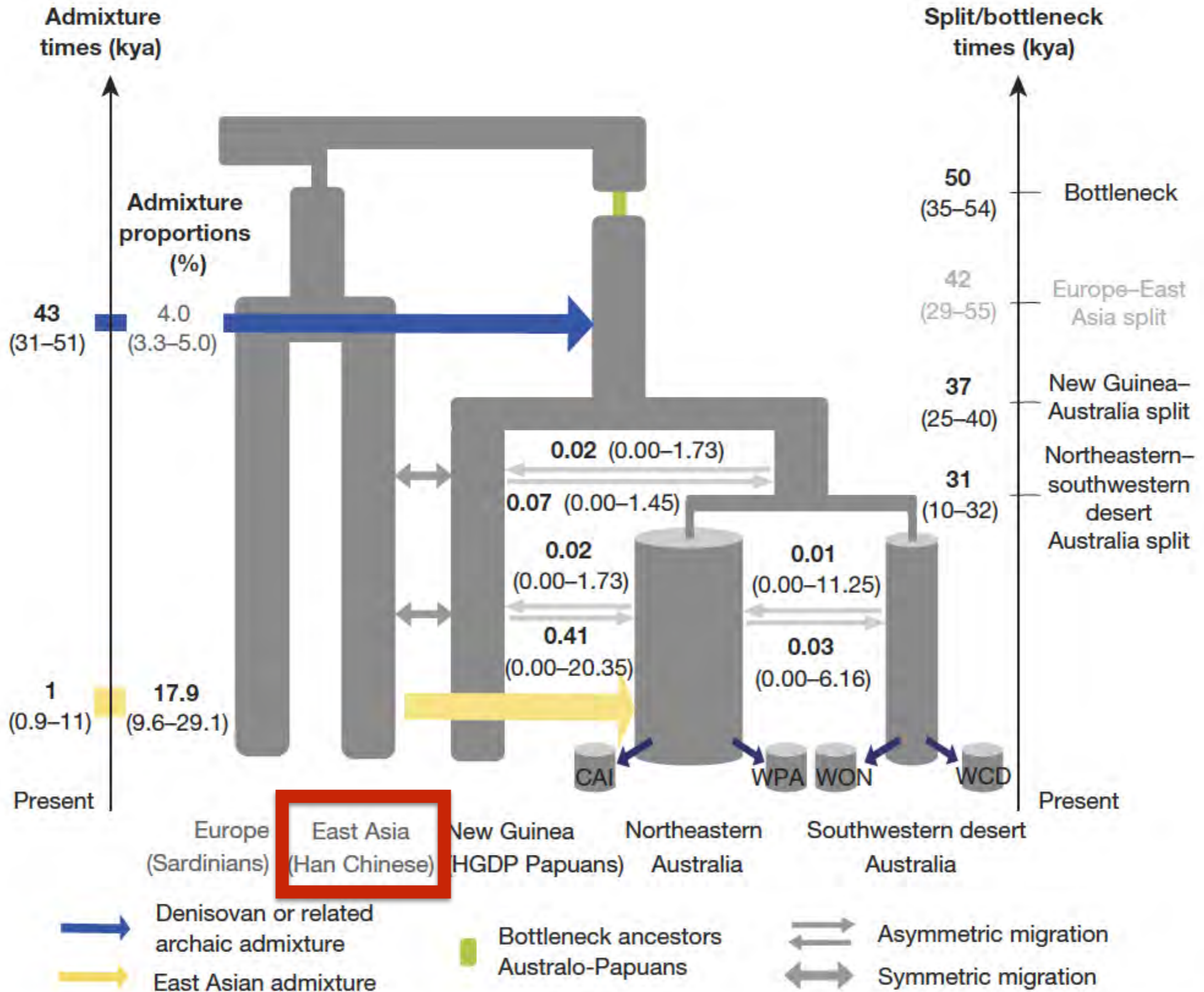


# Divergence time estimates based on SNP data





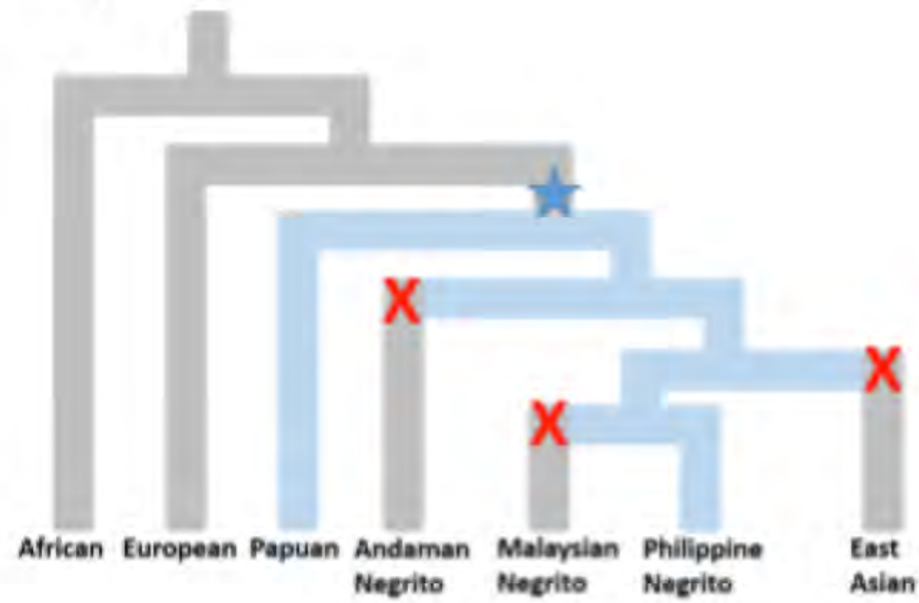
# From Malaspinas et al. (2016)



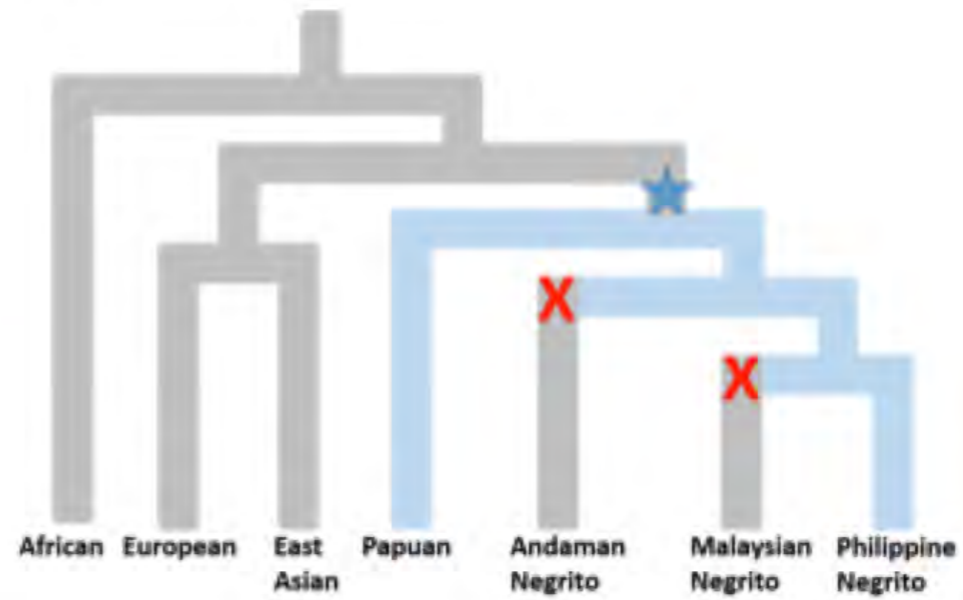
**Figure 3 | Settlement of Australia.** Best supported demographic model of

# Four Possible Scenarios of Denisovan Introgression in Modern Humans

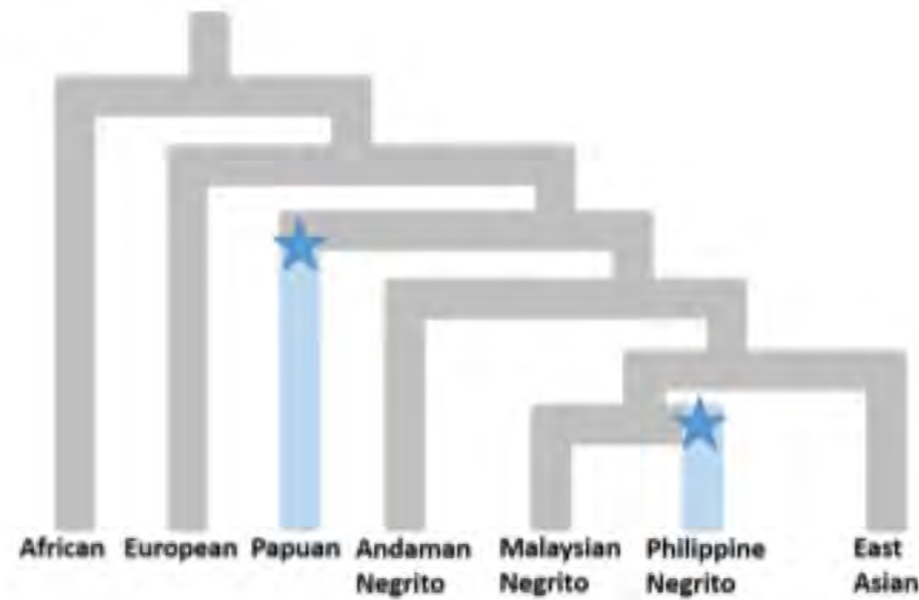
A)



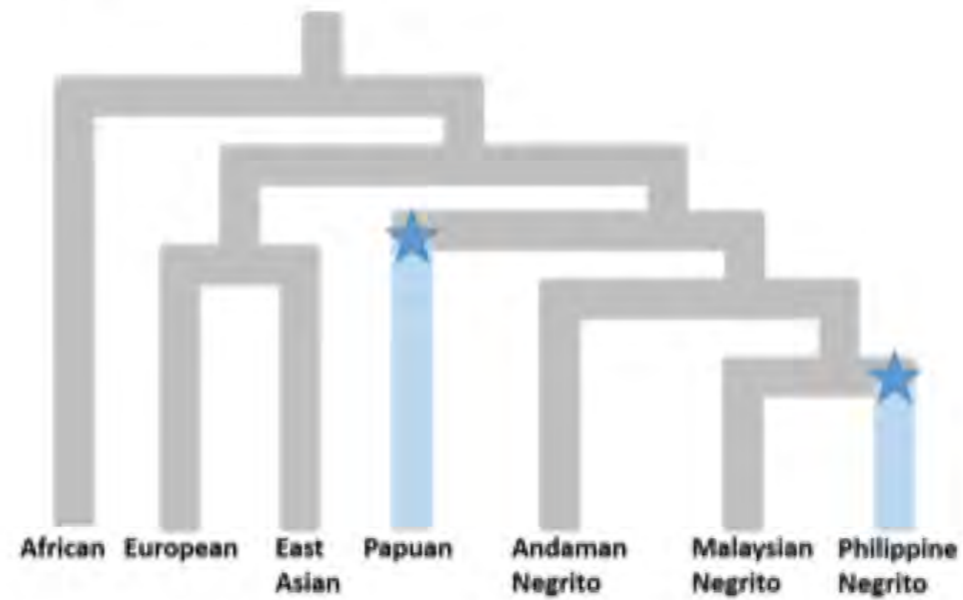
B)



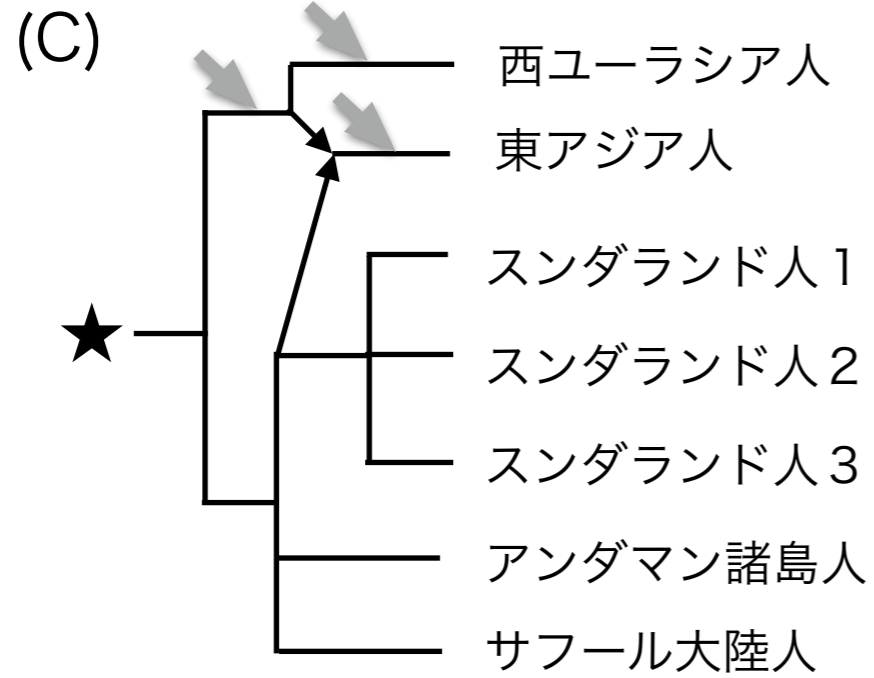
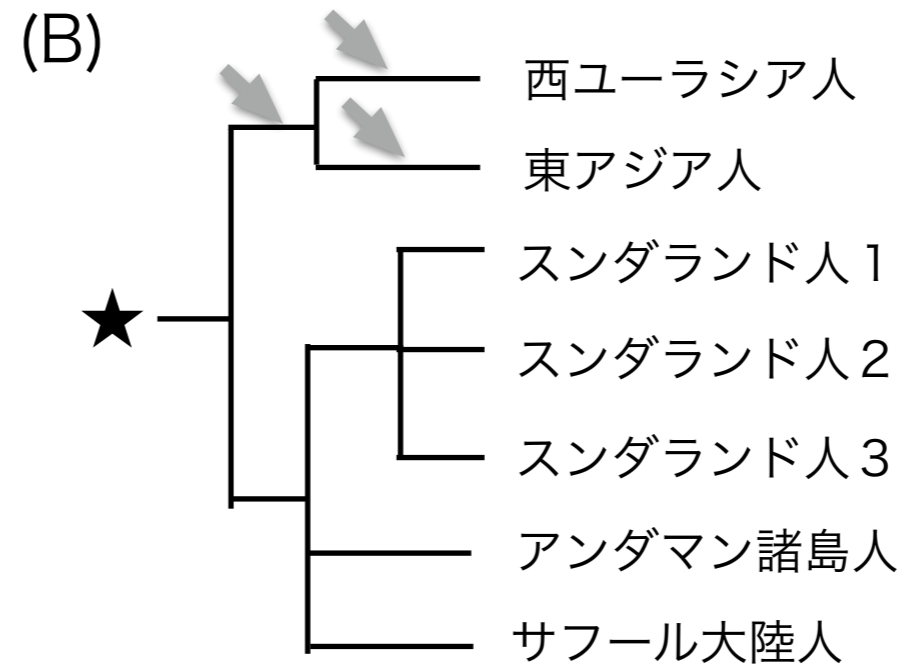
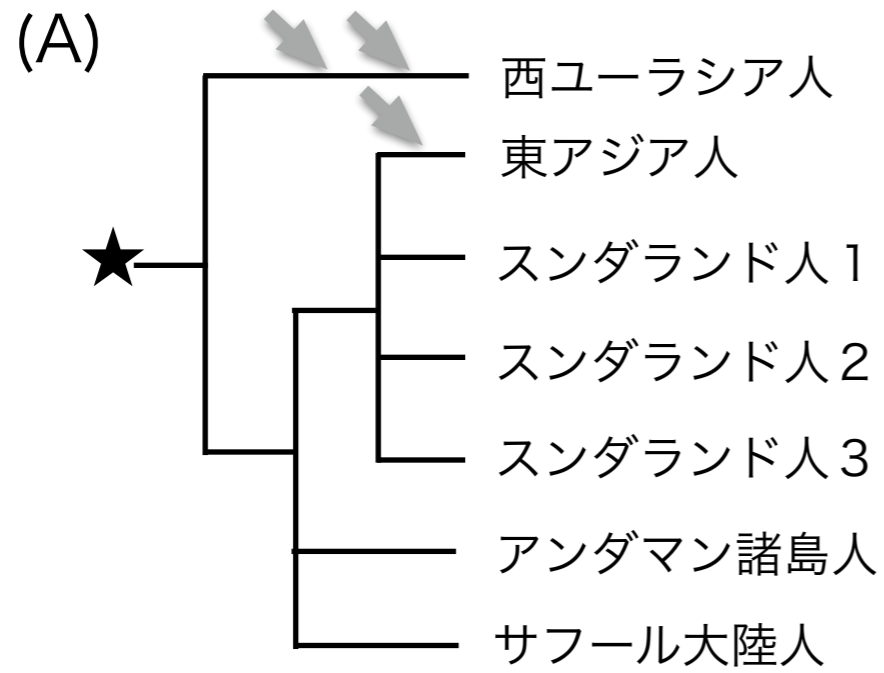
C)



