Roe deer on ice: selection despite limited effective population size through the Pleistocene

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Abstract

Roe deer (Capreolus spp.) are a little odd. They are one of only a few placental mammals — and the only genus among even-toed ungulates — capable of putting embryonic development "on ice", also known as embryonic diapause (Fig. 1). It would seem such an unusual trait is likely the product of natural selection, but a big question is, how does selection for important traits, such as diapause, interact with the historical demography of a species? In a 'From the Cover' article in this issue of Molecular Ecology, de Jong et al. (2020) demonstrate that selection is acting on genes associated with reproductive biology in roe deer, despite heightened genetic drift due to reduced effective population size through the Pleistocene.

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Roe deer on ice: selection despite limited effective population size through the Pleistocene

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Roe deer (*Capreolus* spp.) are a little odd. They are one of only a few placental mammals — and the only genus among even-toed ungulates — capable of putting embryonic development "on ice", also known as embryonic diapause (Fig. 1). It would seem such an unusual trait is likely the product of natural selection, but a big question is, how does selection for important traits, such as diapause, interact with the historical demography of a species? In a 'From the Cover' article in this issue of *Molecular Ecology*, de Jong et al. (2020) demonstrate that selection is acting on genes associated with reproductive biology in roe deer, despite heightened genetic drift due to reduced effective population size through the Pleistocene.

[Emily Warschefsky to provide photo of roe deer from authors]

Figure 1: A European roe deer (**Capreolus capreolus**). *Along with the Siberian roe deer* (C. py-gargus)*, these species are the only two artiodactyls capable of obligate diapause, which allows the timing of the birth of offspring to coincide with favourable environmental conditions.*

The intensive climate fluctuations of the Pleistocene have had an outsized influence on genetic diversity and geographic structure of animal and plant populations (Hofreiter & Stewart, 2009). However, distinguishing whether patterns of contemporary genetic diversity in a population are consistent with the impacts of past/ongoing selection, historical demography, or both, can be a challenging endeavour (**Fig. 2**). The

historical demography of a population also directly influences natural selection: the relative importance of stochastic processes increases with reduced effective population size (N_e), limiting the efficacy of selection (Kimura, Maruyama, & Crow, 1963). Therefore, considering both natural selection and demography together can greatly add to our understanding of evolutionary dynamics in general, and of focal species in particular. In this study, de Jong et al. (2020) not only consider the impact of demography and selection, but how these forces acted at different time scales during the evolution of roe deer (*Capreolus* spp.), through genomic analyses at multiple taxonomic levels (**Fig. 3**).

[Fig. 2 uploaded separately as pdf]

*Figure 2: Some of the forces contributing to patterns of contemporary genetic diversity within populations. A changing environment (e.g. glacial-interglacial periods through the Pleistocene) impacts population size and the relative strength of genetic drift. However, the population can become adapted to new environmental conditions through natural selection, potentially also impacting on population size. Finally, genetic drift will impact the efficacy of natural selection in the population. de Jong et al. (2020) examine the interaction of these forces in roe deer (*Capreolus spp.), focussing particularly on European roe deer (C. capreolus).*

[Fig. 3 uploaded separately as pdf]

Figure 3: de Jong et al. (2020)'s analyses span deep time (against other deer species, right), the divergence of the roe deer genus, Capreolus *(middle right), the divergence of the European and Siberian roe deer (middle left), to patterns found within specific populations of European roe deer (far left). ROH = Runs of homozygosity. LD = Linkage Disequilibrium. MAF = Minor allele frequency. Not shown are analyses finding marked population structure among European roe deer populations, likely also influenced by Pleistocene dynamics.*

After generating a high-quality annotated genome for Siberian roe deer (*Capreolus pygargus*), de Jong et al. (2020) utilise whole exomes across Cervidae to investigate selection within the roe deer genus (*Capreolus*). They further use genomes from both roe deer species to investigate long-term demographic patterns. Finally, they generate Restriction site Associated DNA sequencing (RADseq) data for European roe deer (*C. capreolus*) populations to gain further insights into recent selection and demographic history, including contemporary population structure. By using the roe deer genomes, including the newly generated Siberian roe deer genome, in combination with exome data from other ungulates, de Jong et al. (2020) utilise a phylogenetic framework to demonstrate ongoing positive selection in roe deer on genes linked to reproductive biology. They performed a similar analysis using candidate genes, revealing that *Pard3* , a gene potentially associated with embryonic diapause, showed a particularly high rate of non-synonymous change within the roe deer lineage (**Fig 3**). Interestingly,*Pard3* also interacts with several genes de Jong et al. (2020) found to be under selection in their whole exome analyses. In addition,*g09868* , a gene found to be under positive selection, showed evidence of divergent paralogs (more similar to those found in Cervinae than to other Capreolinae species; **Fig. 3**) retained only in European roe deer, to the exclusion even of Siberian roe deer. These paralogs were retained in European roe deer despite a markedly lowerN_e for the last half-million years in comparison with the Siberian roe deer.

