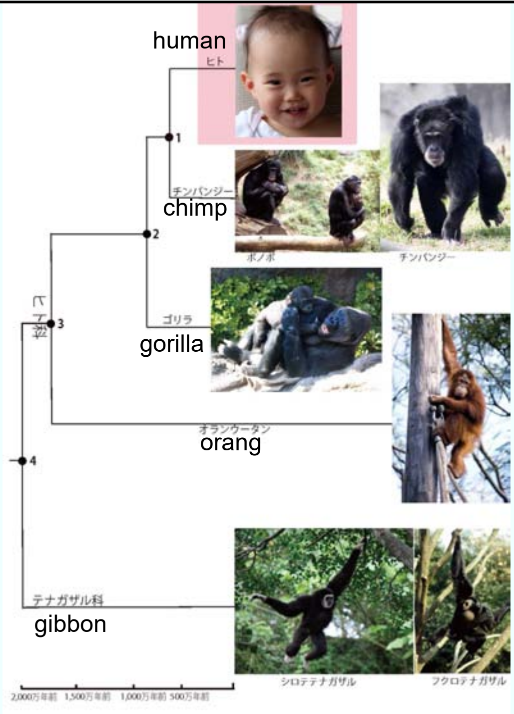
Importance of Population Genetics

Gene trees do not necessarily reflect phylogenetic relationships among species (species tree or population tree).



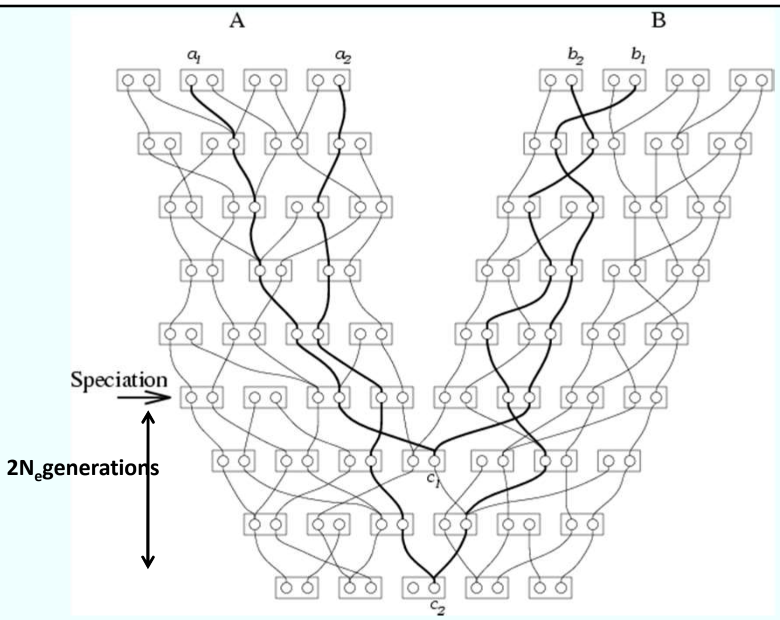
Established Tree of Hominoidea

Chimpanzees and bonobos are closer to humans than to gorillas . This relationship was established by Satoshi Horai in 1990's with the mitochondrial genome analysis, but …

Number of genes significantly (PP>0.95) supporting the 3 topologies among human, chimp and gorilla

	Gene	**%**
((Human,Chimp),Gorilla)	3,814	78.9
((Chimp,Gorilla),Human)	504	10.4*
((Human,Gorilla),Chimp)	492	10.2*

*: Incongruent trees (20.6%)
Modified from Table 3 of Ebersberger et al. (2007, *Mol. Biol. Evol.* 24, 2266-2276)



Under the neutral evolution, a separation in a gene tree is expected to be older than the speciation time by $2N_e$ generations .

Hasegawa, Kishino & Yano (1989)
J. Human Evol. 18, 461-476

Molecular evolution rate

Evolution rate: v (substitutions/site/year)
Effective population size: N_e
Mutation rate: μ
Probability of fixation of the gene: u
$v = 2N_e \mu u = \mu$: Under the neutral theory（$u = 1/2N_e$）

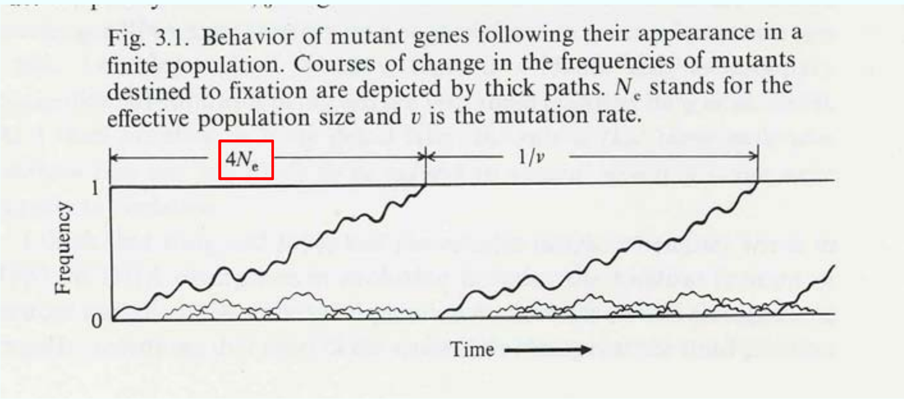
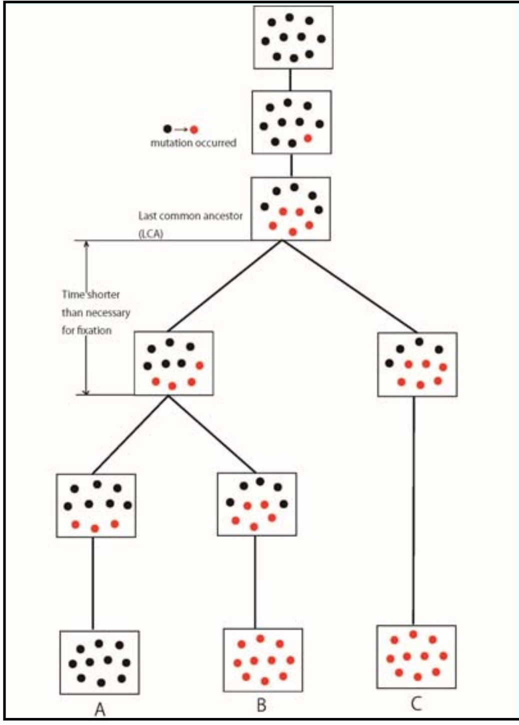
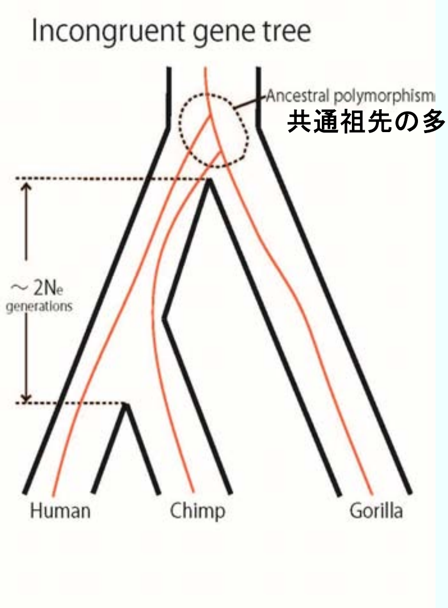


Fig. 3.1. Behavior of mutant genes following their appearance in a finite population. Courses of change in the frequencies of mutants destined to fixation are depicted by thick paths. N_e stands for the effective population size and v is the mutation rate.



