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Genomic analysis of conservation status, population structure, and admixture in local Czech and Slovak dairy goat breeds

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ABSTRACT

Although dairy goat production, characterized by traditional production on small farms, is an important source of income in the Czech Republic and Slovakia, locally adapted breeds have not been fully consolidated over the last 100 yr due to large fluctuations in population size and inconsistent breeding programs that allowed for different crossbreeding strategies. Our main objective in this study was therefore to assess the conservation status of 4 Czech (Alpine Goat, White Shorthair, Brown Shorthair, and Czech Landrace) and 1 Slovak (Slovak White Shorthair) local goat breeds, to analyze their population structure and admixture, and to estimate their relatedness to several neighboring breeds. Our analyses included 142 goats belonging to 5 local breeds genotyped with the Illumina 50K BeadChip, and 618 previously genotyped animals representing 15 goat breeds from Austria and Switzerland (all analyses based on 46,862 autosomal SNPs and 760 animals). In general, the conservation status of the Czech and Slovak local goat breeds was satisfactory, with the exception of the Brown Shorthair goat, as the analyzed parameters (heterozygosity, haplotype richness, runs of homozygosity–based inbreeding, and effective population size) were mostly above the median of 20 breeds. However, for all 5 Czech and Slovakian breeds, an examination of historical effective population size indicated a substantial decline about 8 to 22 generations ago. In addition, our study revealed that the Czech and Slovakian breeds are not fully consolidated; for instance, White Shorthair and Brown Shorthair were not clearly distinguishable. Considerable admixture, especially in Czech Landrace (effective number of parental clusters = 4.2), and low but numerous migration rates from other Austrian and Swiss breeds were found. These results provide valuable insights for future breeding programs and genetic diversity management of local Czech and Slovak goat breeds.

Key words: conservation status, diversity, genomics, goats, population structure

INTRODUCTION

The production of goat milk, mainly processed into cheese and other products, is an important sector of extensive livestock production that can contribute an important source of income on small, nonspecialized farms. The White Shorthair goat (**WSH**) and the Brown Shorthair goat (**BSH**) are local breeds of dairy goat from Czechoslovakia. The WSH is the result of selective breeding of indigenous Czechoslovakian Landraces crossed with Saanen goats from Switzerland and Germany. The WSH bred on Czech or Slovak territory have a different genetic background, as they are descended from various local Landraces. The BSH is the result of selective breeding of a native breed of nonuniform color, mainly light brown to white, which was improved between 1900 and 1930 mainly in the Czech-German border area by crossing with German brown bucks. The 2 goat breeds were recognized as independent breeds in 1954 to 1955, when the WSH population was around 1.5 million animals (Sztankoova and Rychtarova, 2017). Between 1950 and 1990, both breeds were improved by repeated imports of semen for insemination of white German goats for the WSH and brown German goats for the BSH. By 1990, other dairy goat breeds were also included in the improvement process. Due to the very limited artificial insemination and transfer of breeding animals between the Czech and Slovak territories, there were always 2 genetically separate subpopulations of WSH. With the general decline of the

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goat population in the Czech Republic, the number of purebred animals gradually decreased between 1990 and 1996, to about 1,800 animals in the WSH and to a critical level of 180 animals in the BSH. To increase milk yield, crossbreeding between WSH and BSH breeds was carried out between 1990 and 1996, and the hybrids were kept as purebred BSH animals. Since 1994, the breeds have been revitalized via inclusion in genetic resource collections so that they can be used for pure breeding without the influx of genes from other breeds. In the course of the dissolution of Czechoslovakia in 1993, the WSH breed was split into 2 separate breeds, the Czech and Slovak WSH. The Czech Landrace, a local goat breed, is the result of crossing old German Landraces with old Czech Landraces around 1900. Between 1950 and 1980, this breed was systematically crossed with WSH or BSH. The last 10 animals were used to create a new breed in the Czech Republic in the 1990s (Sambraus 2006). The Alpine goat is a French medium-sized breed that was introduced to the Czechia in the 1990s to increase milk production and improve the domestic dairy breeds. The Alpine goat is kept in the Czechia as an independent pure breed with low gene flow from other Alpine goat populations (France, Switzerland, etc.).

Genetic diversity is an important component of any successful breeding program, as the breeder's equation shows (Mueller and Van Eenennaam, 2022). At the same time, even large commercial livestock populations can be genetically small (Cortellari et al., 2022) due to strong selection, high genetic drift (mainly due to unequal utilization of male and female animals), mating of related individuals (increase in inbreeding), a sudden population decline, or a combination of these factors, as well as many others. For example, an increase in inbreeding can lead to inbreeding depression (Ferenčaković et al., 2017; Howard et al., 2017; Doekes et al., 2018) or an increased prevalence of genetic defects, or both, due to the accumulation of deleterious mutations (VanRaden et al., 2011; Cole, 2015). Therefore, as with other livestock species, managing the conservation status of a local goat breed (genetic diversity, inbreeding, effective population size) is an essential component of genetic improvement and adaptation to the local environment. Thus, for example, genes associated with adaptations have already been identified in some goat breeds (Alberto et al., 2018; Manunza et al., 2023a,b). Moreover, our pedigree analyses have already indicated a decline in the genetic diversity of Czech goat breeds, as documented in Vostra-Vydrova et al. (2020), which prompted us to perform additional analyses using genomic information.