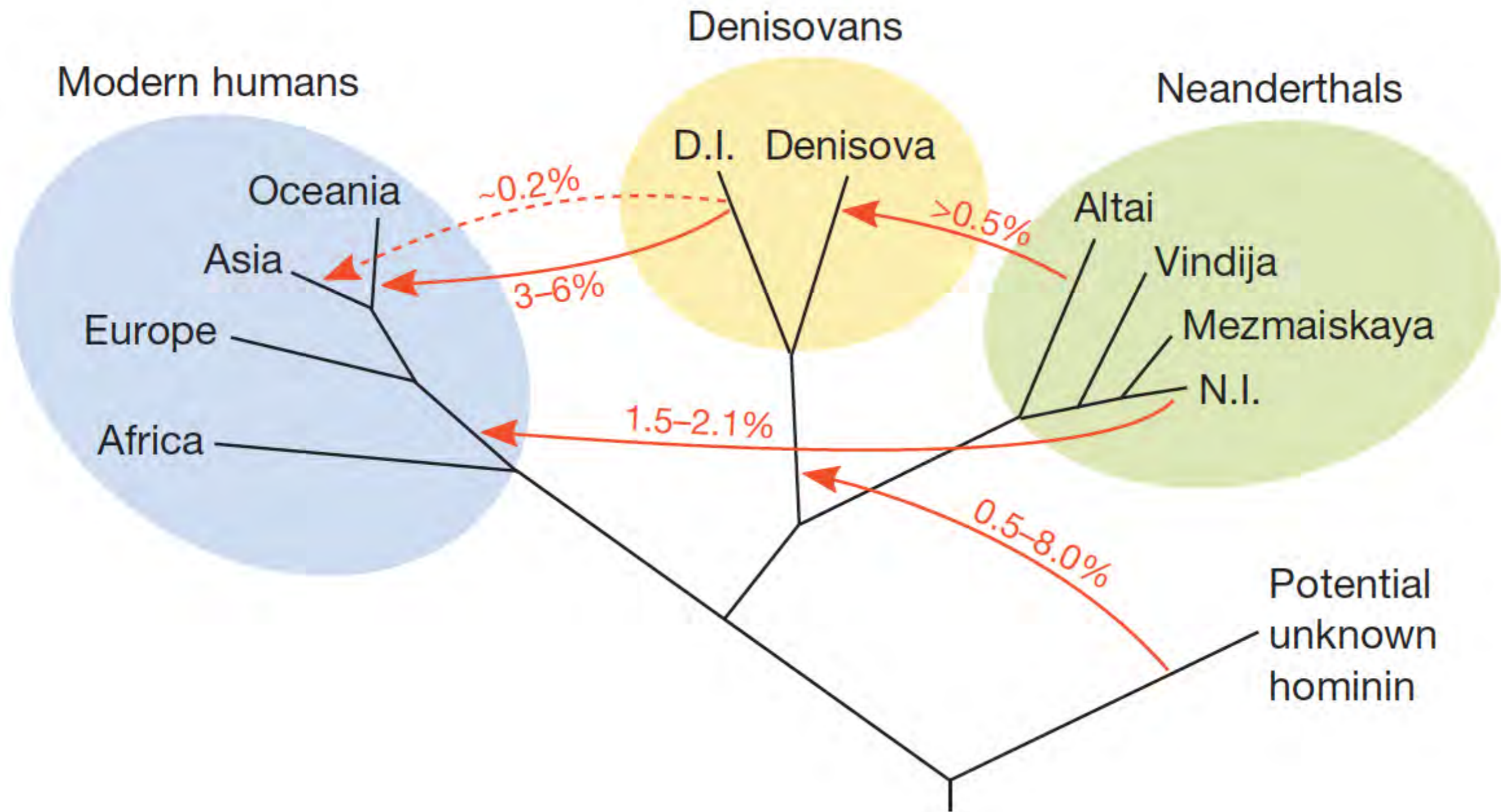
D)



# 東ユーラシア人形成の3モデル

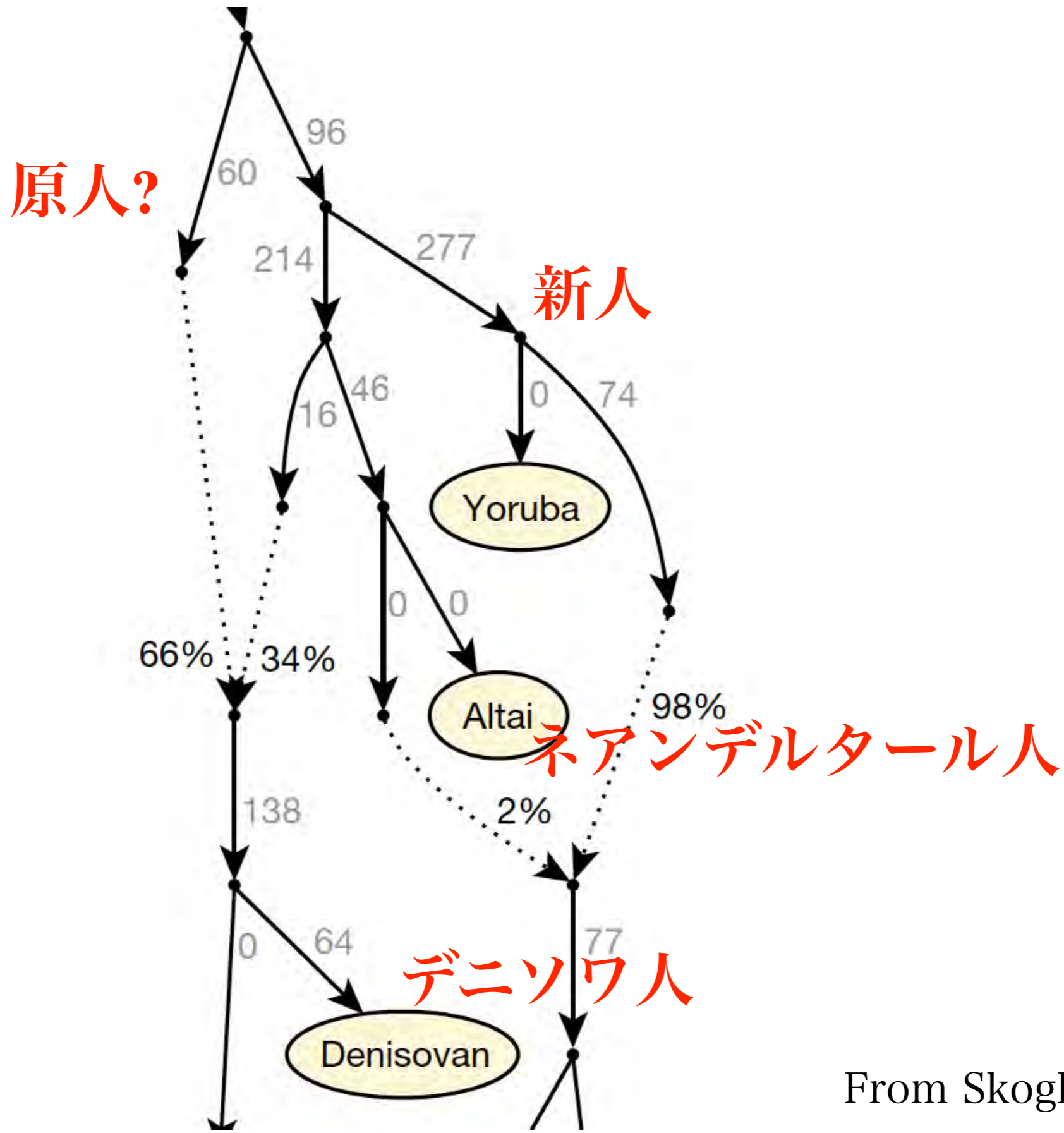


# 重層的な混血が推定される人類進化 (1)



From Prüfer et al. (2008)

# 重層的な混血が推定される人類進化 (2)





アイヌ人

本土人

琉球人



多原かおりさん



故萱野茂氏



酒井美直さん



杉本彩さん



根井正利先生



斎藤のはら



宮里藍さん



下地幹郎氏



南沙織さん

*Japan Review*, 1991, 2: 1-33

# **Dual Structure Model for the Population History of the Japanese**

HANIHARA, Kazuro

*International Research Center for Japanese Studies, Kyoto, Japan*



Photo taken by Saitou, 2000

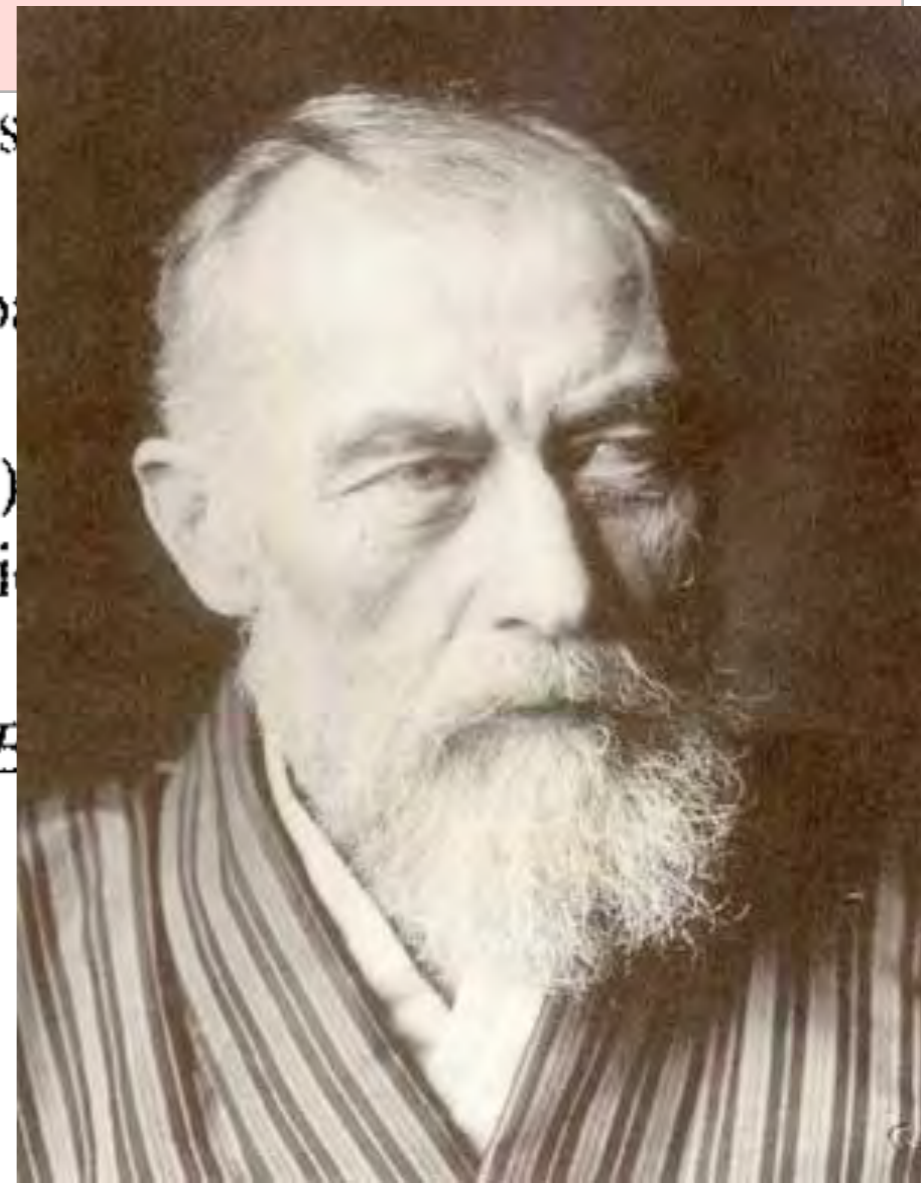
anthropology of the Japanese population. Erwin von Baelz, a pathologist from Germany, found that Japanese could be classified into two different physical types on the basis of somatometry. He called them *Satsuma* and *Choshu* types (Baelz, 1883, 1885, 1901; dual physique theory). *Satsuma* and *Choshu* are, respectively, the old names of Kagoshima and Yamaguchi Prefectures of today. Von Baelz described the *Satsuma* type as being characterized by a broad and low face, large eye openings, double eyelids, wider nose, thick lips and ear lobes, and short stature. The *Choshu* type showed a longer face, narrower eye openings and nose, single eyelids, thin lips and ear lobes, slender body build, and taller stature. He also pointed out that the *Satsuma* type was common in Japanese and the *Choshu* type was predominant in aristocratic families, though small in number. Von Baelz also emphasized the common origin of Ainu and Ryukyus (Okinawa islanders) on the basis of somatological evidence (1911; Ainu-Ryukyu common origin theory).

Edward S. Morse, a zoologist from Boston, discovered in 1877 a shell mound at Omori, Tokyo, and carried out an excavation the same year. This was the first scientific excavation of an archaeological site in Japan. Cultural as well as human skeletal remains unearthed there are now stored at the University of Tokyo as nationally important cultural properties. In his paper on the Omori Shell Mound, the first scientific report published in Japan, Morse (1879) stated, on the basis of comparisons with Amerindian sites, that the Neolithic people had been replaced by



## REFERENCES

- Anutschin, D. (1876): Materialien zur Anthropologie Ost-Asiens. I. Der Ainostamm. Moskau. (cited by Koganei, 1893).
- Baelz, E. von (1883, 1885): Die körperliche Eigenschaften der Japaner. *Mitt. Deutsch. Ges. Natur u. Völkerk. Ostasiens*, **28**:330–359, **32**: 35–103.
- Baelz, E. von (1901): Menschen-Rassen Ost-Asiens mit specieller Rücksicht auf Japan. *Zschr. Ethnol.*, **33**: 166–189, 202–207, 245–249, 393–394.
- Baelz, E. von (1911): Die Riu-Kiu-Insulaner, die Aino und andere kaukasier-ähnliche Reste in Ostasien. *Korres. Blatt. Dtsch. Ges. Anthrop. Ethnol. Urgesch.*, **42**: 187–191.
- Bickmore, A. S. (1868): The Ainos, or hairy men of Yeso. *Trans. Ethnol. Soc. Lond.*, **1**: 353–361.
- Brace, C. L. and Nagai, M. (1982): Japanese tooth size, population and race. *Am. J. Phys. Anthropol.*, **59**: 399–411.
- Brace, C. L., Brace, M. L. and Leonard, W. R. (1989): Japan: A multivariate craniofacial and odontometric study. *Am. J. Phys. Anthropol.*, **78**: 93–113.
- Busk, G. (1868): Description of an Aino-skull. *Trans. Ethnol. Soc. Lond.*, **1**: 109–111.