Incomplete lineage sorting of ancestral polymorphism:

A/B clade in the species tree, but B can be closer to C in a gene tree.

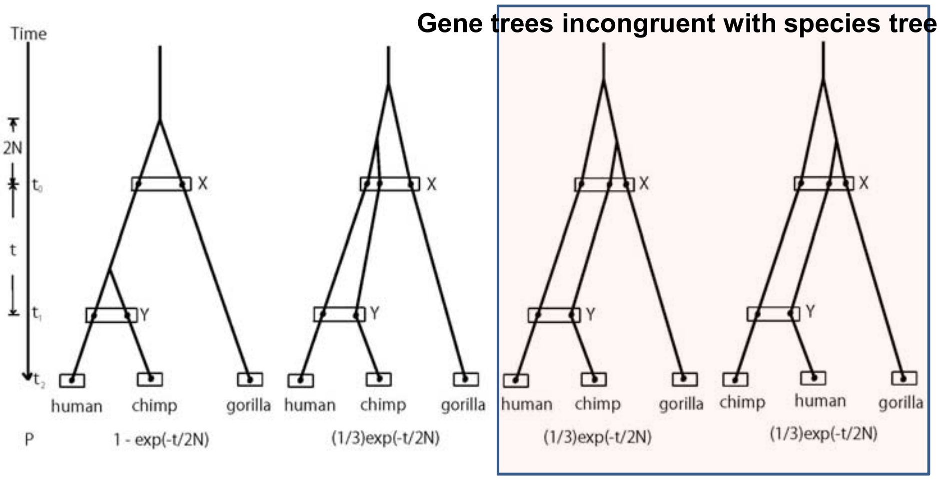


N_e: Effective population size

Incongruent gene tree from the species tree by incomplete lineage sorting of ancestral polymorphism

The time separation the two speciation events is shorter than $2N_e$ generations

Gene trees incongruent with species tree



Relationships between the species tree and the gene tree for the case of the three species of human, chimpanzee and gorilla. Redrawn from Pamilo and Nei (1988).

P：Probability of a gene tree under the neutral theory.

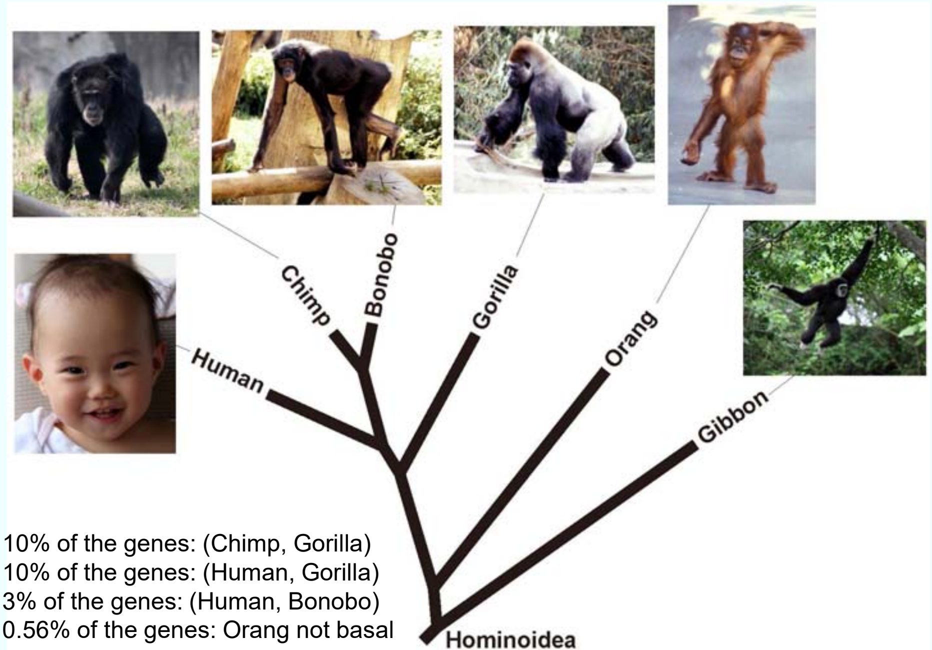
Probabilities of gene trees

$T/2N_e$	((human, chimp), gorilla)	(human,(chimp,gorilla)) + ((human,gorilla),chimp)
	$1-e^{-T/2N_e}$	$(2/3)e^{-T/2N_e}$
0.5	59.6%	40.4%
1	75.5%	24.5%
2	91.0%	9.0%
4	98.8%	1.2%

Number of genes significantly (PP>0.95) supporting the 3 topologies among human, chimp and gorilla

	Gene	**%**
((Human,Chimp),Gorilla)	3,814	78.9
((Chimp,Gorilla),Human)	504	10.4*
((Human,Gorilla),Chimp)	492	10.2*

*: Incongruent trees (20.6%)
Modified from Table 3 of Ebersberger et al. (2007, *Mol. Biol. Evol*. 24, 2266-2276)



10% of the genes: (Chimp, Gorilla)
10% of the genes: (Human, Gorilla)
3% of the genes: (Human, Bonobo)
0.56% of the genes: Orang not basal among great apes

Ebersberger et al. (2007)

Human Genetic Ancestry 2271

% of gene trees rejecting the human/chimp clade

Estimated effective population size of the human/chimp common ancestor

Effective population size of X-chromosome is ¾ of those of autosomes, …

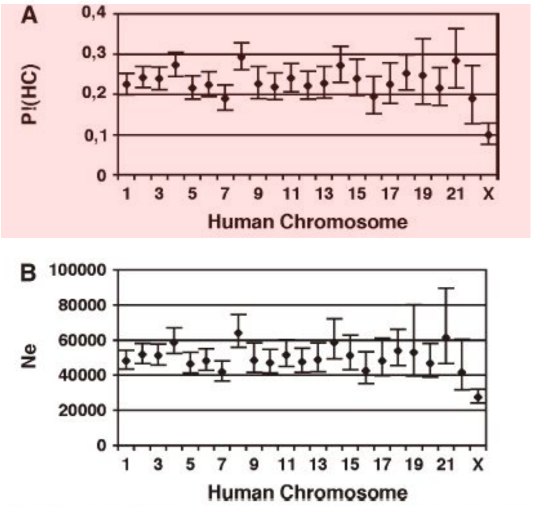
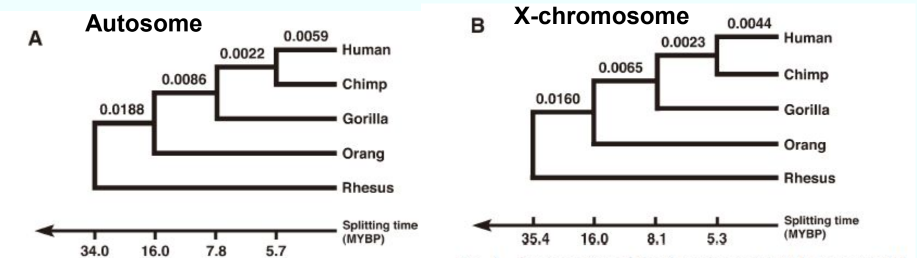


FIG. 3.—(*A*) Variation of the probability to observe a sequence tree significantly rejecting the human–chimp sister group (*P*!(HC)) among the human chromosomes. (*B*) Variation of the effective population size estimate of the human–chimp ancestral species among the human chromosomes. Bars denote the 95% CI.

