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Genetic approach to the sequence of the *prnp* gene in Portuguese cervids species

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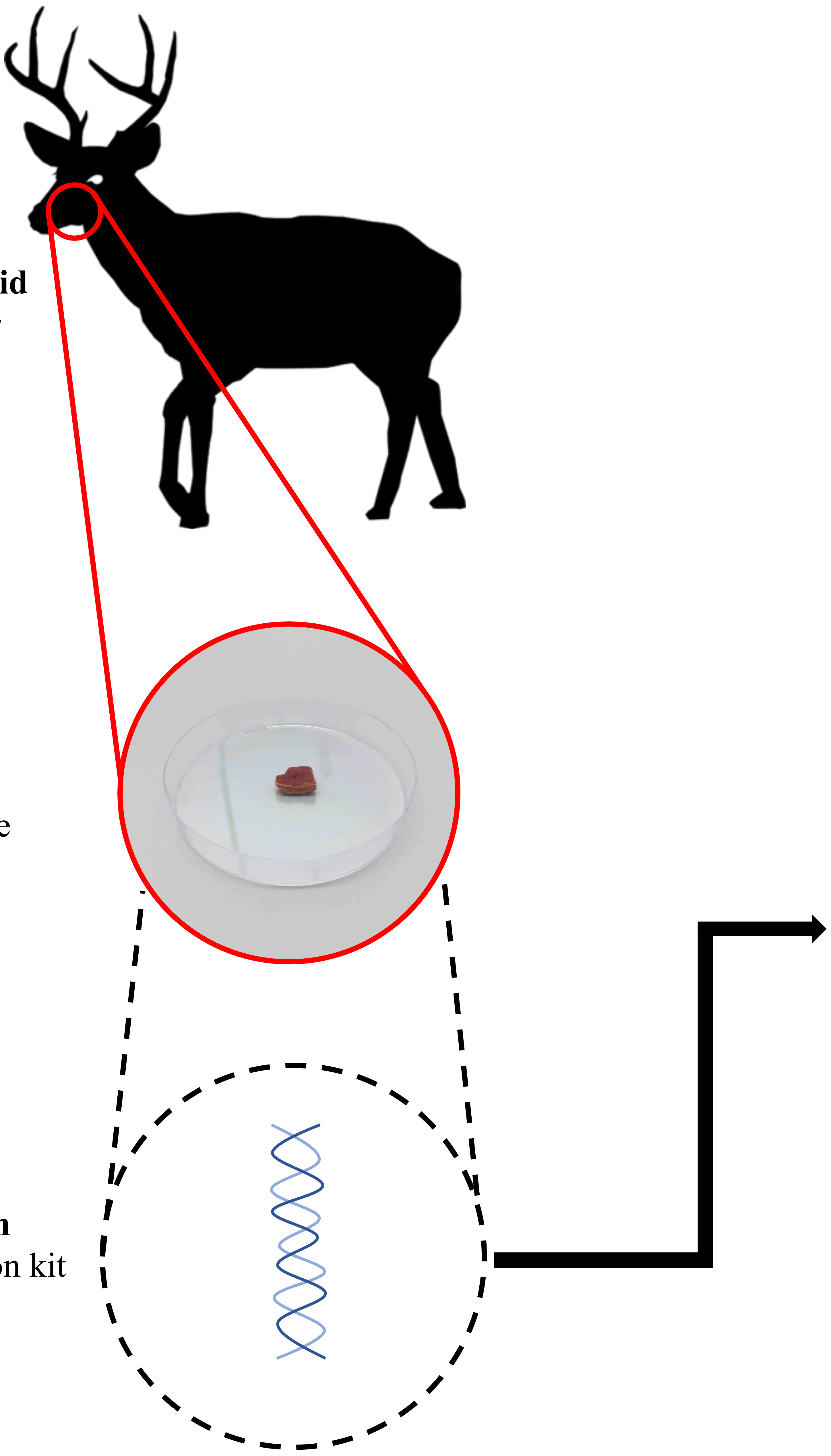
Mutations in **intronic regions** can also have an important effect, similar to the CDS region. In this context, it seems extremely important to genotype and characterize not only the CDS region but extend this characterization to the remain genetic regions, which also have an important value in the disease context. Our group proposes a **different approach**, outlined in this poster, which is overlooked in European and North American researches. Thus, in order to characterize these sequences, new putative primers were designed, based on an alignment between several cervids, to obtain a larger amplicon.