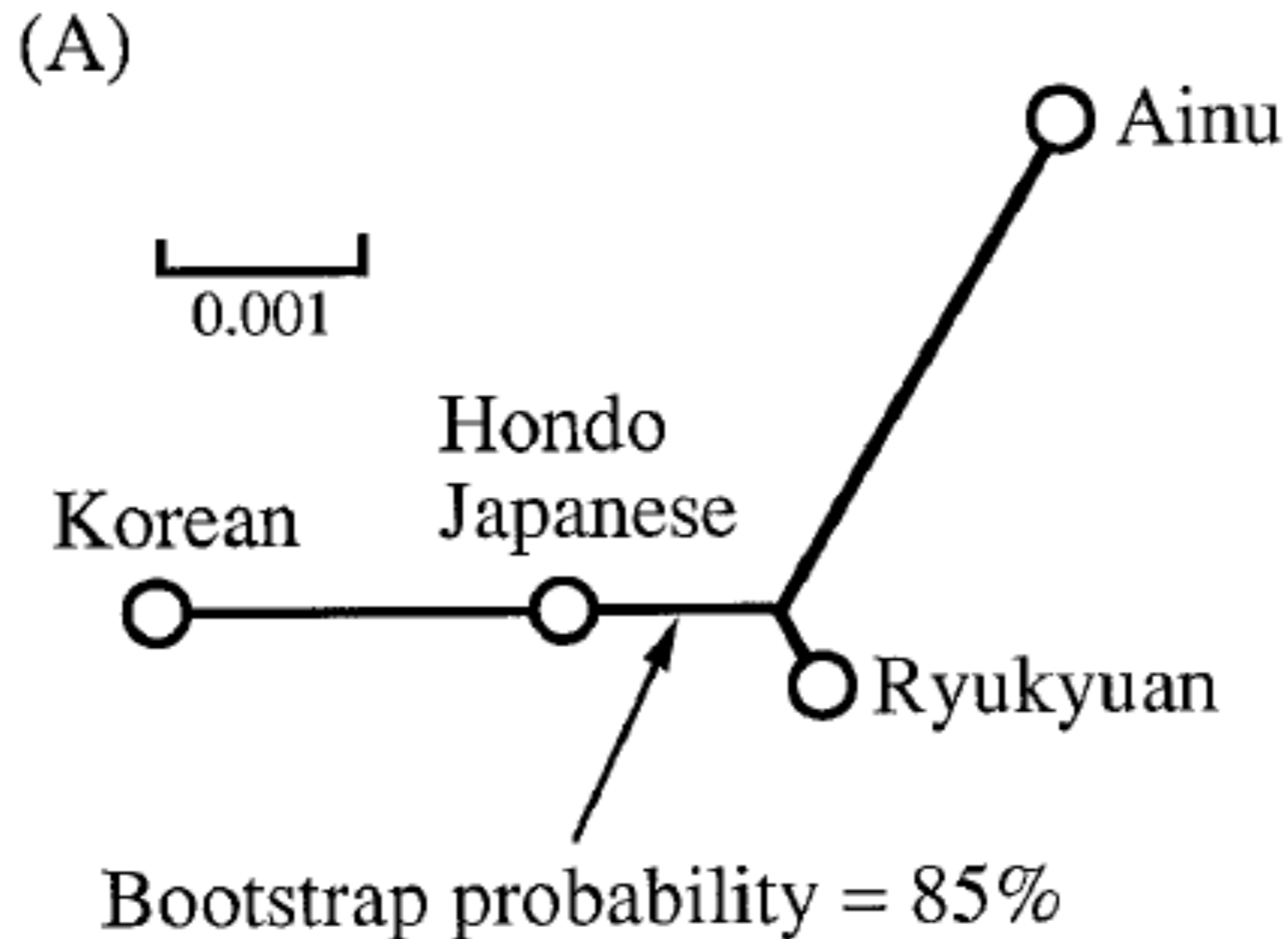


## Genetic Origins of the Japanese: A Partial Support for the Dual Structure Hypothesis

KEIICHI OMOTO<sup>1\*</sup> AND NARUYA SAITOU<sup>2</sup>

<sup>1</sup>*International Research Center for Japanese Studies, Kyoto, 610-11 Japan;*

<sup>2</sup>*National Institute of Genetics, Mishima, 411 Japan*



ORIGINAL ARTICLE

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# The history of human populations in the Japanese Archipelago inferred from genome-wide SNP data with a special reference to the Ainu and the Ryukyuan populations

Japanese Archipelago Human Population Genetics Consortium (Consortium members: Timothy Jinam<sup>1,18</sup>,

# Consortium Members

Timothy Jinam<sup>1</sup>, Nao Nishida<sup>2,+</sup>, Momoki Hirai<sup>3,+</sup>, Shoji Kawamura<sup>3,+</sup>, Hiroki Oota<sup>3,+</sup>, Kazuo Umetsu<sup>4,+</sup>, Ryosuke Kimura<sup>2,+</sup>, Jun Ohashi<sup>2,+</sup>, Atsushi Tajima<sup>5,+</sup>, Toshimichi Yamamoto<sup>6,+</sup>, Hideyuki Tanabe<sup>7,+</sup>, Shuheito Mano<sup>8,+</sup>, Yumiko Suto<sup>9,+</sup>, Tadashi Kaname<sup>10</sup>, Kenji Naritomi<sup>10</sup>, Kumiko Yanagi<sup>10</sup>, Norio Niikawa<sup>11</sup>, Naruya Saitou<sup>1,12,+,\*</sup>, Keiichi Omoto<sup>13,+,\*</sup>, and Katsushi Tokunaga<sup>2,+,\*</sup>

- 1) Dept. Genetics, School of Life Science, SOKENDAI
- 2) Dept. Human Genetics, Graduate School of Medicine, University of Tokyo
- 3) Dept. Integrated Bioscience, Graduate School of Frontier Sciences, University of Tokyo
- 4) Dept. Forensic Science, Yamagata University School of Medicine
- 5) Dept. Human Genetics and Public Health, The University of Tokushima Graduate School
- 6) Dept. Legal Medicine and Bioethics, Nagoya University School of Medicine
- 7) Dept. Evolutionary Studies of Biosystems, School of Advanced Sciences, SOKENDAI
- 8) Institute of Statistical Mathematics
- 9) National Institute of Radiological Sciences
- 10) Dept. Genetic Medicine, Graduate School of Medicine, University of the Ryukyus
- 11) Health Sciences University of Hokkaido
- 12) Dept. Biological Sciences, School of Science, University of Tokyo
- 13) Dept. Anthropology, Faculty of Science, University of Tokyo

+ Members of "Asian Archival DNA Repository Consortium"

\* Corresponding Authors

**Supplementary Table 1:** SNP filtering applied to the Japanese and HapMap dataset

Population	No. samples	Number of SNPs omitted		Remaining SNP
		Genotyping call rate (<95%)	HWE (p<0.001)	
Ainu	36	212,448 <sup>†</sup>	449	655,788
Ryukyu	35	29,874	538	837,845
Mainland Japanese	198	17,169	1888	849,200
CHB	42	3,069	336	864,852
JPT	45	5,004	446	862,807
CEU	89	4,887	514	862,856
YRI	89	4,780	706	862,771

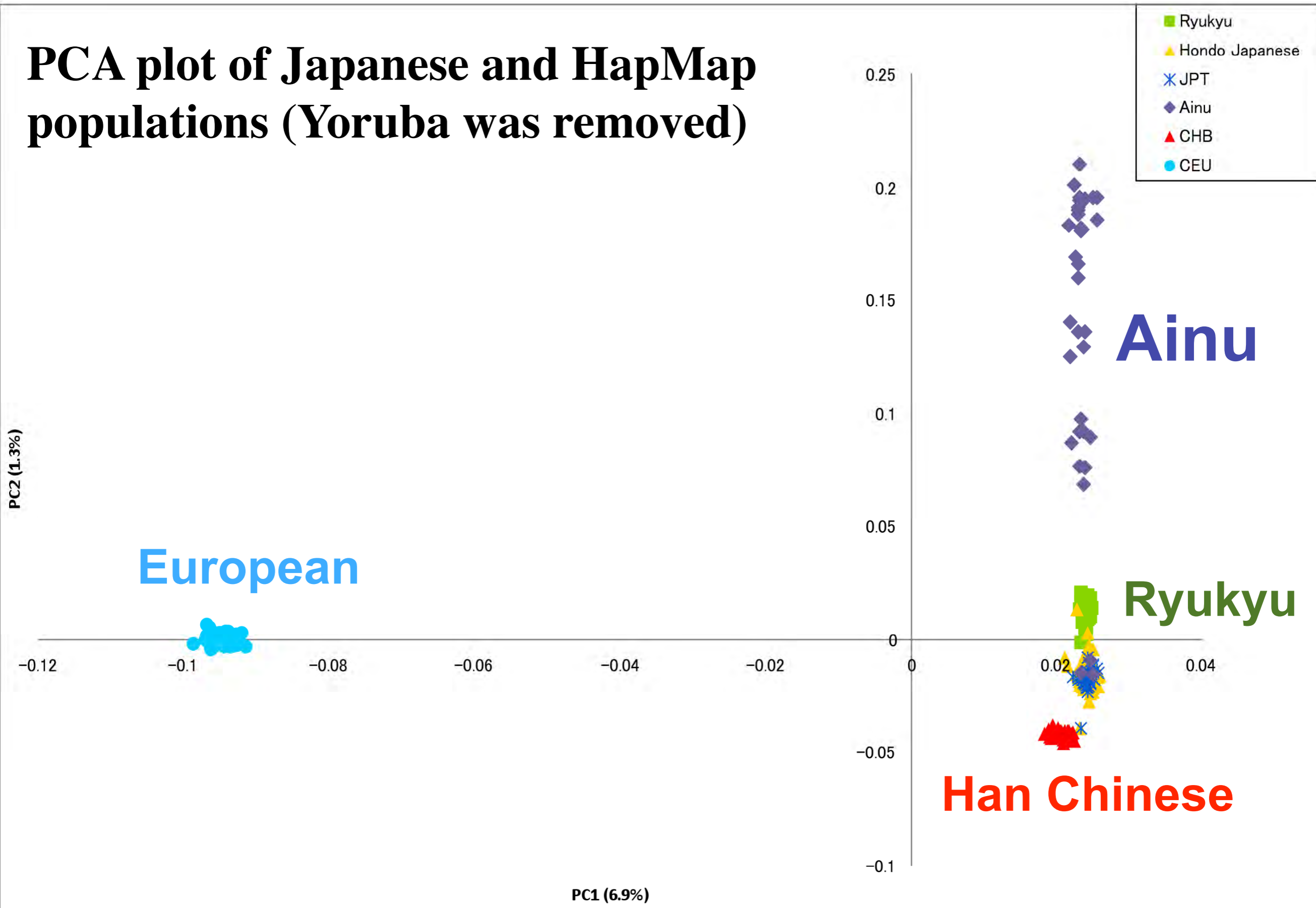
<sup>†</sup>Includes SNP omitted based on confidence scores

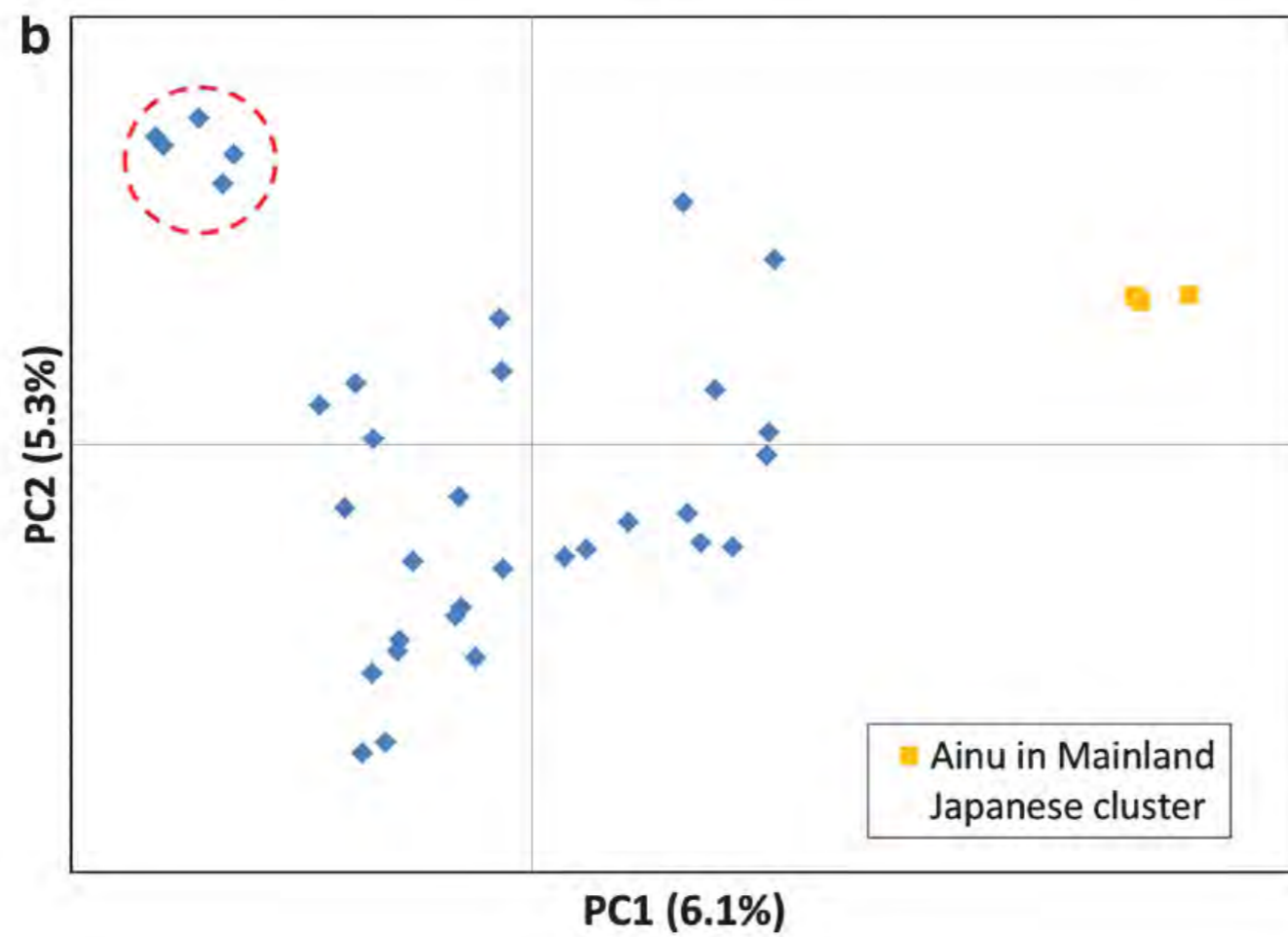
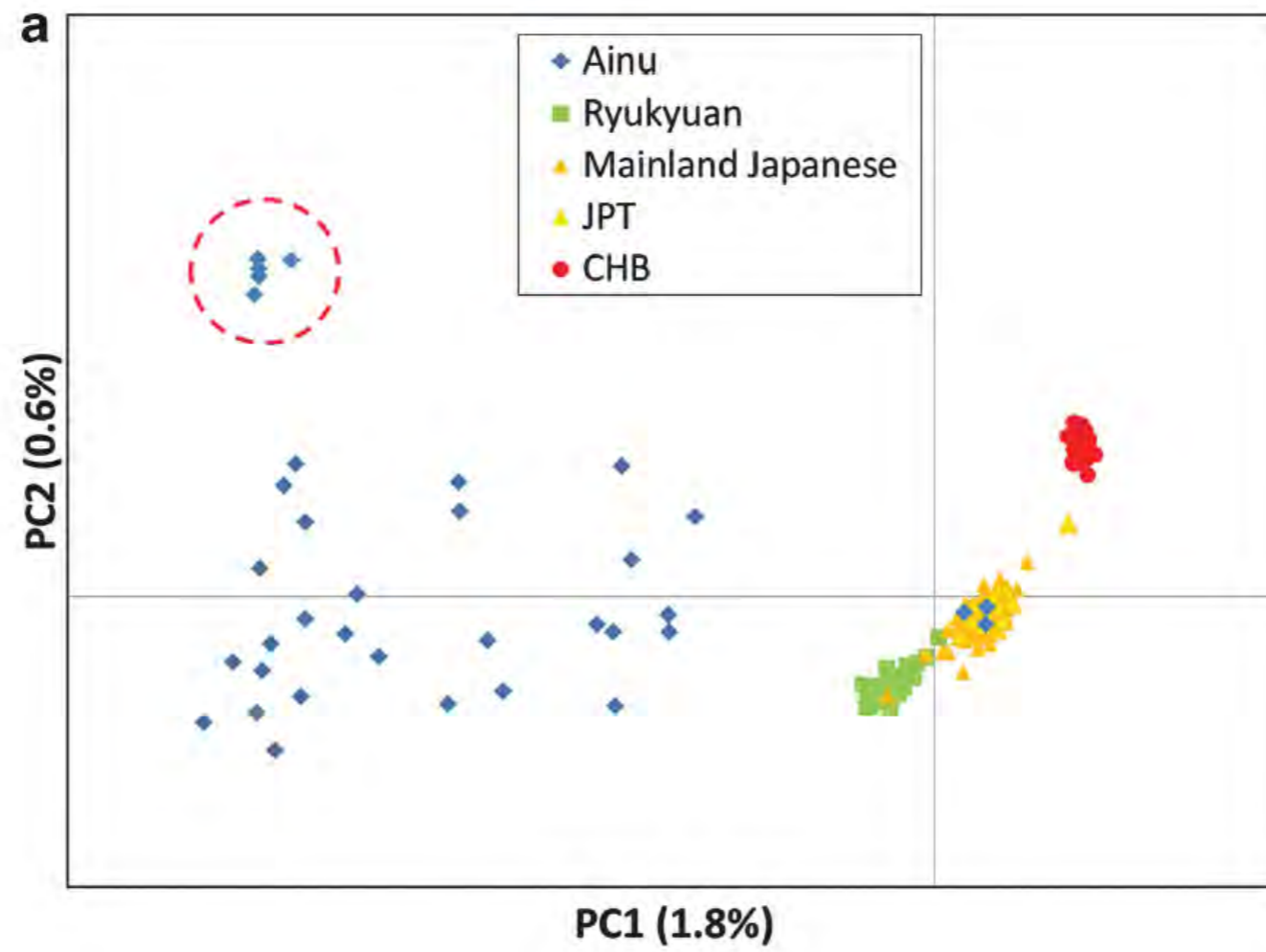
# PC1 vs PC2



