Genomic insights into the relationship between Neolithic Japanese hunter-gatherer, Jomon, and modern East Eurasians

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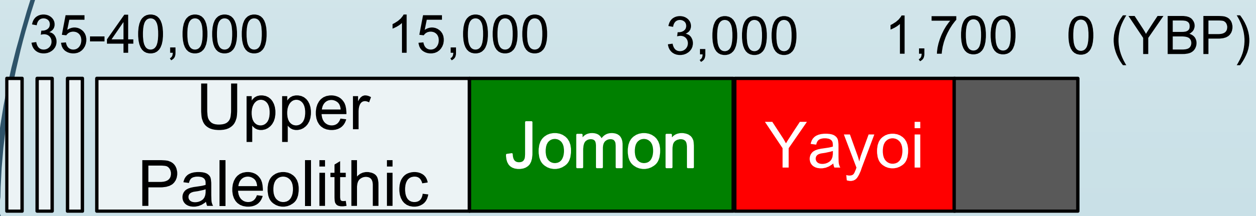
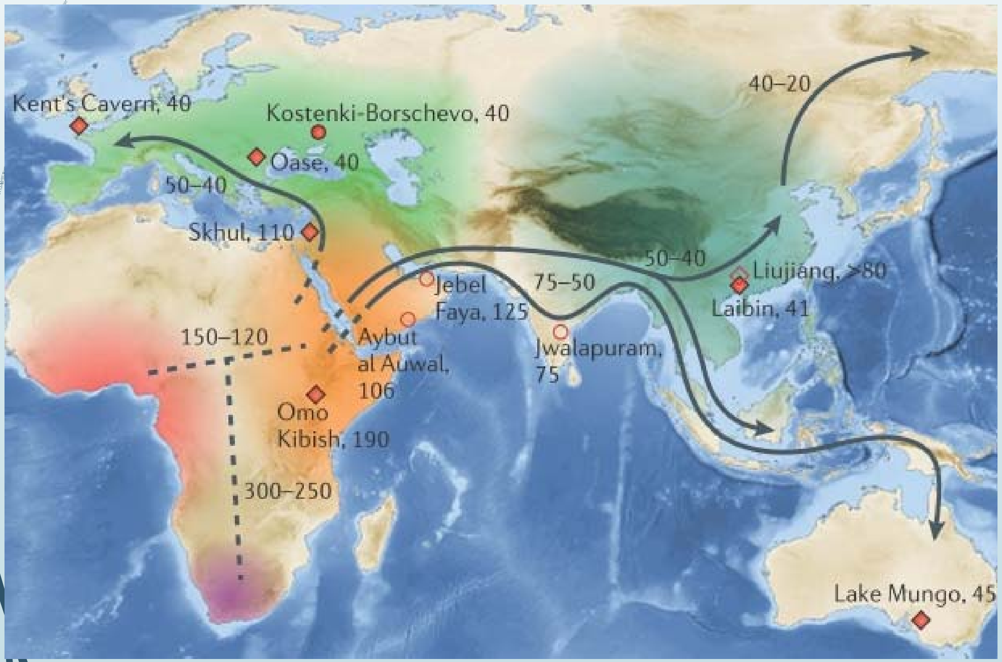

35-40,000 15,000 3,000 1,700 0 (YBP)

Upper Paleolithic | Jomon | Yayoi

Out of Africa

- Homo Sapiens emerged in Africa and radiated from there.
- They entered East Asia 40-50k years ago.