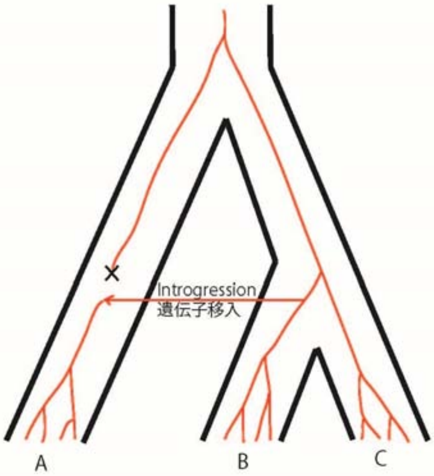


Ebersberger et al. (2007)

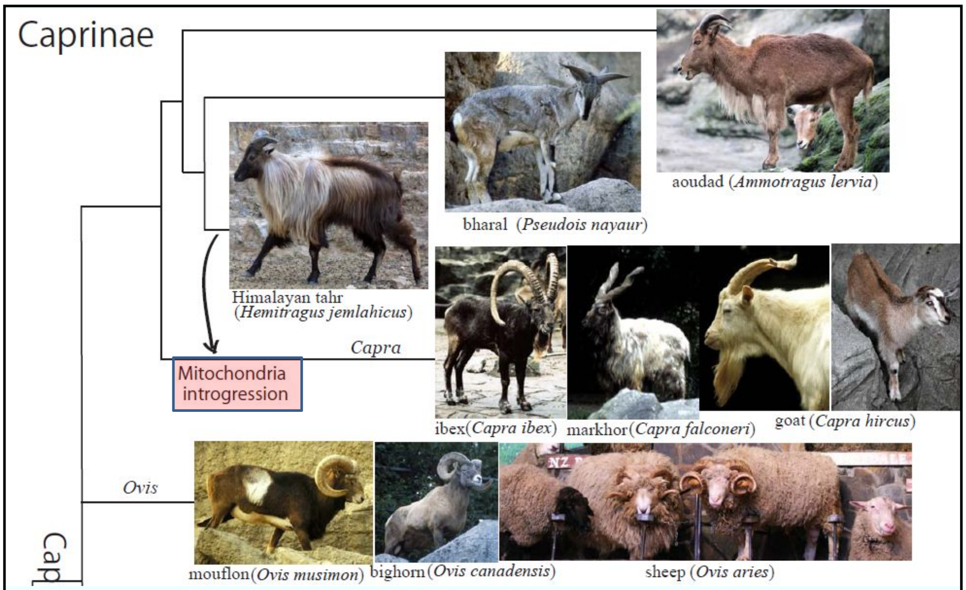
FIG. 4.—Sequence tree of the 5 species under study reconstructed from the autosomal sequences (*A*) and X chromosomal sequences (*B*), respectively. Splitting times were dated assuming the split of the orangutan lineage 16 MYBP. Trees are not drawn to scale.

The branching date estimated from X-chromosome is younger than that from autosomes (closer to species branching?)
→ Effective population size of mitochondria is ¼ of that of autosome, and therefore the branching date estimated from mitogenome may be closer to species divergence than those estimated from nuclear genes, but there is another problem…

Another possibility is introgression by hybridization



Effective population size of mitochondria is ¼ of autosomal genes, and therefore the introgressed haplotype lineage can be fixed more easily in mitochondrial genomes than in nuclear ones.

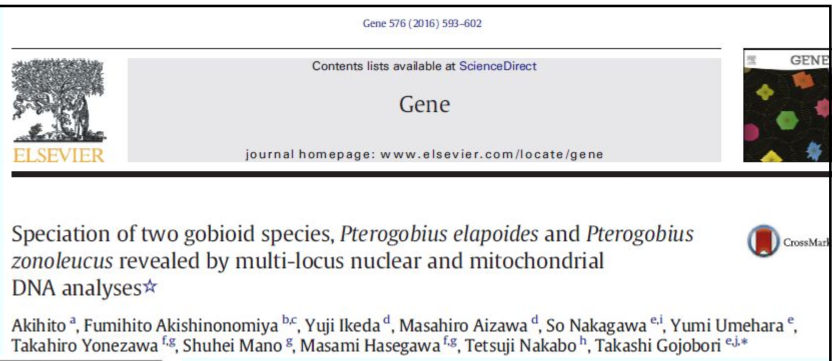


While Himalayan tahr is a sister-group to *Capra* in the mtDNA tree, it is close to bharal in the nuclear gene tree.

Merit and demerit of mitochondrial tree

- Effective population size of mtDNA is ¼ of those of nuclear autosomal genes (haploid and maternal inheritance)

  →Incomplete lineage sorting of ancestral polymorphism is less of a concern for mtDNA tree than for nuclear gene trees.
- On the other hand, small effective population size of mtDNA may facilitate fixation of introgressed haplotype lineage.



Gene 576 (2016) 593–602

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journal homepage: www.elsevier.com/locate/gene

Speciation of two gobioid species, *Pterogobius elapoides* and *Pterogobius zonoleucus* revealed by multi-locus nuclear and mitochondrial DNA analyses☆

Akihito [a], Fumihito Akishinonomiya [b,c], Yuji Ikeda [d], Masahiro Aizawa [d], So Nakagawa [e,i], Yumi Umehara [e], Takahiro Yonezawa [f,g], Shuhei Mano [g], Masami Hasegawa [f,g], Tetsuji Nakabo [h], Takashi Gojobori [e,j,*]

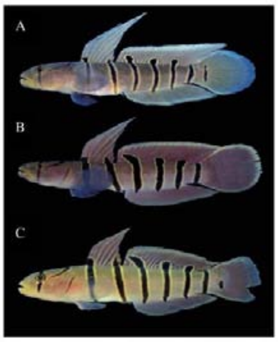


Fig. 1. *Pterogobius elapoides*: (A) The Sea of Japan, Ishikawa Prefecture (BLIH20020439); (B) The Pacific Ocean, Chiba Prefecture (BLIH20030467); (C) The Inland Sea, Ehime

Pterogobius elapoides
キヌバリ

Pterogobius zonoleucus
チャガラ

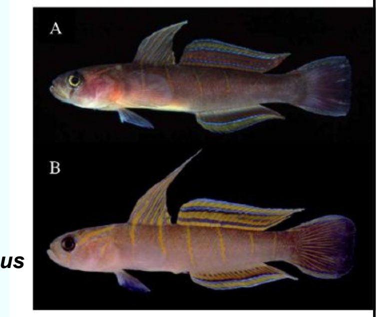


Fig. 2. *Pterogobius zonoleucus*: (A) The Sea of Japan, Ishikawa Prefecture (BLIH200505
(B) The Pacific Ocean, Chiba Prefecture (BLIH20060883).

