

Phylogeny from Transposable Elements

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Transposable elements: “jumping genes”

- ▶ Mobile segments of DNA
- ▶ Transposition: either cut-and-paste or copy-and-paste.
- ▶ Originated as viral infections.
- ▶ There are several types of transposable element, which I won't go into.

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Somatic retrotransposition

- ▶ Transposition happens not only during reproduction, but also during growth and development. You are a mosaic of genetically different cells.
- ▶ Transposition is especially active in brain cells.
- ▶ Associated with neurological problems including PTSD, alcoholism, autism, schizophrenia, and aging.
- ▶ Not known whether transposition is cause or effect.
(Erwin, Marchetto, & Gage (2014))

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Whales

- ▶ aquatic
- ▶ no visible hind limbs
- ▶ yet they are mammals
- ▶ until recently, no transitional fossils

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Why the lack of transitional fossils?

“Such major discontinuities [as that between terrestrial mammal and whale] simply could not, unless we believe in miracles, have been crossed in geologically short periods of time through one or two transitional species occupying restricted geographical areas (Denton, 1986, p. 193).”
Denton argued that if whales evolved from land mammals, the intermediates ought to exist.

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Is it even plausible that land mammals could evolve into whales?

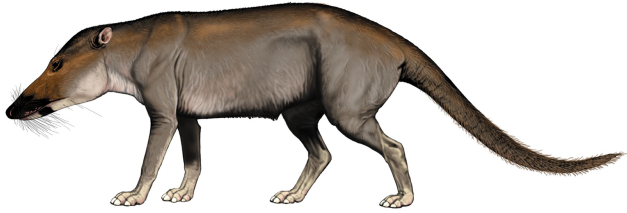
“The gradual transformation of a land animal into a whale or a sea-cow appears to be physically impossible, because the tail could not act as a propeller by vertical motion until the pelvis had been so reduced in size as to render locomotion on land impossible. If such transformation occurred gradually there must have been a long period when the ancestors of these aquatic forms, while yet poor swimmers, were unable to use the hind limbs for locomotion. How could such creatures hold their own in the struggle for existence?”

(Dewar, 1931)

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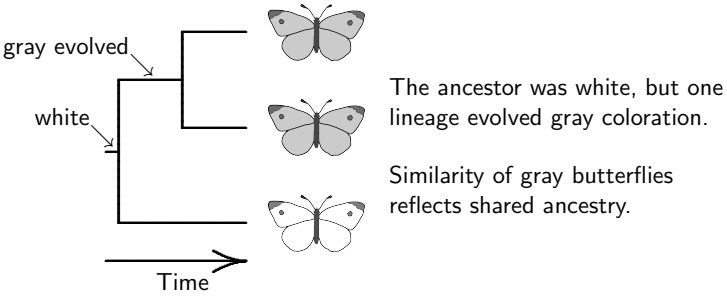
Pakicetus, an Eocene whale from Pakistan

What can genes tell us about the evolution of whales?



Hypothetical phylogenetic tree

Transposable elements



- ▶ Stretches "junk" DNA
- ▶ Copy/paste into random spots in genome.
- ▶ Exceedingly unlikely to insert in same spot twice.
- ▶ Exceedingly unlikely ever to be lost.
- ▶ Those who share a transposable element share an ancestor.
- ▶ All descendants of that ancestor share the transposable element.