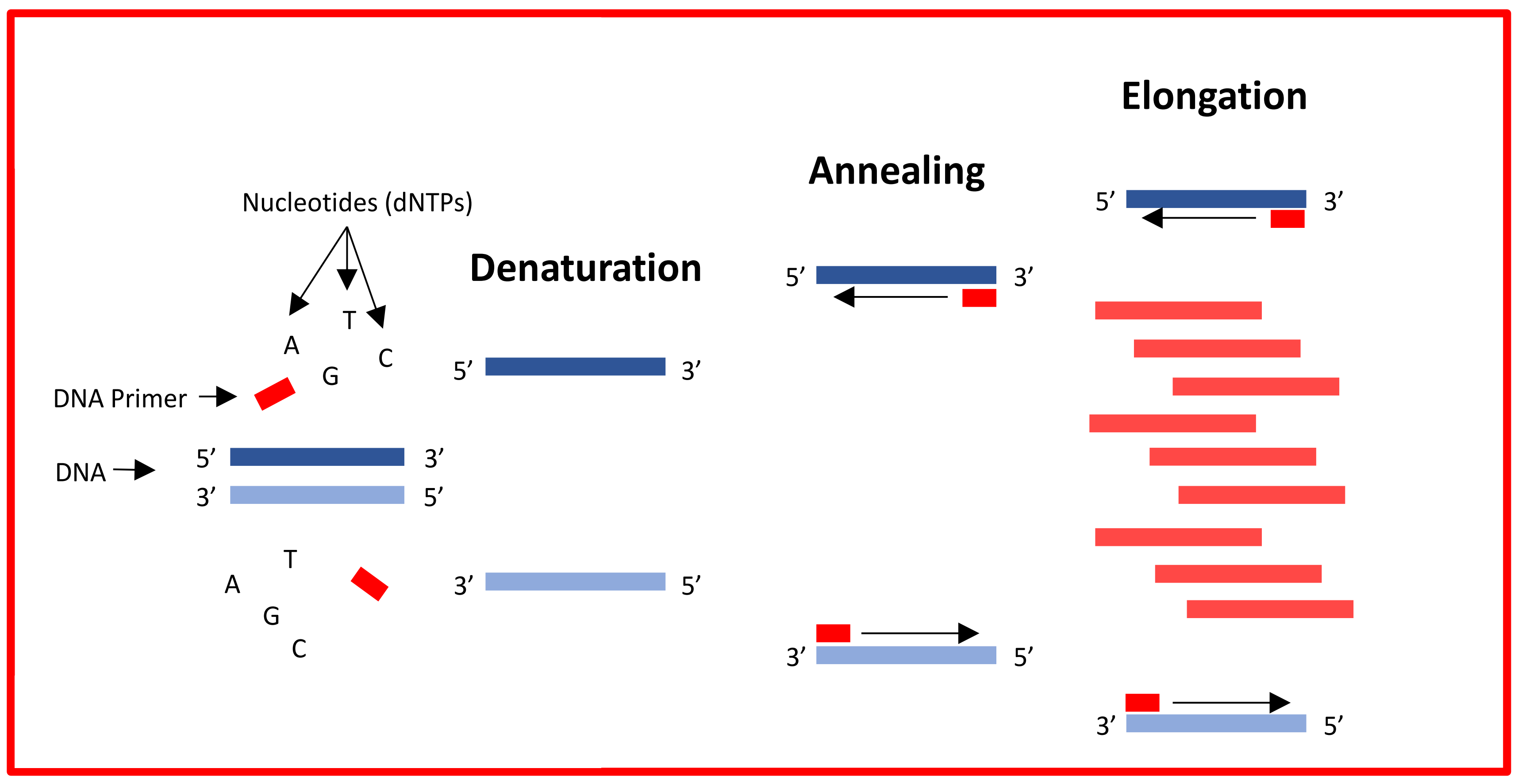


Portuguese cervid
Cervus elaphus

Tissue sample
Masseter muscle

DNA extraction
NZY gDNA isolation kit

PCR amplification



Sequencing

caaaagagac tcattccctg cttacattcc cgcatgagga aagaggatgt gagccaatat
tcagaaaatt atttaatgat ctcagcacct accttggggt ctacccaact ggacattaga
atcaattcca tagggtccat gccagggttc ggaaggttcc aggagctaat atcccttata
acaacccaat aggcagagct tccagtgtcc ccacaagaat aagtccagtt gcaagaatca
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ggaggagaaa gagttgtatt aacactttac ctacaattac tttcgtgaga tgtatggaat
gtgaagaaca tttatgacct agaatattta tagctgatgc cactgctatg cagtcatgca
ttatgctgca gactttaagt gatttttaca tgggcatatg atgctgacac cctctttatt
ttgcagATAA GTCATC**ATGG TGAAAAGCCA CATAGGCAGC TGGATCCTAG TTCTCTTTGT**
GGCCATGTGG AGTGACGTCG GCCTCTGCAA GAAGCGACCA AAACCTGGAG GAGGATGGAA
CACTGGGGGG AGCCGATACC CGGGACAGGG AAGTCCTGGA GGCAACCGCT ATCCACCTCA
GGGAGGGGGT GGCTGGGGTC AGCCCCATGG AGGTGGCTGG GGCCAACCTC ATGGAGGTGG
CTGGGGTCAG CCCCATGGTG GTGGCTGGGG ACAGCCACAT GGTGGTGGAG GCTGGGGTCA
AGGTGGTACC CACAGTCAGT GGAACAAGCC CAGTAAACCA AAAACCAACA TGAAGCATGT
GGCAGGAGCT GCTGCAGCTG GAGCAGTGGT AGGGGGCCTC GGTGGCTACA TGCTGGGAAG
TGCCATGAGC AGGCCTCTTA TACATTTTGG CAATGACTAT GAGGACCGTT ACTATCGTGA
AAACATGTAC CGTTACCCCA ACCAAGTGTA CTACAGGCCA GTGGATCAGT ATAATAACCA
GAACACCTTT GTGCATGACT GTGTCAACAT CACAGTCAAG CAACACACAG TCACCACCAC