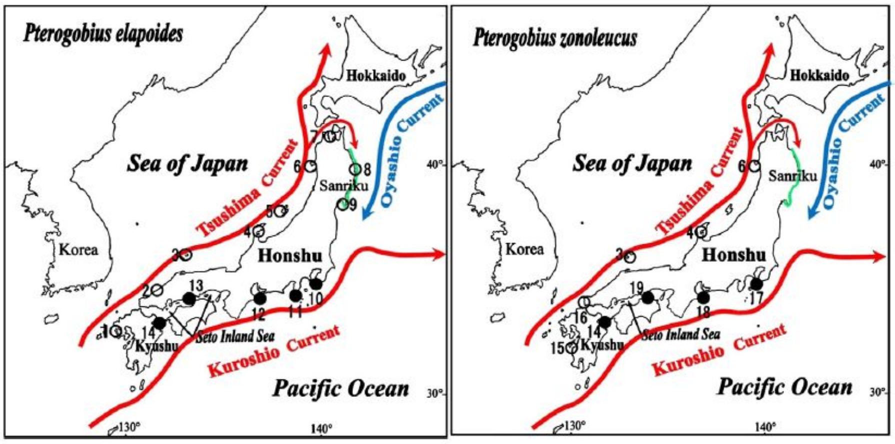
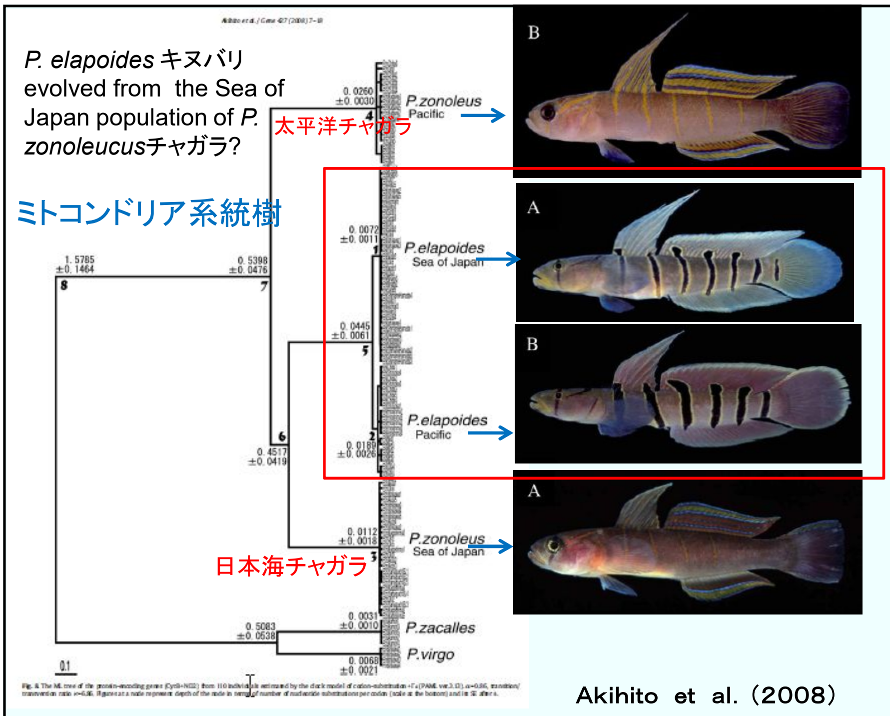


Fig. 4. The sampling localities of *Pterogobius elapoides* and *Pterogobius zonoleucus*: The Sea of Japan population(○), The Pacific Ocean population (●); each number of localities are shown in Table 1. 🟩shows the coast of the Sanriku District; 🟥show the warm currents, 🟦shows the cold current.

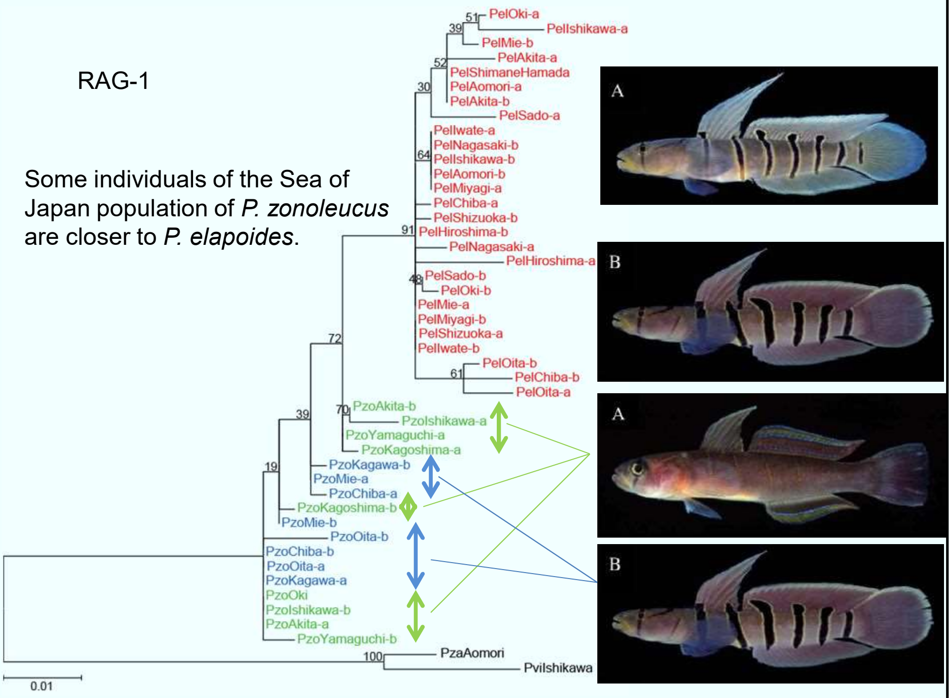
Akihito et al. (2008)



P. elapoides キヌバリ evolved from the Sea of Japan population of *P. zonoleucus*チャガラ?

ミトコンドリア系統樹

Akihito et al.（2008）

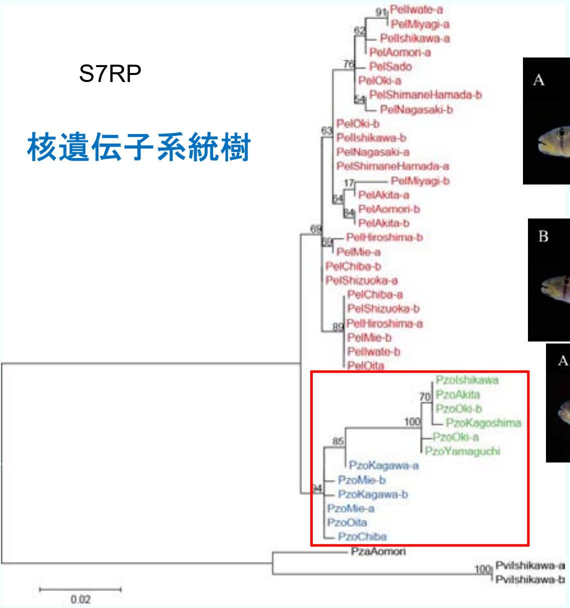


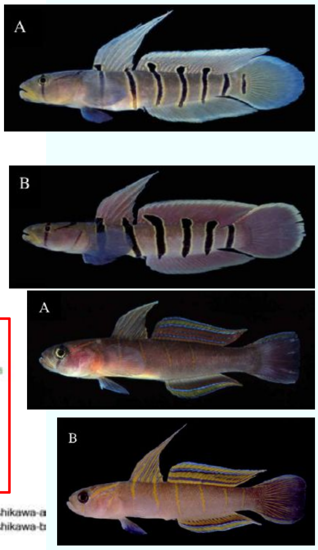
RAG-1

Some individuals of the Sea of Japan population of *P. zonoleucus* are closer to *P. elapoides*.

