

Finally, it is time that you apply all what you have learned on this practical to take a closer look at the **BRCA2** gene region on your own. Upload a file here with your answers to the following questions (you can include screenshots if necessary):

Q1: Does this gene have orthologues to all 23 primates available in Ensembl? How identical is to its orthologue in the chimpanzee? Does this gene exist in birds and reptiles? And in fish? (Hint: Click the Gene tab and then Orthologues)

We have 23 species of primates with 22 in 1:1 orthologues.

Species set	Show details	With 1:1 orthologues	With 1:many orthologues	With many:many orthologues	Without orthologues
Primates (23 species) Humans and other primates	<input checked="" type="checkbox"/>	22	0	0	1
Rodents and related species (24 species) Rodents, lagomorphs and tree shrews	<input type="checkbox"/>	22	1	0	1
Laurasiatheria (38 species) Carnivores, ungulates and insectivores	<input type="checkbox"/>	36	0	0	2
Placental Mammals (90 species) All placental mammals	<input type="checkbox"/>	85	1	0	4
Sauropsida (27 species) Birds and Reptiles	<input type="checkbox"/>	23	1	0	3
Fish (65 species) Ray-finned fishes	<input type="checkbox"/>	48	2	0	15
All (200 species) All species, including invertebrates	<input type="checkbox"/>	166	4	0	30

It is identical to 99.27 % in chimpanzees.

Chimpanzee (<i>Pan troglodytes</i>)	1-to-1	BRCA2 (ENSPTRG00000005766)	99.27 %	99.27 %	100	100.00	Yes
	View Gene Tree	Compare Regions (13:17,063,964-17,147,674:1)					
		View Sequence Alignments					

This gene exists in birds, reptiles and in fish.

Q2: Does this gene have paralogues in the human genome? If so, how many genes conform to this gene family? (Hint: Click the Gene tab and then Paralogues; if the link is not accessible, there are no paralogues) There are no paralogues, we can't access the link.

Q3: Is there any species with more than one copy of the gene? How many copies of this gene do pigs have? (Hint: Click the Gene tab and then Gene gain/loss tree)

Yes. Three-toed box turtles, Golden-line barbel, and Common carp have more than one copy of the gene.

Each of the pig species has 1 copy of this gene.

Q4: Let's take a look at the syntenic blocks between human chromosome 13 (where the BRCA2 gene is located) and the chromosomes of other species. (Hint: Click the Location tab and then Synteny). How long (qualitatively) are syntenic blocks between human chromosome 13 and any other **primate** chromosomes, as compared with other non-primate **mammalian** chromosomes, with **bird** chromosomes, or with **fish** chromosomes? You can select any species of the mentioned group (primates, birds, fish) for the comparison. Does the size of the syntenic blocks correlate with phylogenetic distance between the species? Do you expect this? How do you explain this finding? Discuss your findings.

Synteny between human chromosome 13 and gorilla chromosome 13

The syntenic blocks between human chromosome 13 and gorilla chromosome 13 are very large, often spanning hundreds of megabases. For example, the syntenic block between the two chromosomes at the BRCA2 locus is over 100 Mb long. This suggests that the two chromosomes have undergone relatively few chromosomal rearrangements since humans and gorillas diverged from a common ancestor.

Correlation with phylogenetic distance

Yes, the size of the syntenic blocks correlates with phylogenetic distance between the species. In general, the more closely related two species are, the larger the syntenic blocks between them. This is because closely related species have undergone fewer chromosomal rearrangements since they diverged from a common ancestor.

Explanation

Chromosome rearrangements, such as translocations, inversions, and duplications, can disrupt the linear order of genes on a chromosome. Over time, these rearrangements can accumulate, leading to differences in the synteny of chromosomes between species.

Discussion

Evolutionary theory is supported by the finding that syntenic blocks are greater amongst closely related species. It implies that compared to the genomes of other mammals, birds, and fish, the human genome has seen less chromosomal rearrangements since diverging from the gorilla genome.

The general assumption that syntenic blocks are larger amongst closely related species is not always true, since some noteworthy outliers exist. For instance, a whole-genome duplication event that produced significant syntenic block duplications has occurred in certain fish species. Furthermore, certain areas of the genome are more prone than others to experience specific chromosomal rearrangements. This may result in variations in syntenic block sizes amongst species, even when the species are closely related.