

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*)

This gene has orthologues to all 23 primates in Ensembl

Orthologues ?

[Download orthologues](#)

Summary of orthologues of this gene [Hide](#)

Click on 'Show details' to display the orthologues for one or more groups of species. Alternatively, click on 'Configure this page' to choose a custom list of species.

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 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

It is identical in a 99.27%

Chimpanzee (<i>Pan troglodytes</i>)	1-to-1 View Gene Tree	BRCA2 (ENSPTRG00000005766) Compare Regions (13:17,063,964-17,147,674:1) View Sequence Alignments	99.27 %	99.27 %
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This gene does exist in birds, reptiles and fish

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

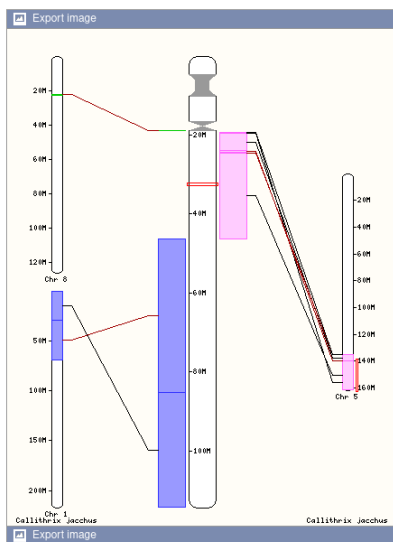
Q2: Does this gene have paralogues in the human genome? If so, how many genes conform this gene family? (*Hint: Click the Gene tab and then Paralogues; if the link is not accessible, there are no paralogues*)

It has no paralogues, as the paralogue link in the menu is grayed out.

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*)

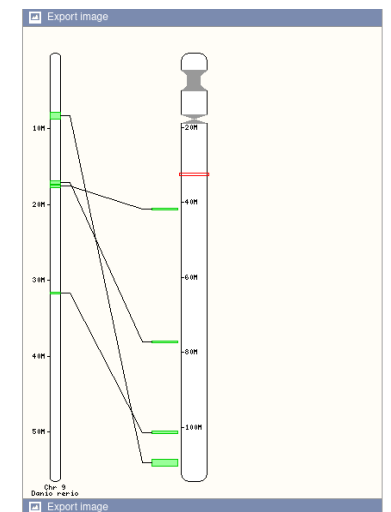
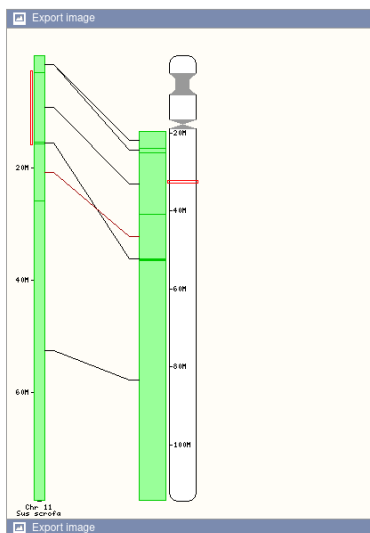
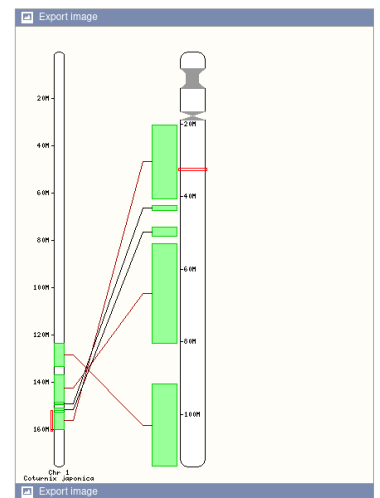
The Common carp, Golden-line barbel and Three-toed box turtle have more than one copy of the gene. Pigs have one copy of the gene

Q4: Let's take a look at the syntenic blocks between human chromosome 13 (where *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, fishes) 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.



The Top Left image corresponds to the synteny between human chr13 and White-tufted-ear marmoset (primate), the bottom left image corresponds to the synteny between human chr13 and pig (not primate mammal), the top right image corresponds to the synteny between human chr13 and the japanese quail (bird) and the bottom right image is the synteny between human chr13 and the zebrafish (fish).

The biggest syntenic block in comparison to human chr13 is from primates, where even though the blocks come from different chromosomes in primates, are the ones that have the most "coverage" of similarity with human chr13, then come the mammals, where we see that the pig's chr11 is almost identical to the human chr13, followed by the bird and then the fish. We did indeed expect these results because phylogenetically, this is the order in which this species are closer to the human, so it's normal that the mammals have the most similar chromosome and the fish the least.



The only finding that we find a bit confusing is that in primates, the different syntenic blocks come from different chromosomes while in non-primate mammals the whole coverage come from the same chromosome.

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