S7RP

核遺伝子系統樹





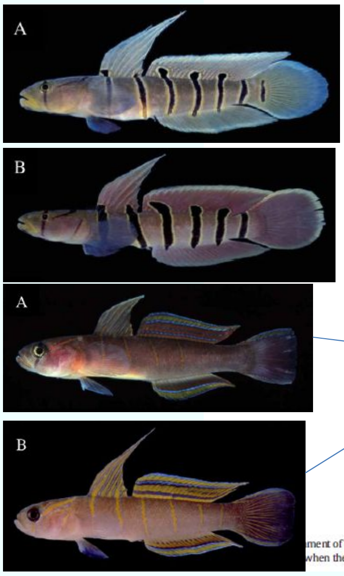
日本海
キヌバリ

太平洋
キヌバリ

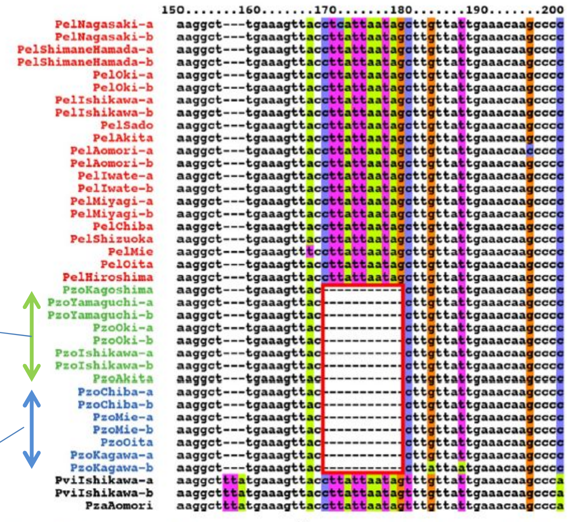
日本海
チャガラ

太平洋
チャガラ

TBR1

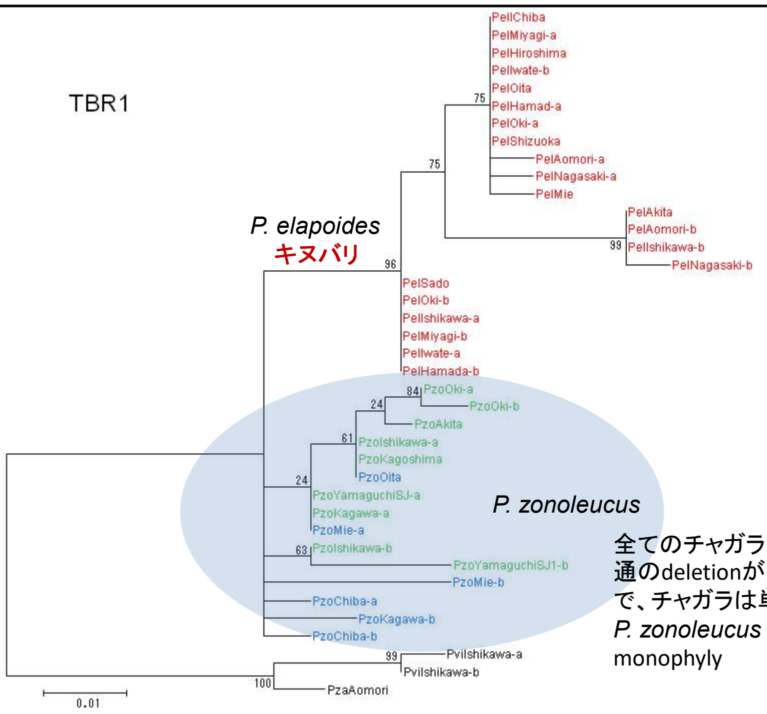


Akihito et al. / Gene 576 (2016) 593–602

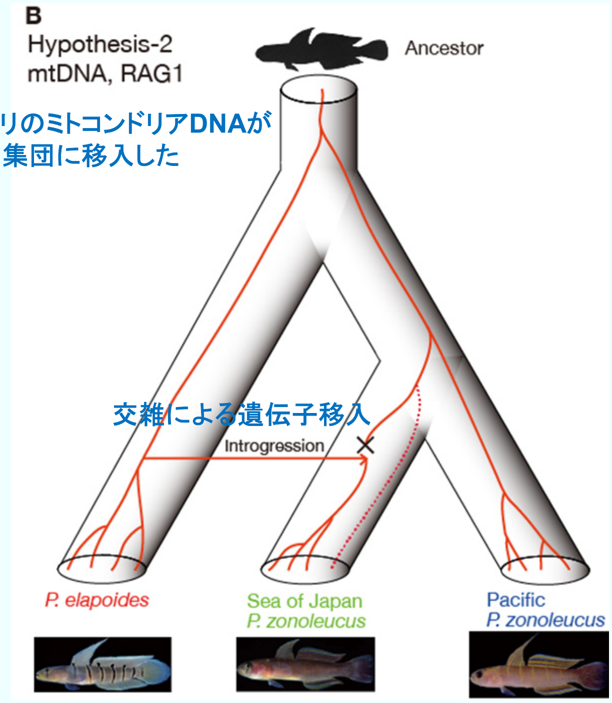


```
                  150.......160.......170.......180.......190.......200
  PelNagasaki-a   aaggct---tgaaagttacctcattaatagcttgttattgaaacaagcccc
  PelNagasaki-b   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
PelShimaneHamada-a aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
PelShimaneHamada-b aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
        PelOki-a   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
        PelOki-b   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
  PelIshikawa-a   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
  PelIshikawa-b   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
         PelSado   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
        PelAkita   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
    PelAomori-a   aaggct---tgaaagttaccttattaatagcttgttattgaaacaaccccc
    PelAomori-b   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
     PelIwate-a   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
     PelIwate-b   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
    PelMiyagi-a   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
    PelMiyagi-b   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
        PelChiba   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
    PelShizuoka   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
          PelMie   aaggct---tgaaagtttccttattaatagcttgttattgaaacaagcccc
         PelOita   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
    PelHiroshima   aaggct---tgaaagttaccttattaatagcttgttattgaaacaagcccc
    PzoKagoshima   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
  PzoYamaguchi-a   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
  PzoYamaguchi-b   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
        PzoOki-a   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
        PzoOki-b   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
   PzoIshikawa-a   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
   PzoIshikawa-b   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
        PzoAkita   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
      PzoChiba-a   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
      PzoChiba-b   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
        PzoMie-a   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
        PzoMie-b   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
         PzoOita   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
     PzoKagawa-a   aaggct---tgaaagttac-----------cttgttattgaaacaagcccc
     PzoKagawa-b   aaggct---tgaaagttac-----------cttattaatgaaacaagcccc
   PviIshikawa-a   aaggctttatgaaagttaccttattaatagtttgttattgaaacaagccca
   PviIshikawa-b   aaggctttatgaaagttaccttattaatagtttgttattgaaacaagccca
       PzaAomori   aaggctttatgaaagttaccttattaatagtttgttattgaaacaagccca
```

ment of TBR1 nucleotide sequence positions from 150 to 200. The red open rectangular box indicates a deletion observed only in *P. zonoleucus*
when the nucleotide was not conserved at a position.