Admixing different breeds (populations) can have both positive and negative effects on the genetic improvement of local populations, whereas the negative consequences of reduced genetic diversity are well known. Thus, crossbreeding can be beneficial if done carefully to introduce desirable traits, increase genetic diversity, or improve overall fitness through the phenomenon known as heterosis (Jaafar et al., 2022). In contrast, the introduction of foreign "blood" can lead to the disintegration of the unique and often advantageous gene combinations that may be important for adaptation (e.g., survival, productivity) in a given environment (Frankham et al., 2011; Jaafar et al., 2022). Uncontrolled gene flow can also lead to a loss of genetic purity, which can be a problem if the purity of a particular breed or wild population reduces its homogeneity (i.e., the ability to maintain uniform inheritance or transmission of desirable traits and characteristics within a selected or well-adapted population; Cardoso et al., 2021). Finally, admixture can lead to mitonuclear incompatibility, resulting in conflicts between genes in the nuclear genome and those in the mitochondrial genome (Kwon et al., 2022; Ward et al., 2022). The presence of uncontrolled admixture should therefore be interpreted with caution, as negative consequences for the breeding population are also possible.

Considering the possibility of obtaining high-throughput genotype information for Czech and Slovak goat populations and observed population decline in the pedigree analysis (Vostra-Vydrova et al., 2020), estimation of genetic diversity, effective population size, and inbreeding, together with assessment of the degree of admixture, was an important main goal of this study for further improvement of goat breeding programs in the Czechia and Slovakia. In addition, we analyzed the genetic relatedness of Czech and Slovak goat breeds with neighboring breeds from Switzerland and Austria. Our study has a small methodological component, as we used several recently developed approaches that have been shown to be effective in humans but have not yet been applied in livestock populations.

MATERIALS AND METHODS

Sampling, Genotyping, Analyzed Data, and Quality Control

The analyzed data set contained genome-wide SNP data for 4 indigenous Czech and Slovak dairy goats: Czech White Shorthair (**CZWSH**), Slovak White Shorthair (**SKWSH**), Czech Brown Shorthair (**CZBSH**), and Czech Landrace (**CZLAN**), as well as an international Alpine goat breed (**CZALG**) with 140 individuals kept in the Czechia. The genotyped individuals of the CZWSH, CZBSH, and CZALG breeds originate from farms in the central and western Czech Republic, and the individuals of the CZLAN breed originate from farms in the southwest of the Czech Republic. Individuals of the SKWSH breed were sampled in central Slovakia. These locations are the most important breeding areas of the analyzed breeds. The selection of genotyped individuals was randomized based on the FAO practical guide (Ajmone-Marsan et al., 2023), with only 1 individual selected from each breeding unit (farm), to avoid sampling related individuals. The minimum number of genotyped animals, especially for small breeds such as CZLAN (20 individuals), fulfilled the requirement for a reliable estimation of allele frequencies of biallelic markers such as SNP (Hein et al., 2005). To compare the genetic relatedness between Czech and Slovak breeds and to analyze the population structure and admixture patterns, we included an additional 620 publicly available genotypes. Specifically, genotypes of 5 breeds from Austria were included in our analyses: Blobe (**ATBLB**), Chamois Colored (**ATCHA**), Pinzgau (**ATPNZ**), Styrian Pied (**ATSTP**), and Tauern Pied (**ATTAP**); as well as the following 10 breeds from Switzerland: Appenzell (**CHAPP**), Grisons Striped (**CHGST**), Chamois Colored (**CHCHA**), Nera Verzasca (**CHNVR**), Peacock (**CHPEA**), Saanen (**CHSAA**), Booted (**CHBOT**), Tessin Gray (**CHTSG**), Toggenburg (**CHTGB**), and Valais (**CHVAL**). Further details on the Austrian and Swiss goat genotypes can be found in Burren et al. (2016) and Pogorevc et al. (2021). The number of genotyped animals for each breed is listed in Table 1, and the summary statistics of the relatedness structure, calculated from the off-diagonal elements of the relationship matrix estimated using the genome function PLINK v 1.9 (Chang et al., 2015), are presented in Supplemental Table S1 (see Notes). Due to the changes of political borders and the known breed descriptions, we expected some historical gene flow between the indigenous breeds of the Czechia and Slovakia. For all Czech and Slovakian goat samples, DNA extraction from buccal swabs was performed at Neogene Genomics (Ayr, Scotland, UK), together with genotyping using the GoatSNP50 Illumina Beadchip (Illumina Inc., San Diego, CA). Quality control of the genotyping data was performed with PLINK v 1.9 (https://zzz.bwh.harvard.edu/plink/), using the following quality control parameters: only autosomal SNPs with known chromosomal positions were taken, to exclude the bias between male and female individuals, and all individuals and SNPs with call rate below 0.9 were excluded. After quality control, the final database comprised 46,862 SNP markers and 760 animals.

Conservation Status

Genetic Diversity and Haplotype Richness. To reduce the ascertainment bias of the Illumina 50 K BeadChip, short haplotypes were used instead of single SNPs, as recommended by Simčič et al. (2015). Therefore, the SNP data were divided into nonoverlapping blocks of 4 SNP genotypes (block) with an intermarker distance of

Table 1. Genomic observed (H_O) and expected (H_E) heterozygosity, Wright's genomic inbreeding coefficient (F_{IS}), haplotype richness (HR), rarefacted ($2n = 40$)[1] haplotype richness (HRR), and ROH genomic inbreeding ($F_{ROH\ >2Mb}$) estimates (± SE) for 20 Central European goat breeds