Transposable elements in whales

After shuffling each column

	Transposable element															
	a	b	c	d	e	f	g	h	i	j	k	l	m	n	o	p
Bottlenose dolphin	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Narwhal	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Sperm whale	●	●	●	●	●	○	○	○	○	○	○	○	○	○	○	○
Humpback whale	●	●	○	○	○	○	○	○	○	○	○	○	○	○	○	○

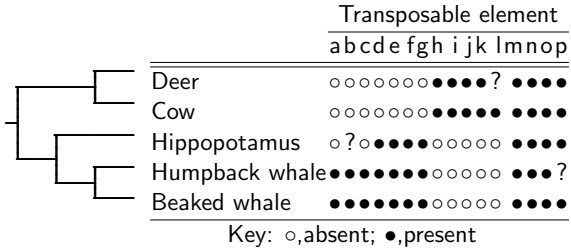
Key: ●, present; ○, absent

	Transposable element															
	a	b	c	d	e	f	g	h	i	j	k	l	m	n	o	p
Bottlenose dolphin	●	●	●	○	○	○	○	○	○	○	○	○	○	○	○	○
Narwhal	●	●	●	●	●	●	●	●	○	○	○	○	○	○	○	○
Sperm whale	●	●	○	○	○	○	○	○	○	○	○	○	○	○	○	○
Humpback whale	●	●	○	○	○	○	○	○	○	○	○	○	○	○	○	○

Key: ●, present; ○, absent

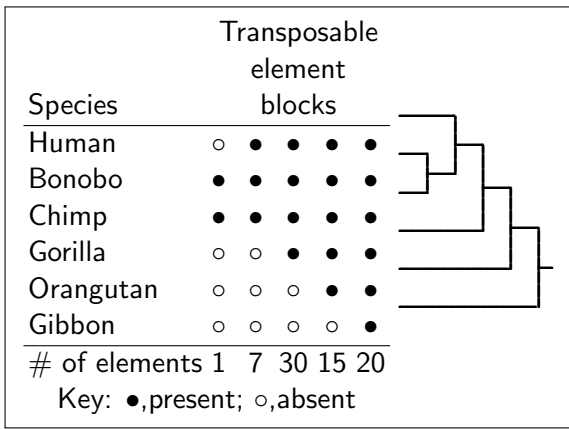
Not consistent with any tree.

Transposable elements in whales and relatives

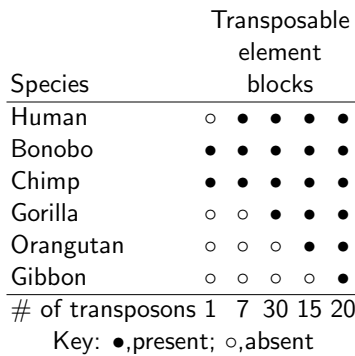


What can genes tell us about human evolution?

Transposable elements in humans and apes



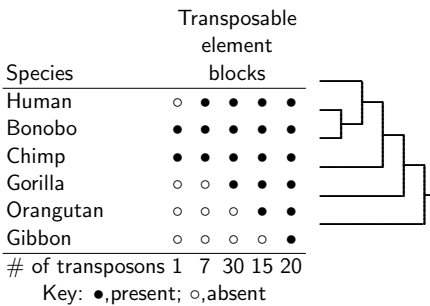
Could unrelated species share transposable elements by chance?



- ▶ Same element in 2 unrelated species exceedingly unlikely.
- ▶ In 3? verging on a miracle.
- ▶ In 4, 5, or 6? No way!

Data provide strong evidence of common ancestry.

Could nested hierarchy arise by chance?



- ▶ Suppose that 1 transposable element *did* insert into 2 species, 7 into 3 species, etc.
- ▶ How likely is it that they would fit onto a tree?
- ▶ With whales, prob was ≈ 0 with 17 elements.
- ▶ Our data have 73!

Had these elements inserted into random species, they would *not* have formed a nested hierarchy.

Summary

- ▶ Transposable elements are mobile segments of DNA that can copy-and-paste (or cut-and-paste) themselves into
- ▶ Make up a large fraction of human genome.
- ▶ Copy-and-paste elements are ideal for phylogenetic studies because
 1. Each element is unique.
 2. Once inserted, never lost (cleanly).
 3. Individuals who share an element must also share an ancestor.
- ▶ Whales are artiodactyls, related to hippos.
- ▶ Humans are apes.