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Chronic wasting disease risk assessment in Portugal: analysis of variability and genetic structure of the Portuguese roe deer population

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Aims: Among the Transmissible Spongiform Encephalopathies, Chronic Wasting Disease (CWD) in cervids is now the rising concern in wildlife within Europe after the first case detected in Norway in 2016. CWD shows a notable horizontal transmission, affecting both free-ranging and captive cervids. Furthermore, several genetic variants in the Prion Protein (*PRNP*) gene coding sequence of the cervid were identified, which increase the susceptibility to the disease.

Roe deer has a widespread geographical distribution which extends from the Iberian Peninsula to the north of Scandinavia, being found as well in Turkey, Israel, and Jordan. In recent centuries, a significant number of fluctuations have occurred in these deer populations, leading to a decline in deer population and distribution. These alterations combined with translocations of animals of the same species, can drive to meaningful consequences on the genetic structure, diversity, and fitness of populations.

Material and Methods: In this study we aimed to outline the genetic diversity and structure of Western Iberian roe deer populations using mitochondrial DNA (mtDNA) molecular analysis and link to sequence diversity of *PRNP* gene performed to evaluate the susceptibility to CWD of this population.

Results: The preliminary results from the analysis of 60 roe deer samples from different Western Iberian areas (wild

and fenced populations) allowed the confirmation of the *consensus PRNP* gene CDS sequence and revealed the presence of 13 haplotypes by mtDNA molecular analysis.

Conclusions: These studies are of great importance to obtain information about the phylogeography, which can be used to design appropriate strategies for the conservation and management of populations, but as well to maintain the genetic heritage of roe deer in Europe. Moreover, the establishment of risk assessment projects, even in countries with no cases of CWD is very important to predict the effects of possible contamination in the future.

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DTI Abnormalities in Healthy E200K Carriers May Serve as an Early Biomarker for Genetic Creutzfeldt-Jakob Disease (gCJD)

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Aims: To investigate microstructural changes in healthy E200K carriers using diffusion tensor imaging (DTI).

Material and Methods: Seven symptomatic gCJD patients and N = 60 healthy relatives of gCJD patients were included. Participants underwent genetic testing for the E200K mutation, MRI scans at 3T, and a lumbar puncture (LP) for total Tau protein levels (t-Tau). Diffusion tensor imaging (DTI) metrics including; fractional anisotropy (FA), mean diffusivity (MD), radial diffusivity (RD) and axonal diffusivity (AD) were calculated along 45 WM tracts.