Breed, by country of origin	N	H_O	H_E	HR	HRR	F_{IS}	$F_{ROH\ >2Mb}$
Austria (AT)							
Blobe (ATBLB)	34	0.817 ± 0.001	0.819 ± 0.001	9.67 ± 0.03	9.00 ± 0.03	0.003 ± 0.001	0.060 ± 0.014
Chamois Colored (ATCHA)	22	0.807 ± 0.002	0.809 ± 0.001	8.28 ± 0.03	8.59 ± 0.03	0.003 ± 0.001	0.055 ± 0.006
Pinzgau (ATPNZ)	28	0.791 ± 0.001	0.803 ± 0.001	8.81 ± 0.03	8.58 ± 0.03	0.014 ± 0.001*	0.082 ± 0.013
Styrian Pied (ATSTP)	33	0.847 ± 0.001*	0.852 ± 0.001*	10.82 ± 0.03*	10.15 ± 0.03*	0.007 ± 0.001	0.030 ± 0.012
Tauern Pied (ATTAP)	28	0.762 ± 0.002	0.746 ± 0.001	6.84 ± 0.02	6.70 ± 0.02	−0.022 ± 0.001	0.115 ± 0.007
Czech Republic (CZ)							
Alpine (CZALG)	28	0.812 ± 0.002	0.803 ± 0.001	8.28 ± 0.02	8.11 ± 0.02	−0.011 ± 0.001	0.062 ± 0.005
Brown Shorthair (CZBSH)	34	0.783 ± 0.002	0.782 ± 0.001	8.28 ± 0.03	7.75 ± 0.02	−0.001 ± 0.001	0.094 ± 0.010
Czech Landrace (CZLAN)	20	0.833 ± 0.002	0.820 ± 0.001	8.14 ± 0.02	8.79 ± 0.03	−0.017 ± 0.001	0.034 ± 0.010
White Shorthair (CZWSH)	36	0.843 ± 0.001	0.846 ± 0.001	10.65 ± 0.03	9.83 ± 0.03	0.003 ± 0.001	0.025 ± 0.006*
Slovakia (SK)							
White Shorthair (SKWSH)	24	0.845 ± 0.001	0.842 ± 0.001	9.43 ± 0.03	9.56 ± 0.03	−0.004 ± 0.001	0.027 ± 0.010
Switzerland (CH)							
Appenzell (CHAPP)	29	0.722 ± 0.002*	0.713 ± 0.002*	6.33 ± 0.02*	6.16 ± 0.02	−0.014 ± 0.001	0.141 ± 0.006*
Booted (CHBOT)	23	0.771 ± 0.002	0.758 ± 0.001	6.85 ± 0.02	6.97 ± 0.02	−0.018 ± 0.001	0.096 ± 0.010
Grisons Striped (CHGST)	49	0.795 ± 0.001	0.783 ± 0.001	8.66 ± 0.03	7.67 ± 0.02	−0.014 ± 0.001	0.078 ± 0.008
Chamois Colored (CHCHA)	124	0.803 ± 0.001	0.799 ± 0.001	10.43 ± 0.03	8.18 ± 0.02	−0.005 ± 0.001	0.059 ± 0.003
Nera Verzasca (CHNVR)	42	0.787 ± 0.002	0.792 ± 0.001	8.92 ± 0.03	8.01 ± 0.03	0.006 ± 0.001	0.059 ± 0.007
Peacock (CHPEA)	31	0.809 ± 0.001	0.791 ± 0.001	8.04 ± 0.03	7.73 ± 0.03	−0.023 ± 0.001*	0.046 ± 0.005
Saanen (CHSAA)	64	0.764 ± 0.001	0.759 ± 0.001	8.21 ± 0.03	7.02 ± 0.02	−0.008 ± 0.001	0.093 ± 0.007
Toggenburg (CHTGB)	31	0.724 ± 0.002	0.717 ± 0.002	6.36 ± 0.02	6.13 ± 0.02*	−0.011 ± 0.001	0.124 ± 0.006
Tessin Gray (CHTSG)	37	0.809 ± 0.001	0.809 ± 0.001	9.49 ± 0.03	8.68 ± 0.03	−0.001 ± 0.001	0.042 ± 0.008
Valais (CHVAL)	43	0.728 ± 0.002	0.728 ± 0.002	7.11 ± 0.03	6.45 ± 0.02	0.000 ± 0.001	0.128 ± 0.009
Median		0.799	0.796	8.28	8.06	−0.005	0.061

[1] For 2n = 40, n is the number of individuals in the smallest sample with 20 individuals.

*The most extreme values (minimum and maximum) are marked with asterisks.

less than 50 kb for neighboring SNPs, with the maximum length of each block being less than 150 kb. A total of 5,136 blocks were used for further analysis. These blocks as multiallelic markers allow a better analysis of genetic diversity than biallelic SNPs. The haplotype blocks and SNPs were used to assess genetic diversity based on a comparison of observed (**H_O**) and expected heterozygosity (**H_E**) and individual inbreeding coefficients (**F_{IS}**). The F_{IS} indicate the deviation from Hardy-Weinberg equilibrium. Negative values are usually the result of outcrossing or avoidance of inbreeding, whereas positive values indicate inbreeding (Wright, 1965). The estimated haplotype richness (**HR**), which here replaces the allele richness used for loci with multiple alleles, was also calculated as rarefacted haplotype richness (**HRR**) to account for differences in sample size when comparing different breeds. All these parameters were calculated using the R package PopGenReport (Adamack and Gruber, 2014), with HRR calculated using ADZE software (Szpiech et al., 2008).

Runs of Homozygosity Genomic Inbreeding. The proportion of the genome in autozygosity was estimated by identifying runs of homozygosity (**ROH**) segments using detectRUNS and the consecutive runs method (Biscarini et al., 2018). Runs of homozygosity were detected separately for each individual with the following criteria: the minimum number of SNPs in ROH was set to 15; the maximum gap between adjacent SNPs was set to 1 Mb; the minimum SNP density per ROH was set to 1 SNP per 1 Mb. The minimum length of the ROH was set to 2 Mb. To account for genotyping errors, ROH were calculated separately for each of the 5 categories defined by ROH length (>2 Mb, 2–4 Mb, 4–8 Mb, and >8 Mb). Based on the ROH length, the numbers of allowed heterozygotes and missing genotypes were adjusted according to Ferenčaković et al. (2013). Our value of 15 for the minimum number of SNPs was calculated according to (Lencz et al., 2007) to minimize false-positive ROH:

$$l = \ln[\alpha/(n_s \times n_i)]/\ln(1 - het),$$

where n_s is the number of genotyped SNPs per individual; n_i is the number of genotyped individuals of the breed; α is the percentage of false-positive ROH (set to 0.05 in this study); and het is the average SNP heterozygosity. The F_{ROH} (runs of homozygosity genomic inbreeding coefficient) was estimated as the fraction of the autosome in ROH covering 29 chromosome pairs ($F_{ROH} = L_{ROH}/L_{AUTOSOME}$, where L_{ROH} is the length of all ROH in the genome of the individual and $L_{AUTOSOME}$ is the specific length of the autosomal genome).