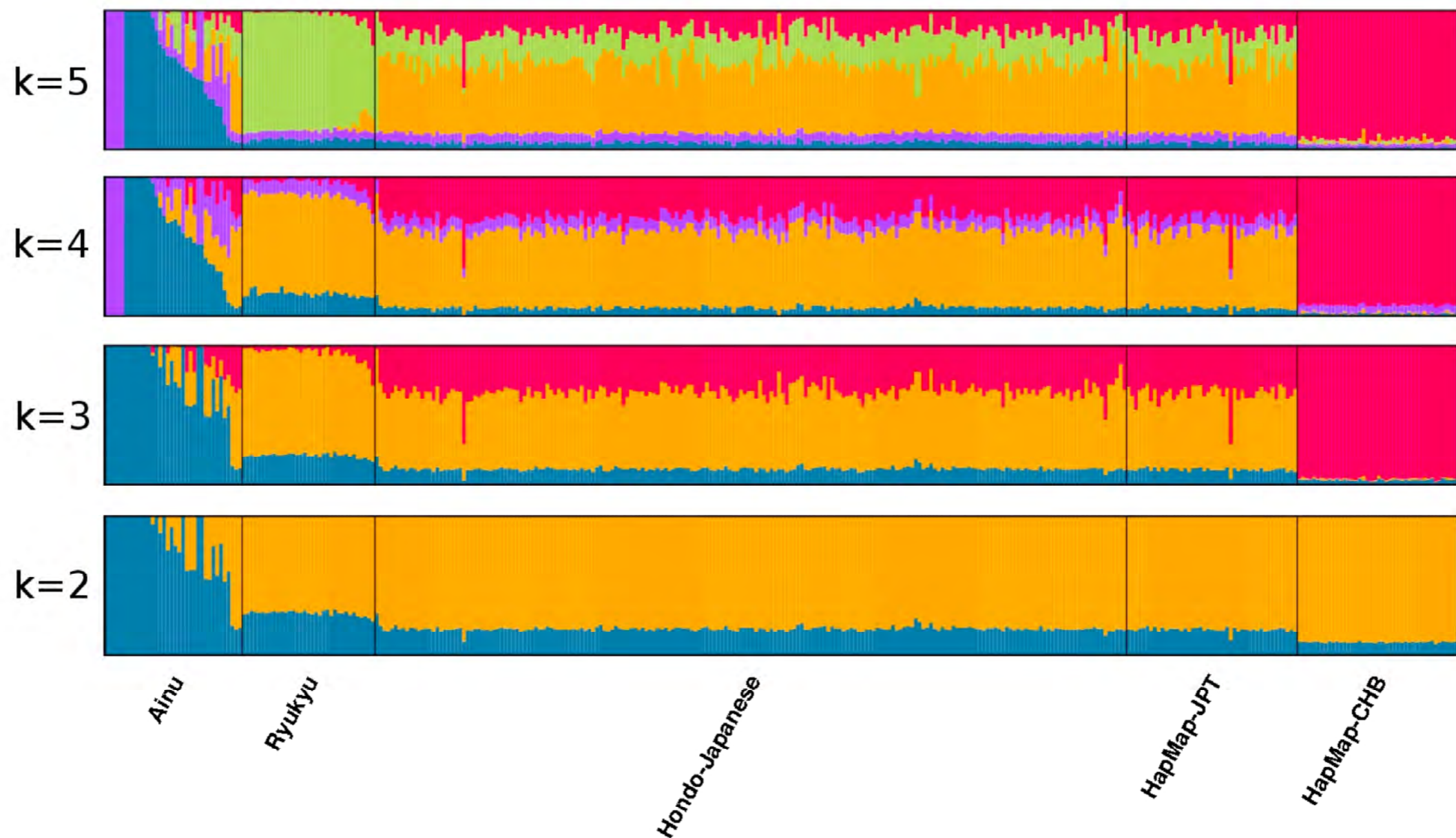
# PCA plot of Japanese and HapMap populations

# PCA plot of Japanese and HapMap populations (Yoruba was removed)

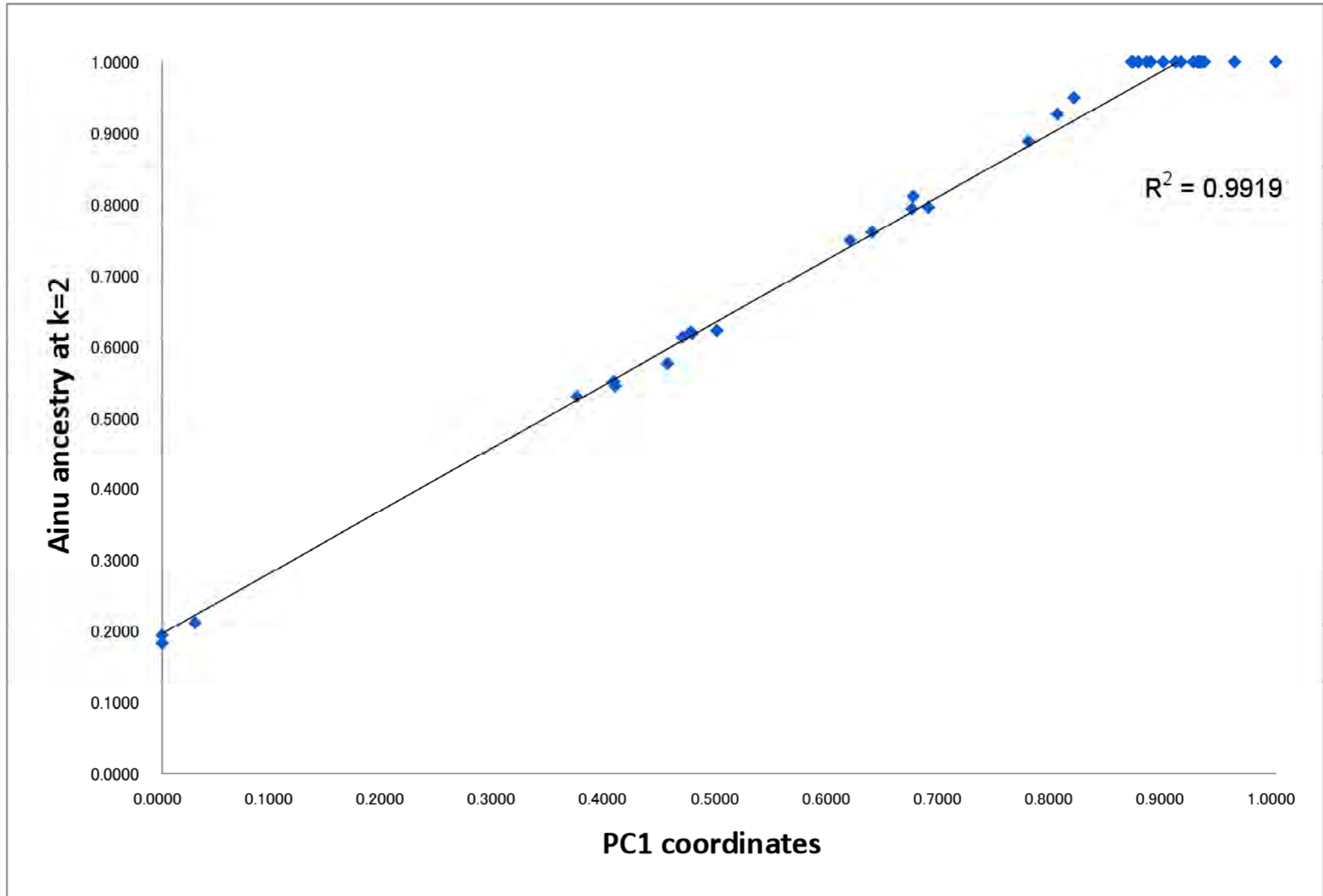






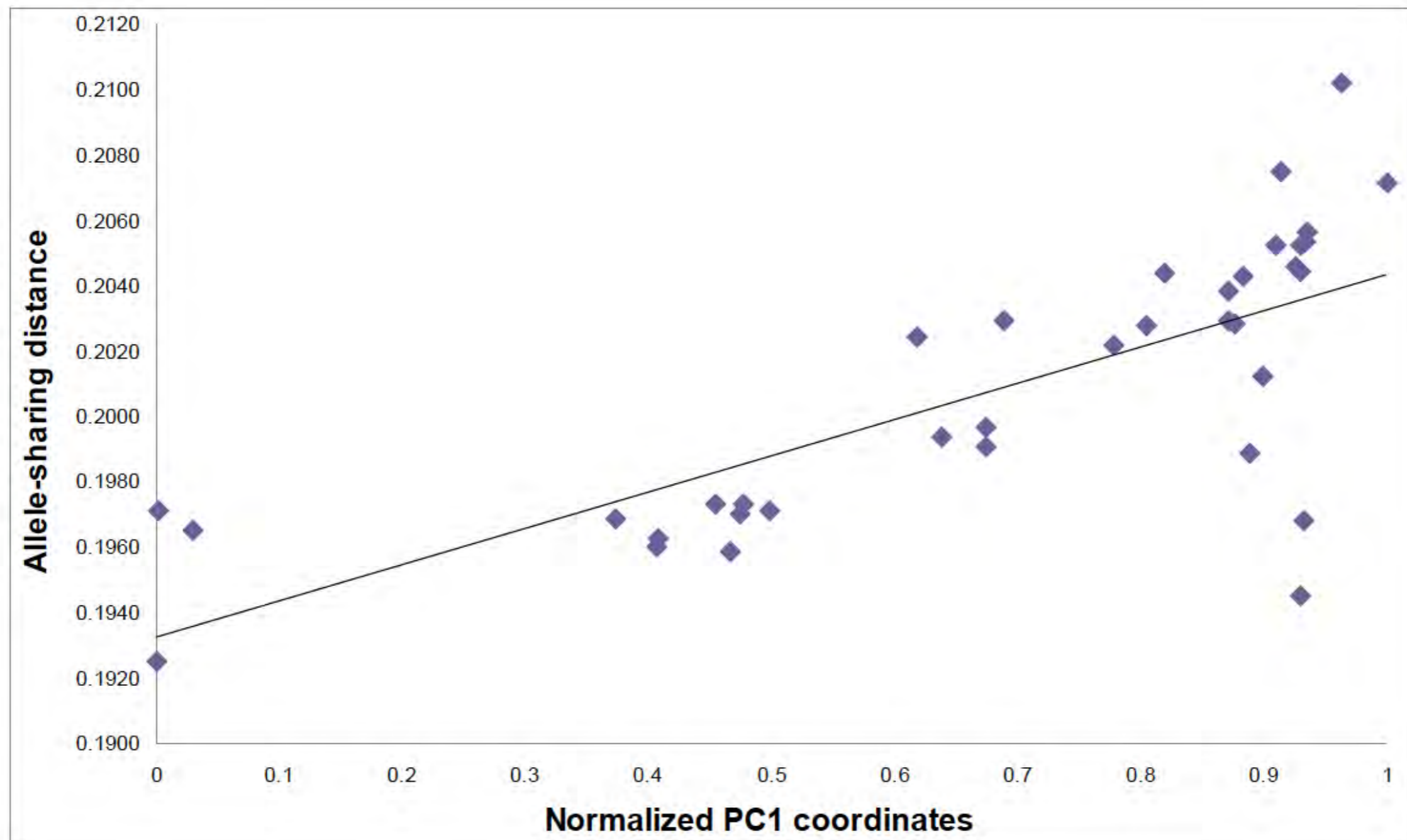


Results of *frappe* analysis for  $k=2$  to  $k=5$  in three Japanese and HapMap East Asia populations. Each vertical bar represents an individual and each color represents ancestry components corresponding to assumed number of ancestral populations, denoted by  $k$ .

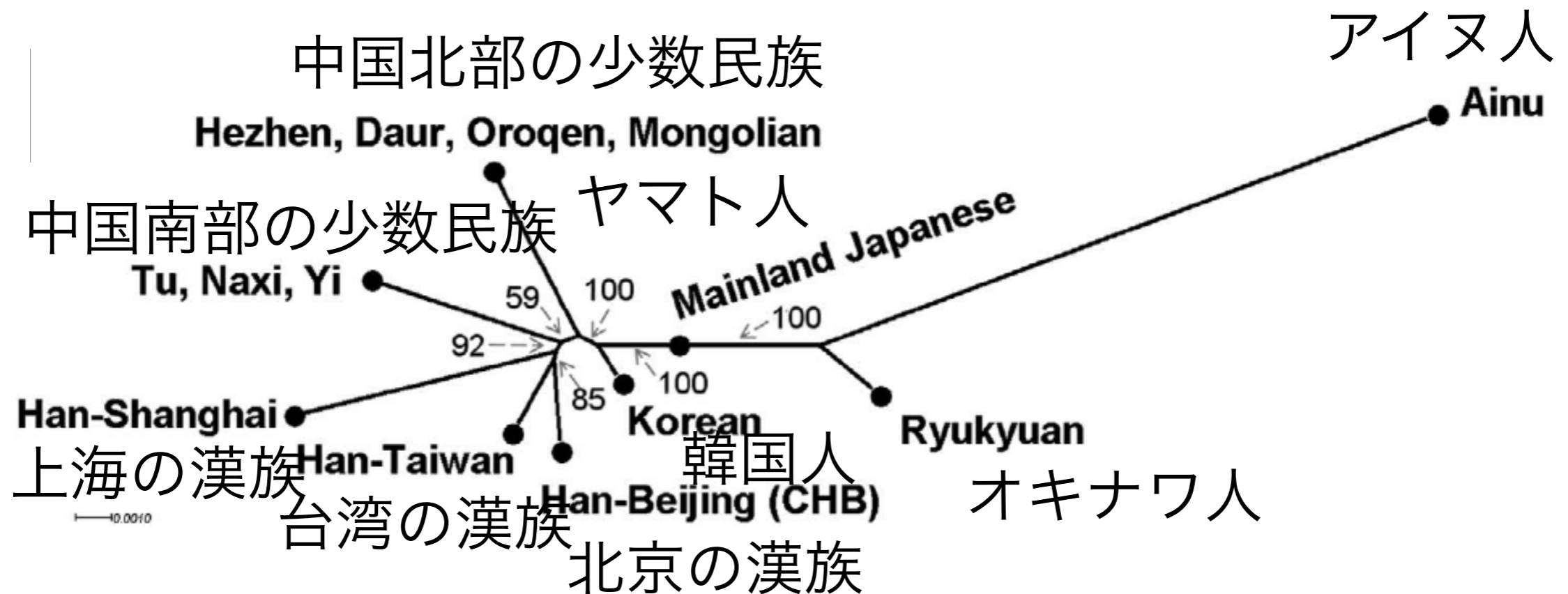


Correlation between Principal Component 1 (PC1) coordinates from Figure 1A with proportion of Ainu ancestry from *frappe* analysis, assuming k=2. PC1 coordinates have been normalized to range from 0 to 1.

**Supplementary Figure 3.** Correlation between PC1 coordinates from Figure 1A (X-axis) and allele sharing distances (Y-axis) between Ainu and Mainland Japanese individuals. PC1 coordinates have been normalized to range from 0 to 1, so that 0 represents the closest proximity to the Mainland Japanese cluster. Pairwise allele sharing distance between Ainu and Mainland Japanese individuals was computed using AWclust<sup>1</sup> and was averaged to obtain values on the y-axis.