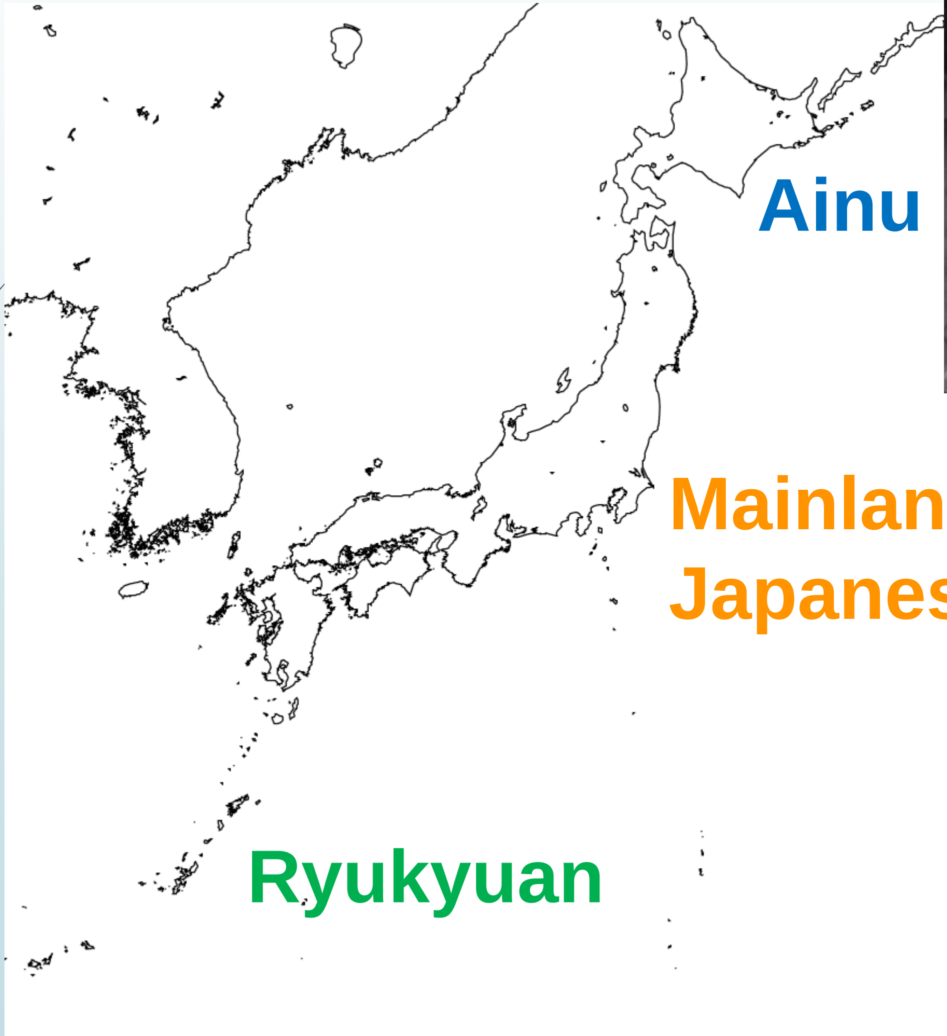
Scally and Durbin, 2012

Paleoanthropology

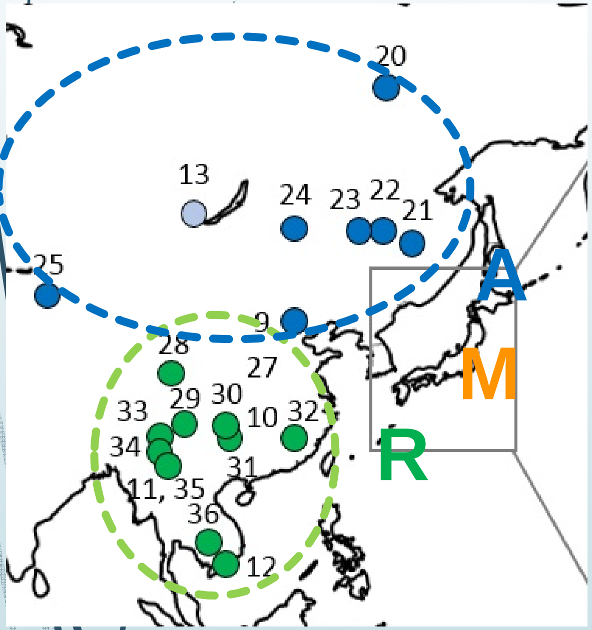
Archeology

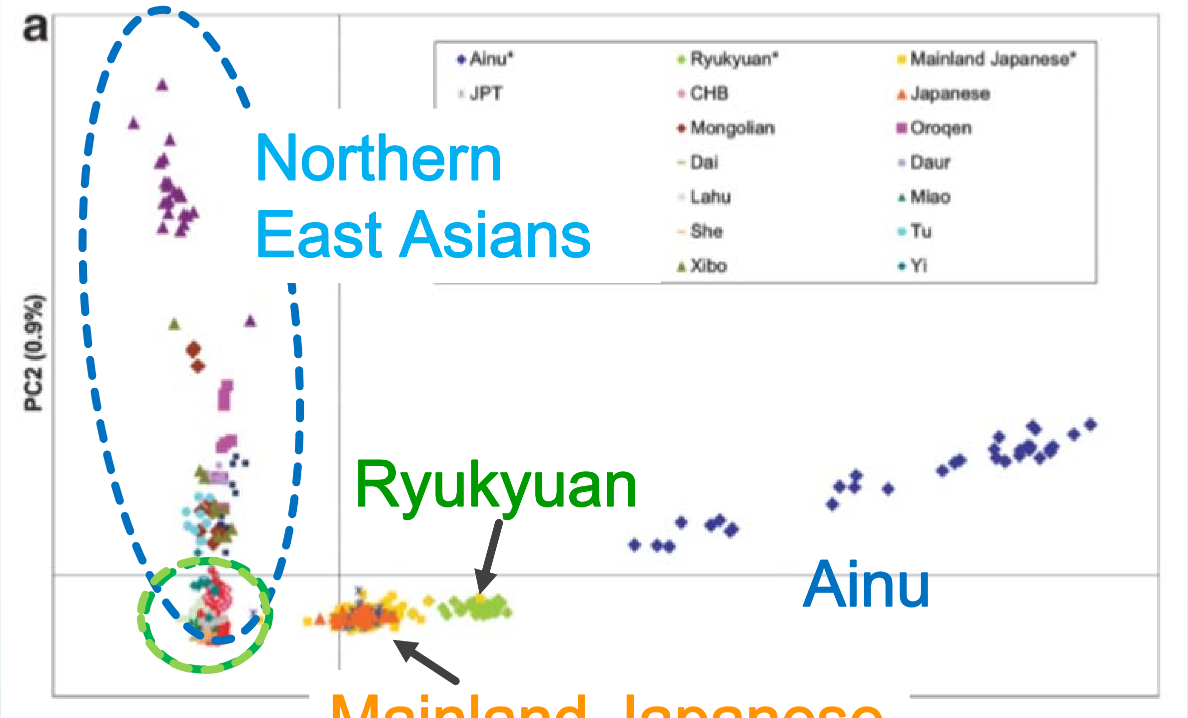
Genetics

Ainu, Mainland Japanese, and Ryukyuan



Uniqueness of **Ainu** in East Eurasians





Northern East Asians

Ryukyuan

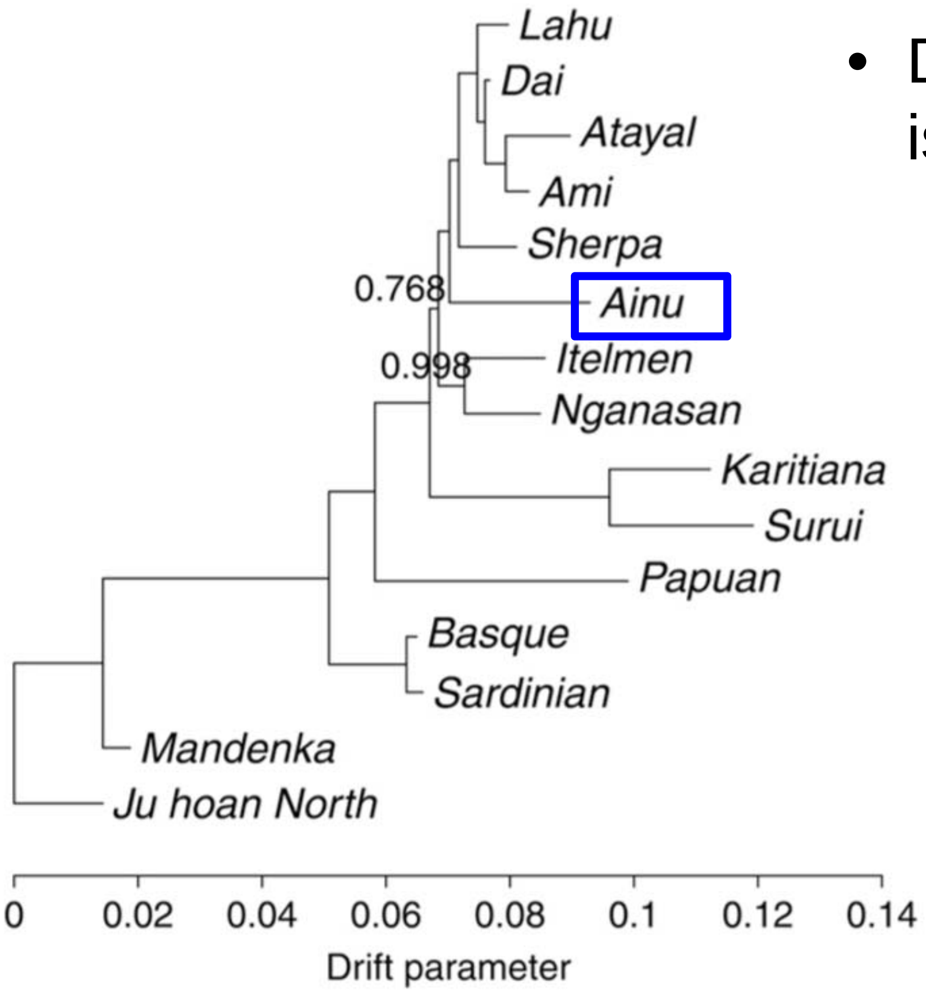
Ainu

Mainland Japanese

Southern East Asians

The Japanese Archipelago Human Population Genetics Consortium, 2012

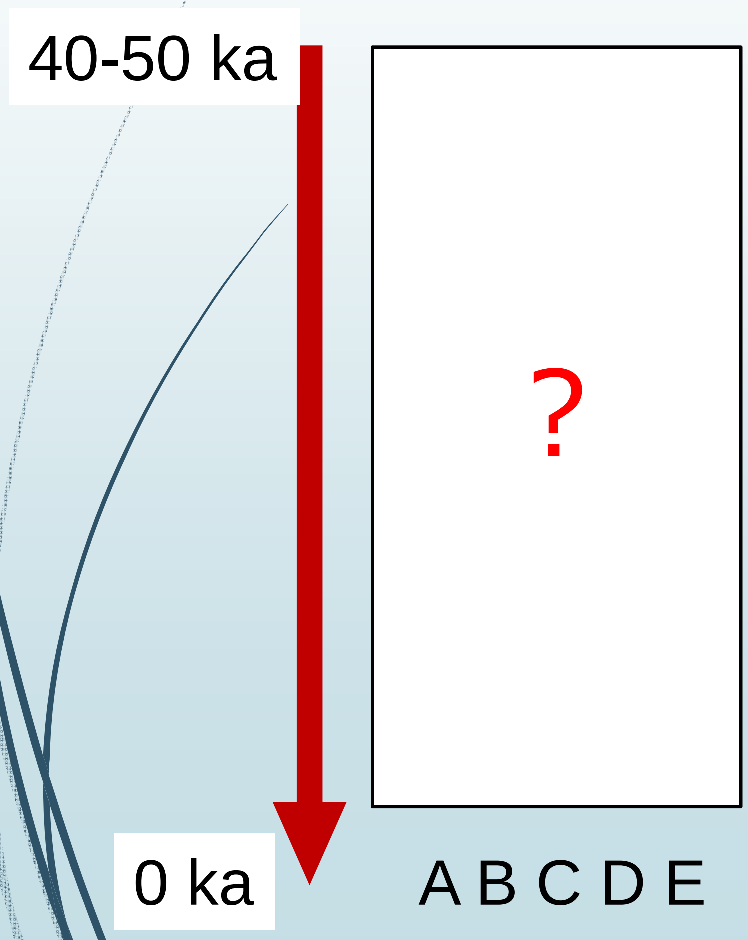
Ainu is outgroup of all East Asian farmers