Current and Historical Effective Population Sizes. Two approaches were used to determine the contemporary effective population size (**Ne_{LD}**) of the populations analyzed. In the first approach, the Ne_{LD} was based on a method described by Waples and Do (2008), which was implemented in NeEstimator v.2 (Do et al., 2014). This approach is based on gamete disequilibrium and estimates a 95% confidence interval in the jackknife method, removing SNPs with a frequency of less than 5%. In the second approach, Ne_{LD} was estimated by the approach implemented in the GONE software (Santiago et al., 2020). The estimation approach developed in GONE is based on the functional relationship between gamete or linkage disequilibrium patterns and effective population size (Hill, 1981), but is calculated using a complex modeling approach. A genetic algorithm implemented in the GONE software (Mitchell, 1998) derives the historical set of effective population size series that best minimizes the sum of squared differences between the observed D^2 values (averaged squared correlations between 2 loci allele frequencies, weighted by their variance) of the bins and those predicted at corresponding different demographic trajectories (Saura et al., 2021). In this study, we have labeled all our estimates as Ne_{LD} to indicate that the estimates are based on gamete/linkage disequilibrium. The Ne_{LD} is the size of an idealized population, often considered a Wright-Fisher population, that exhibits the same degree of genetic drift and change in gamete/linkage disequilibrium as the population under consideration. Further information on Ne_{LD} and potential biases can be found in Waples et al. (2014), Ryman et al. (2019), and Waples (2021). In addition, GONE software was used to estimate contemporary and historical Ne_{LD} using phased and unphased genotypic information. Phasing was performed using the genetic model of coalescence with recombination and with the default options implemented in the SHAPEIT2 software (Delaneau et al., 2013), as these provided the best fit. Our historical estimates extended back 40 generations, which corresponds to up to 160 yr, considering that the estimated generation interval of our goats is about 4 yr (Vostra-Vydrova et al., 2020).

Genomic Relatedness, Population Structure, and Admixture

Genomic Relatedness. The phylogenetic relatedness between analyzed goat breeds (populations) was represented by the Neighbor-Net network, inferred from Reynolds's pairwise genetic distances (Reynolds et al., 1983). Reynolds's pairwise genetic distances were calculated using the StAMPP package (Pembleton et al., 2013). The phylogenetic network (Neighbor-Net) was created and drawn using the software SplitsTree5 (Huson and Bryant, 2006). Discriminant analysis of principal components (**DAPC**), implemented in the R package adegenet (Jombart and Ahmed, 2011), was used to determine the genetic structure and differentiation between the goat populations. The optimal number of principal components reflecting the highest proportion of variance in the database was tested based on the a-score (Jombart and Ahmed, 2011). In addition, DAPC was used to assign individuals and obtain the affiliation probability representing the total genetic background of an individual. Relatedness between goat populations was also inferred using variational autoencoders (**VAE**), an unsupervised machine learning modeling approach based on a neural network. In this study, feed-forward networks were used for the encoder and decoder parts to regenerate the data and provide a visualization of the genetic data in a latent space. Variational autoencoders consist of a dual set of deep neural networks designed to effectively reduce the high-dimensional complexity of genomic data. The first network, called the encoder, converts the input data into a probability distribution in a latent space. At the same time, the second network, called the decoder, attempts to reconstruct the input data based on a given set of latent coordinates (Kingma and Welling, 2013). Unlike some other methods developed for large data sets (Battey et al., 2021), the algorithm implemented in the POPVAE software (https://github.com/kr-colab/popvae) has a lower dimensionality than principal component analysis while preserving the geometry.

Population Structure. Haplotype blocks of 4 SNPs were used to analyze the population structure of the breeds by the clustering algorithm based on the Bayesian model implemented in the STRUCTURE program (Pritchard et al., 2000). For all STRUCTURE runs, a model assuming admixture and correlated allele frequencies was used, with a burn-in of 10^5 and subsequent million Markov chain Monte Carlo iterations. Runs were repeated 10 times for each assumed K (the K-value is the number of (sub)populations or clusters assumed to exist in the analyzed dataset), starting with K = 1 to K = 22. The most likely K-value in the data set was determined using the rate of change of Ln Pr(G|K) (the log likelihood of the probability of genomic information given a certain K) between successive K-values, as suggested by Evanno et al. (2005), with calculations performed using the STRUCTURE HARVESTER program (Earl and von Holdt, 2012). Visualization of the STRUCTURE results was carried out using the web-based tool StructureSelector software (Li and Liu, 2018). The "same" STRUCTURE analysis was also performed for 17,800 biallelic SNPs obtained after pruning with a threshold for linkage disequilibrium between pairwise SNPs of 0.05 (r^2), a window of 50 SNPs, and a step size of 5 SNPs. However, the resulting population structure was less informative than that obtained with 5,136 polymorphic haplotype loci and is therefore not presented further.