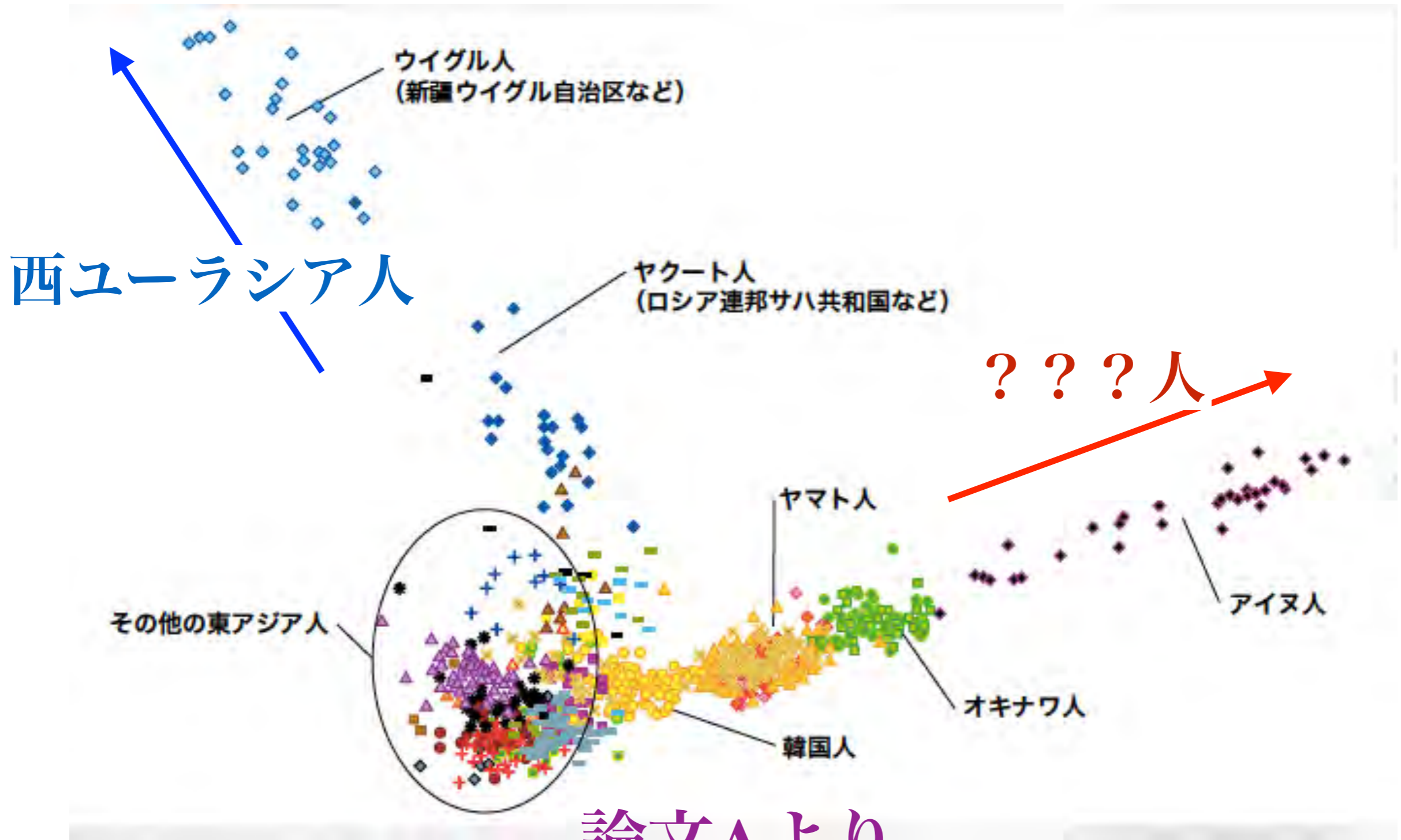


# 東アジア人の遺伝的系統関係



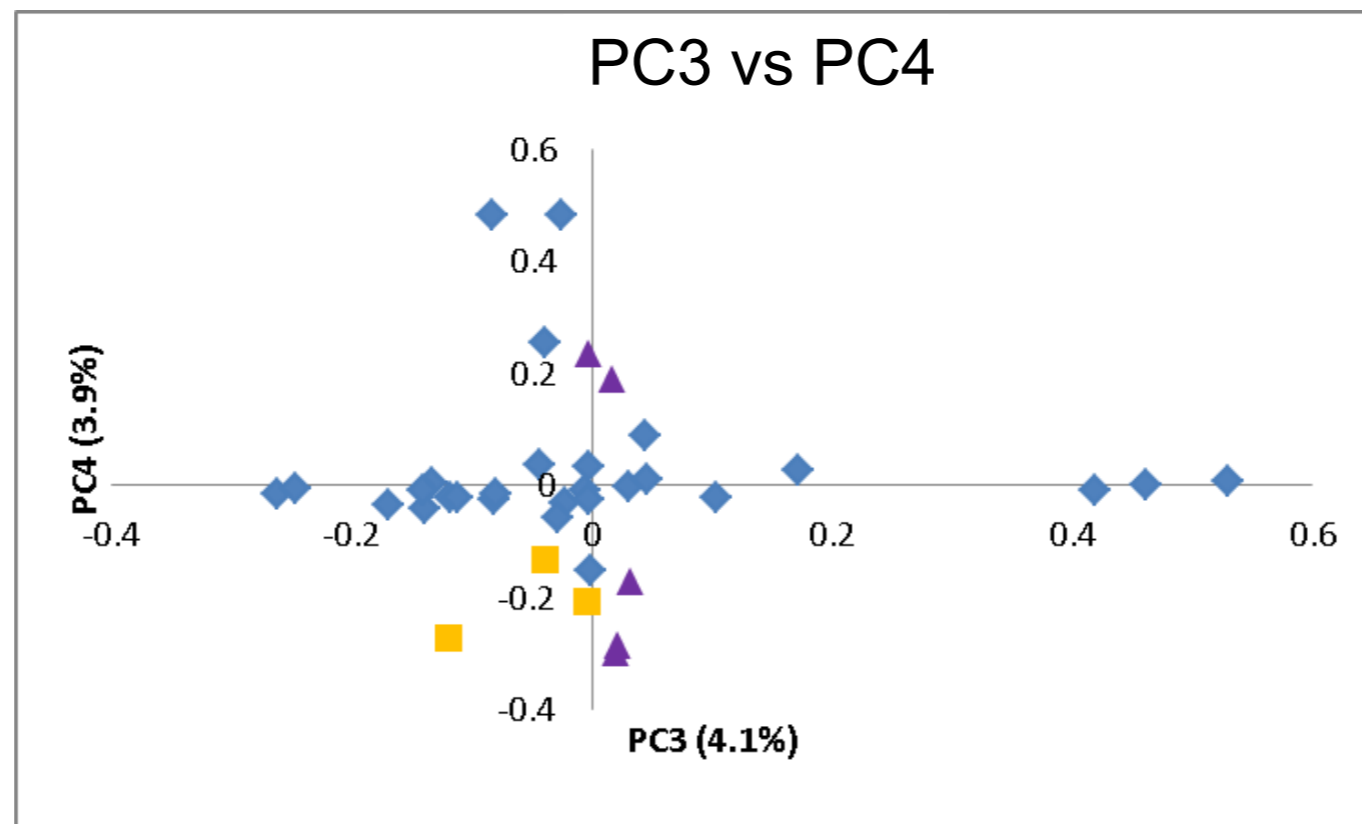
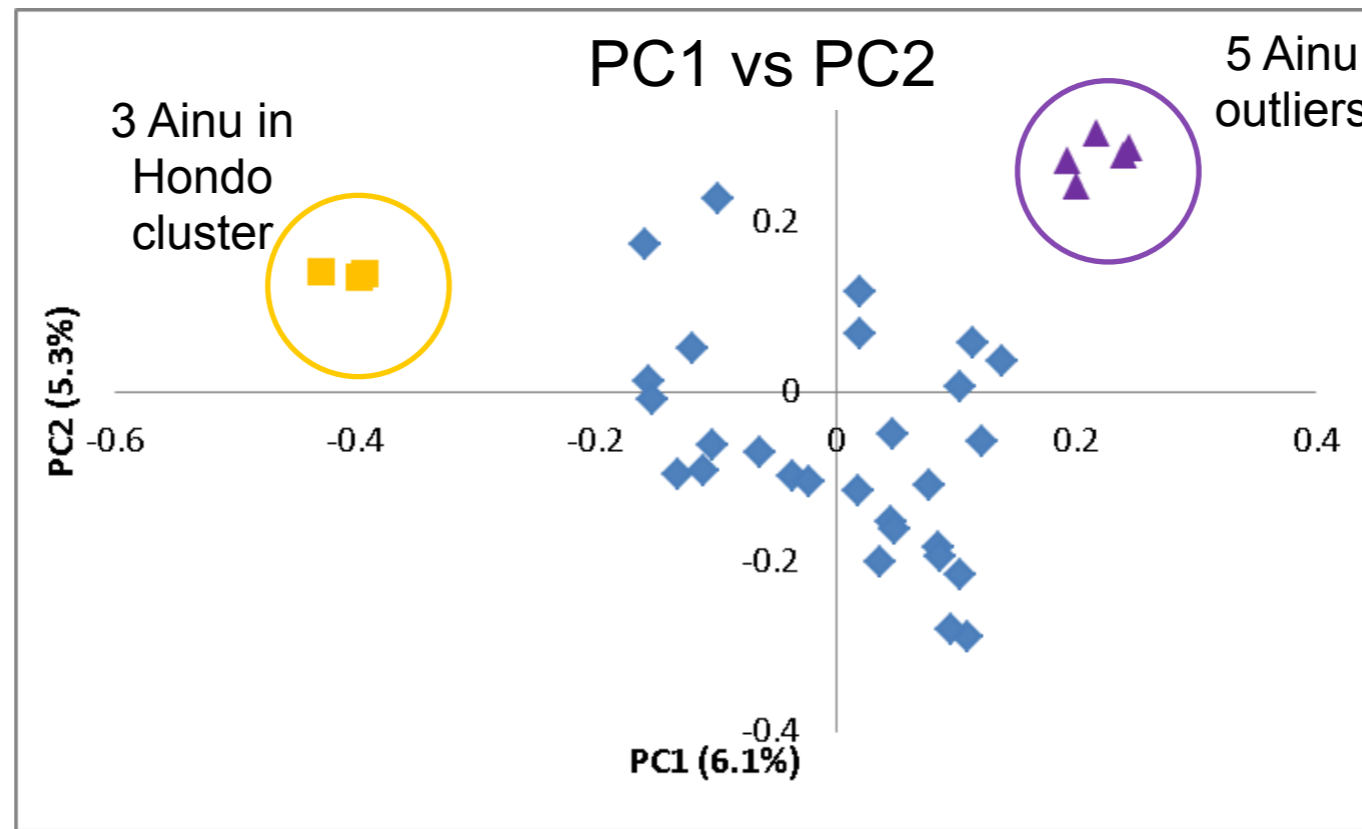
論文Aより

# 他の東アジア人との比較



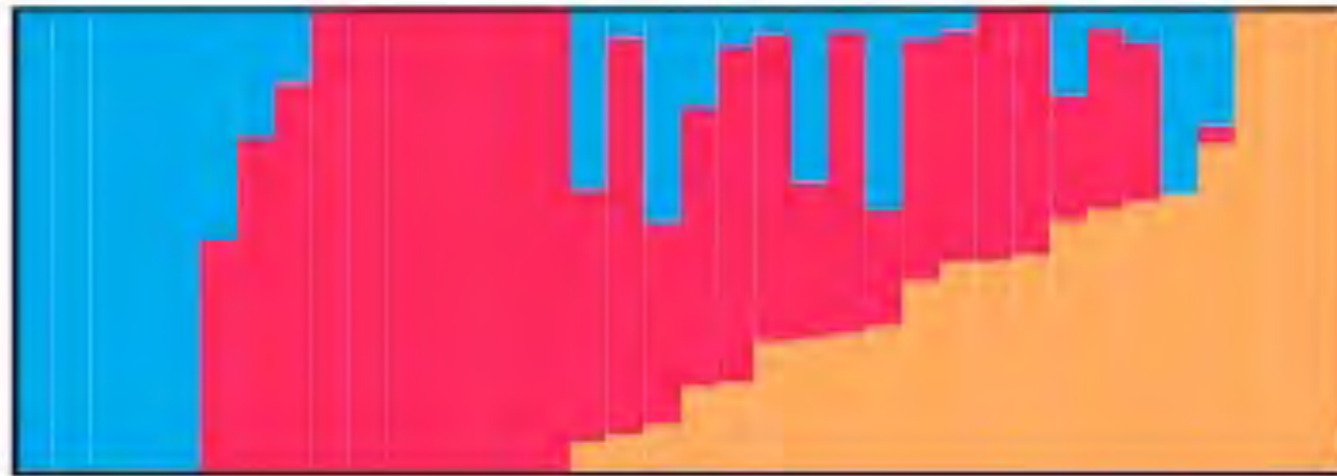
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# PCA for Ainu only

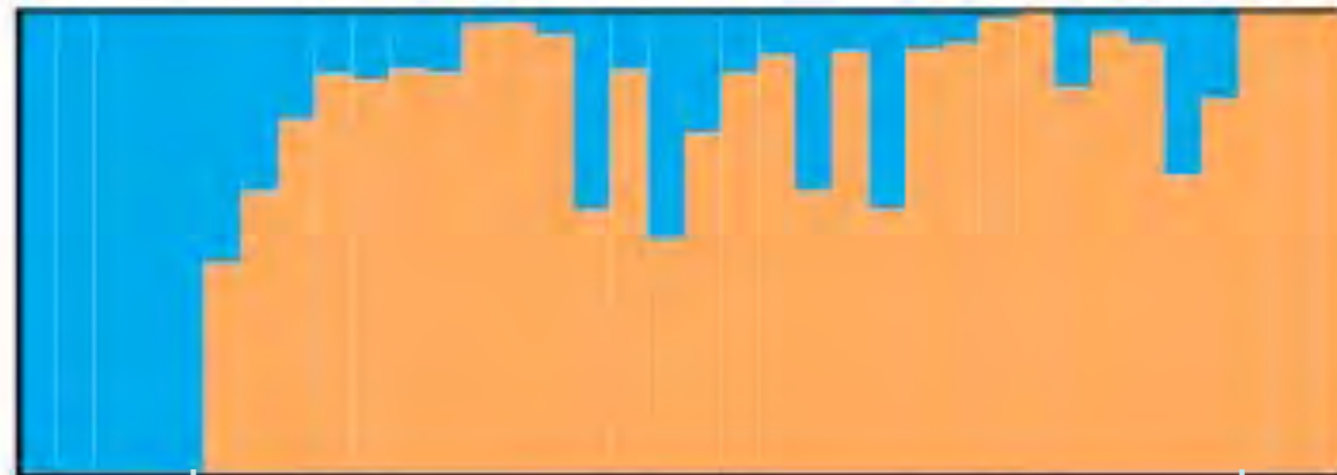


# Ainu frappe results

$k = 3$



$k = 2$



5 Ainu outlier individuals

3 Ainu individuals in Hondo cluster

# アイヌ人の遺伝的特徴を詳細に解析 (2015; 論文B)

*Journal of Human Genetics* (2015) **60**, 565–571; doi:10.1038/jhg.2015.79; published online 16 July 2015

## Unique characteristics of the Ainu population in Northern Japan

Timothy A Jinam<sup>1,2,3</sup>, Hideaki Kanzawa-Kiriyama<sup>2,4</sup>, Ituro Inoue<sup>2,3</sup>, Katsushi Tokunaga<sup>5</sup>, Keiichi Omoto<sup>6</sup> and Naruya Saitou<sup>1,2,7</sup>

<sup>1</sup>Division of Population Genetics, National Institute of Genetics, Mishima, Japan

<sup>2</sup>Department of Genetics, School of Life Science, Graduate University for Advanced Studies, Mishima, Japan

<sup>3</sup>Division of Human Genetics, National Institute of Genetics, Mishima, Japan

<sup>4</sup>Department of Anthropology, National Museum of Nature and Science, Tsukuba, Japan

<sup>5</sup>Department of Human Genetics, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan

<sup>6</sup>Department of Anthropology, Faculty of Science, The University of Tokyo, Tokyo, Japan

<sup>7</sup>Department of Biological Sciences, Graduate School of Science, The University of Tokyo, Tokyo, Japan

Correspondence: Professor N Saitou, Division of Population Genetics, National Institute of Genetics, 1-1-1 Higashi, Yatai, Mishima, Shizuoka 411-8541, Japan

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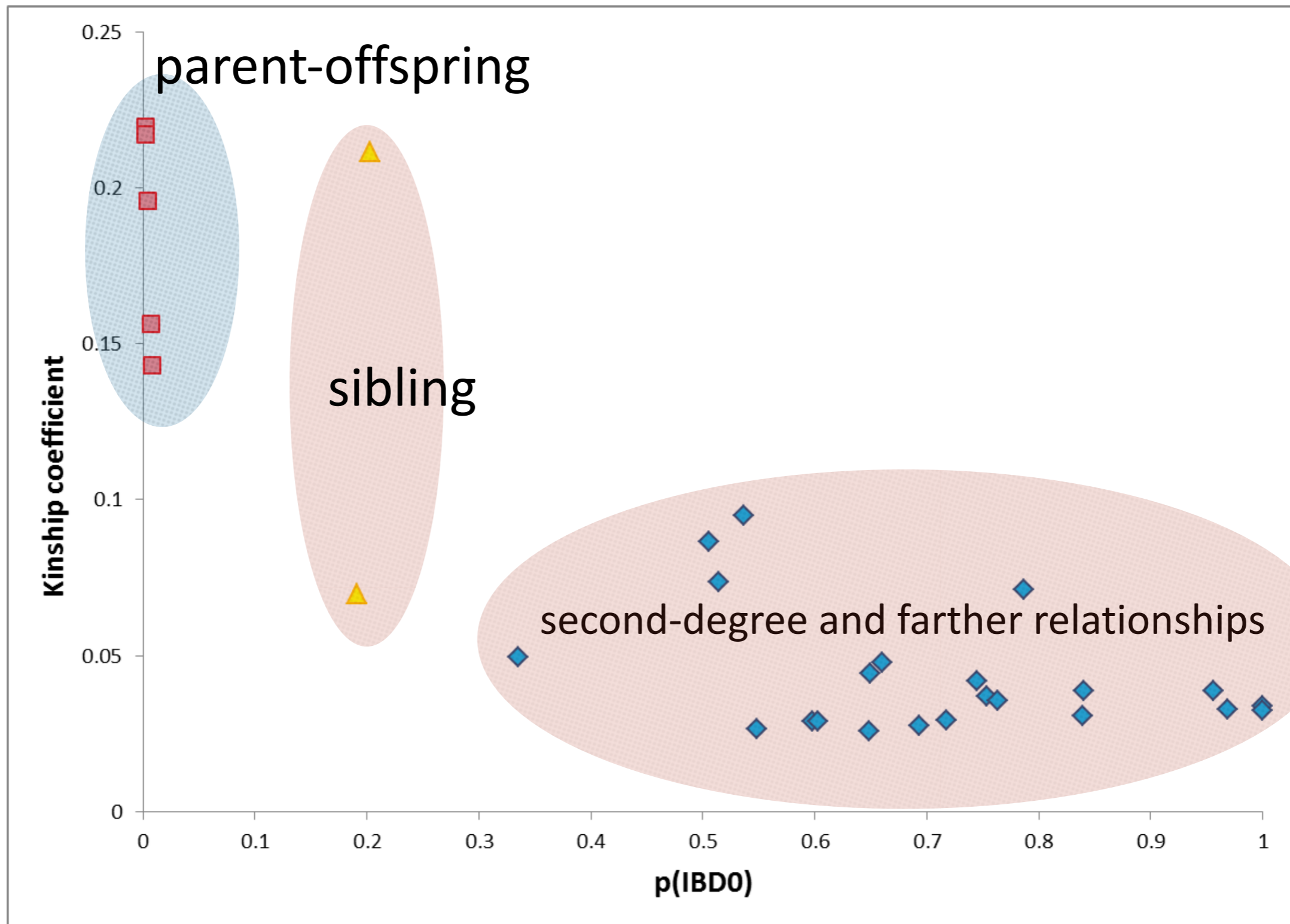
Advance online publication 16 July 2015