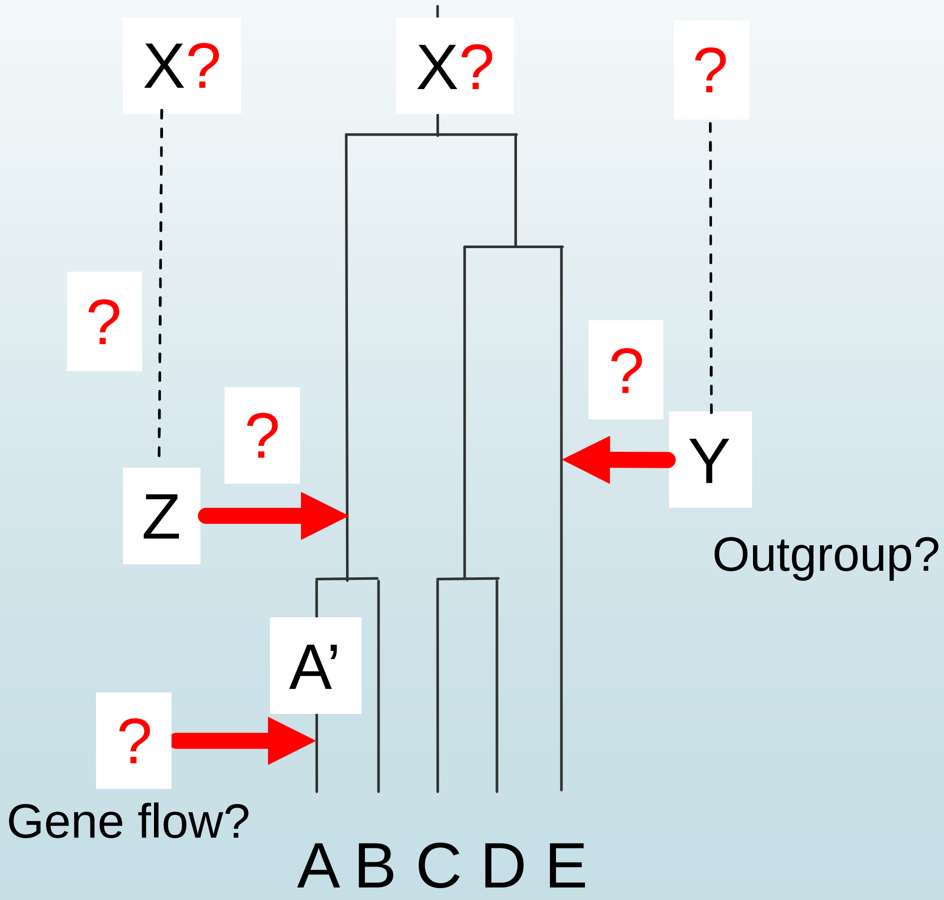


- Deep divergence and isolation in Ainu people

Jeong et al., 2016

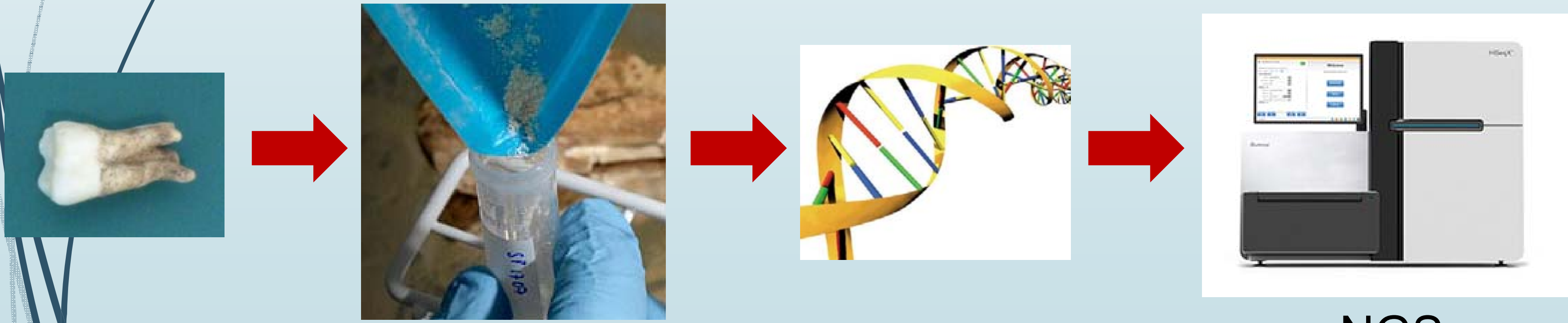
Modern and Ancient populations





Ancient DNA analysis

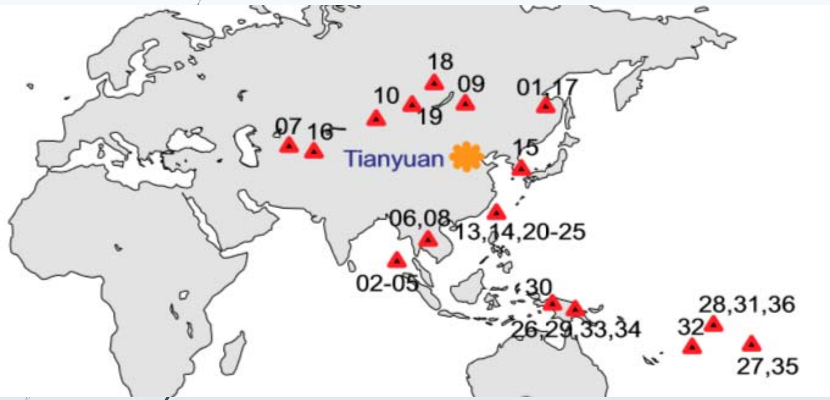
- Sequencing DNA extracted from ancient teeth/bone
- Direct evidence of ancient genotype



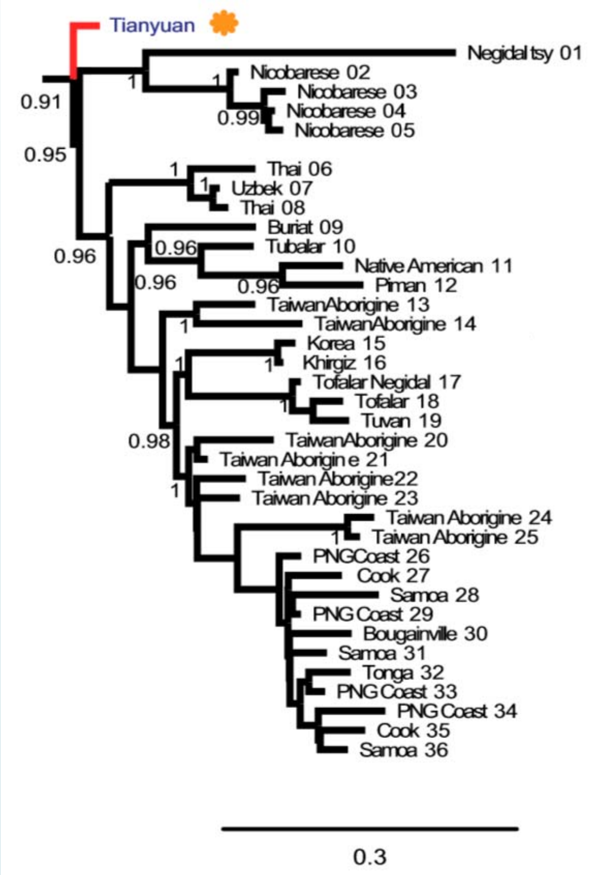
NGS
(Next Generation
sequencer)

Genetic continuity from earliest modern humans in East Asia

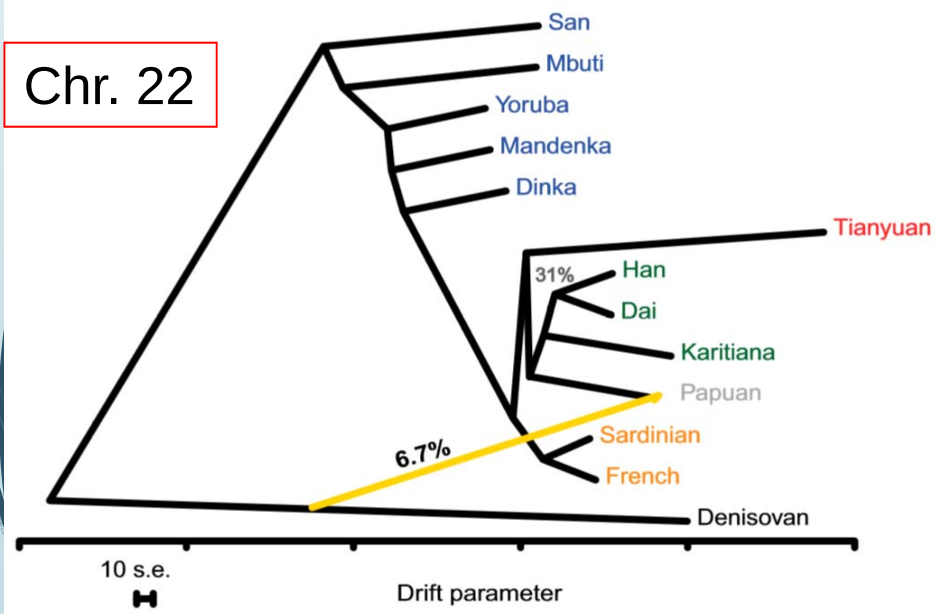
40,000-y old human remains from Tianyuan, Beijing (Fu et al., 2013)



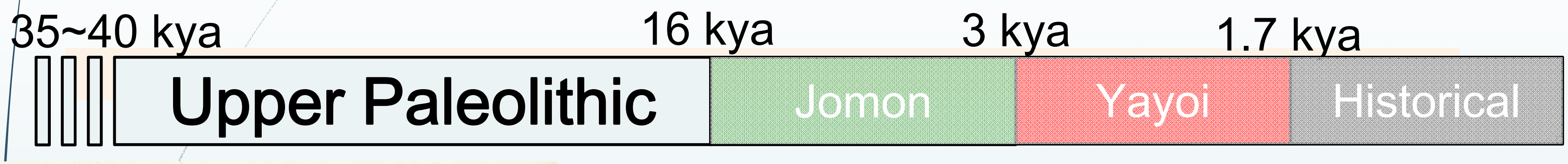
Tree of mtDNA haplogroup B

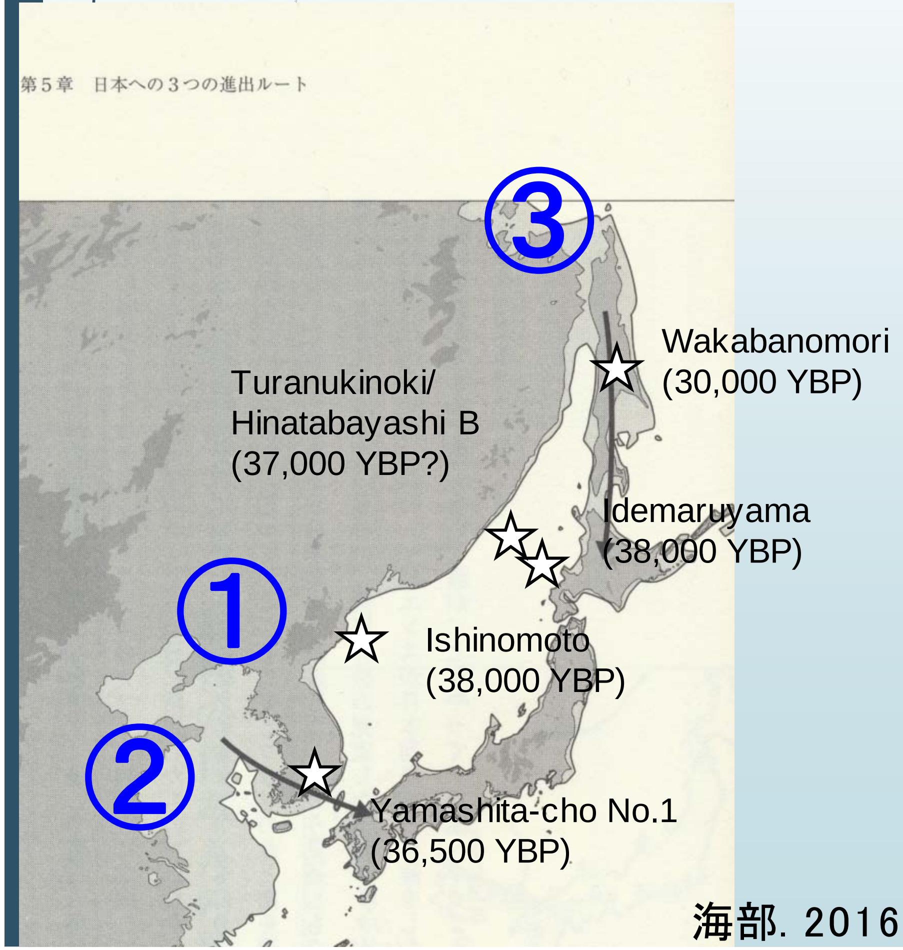


Chr. 22



First modern humans in Japan





海部. 2016

- Stone tools from everywhere in Japan from Hokkaido to Okinawa
- Dating to younger than 40,000 years old