Admixture Quantified by Effective Number of Parental Clusters. The STRUCTURE program can be used to estimate the admixture coefficient (Q-membership matrix) for each individual and provides information on the membership of the observed clusters within a particular breed or population. To distill the complexity of admixture into a single, informative metric for each breed or individual, we introduce a novel measure, termed effective number of parental clusters (**ENPC**). The ENPC serves as a quantitative indicator that captures the extent to which parental clusters contribute uniformly and effectively to the formation of a given breed or individual. It is calculated by deriving the inverse of the squared sum of all parental cluster proportions that contribute to the genetic composition of the breed or individual in question. For example, if only 2 parental clusters contribute 0.5 each to a breed, the resulting ENPC would be 2.0. In a more complex scenario where parental clusters A, B, C, D, and E contribute to a breed with proportions of 0.5, 0.3, 0.1, 0.1, and 0.0, respectively, the resulting ENPC would be 2.8. This implies that 2.8 parental clusters, each contributing equally, characterize the genetic composition of that particular breed. Notably, when ENPC is equal to 1.0, this means that the breed is 100% purebred, as only 1 parental cluster contributes exclusively to its genome. The ENPC thus provides a concise and informative measure to quantify the level of admixture and to assess the purity of breeds in terms of parental cluster contributions.

Gene Flow Between Breeds. Contemporary gene flow between goat breeds was analyzed by assessing migration rates (**m**) using the BayesAss v3 assignment test (Wilson and Rannala, 2003). The evaluation comprised 20 replicates with different random seeds, each subjected to Markov chain Monte Carlo simulations with up to 20 million iterations, with the first 2 million iterations discarded during the burn-in process. In 10 iterations, the mixing parameters—delta allele frequency, delta migration rate, and delta inbreeding coefficient—were set to 0.1, whereas these parameters were set to 0.2 in the remaining 10 replicates. In this way, we were able to slightly improve the reliability of our estimates. The resulting log outputs were analyzed using Tracer (Rambaut et al., 2018), and Bayesian deviance was computed following the R script provided in Meirmans (2014). The migration estimates from the 3 runs with the lowest Bayesian deviance were merged to create a posterior distribution encompassing the estimated migrations. Subsequently, we calculated the median and a 95% credibility interval from this distribution. A comprehensive overview can be found in Supplemental Table S2 (see Notes) and in Supplemental Figure S1 (see Notes).

RESULTS AND DISCUSSION

Conservation Status

Genetic Diversity and Haplotype Richness. Our analyses referred to the indigenous goat breeds that were not used for SNP array formation. To reduce the impact of ascertainment bias, which can lead to biased conclusions (Gautier et al., 2010), we used haplotype blocks, as suggested by Simčič al. (2015) and Pogorevc et al. (2024).

The estimates of genetic diversity (H_O, H_E, F_{IS}, HR, and HRR) based on haplotype blocks are shown in Table 1. Heterozygosity of haplotype blocks (H_O and H_E) for 20 goat breeds analyzed ranged from 0.722 (CHAPP) to 0.847 (ATSTP) for H_O and from 0.713 (CHAPP) to 0.852 (ATSTP) for H_E. With the exception of CZBSH, where the values for H_O and H_E were below the median, the other 4 Czech and Slovakian goats showed high heterozygosity, especially CZLAN, CZWSH, and SKWSH. The heterozygosity values of the haplotype blocks (H_O and H_E) were consistent with those of Pogorevc et al. (2024), where some of the investigated breeds overlapped with our data set and the analyses were also performed on 4 SNP blocks. We also estimated genetic diversity based on biallelic single-SNP loci; the results are shown in Supplemental Table S3 (see Notes). In 20 goat breeds analyzed, SNP-based heterozygosity was lower compared with estimates based on haplotype blocks, which is consistent with the phenomenon observed in other studies using both SNP-based and haplotype block-based estimates (Amador et al., 2014; Simčič et al., 2015; Manunza et al., 2023b). In this study, the SNP-based H_O ranged from 0.352 (CHTSG and CHAPP) to 0.418 (ATSTP), whereas the H_E ranged from 0.341 (CHAPP) to 0.415 (ATSTP) and were comparable to the results of other studies (Colli et al., 2018; Berihulay et al., 2019; Michailidou et al., 2019; Deniskova et al., 2021; Zhong et al., 2023; Drzaic et al., 2024). Although the magnitude of heterozygosity estimated by different methods was considerable, the linear correlation between heterozygosity estimates based on biallelic single SNPs or on haplotype block loci (4 SNPs) was very high (0.98) in 20 breeds.

Haplotype richness ranged from 6.33 (CHAPP) to 10.82 (ATSTP), which was comparable to the values determined by Pogorevc et al. (2024). Although the difference was small, more comparable values were obtained when HRR was corrected for sample size (2n = 40, where n is number of individuals in the smallest sample with 20 individuals). Although in 20 goat breeds the HRR was between 6.13 (CHTGB) and 10.15 (ATSTP) haplotypes (alleles), all other indigenous Czech and Slovakian breeds except CZBSH had very high HRR (Table 1).

In general, the heterozygosity and allelic richness of the 4 SNP haplotype blocks were comparable to those observed in microsatellites (Xiang-Long and Valentini, 2004; Cañón et al., 2006; Whannou et al., 2023), suggesting that they are roughly equivalent in measuring diversity (1 microsatellite corresponds to approximately 4 SNP haplotype blocks). At the same time, the Pearson correlation of 0.85 between H_O and HR was strong. Wright's inbreeding coefficient (F_{IS}) quantifies the deviation of the observed proportion of homozygotes within a population from the expected proportion in random mating and is an important parameter in population genetics. Because a high linear Pearson correlation (r = 0.913) was found between the estimates of haplotype blocks (4 SNPs) and the estimates for individual SNPs, we have presented only the F_{IS} estimates of haplotype blocks in Table 1. The F_{IS} estimates for the SNPs are shown in Supplemental Table S2. Negative F_{IS} values are interpreted as a preference for outbreeding or avoidance of mating with close relatives, and positive F_{IS} values indicate inbreeding or mating between close relatives. Of the 20 goat breeds analyzed, negative F_{IS} values (11 significantly different from 0) were found in 14 breeds, indicating avoidance of mating with close relatives, whereas 6 positive F_{IS} values (all significant) indicated the practice of inbreeding as a mating strategy (Table 1). Negative F_{IS} values have also been observed in other goat breeds (Colli et al., 2018; Drzaic et al., 2024), which seems to be a characteristic of goat populations. Of the 5 Czech and Slovakian breeds, only CZWSH showed slightly positive F_{IS} estimates, whereas CZALG and CZLAN indicated strong avoidance of mating with close relatives.