Dr. Jinam (2016年4月)

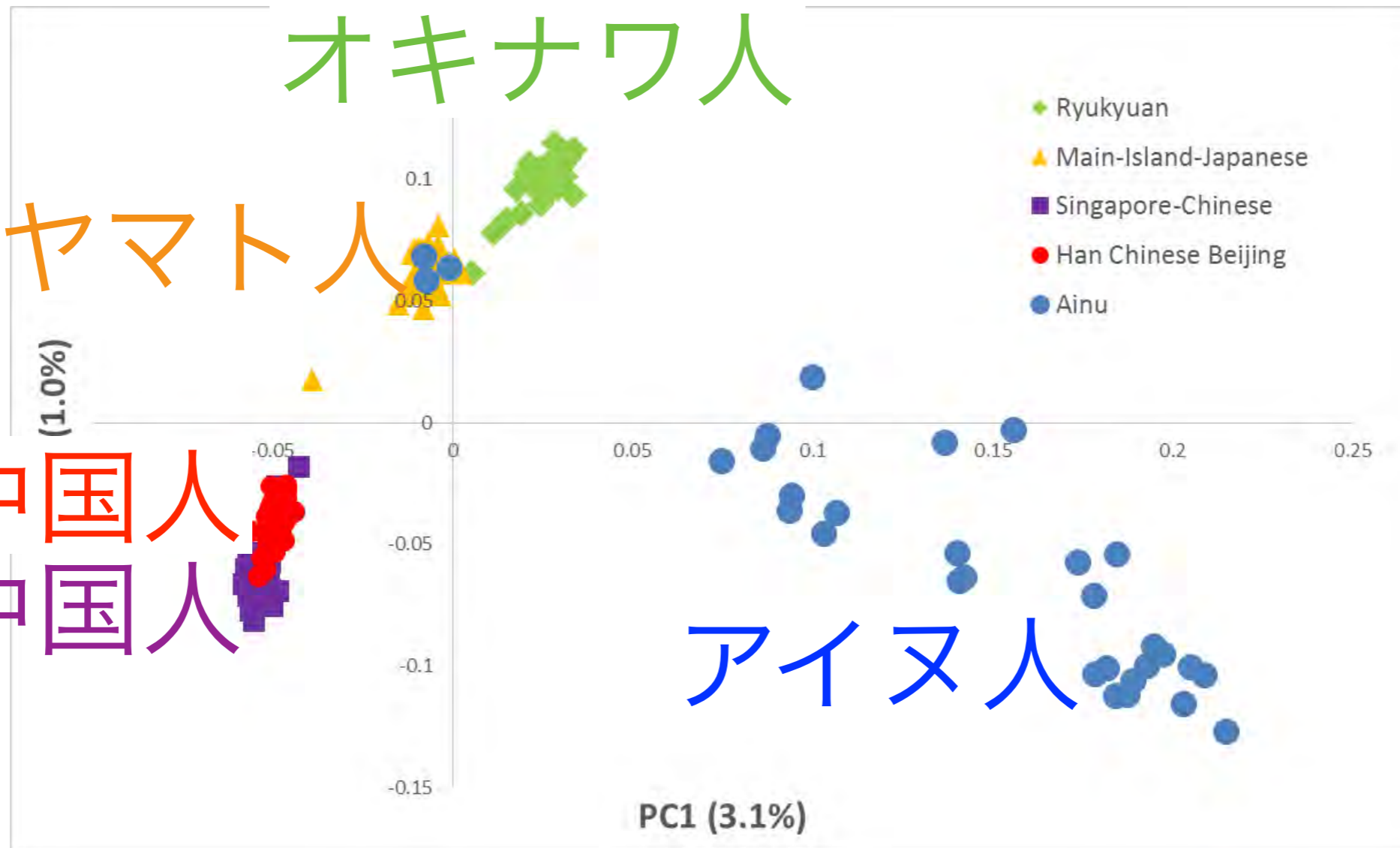




# Estimation of kinship within Ainu

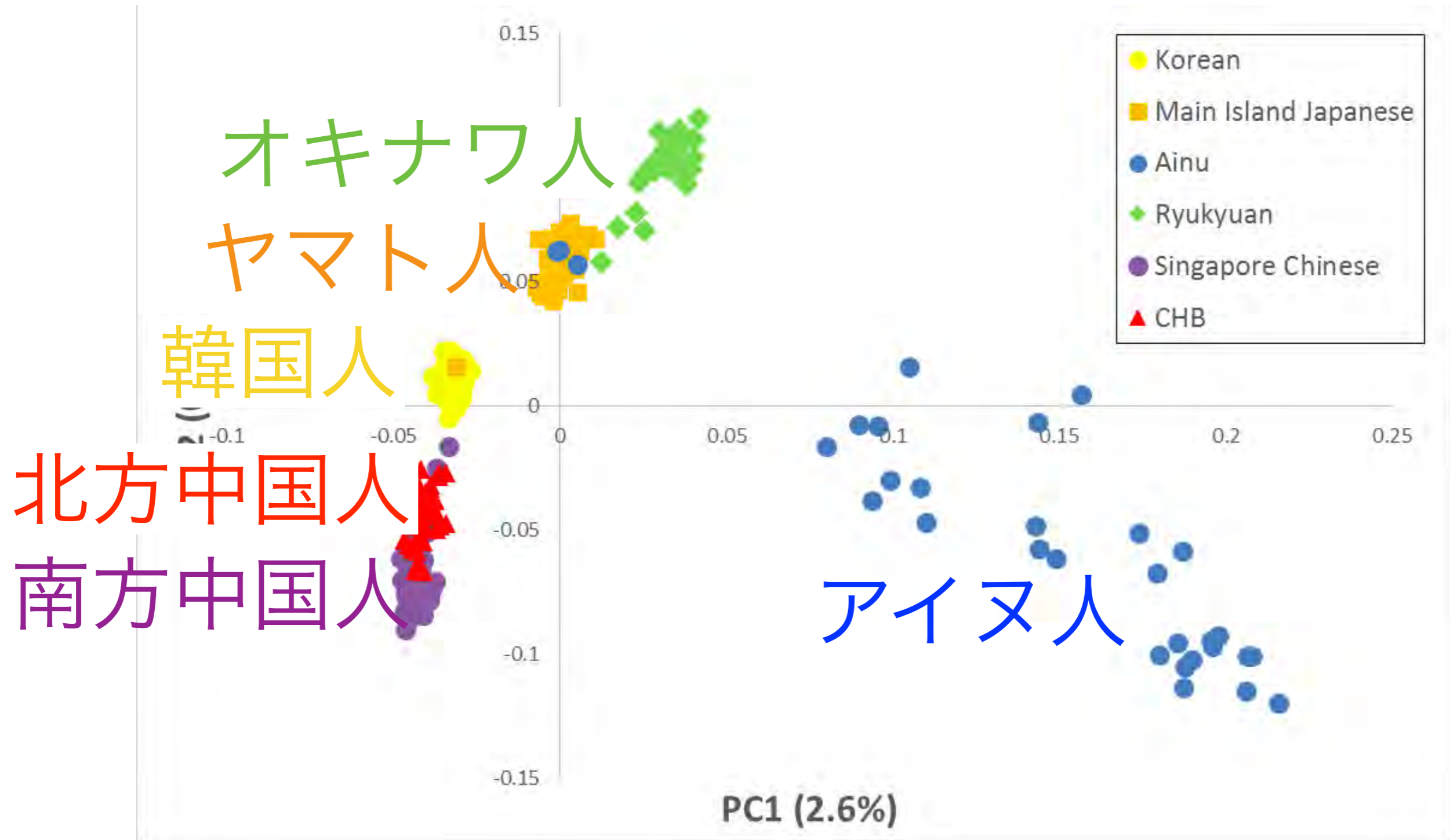


# 東アジアの5人類集団のあいだの遺伝的関係

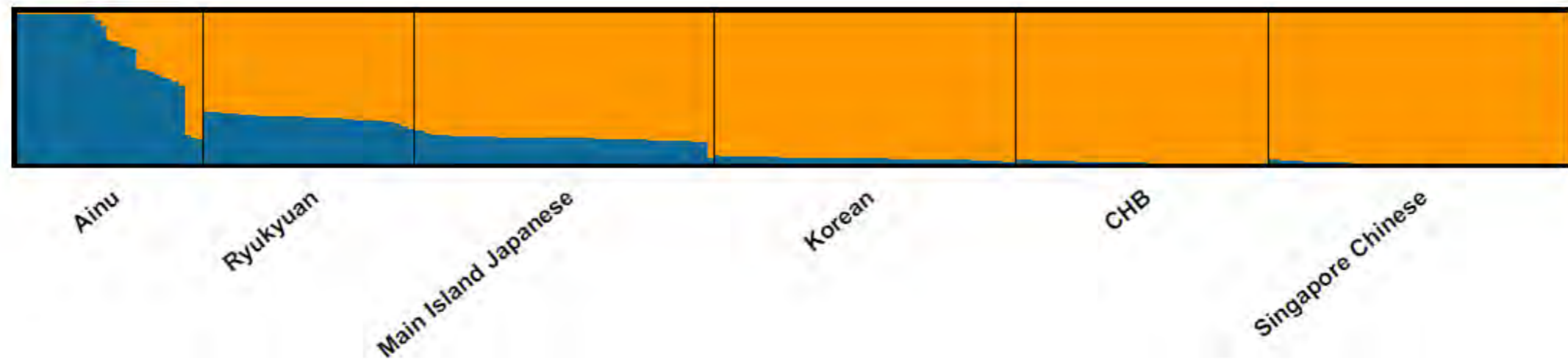
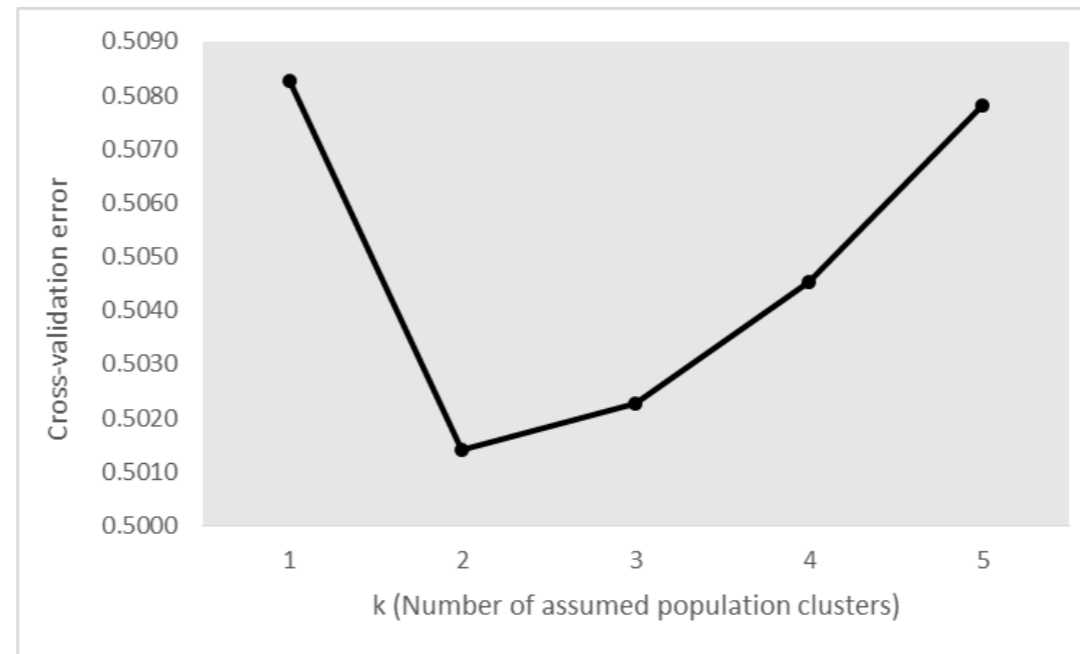


Jinamら (2015)