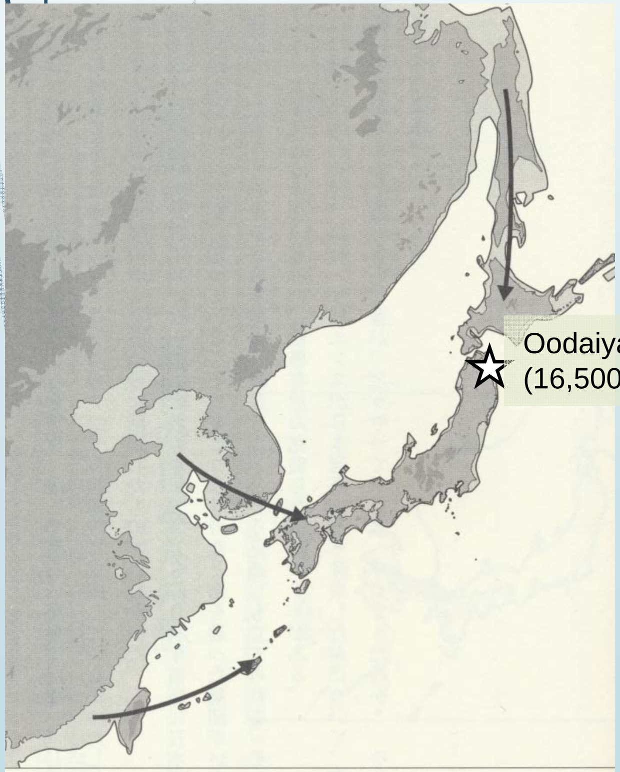
Upper Paleolithic human bones are quite rare.

From Japanese Palaeolithic Research Association

Neolitic hunter-gathers; Jomon

35~40 kya
16 kya
3 kya
1.7 kya

Upper Paleolithic
Jomon
Yayoi
Historical



Oodaiyamamoto No.1
(16,500 YBP)

図5-6　3万8000年前頃の日本列島への3つの可能な渡来ルート

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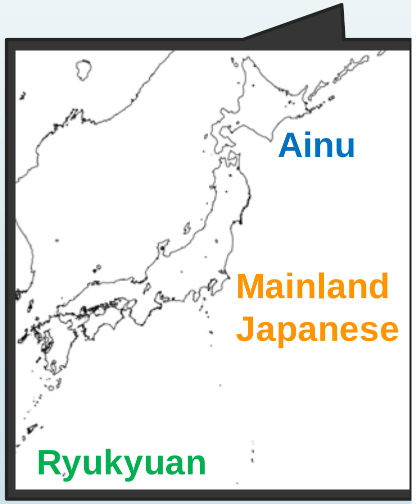
Hunting and Gathering

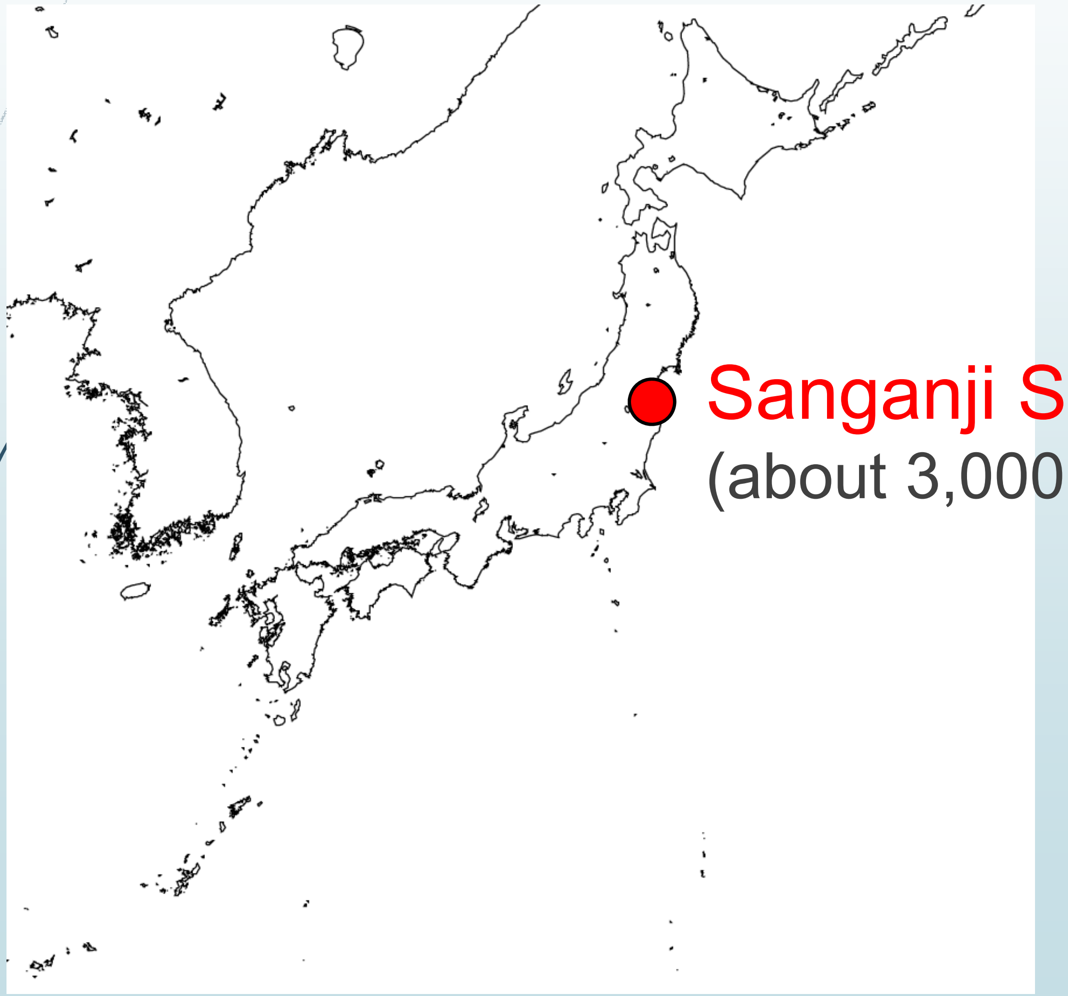


Q1: Genetic relationship with modern humans

Q2: Origin of Jomon

国立科学博物館
National Museum of Nature and Science

Investigating Jomon Nuclear Genome



Sampling to sequencing

http://www.nhk.or.jp/zero/contents/dsp540.html

GAIIx/HiSeq 2000
(120/100bp paired end)

Data analysis

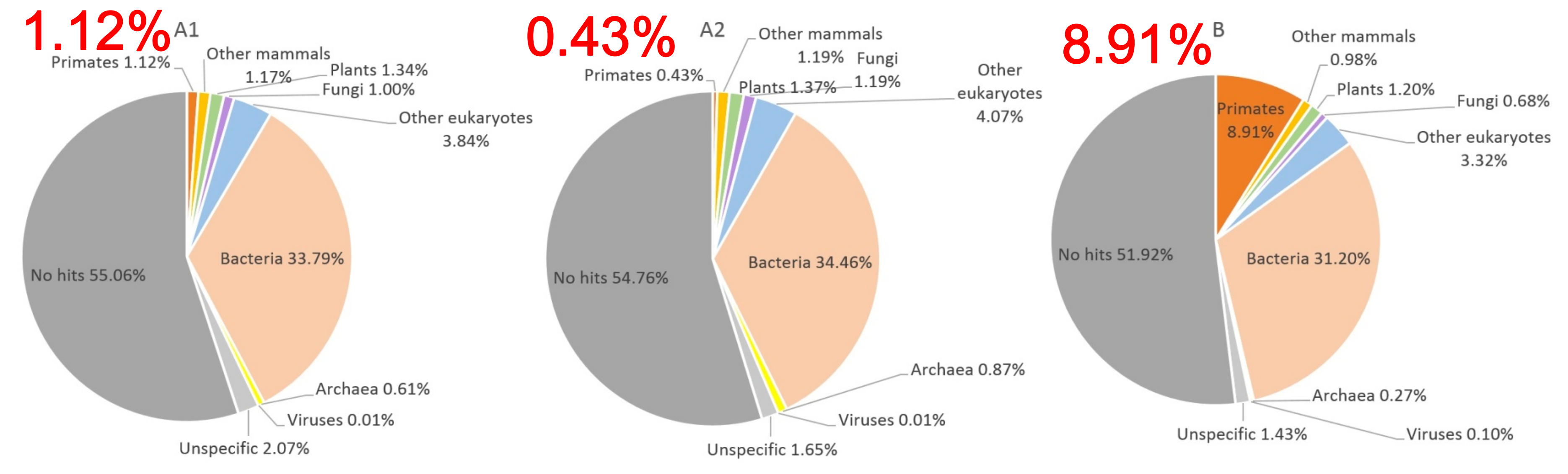
Fastq file

- Adapter trimming, filtering low quality bases (FASTX-Toolkit)
- Merging plus and minus strand (fastq-join)

0.4% ~ 9% of sequence data were from Jomon

Metagenomic analysis for three Sanganji Jomons using BLASTN (parameters were –evalue 3.80e-2, -dbsize 3,200,000,000)