Runs of Homozygosity Genomic Inbreeding. Quite large differences in the estimated ROH-based genomic inbreeding levels (ROH >2 Mb) among 20 goat breeds, ranging from 0.025 (CZWSH) to 0.141 (CHAPP), were observed (Table 1). Similar inbreeding levels for $F_{ROH>2Mb}$ were observed in French (Oget et al., 2019) and Italian (Cortellari et al., 2021) goat breeds, whereas African goat breeds had somewhat lower inbreeding, with the third quartile equal to 0.049 (Nandolo et al., 2019). Two Czech breeds (CZWSH and CZLAN) and one Slovakian (SKWSH) breed had inbreeding levels much lower than the median inbreeding level, close to CZALG, over 20 breeds. The low F_{ROH} values observed in CZWSH and SKWSH may be attributed to their common history until 1993. During this time, these 2 breeds were kept as a unified breed, intermittently enriched with genetic material from other high-milking dairy goat breeds, in particular the Saanen goat. In contrast, although CZBSH have a similar breeding history to CZWSH and SKWSH, a high level of inbreeding has been observed (Table 1).

A more comprehensive examination of inbreeding patterns and their temporal origins (categorized as remote, intermediate, or recent inbreeding) can be found in Figure 1. Figure 1A shows that, with the exceptions of

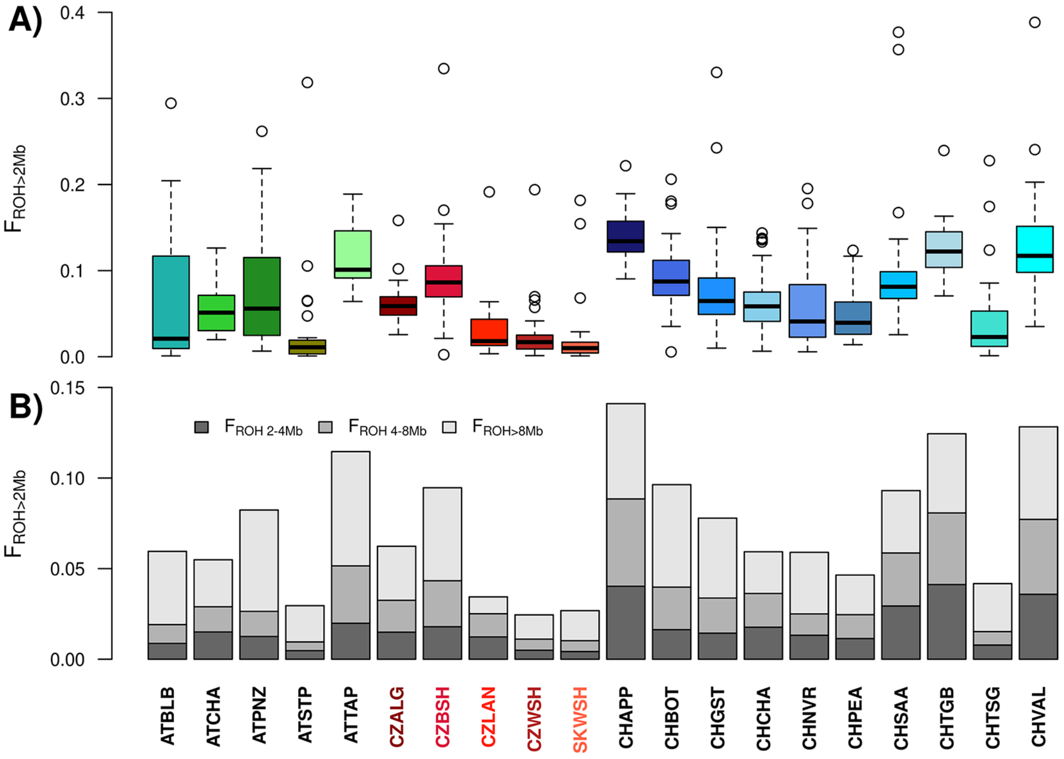

Figure 1. Distribution of ROH genomic inbreeding coefficient for 20 Central European goat breeds. (A) Box plot presentation of genomic ROH inbreeding coefficients ($F_{ROH\ >2Mb}$), where the upper and lower edges of boxes represent the lower to the upper quartile of the distribution, midlines show the median of the distribution, whiskers represent 1.5 × the interquartile range value, and dots are considered potential outliers. (B) Stacked-bar presentation of remote ($F_{ROH\ 2–4Mb}$), intermediate ($F_{ROH\ 4–8Mb}$), and recent ($F_{ROH\ >8Mb}$) autozygosity origins. The breeds indicated by the red text on the x-axis are the target breeds in this study. Austrian breeds (AT): ATBLB (Blobe), ATCHA (Chamois Colored), ATPNZ (Pinzgau), ATSTP (Styrian Pied), ATTAP (Tauern Pied). Czech breeds (CZ): CZWSH (White Shorthair), CZBSH (Czech Brown Shorthair), CZLAN (Czech Landrace), CZALG (Alpine). Slovak breed (SK): SKWSH (Slovak White Shorthair). Swiss breeds (CH): CHAPP (Appenzell), CHGST (Grisons Striped), CHCHA (Chamois Colored), CHNVR (Nera Verzasca), CHPEA (Peacock), CHSAA (Saanen), CHBOT (Booted), CHTSG (Tessin Gray), CHTGB (Toggenburg), CHVAL (Valais)..