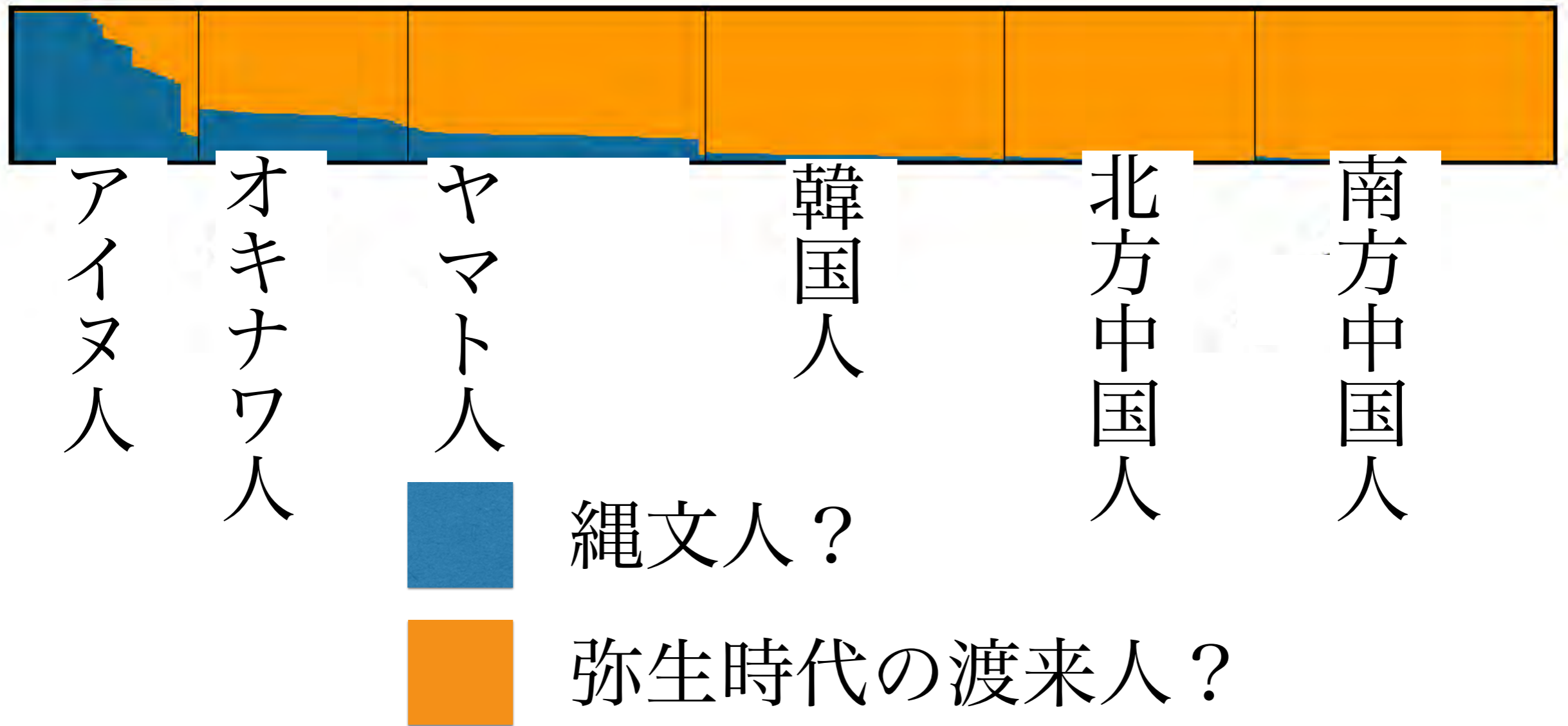
# 東アジアの6人類集団のあいだの遺伝的関係



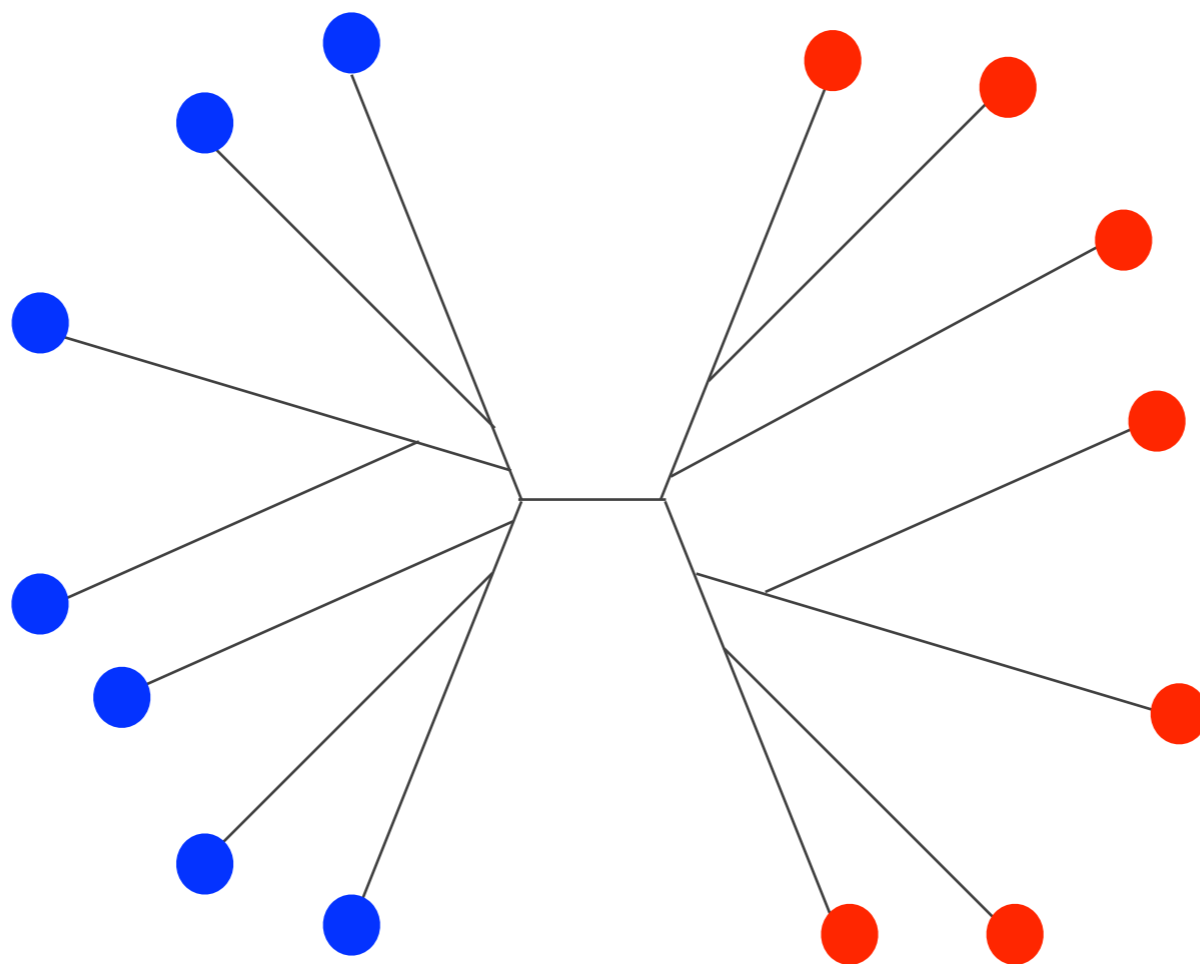
# ADMIXTURE results after omission of closely related individuals in Ainu using approximately 65k SNPs



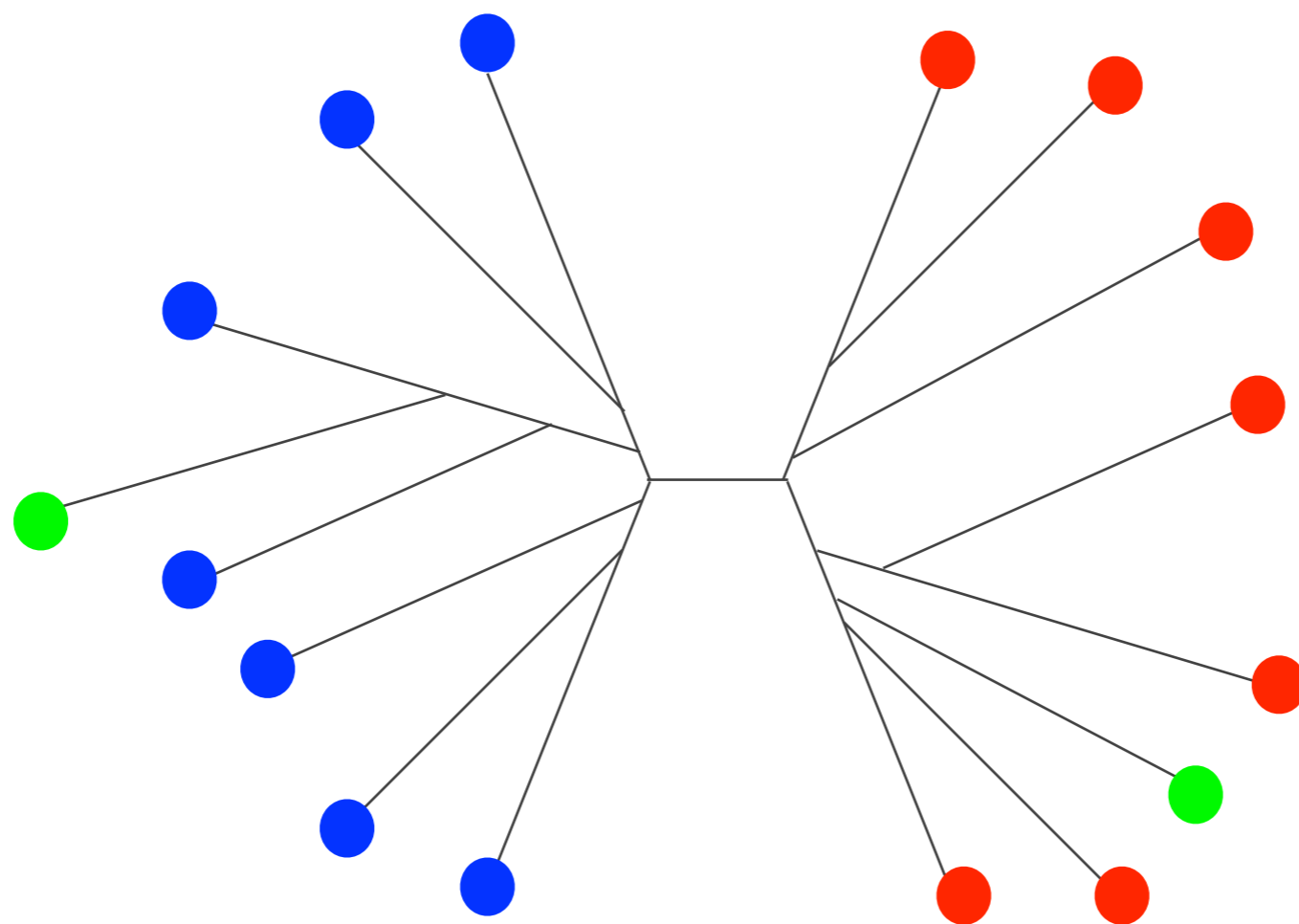
ひとりひとりを2集団の混血と仮定すると・・・



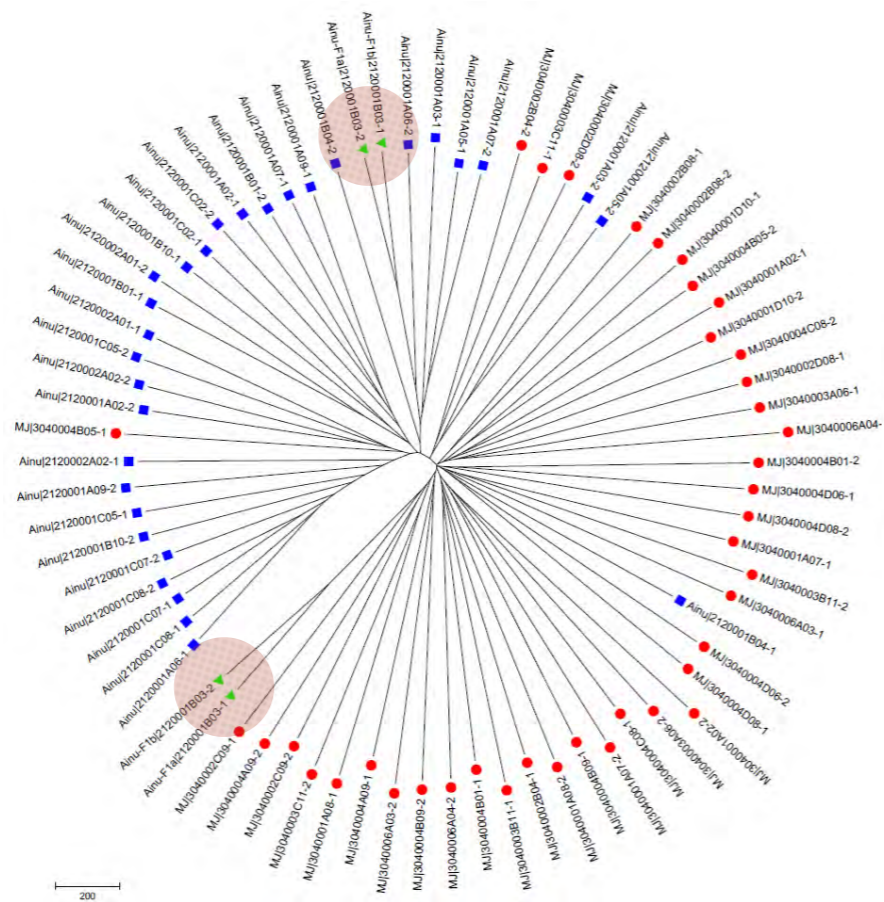
# アイヌ人と 本土人の核DNAハプロタイプの期待される系統関係



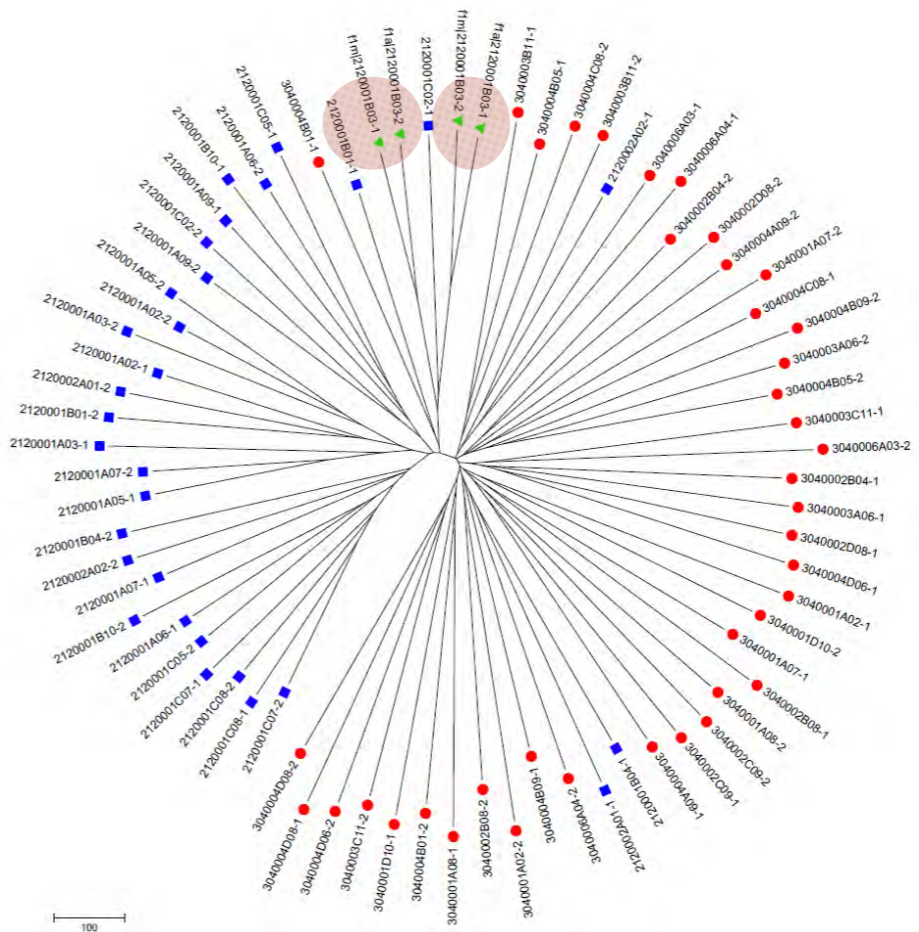
# アイヌ人と 本土人の核DNAハプロタイプの期待される系統関係



● 混血個体のハプロタイプ



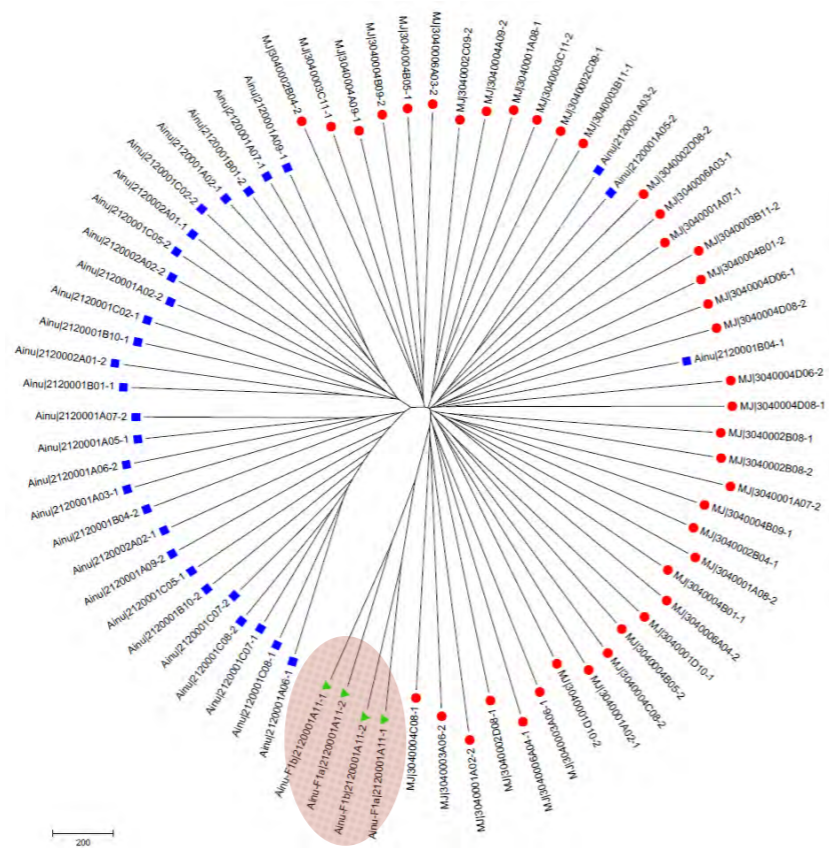
fastphase



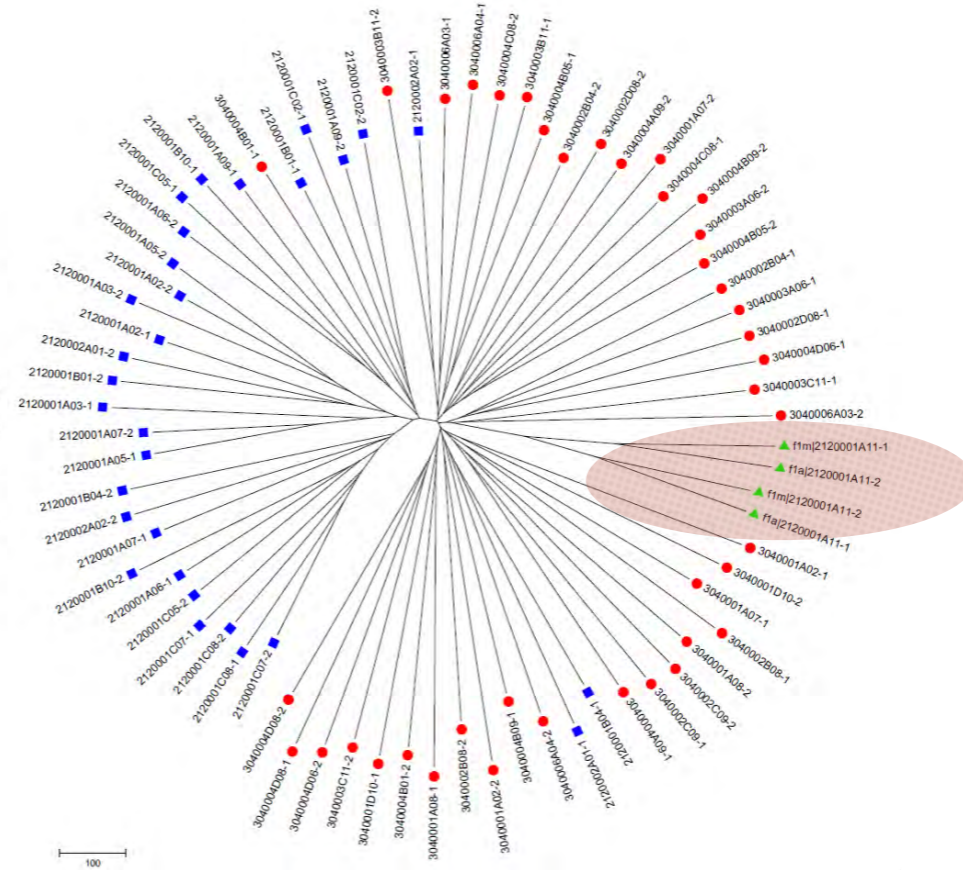
beagle

Jinam・斎藤ら (未発表)