The authors also introduce an elegant new method for estimating divergence times based on genomic data. They constructed a Markovian model relying on an initial value of similarity between two sister species (the theta of the ancestral population), a given mutation rate, and transition probabilities for moving between "same" and "different" nucleotide states between the two species through time. They use this model to solve for the number of generations it would take to achieve the levels of similarity seen between the two species currently. After validating this method with well-characterised divergences within Hominidae, they estimated the similarity between the European and Siberian roe deer by mapping raw reads from one species to another, and used this method to estimate the divergence time of European and Siberian roe deer at 0.9-1.35 million years ago, consistent with the Mid-Pleistocene Transition.

To investigate more recent demographic history of European roe deer, the authors included multiple popula-

tions sampled using RADseq data (**Fig. 3**). In doing so, they included a particularly interesting population: East Anglia. European roe deer were extirpated from throughout much of England by the late 18th century (Chapman, 1993). Around 1884, the East Anglia population was anthropogenically founded. Given the known source (Germany), timing, and size of the founding group (three to six pairs of deer; Chapman 1993), this relatively isolated population was a particularly powerful data point for investigating demography and whether selection could overcome even a pronounced bottleneck associated with an anthropogenic founding event. de Jong et al. (2020) uncover consistent patterns of putative selection in this population and others included in this study (Scotland, France, Germany). However, it is important to note that these consistent signatures of apparent selection could also be due to selection in the shared ancestral population or associated with patterns of diversity linked to shared genomic features (Burri et al., 2015; Cruickshank & Hahn, 2014). While the evidence for selection in this case might be somewhat equivocal, anthropogenic founding events such as the reintroduction of European roe deer to East Anglia could present a powerful opportunity for calibrating mutation rates for both methods (e.g. site frequency spectra analyses) and datasets (e.g. the mutational spectra of RADseq loci versus whole genome sequencing datasets), allowing demographic events inferred from genomic data to be more robustly anchored to realistic timeframes.

In summary, de Jong et al. (2020) use genomic data from different taxonomic datasets to provide a window into the interaction of selection and demography at different time scales. In doing so, they demonstrate that selection is a consistent feature influencing the genomes of roe deer from past to present, even in the face of pronounced genetic drift. More broadly, they provide a comprehensive overview of how considering demographic history and natural selection at multiple transition points can provide richer information about the main driving forces shaping genetic diversity and differentiation within a species. As genomic data become more readily available for non-model species, this study elegantly demonstrates how combining comparative genomics analyses with reduced representation sequencing at a population scale can lead to insights at different time points of the evolutionary process.

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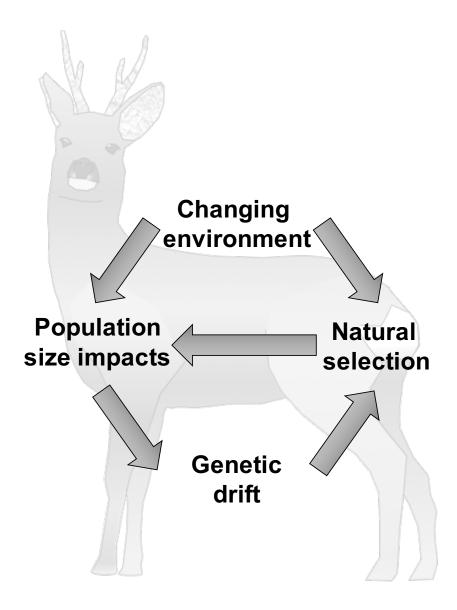
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Data	Restriction site associated DNA sequencing (RAD-Seq)	Whole genome sequence		
Analyses of historical demography	Site-frequency spectra LD MAF	PSMC (N_e) ROH Genome-wide diversity/ROH		
Analyses of selection	Tajima's D/ ROH/ Watterson's theta	dN/dS (CodeML)/TreeSAAP Paralogs		
Evolutionary transitions	European roe deer populations	Population differentiation	European roe deer vs. Siberian roe deer	Diverging of roe deer species	Genus *Capreolus* (roe deer)	Diverging of *Capreolus*	Family Cervidae (deer) Capreolinae ("New World") Reindeer Moose Mule deer *Roe deer* Cervinae ("Old World") Munjac Elk/Wapiti Chital Fallow deer *Outgroup* *Cow*
Time	Present		Past	