Primates



1.12%

0.43%

8.91%

Kanzawa-Kiriyama et al. 2016

Data analysis

Fastq file

- Adapter trimming, filtering low quality bases (FASTX-Toolkit)
- Merge plus and minus strand (fastq-join)

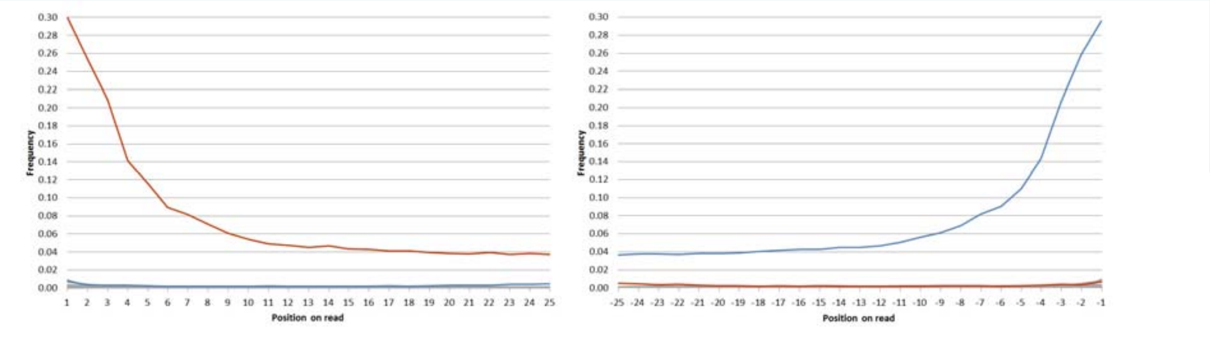
- Mapping to hg19 (BWA, aln, -l 1000)
- Mapping quality >= 30 (SAMtools), length >= 35bp
- Remove PCR depulicates (Picking Bases)

- Checking post-mortem changes
- Contamination estimates

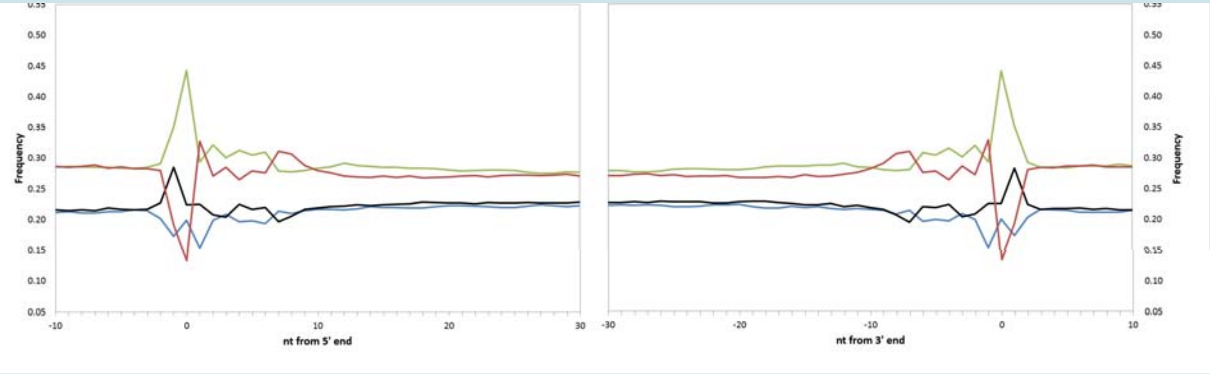
Post-mortem misincorporation and depurination

- Characteristic to ancient DNA

C to T misincorporation

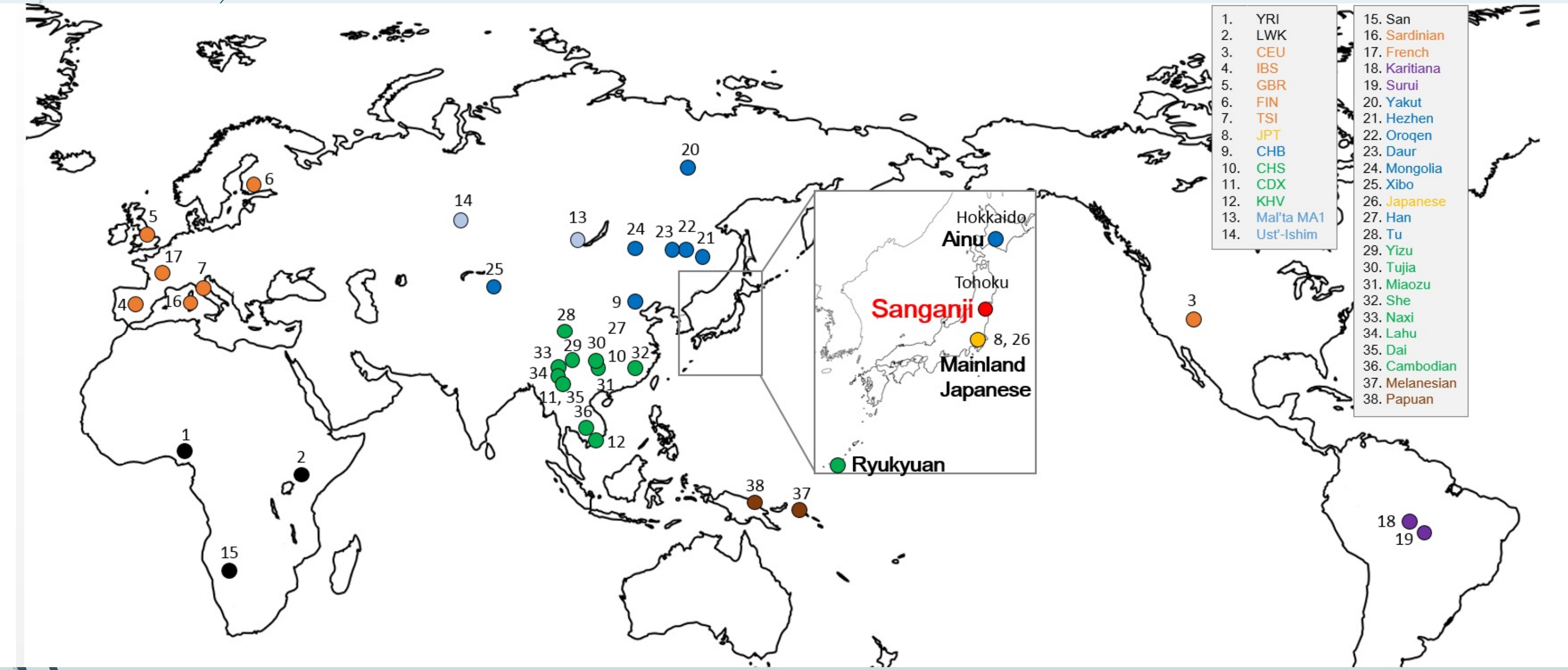


Depurination



Comparison with worldwide populations

- HapMap project (The International HapMap Consortium, 2005)
- Human Genome Diversity Project (HGDP) (Li et al., 2008)
- 1000 Genomes (1000 Genomes Project Consortium, 2012)
- Japanese populations (Japanese Archipelago Human Population Genetics Consortium, 2012)



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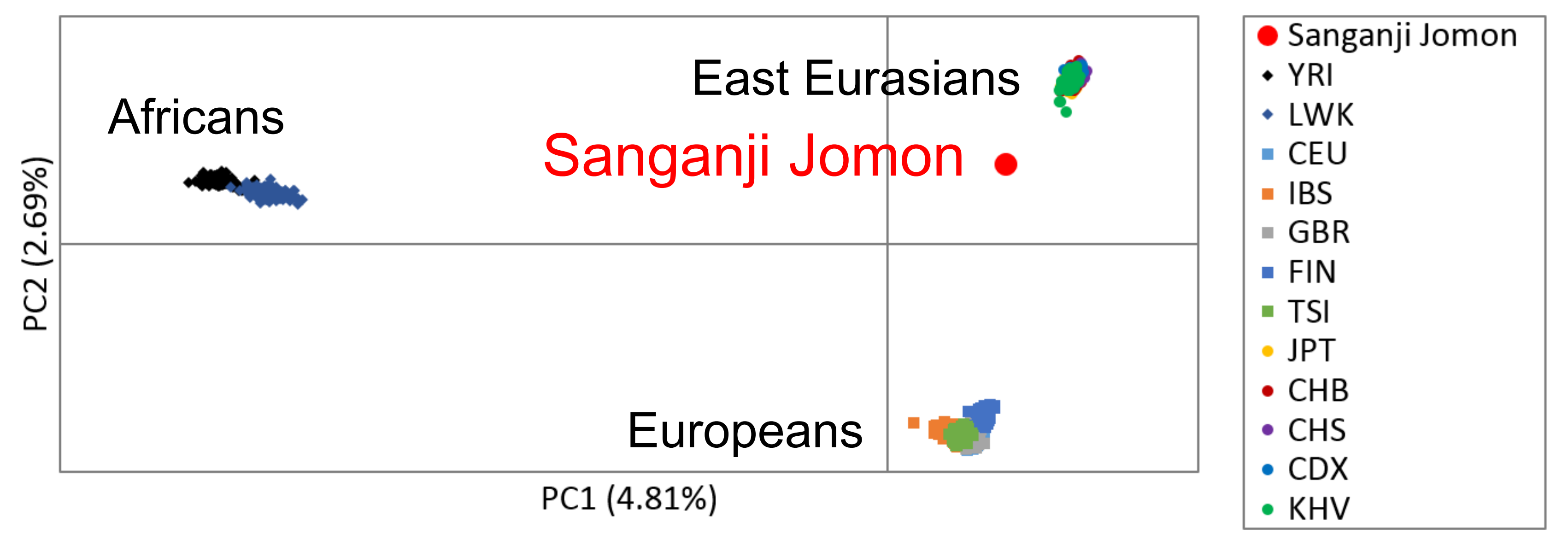
Jomon and 1KG worldwide populations