ATCHA and ATTAP (note that the ATTAP breed is not a good example, as its inbreeding level of 0.115 was very high), all other breeds have outliers characterized by significantly high individual inbreeding. In particular, although rare, outliers with exceptionally high inbreeding coefficients (mostly even above 0.125) were found in all Czech breeds and 1 Slovakian goat breed, highlighting the potential for enhanced mating management strategies to minimize the risk of closely related individuals mating. Figure 1A unequivocally shows that the pronounced inbreeding in CZBSH and the moderate inbreeding in CZALG are clearly of recent origin, as evidenced by the observation of a high $F_{ROH\ >8Mb}$. These patterns emphasize the need for sound mating management in these 2 populations, leading to a reduction in inbreeding.

Effective Population Size. Effective population size is an important parameter in domestic animal genetic improvement programs, as it is functionally linked to genetic diversity, avoidance of inbreeding, response to selection, adaptability, and long-term viability of populations. Our estimates of Ne_{LD} for 20 goat breeds obtained using NeEstimator v2 (Do et al., 2014) and GONE (Santiago et al., 2020) software are presented in Table 2 (GONE results are presented for both phased and unphased genomic information). No significant differences were found between the phased and unphased Ne_{LD} estimates, as the confidence intervals of the GONE estimates overlap (see Table 2). However, the estimates obtained with GONE based on phased or unphased genomic information were higher than those obtained with NeEstimator v2, with significant differences in 11 out of 20 breeds where the confidence intervals did not overlap. Unfortunately, the empirical results showing that NeEstimator v2 estimates are lower compared with those of GONE are neither consistent nor conclusive. For example, the GONE estimates (without phased genomic information) obtained in Vostry et al. (2023) were significantly higher compared with the estimates obtained with NeEstimator v2 in only 2 of 13 Holstein cattle subpopulations ("breeds" of different origins), which was not the case in this study. According to simulations performed in several studies (Santiago et al., 2020; Reid and Pinsky, 2022; Novo et al., 2023) Ne_{LD}

Table 2. Genomic contemporary $\mathrm{Ne}_{\mathrm{LD}}$ estimates (95% CI) for 20 Central European goat breeds from Austria (AT), the Czech Republic (CZ), Slovakia (SK), and Switzerland (CH)

Breed	NeEstimator v2.0[1]	GONE unphased[2]	GONE phased[2]	ENPC[3]
Blobe (ATBLB)	28 (19–46)	64 (49–83)*	89 (67–118)*	2.5
Chamois Colored (ATCHA)	185 (112–492)	270 (166–440)	314 (189–521)	2.1
Pinzgau (ATPNZ)	59 (40–105)	110 (78–154)*	162 (112–235)*	1.7
Styrian Pied (ATSTP)	140 (98–237)	255 (176–367)	277 (190–403)	2.1
Tauern Pied (ATTAP)	95 (63–183)	166 (114–241)	106 (76–148)	1.1
Alpine (CZALG)	44 (28–89)	127 (89–180)	158 (109–228)*	1.1
Czech Brown Shorthair (CZBSH)	46 (31–82)	127 (93–173)*	159 (115–220)	1.2
Czech Landrace (CZLAN)	21 (13–29)	116 (74–179)*	73 (47–107)*	4.2
White Shorthair (CZWSH)	105 (68–205)	254 (179–358)	283 (198–404)	1.3
Slovak White Shorthair (SKWSH)	37 (23–78)	118 (79–173)*	149 (100–223)*	1.4
Appenzell (CHAPP)	64 (38–157)	142 (99–200)	189 (130–275)	2.0
Booted (CHBOT)	54 (33–129)	100 (82–120)	121 (82–180)	1.1
Grisons Striped (CHGST)	37 (28–54)	75 (60–91)*	88 (71–110)*	1.9
Chamois Colored (CHCHA)	80 (63–103)	159 (138–181)*	182 (158–209)*	1.8
Nera Verzasca (CHNVR)	46 (32–72)	97 (75–125)*	124 (95–162)*	1.8
Peacock (CHPEA)	56 (36–110)	182 (114–287)*	115 (84–158)	1.5
Saanen (CHSAA)	38 (27–58)	128 (90–182)*	117 (96–143)*	1.2
Toggenburg (CHTGB)	75 (45–173)	127 (91–175)	184 (129–263)	1.0
Tessin gray (CHTSG)	58 (40–95)	126 (94–167)	118 (89–157)	2.1
Valais (CHVAL)	37 (26–58)	111 (85–142)*	131 (101–171)*	1.1
Median	55 (35–99)	127 (91–177)	140 (101–195)	1.6

[1] NeEstimator v2.0 software estimates; Do et al. (2014).

[2] GONE software estimates; Santiago et al. (2020).

[3] Effective number of parental clusters derived from STRUCTURE analysis at K = 19; Pritchard et al. (2000).