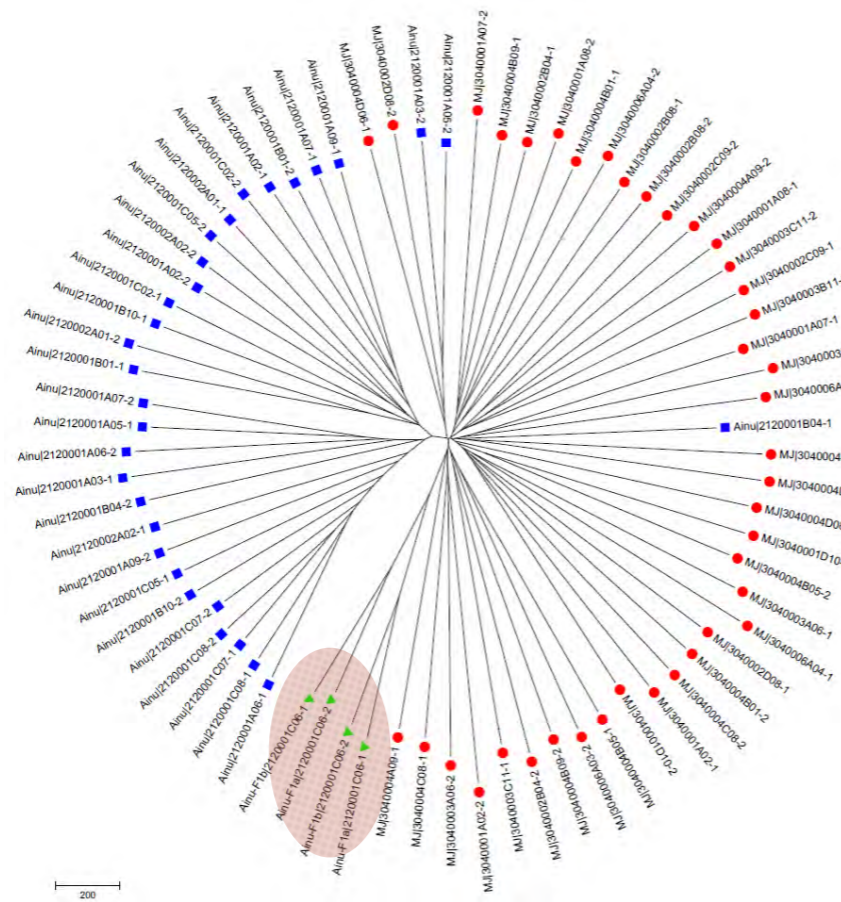


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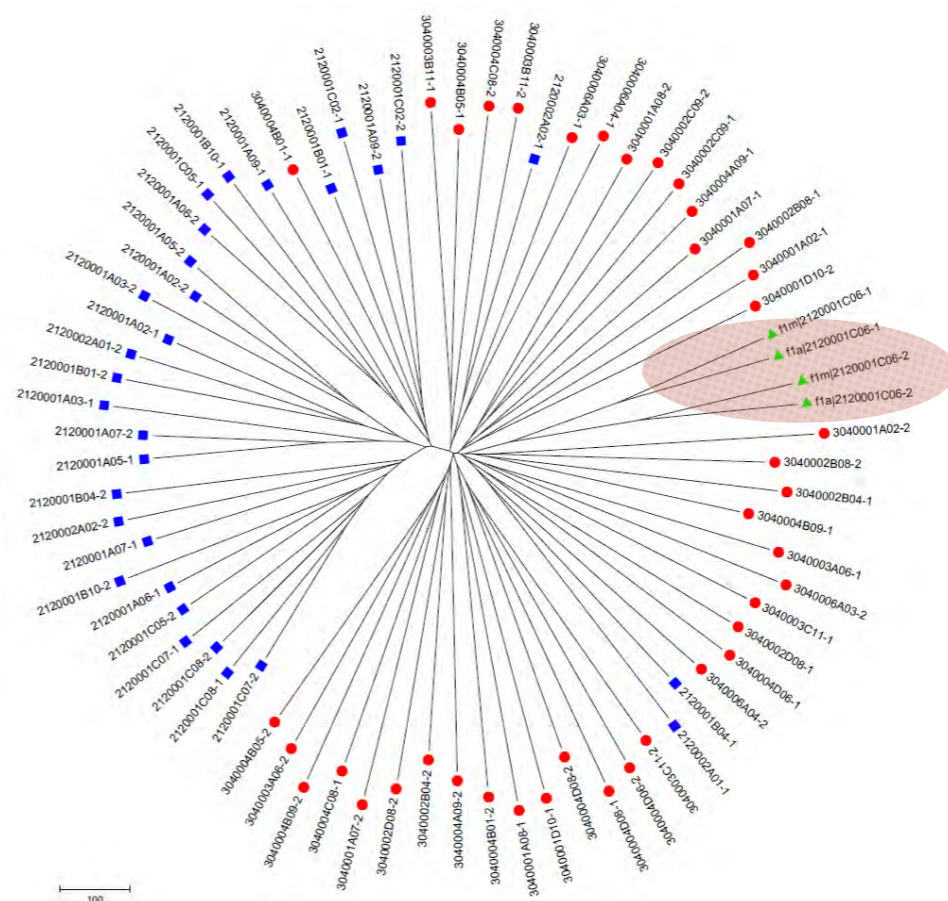


beagle

Jinam・斎藤ら (未発表)

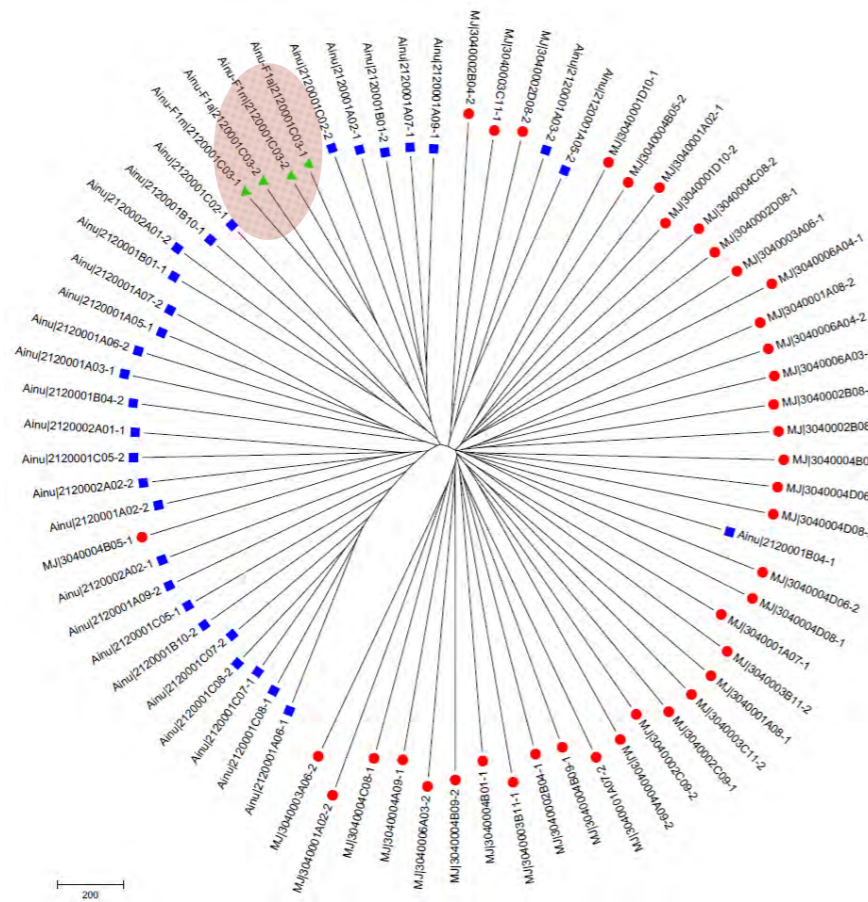


fastphase

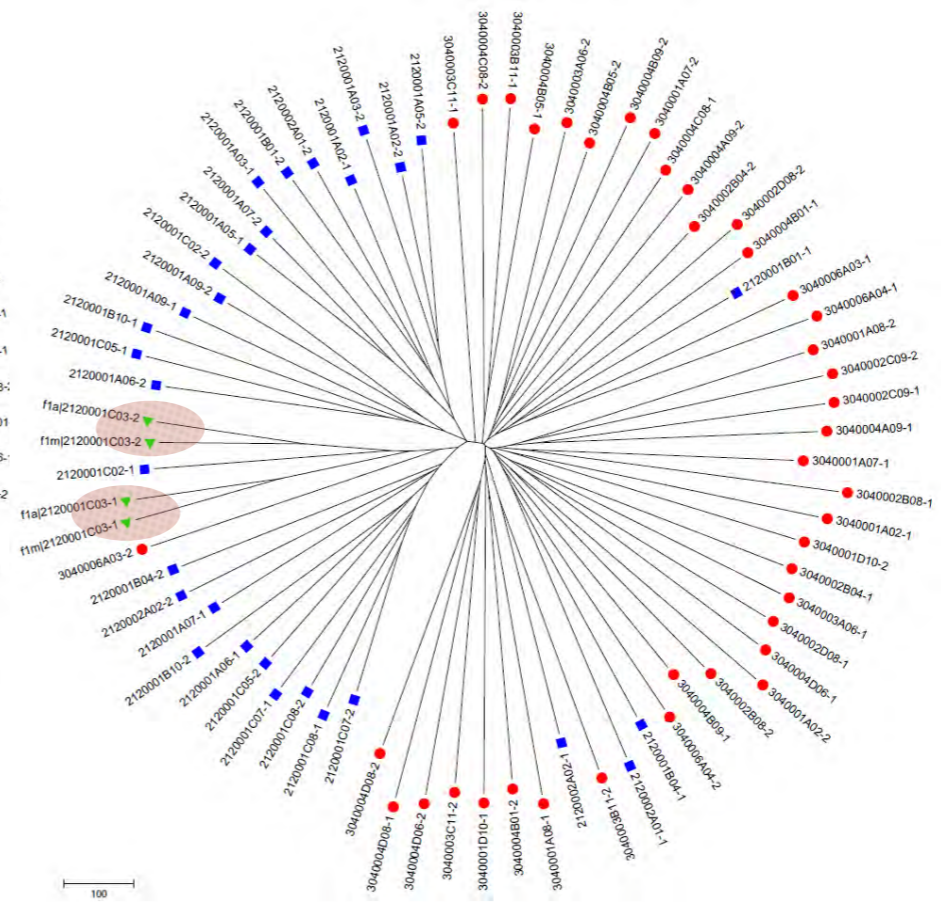


beagle

Jinam・斎藤ら (未発表)

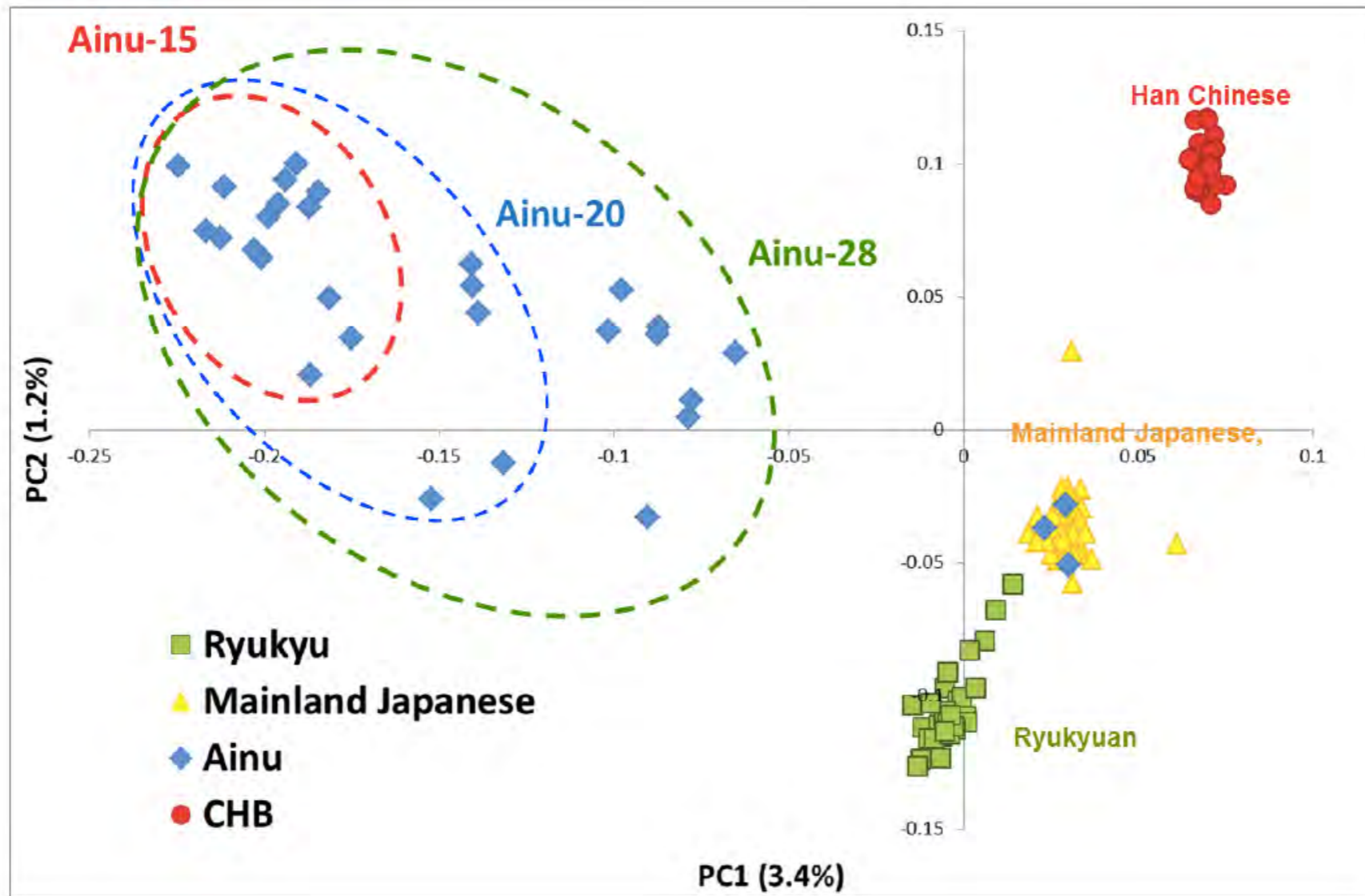


fastphase



beagle

Jinam・斎藤ら (未発表)



論文Bより

# アイヌ人祖先集団と本土人祖先集団が混血をはじめた時期の推定

Ainu dataset	Ainu ancestry (frappe results at k=2)	Time since admixture	
		Generations	Years*
Ainu-15	98.2%	58	1,450
Ainu-20	92.9%	56	1,400
Ainu-28	82.5%	55	1,375

\*A generation time of 25 years was used.

およそ紀元6~7世紀

論文Bより



# 理化学研究所のデータとの重ね合わせ

北京の漢族

関東ヤマト人

オキナワ人

えみしの子孫？

アイヌ人

論文Bより



# Distribution of differentiated SNP in the Ainu for the five genes associated with facial morphology in Europeans and for hair and tooth morphology (EDAR)