- Creating homozygous diploid genotypes
- HWE p<1e-10, minor allele <0.01, genotype call <0.95 (PLINK)
- PCA (EIGENSOFT, smartpca)

- 68,542 SNPs



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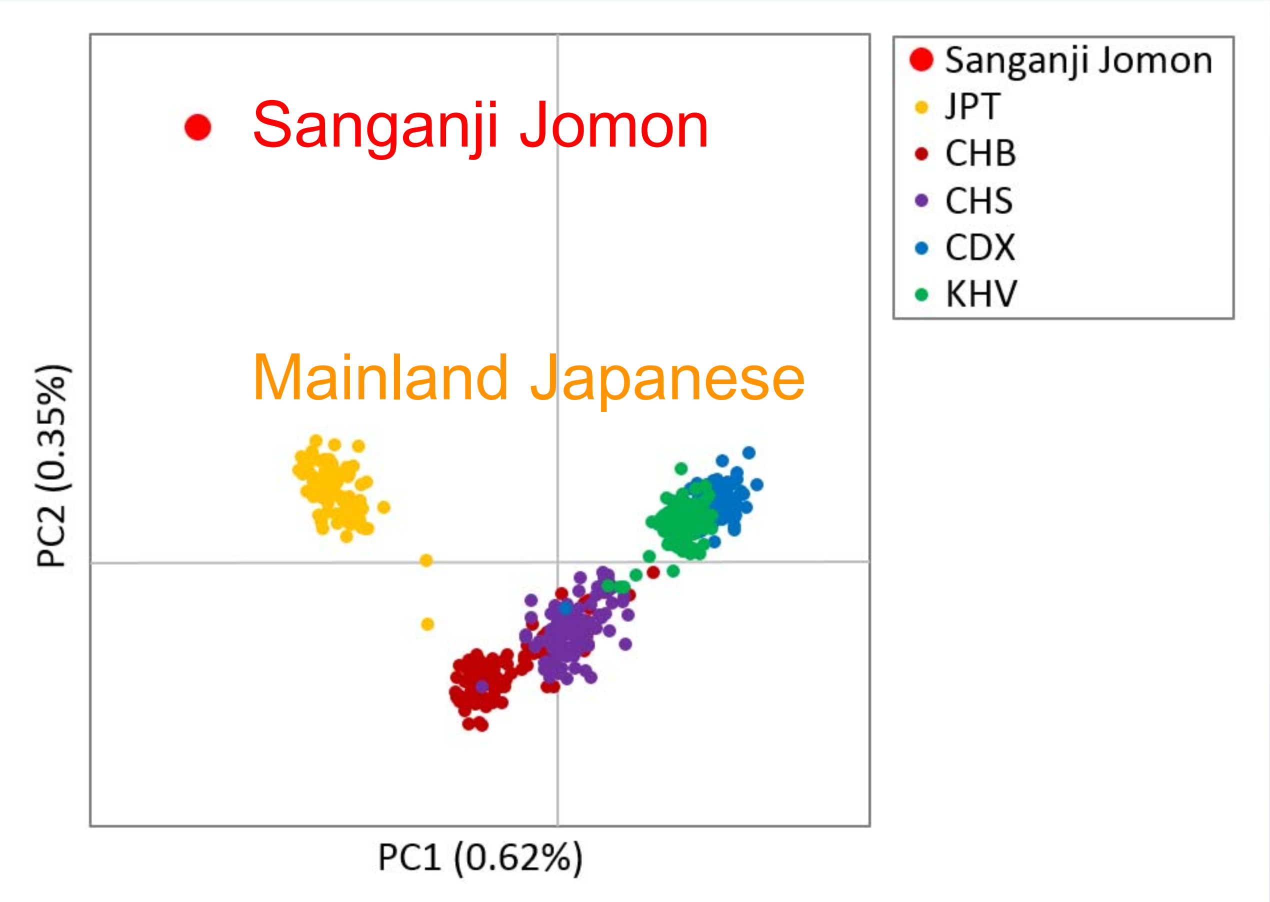
Jomon and 1KG East Eurasians

- 46,158 SNPs



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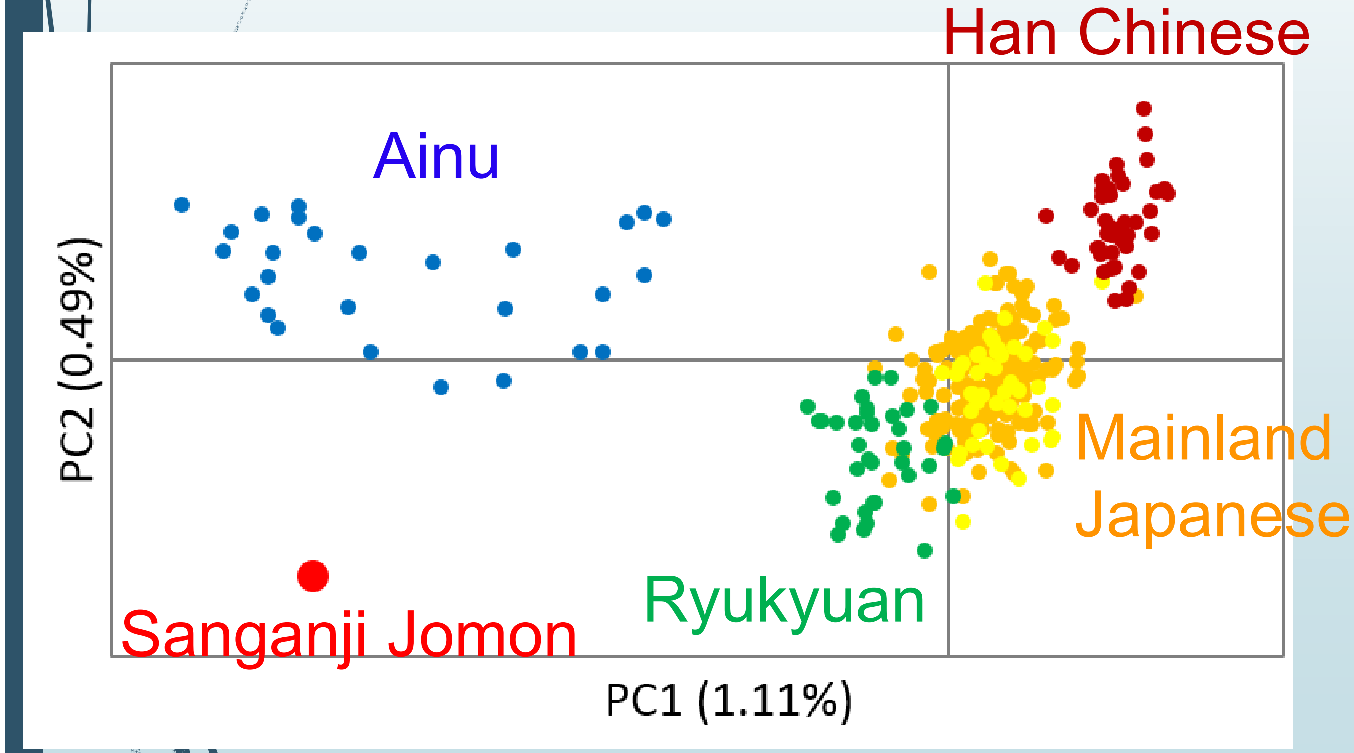
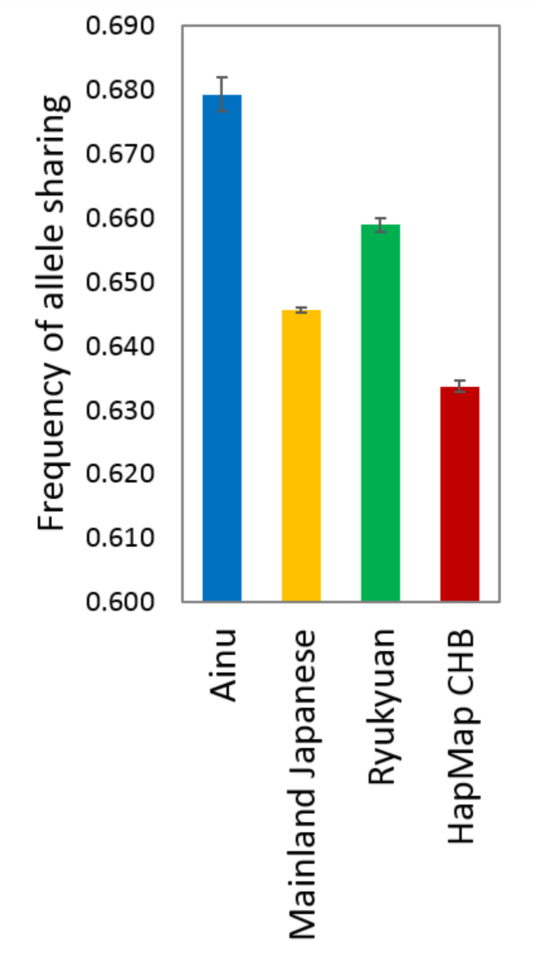
Comparison with Ainu, Mainland Japanese, and Ryukyuan