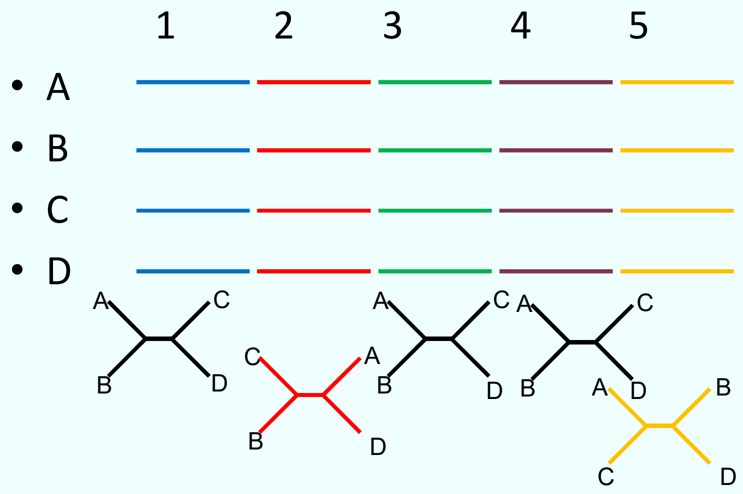
全てのチャガラに共通のdeletionがあるので、チャガラは単系統
P. zonoleucus
monophyly



交雑によりキヌバリのミトコンドリアDNAが日本海チャガラの集団に移入した

交雑による遺伝子移入

Analysis of concatenated sequences



Since gene trees can be incongruent among different genes, it cannot be justified to analyze concatenated sequences.

Problems of the "concatenated sequence" analyses

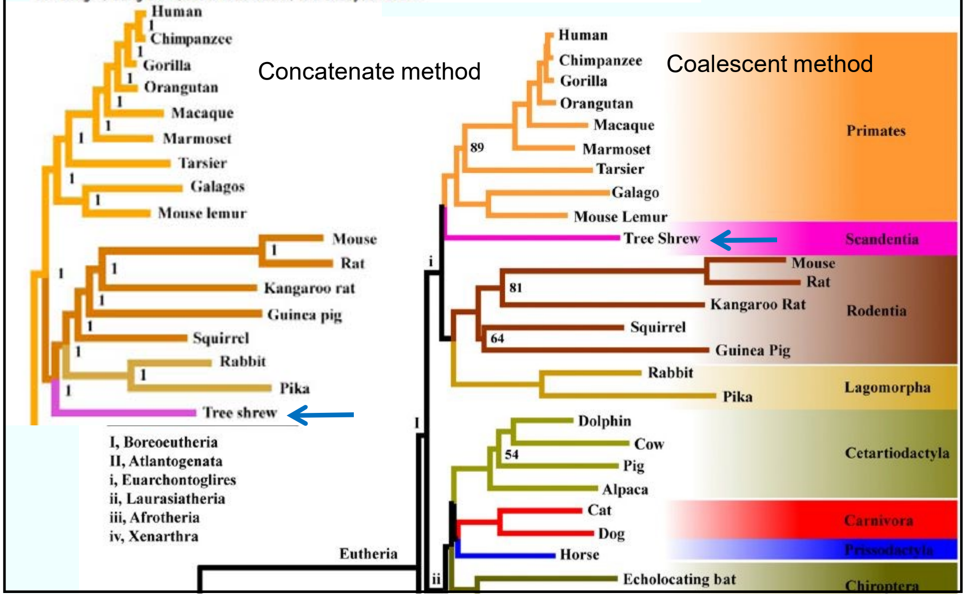
- Each gene has its own history, and gene trees can be different among different genes.
- Concatenation of different genes ignores this.
- Gene tree vs. Species tree

Importance of population genetics

Resolving conflict in eutherian mammal phylogeny using phylogenomics and the multispecies coalescent model

Sen Song[a,1], Liang Liu[b,1], Scott V. Edwards[c], and Shaoyuan Wu[b,d,2]

PNAS 2012


Concatenate method

Coalescent method

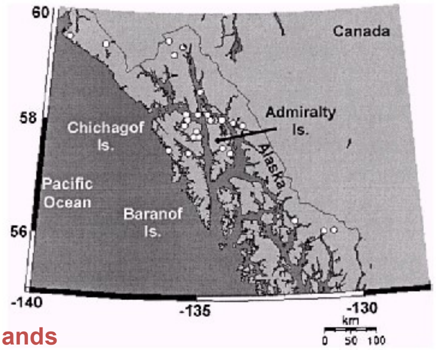
Molecular Phylogenetics and Evolution
Vol. 15, No. 2, May, pp. 319–326, 2000
doi:10.1006/mpev.1999.0730, available online at http://www.idealibrary.com on IDEAL®

Phylogeography of Mitochondrial DNA Variation in Brown Bears and Polar Bears

Gerald F. Shields, Deborah Adams, Gerald Garner,*,[1] Martine Labelle, Jacy Pietsch, Malcolm Ramsay,† Charles Schwartz,‡ Kimberly Titus,§ and Scott Williamson

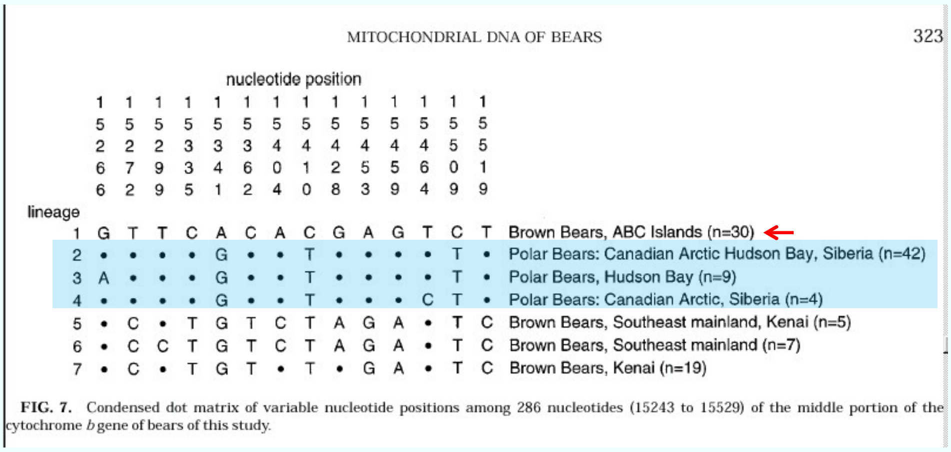
of Biology and Wildlife,
e, Alaska Science Cente
atoon, Saskatchewan S
n, Soldotna, Alaska 99
of Wildlife Conservatio

Received June 4, 1999; re



ABC Islands

FIG. 1. Locations (open circles) on Admiralty, Baranof, and Chichagof islands and on the coastal mainland of southeast Alaska where tissues of brown bears were obtained.