CACCAAGGGG GAGAACTTCA CCGAAACTGA CATCAAGATG ATGGAGCGAG TTGTGGAGCA
AATGTGCATC ACCCAGTACC AGAGAGAATC CGAGGCTTAT TACCAAAGAG GGCAAGTGT
GATCCTCTTC TCCTCCCCTC CTGTGATCCT CCTCATCTCT TTCCTCATTT TTCTCATAGT
AGGATAGGGG CAACCTTCCT GTTTTCATTA TCTTCT

Partial sequence of the *Cervus elaphus prnp* gene (GenBank accession: FJ590751.1), including part of the intron 2 in lowercase, the exon 3 in uppercase and the coding sequence (CDS), bolded, within the exon 3. For the amplification of this region, the primers forward (royal blue) and reverse (red) were used. These primers were designed by our group to amplify only the CDS region of the *prnp* functional gene, avoiding the amplification of the pseudogene, and to increase the genetic information of the intronic region. The variations found in Portuguese population of *Cervus elaphus* are marked in red.

Conclusions:

- ❖ Mutations in **CDS region** can have a direct effect in the protein and consequently predispose the individuals to disease. The *prnp* CDS region is highly conserved among several species, and some variations reported in this region can influence the susceptibility to prion infection;
- ❖ **Untranslated sequences** play an important role in gene regulation. Mutations in intronic regions are related to some diseases in several species and the *prnp* gene should not be an exception.

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