- 5,392 SNPs





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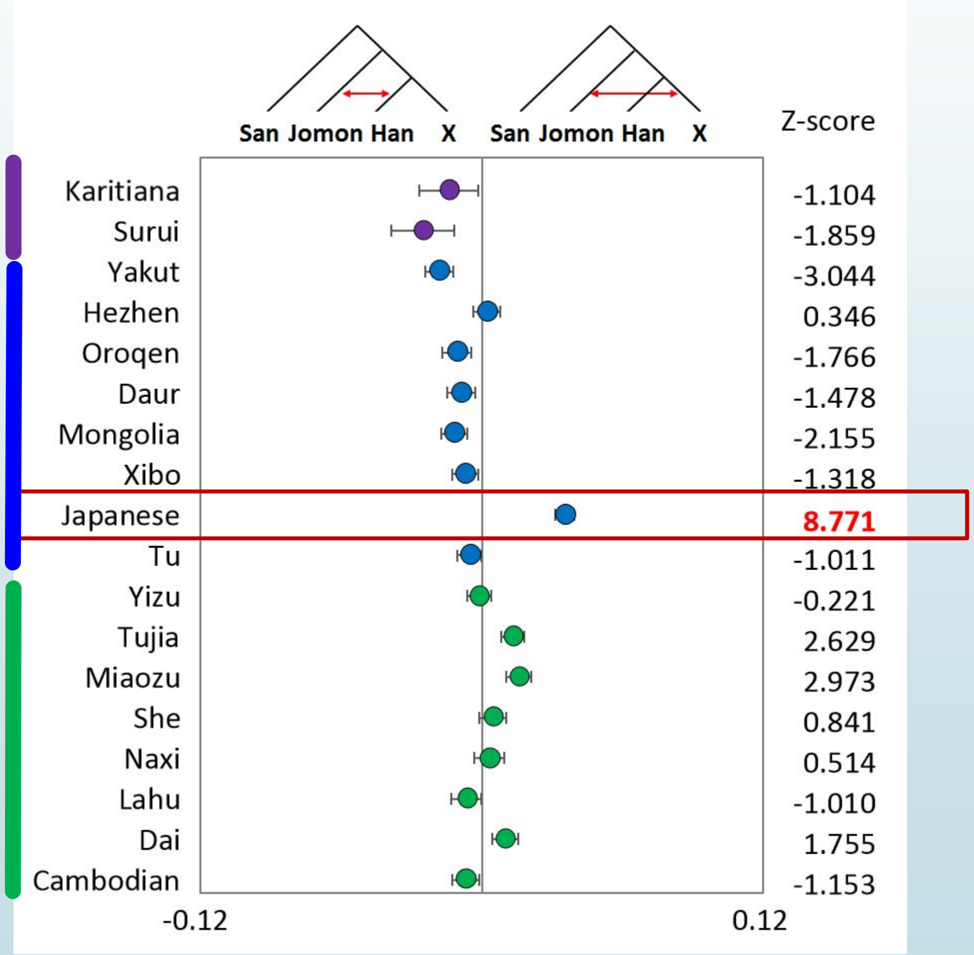
D-statistics to detect **gene flow**

- 7,529 SNPs

Native Americans

Northern East Asians

Southern East Asians



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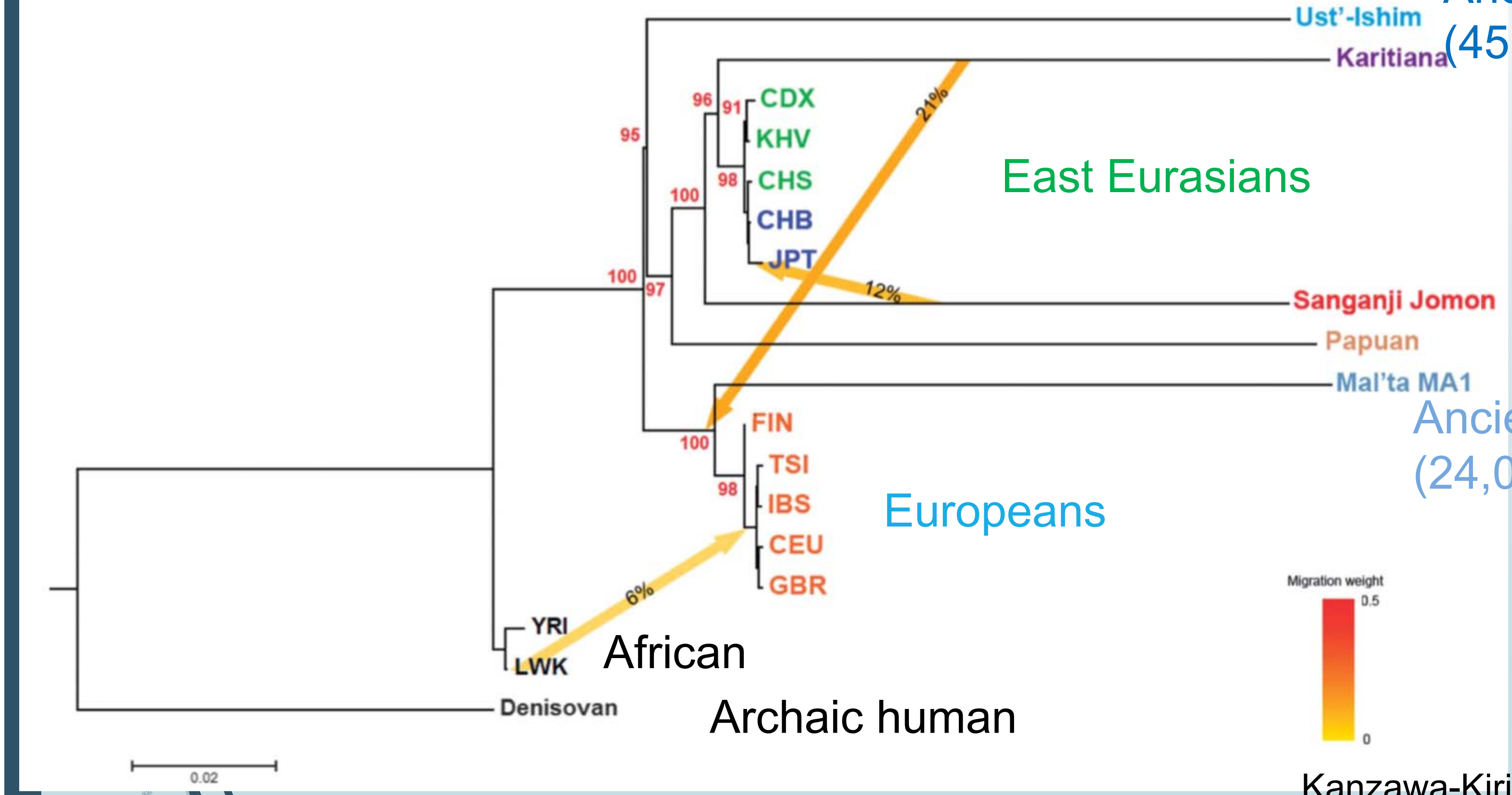
Jomon diverged from ancestor of East Eurasians

- ML tree with 3 gene flow events base on 43,310 SNPs using TreeMix



Ancient W. Siberian (45,000 YBP)

East Eurasians

Ancient S.C. Siberian (24,000 YBP)

Europeans

African

Archaic human

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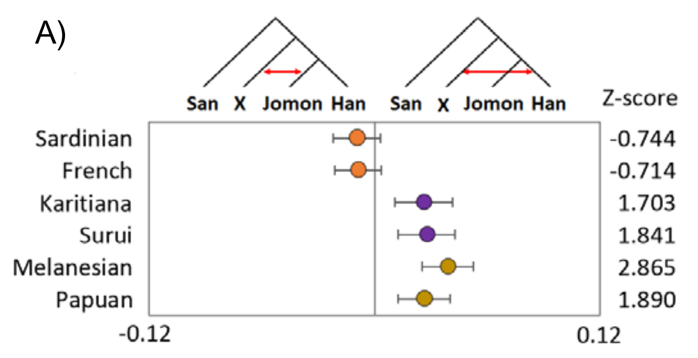
No gene flow with other modern/ancient Eurasians

A) 7,529 SNPs



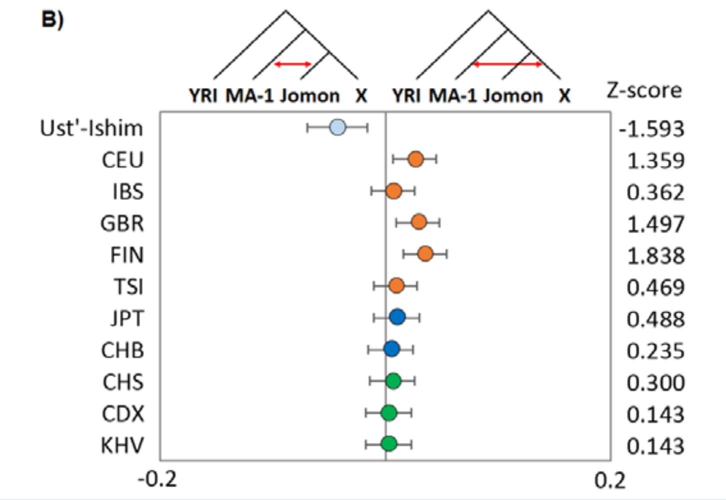
Comparison with

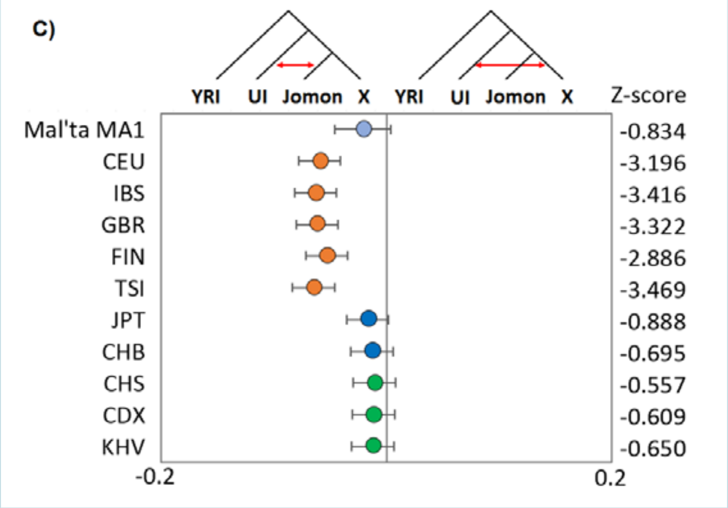
A) Europeans, Native Americans, and Oceanians