MITOCHONDRIAL DNA OF BEARS 323

nucleotide position

lineage	15266	15272	15299	15335	15341	15362	15404	15410	15428	15453	15459	15464	15509	15519	
1	G	T	T	C	A	C	A	C	G	A	G	T	C	T	Brown Bears, ABC Islands (n=30) ←
2	•	•	•	•	G	•	•	T	•	•	•	•	T	•	Polar Bears: Canadian Arctic Hudson Bay, Siberia (n=42)
3	A	•	•	•	G	•	•	T	•	•	•	•	T	•	Polar Bears, Hudson Bay (n=9)
4	•	•	•	•	G	•	•	T	•	•	•	C	T	•	Polar Bears: Canadian Arctic, Siberia (n=4)
5	•	C	•	T	G	T	C	T	A	G	A	•	T	C	Brown Bears, Southeast mainland, Kenai (n=5)
6	•	C	C	T	G	T	C	T	A	G	A	•	T	C	Brown Bears, Southeast mainland (n=7)
7	•	C	•	T	G	T	•	T	•	G	A	•	T	C	Brown Bears, Kenai (n=19)

FIG. 7. Condensed dot matrix of variable nucleotide positions among 286 nucleotides (15243 to 15529) of the middle portion of the cytochrome *b* gene of bears of this study.

MtDNA of brown bear from ABC Islands is closer to those of polar bears than to those of brown bears from other areas.

SHIELDS ET AL. (2000) *MPE* 15:319-326

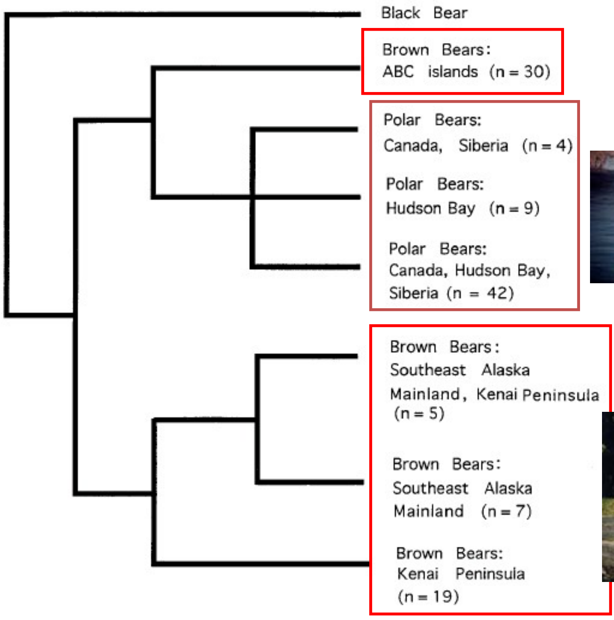
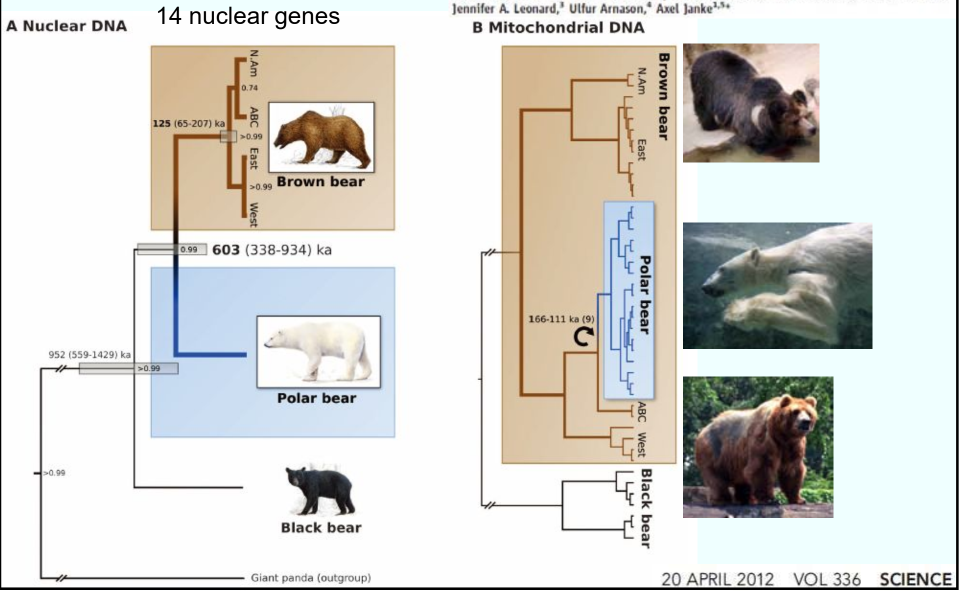


FIG. 8. Unweighted maximum-parsimony tree for the 286 nucleotides of the seven mtDNA lineages of bears of this study.

Nuclear Genomic Sequences Reveal that Polar Bears Are an Old and Distinct Bear Lineage

Frank Hailer,[1*] Verena E. Kutschera,[1] Björn M. Hallström,[1] Denise Klassert,[1] Steven R. Fain,[2] Jennifer A. Leonard,[3] Ulfur Arnason,[4] Axel Janke[1,5*]

14 nuclear genes



20 APRIL 2012 VOL 336 SCIENCE

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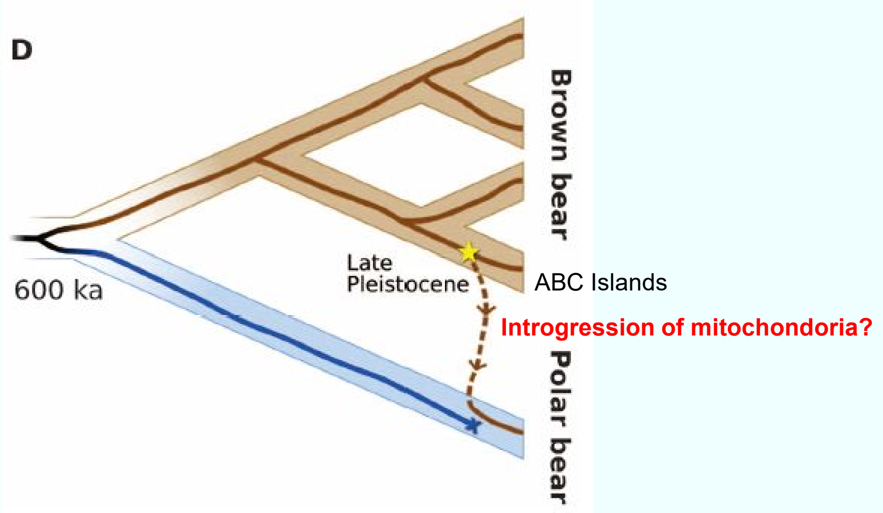

ABC Islands

Introgression of mitochondria?