B) Mal'ta MA1 (24,000 YBP Siberians)

C) Ust'-Ishim (45,000 YBP Siberians)

B), C) 15,549 SNPs (only Tv)





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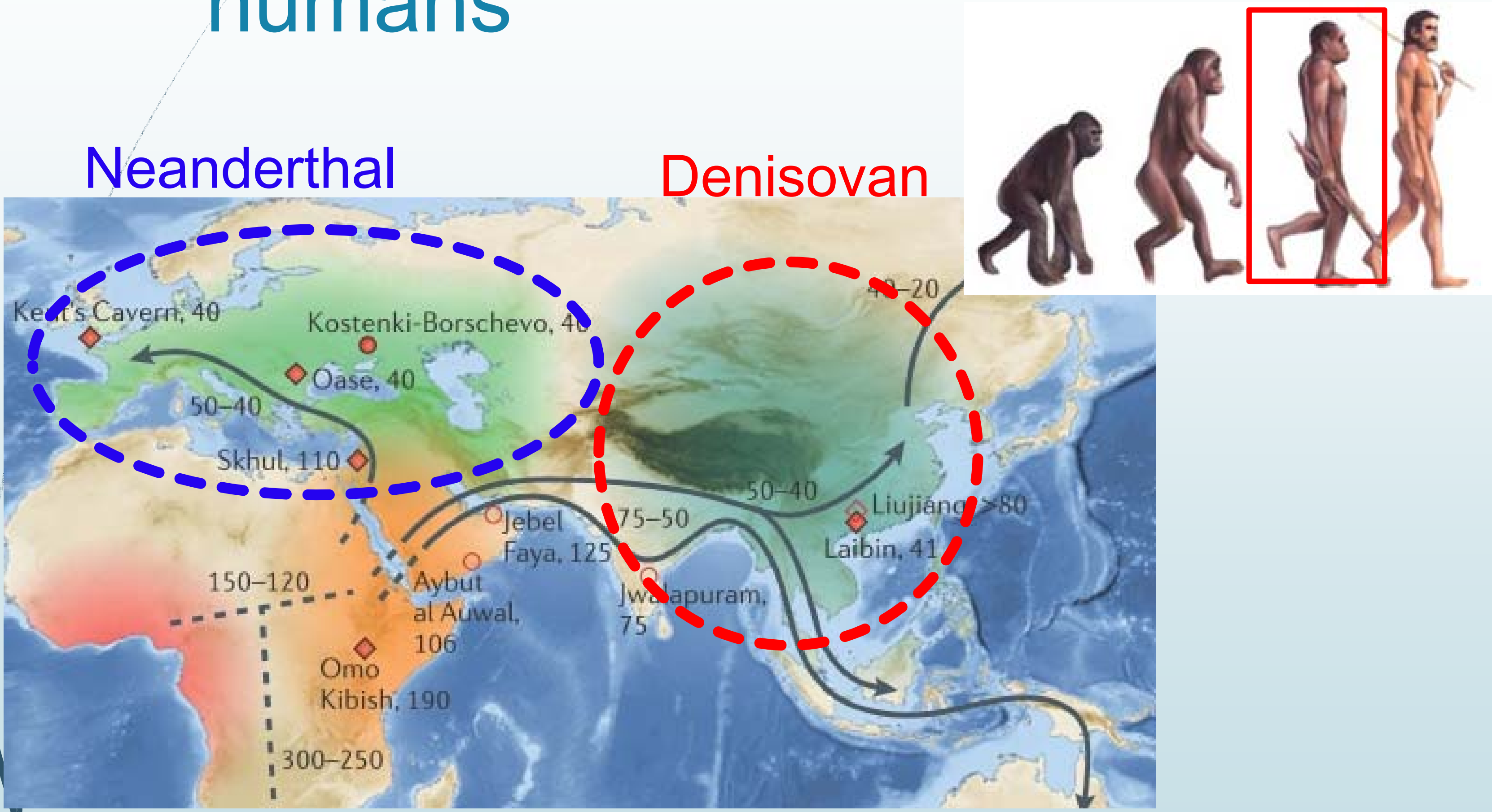
Comparison with Archaic humans

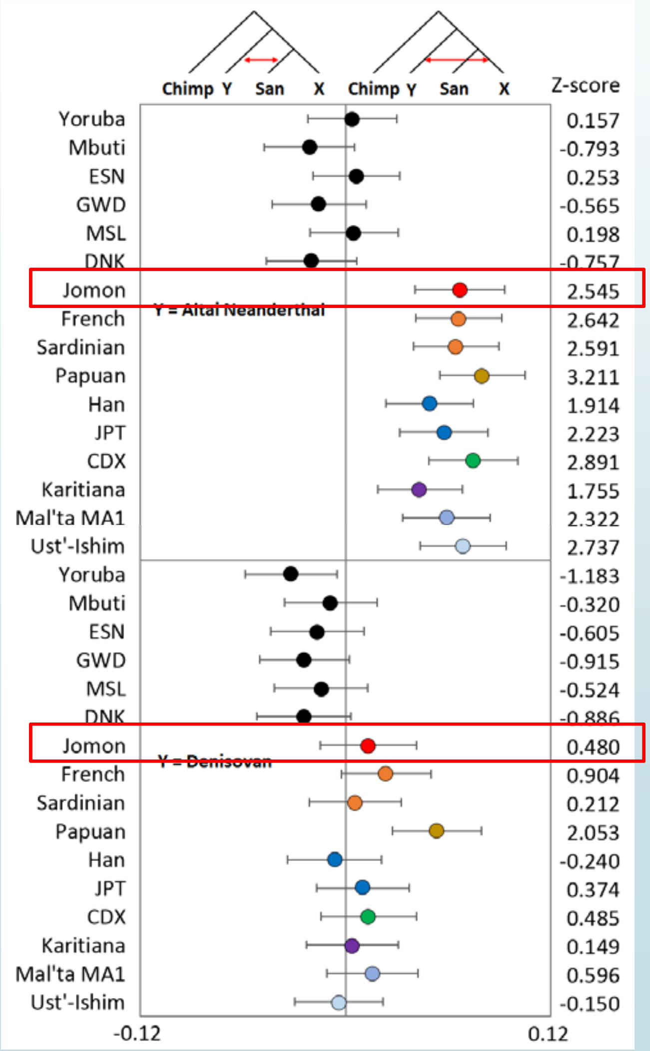


Admixture : Neanderthal ― Non Africans
Denisovan ― Oceanians

No additional gene flow with Archaic humans

Neanderthal

Denisovan



Africans

Non-Africans

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Conclusion

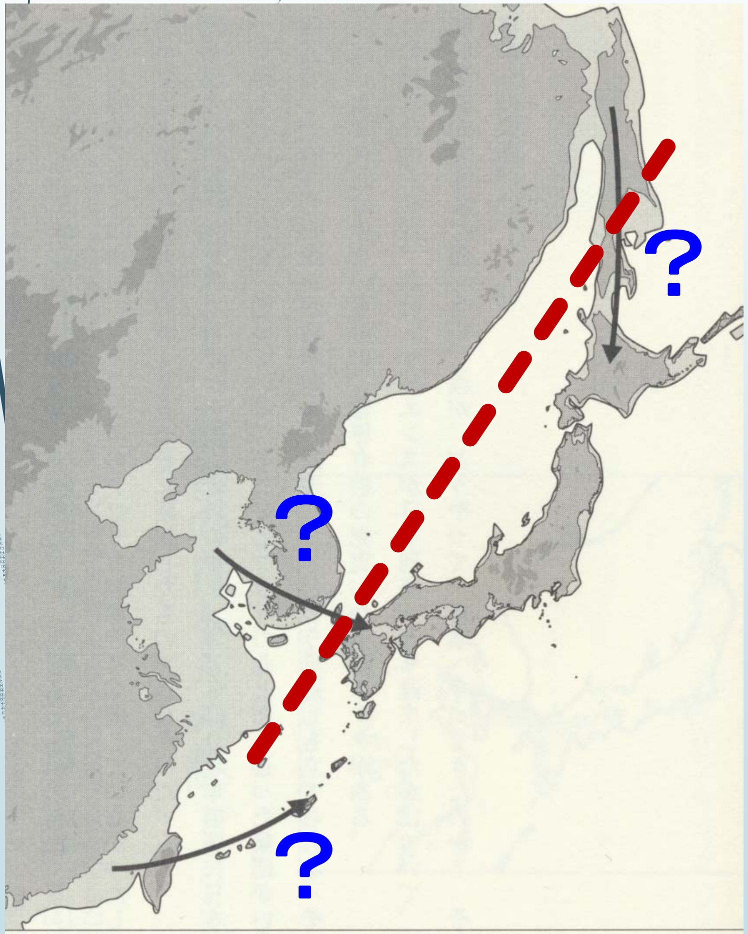

図5-6　3万8000年前頃の日本列島への3つの可能な渡来ルート

Q1: Genetic relationship with modern humans

- Deep divergence
- Genetic Isolation from continental populations after the divergence
- Affinity to three Japanese populations

Q2: Origin of Jomon

- No answer…

Thank you for your listening

2014.08.03
Shitsukari-Abe cave