20 APRIL 2012 VOL 336 SCIENCE

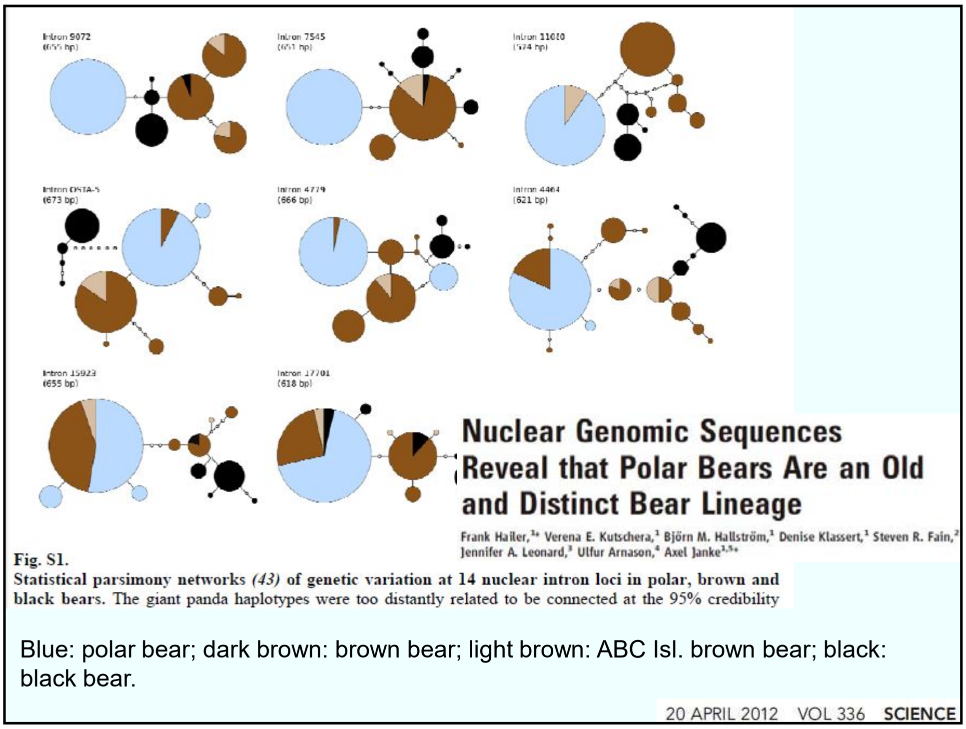

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Fig. S1.
Statistical parsimony networks *(43)* **of genetic variation at 14 nuclear intron loci in polar, brown and black bears.** The giant panda haplotypes were too distantly related to be connected at the 95% credibility

Blue: polar bear; dark brown: brown bear; light brown: ABC Isl. brown bear; black: black bear.

20 APRIL 2012 VOL 336 **SCIENCE**

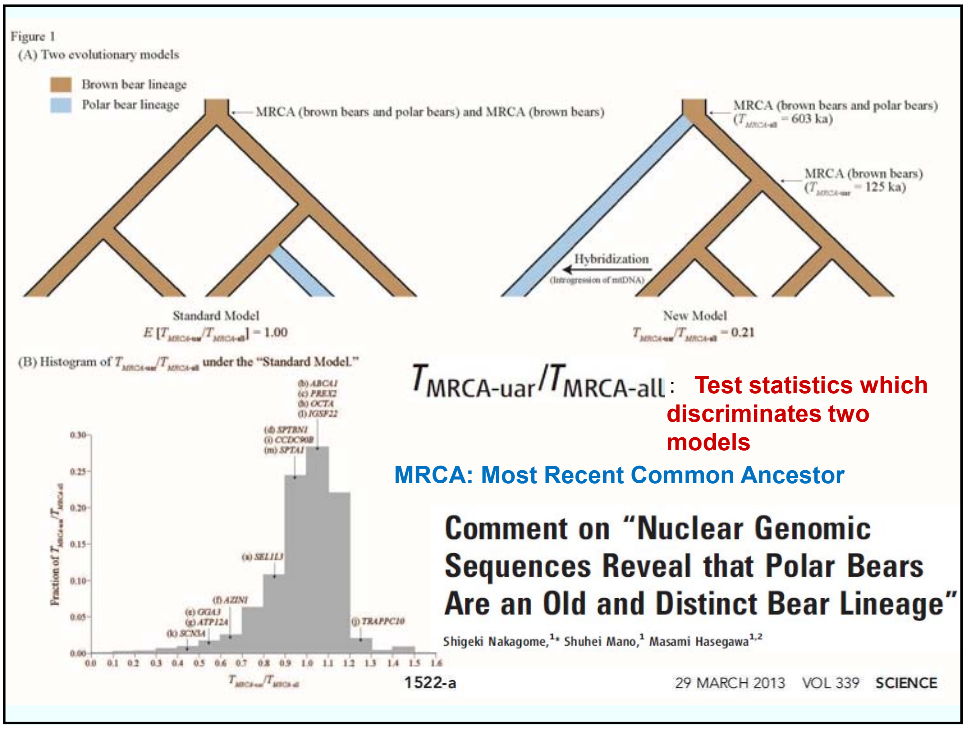

Figure 1
(A) Two evolutionary models

(B) Histogram of $T_{MRCA\text{-}uar}/T_{MRCA\text{-}all}$ under the "Standard Model."

$T_{\text{MRCA-uar}}/T_{\text{MRCA-all}}$: **Test statistics which discriminates two models**

MRCA: Most Recent Common Ancestor

Comment on "Nuclear Genomic Sequences Reveal that Polar Bears Are an Old and Distinct Bear Lineage"

Shigeki Nakagome,[1*] Shuhei Mano,[1] Masami Hasegawa[1,2]

1522-a

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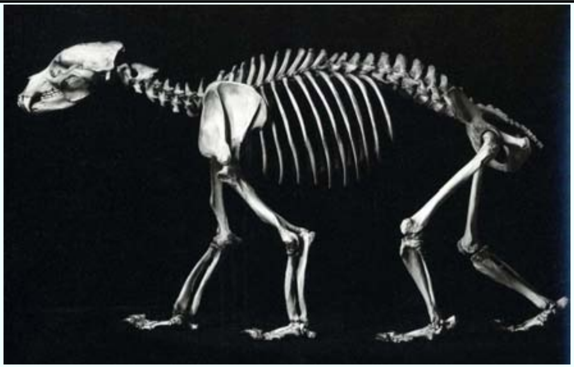
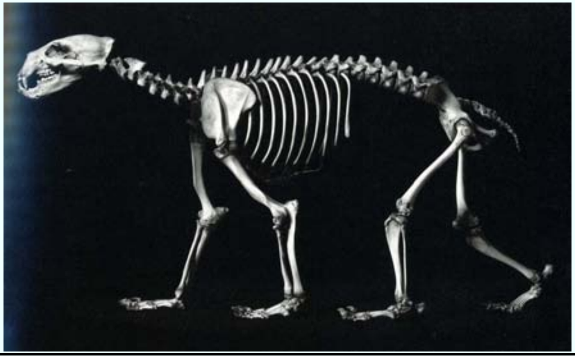


Brown bear (Grizzly)



Polar bear

「骨から見る生物の進化」
J.-B. de Panafieu