*Significantly different estimates, GONE vs. NeEstimator v2.0, are marked with asterisks.

estimates obtained with GONE are generally considered more precise, although the simulations did not take into account all possible situations that could occur in the empirical populations, such as admixture or population structure, which is known to affect $\mathrm{Ne}_{\mathrm{LD}}$ estimates (Santiago et al., 2020). As the presence of admixed individuals and the introgression of wild goats into domestic goat breeds is well documented (Cortellari et al., 2021; Pogorevc et al., 2024), some estimates in this study should be interpreted with caution. Additionally, the existence of overlapping generations introduces another layer of potential bias in $\mathrm{Ne}_{\mathrm{LD}}$ estimates, as noted by Waples et al. (2014). However, higher estimates of 73 (CZLAN) to 314 (ATCHA) in the phased $\mathrm{Ne}_{\mathrm{LD}}$ or 64 (ATBLB) to 270 (ATCHA) in the unphased $\mathrm{Ne}_{\mathrm{LD}}$ were considered as acceptable. For example, many of the $\mathrm{Ne}_{\mathrm{LD}}$ estimates obtained with NeEstimator v2.0 were extremely low, with median of 55, which we consider unlikely given the population history and degree of admixture. With the exception of CZLAN, all GONE phased $\mathrm{Ne}_{\mathrm{LD}}$ estimates for Czech and Slovakian breeds were above the median value of 127 (unphased genotypes) or 140 (phased genotypes), whereas a particularly high $\mathrm{Ne}_{\mathrm{LD}}$ (283) was found in CZWSH. In 13 South African goat breeds, the estimated $\mathrm{Ne}_{\mathrm{LD}}$ was above 150 (Mdladla et al., 2016), which is close to the median observed for 20 breeds in this study. Inbreeding and gamete/linkage disequilibrium effective population size estimates may diverge, especially in the presence of significant migrations (Ryman et al., 2019).

However, the inbreeding effective population size, based on inbreeding rate per year, estimated from genome-wide data in French breeds, ranged from 114 to 356 (Rodríguez-Ramilo et al., 2019), which was fully consistent with the results of GONE for breed analyzed in this study (see Table 2). The estimates of effective population size obtained from pedigree data were in a similar range. For example, the estimated inbreeding effective population size was 244 for CZWSH and 82 for CZBSH (Vostra-Vydrova et al., 2020) or 149 for Alpine goats, 129 for Saanen goats, and 76 for Angora goats (Danchin-Burge et al., 2012).

In contrast, the estimation of the historical effective population size revealed a significant decline from about 12 to 8 generations ago in the CZLAN and SKWSH breeds, from 27 to 21 generations ago in CZBSH, and from 32 to 21 generations ago in CZALG. In CZWSH, a gradual but steady decline from 40 to 14 generations ago was observed. Please note that, according to Vostra-Vydrova et al. (2020), estimated generation interval was 4.16 for CZWSH and 3.92 for CZBSH. A visual representation of this trend can be found in Figure 2. Although there is no immediate need for drastic conservation measures for any of the breeds analyzed, the observed decline prompted us to reconsider our previous breeding policy decisions and the circumstances that caused this decline.

Relationship Among Parameters Defining Conservation Status. The relationships between genomic inbreed-

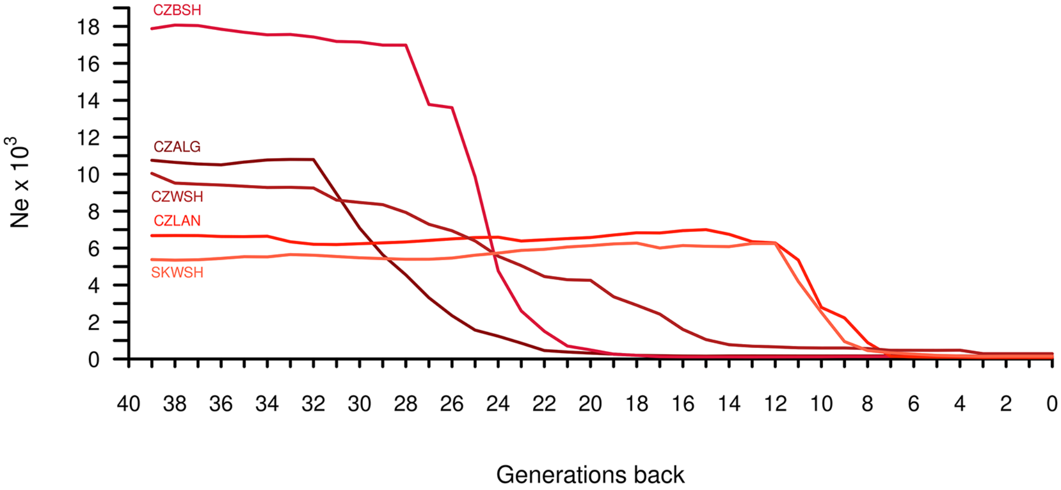

Figure 2. Historical gametic/linkage effective population size (Ne_{LD}) of Czech and Slovak goat breeds. Czech breeds are represented by CZWSH (White Shorthair), CZBSH (Czech Brown Shorthair), CZLAN (Czech Landrace), and CZALG (Alpine goat), and SKWSH (Slovak White Shorthair) represents the Slovak breed.

ing coefficient ROH, Wright's inbreeding coefficient (F_{IS}), and gamete/linkage population size (Ne_{LD}) are shown in Figure 3. Genomic ROH inbreeding within the goat breeds studied was primarily due to combination of low effective population size and deliberate avoidance of mating with close relatives. Consequently, breeds with very high genomic inbreeding, such as CHAPP, ATTAP, and CHTGB, have displayed negative F_{IS} values. This shows that, despite the observed high genomic inbreeding, mating with close relatives was avoided, as the F_{IS} values of these breeds were significantly negative, indicating that breeders are aware of the high inbreeding (Table 1; Figure 3). In contrast, no signs of inbreeding mitigation ($F_{IS} = 0$) were observed in CHVAL. The high genomic inbreeding with significant positive F_{IS} values observed in ATPNZ indicates the need for an effective diversity management strategy (e.g., the prevention of mating with close relatives, which can reduce the high inbreeding). Breeders should also be aware that the high positive F_{IS} values found in ATSTP and CHNVR could lead to high inbreeding in the long term, even if this is not currently a problem. In general, breeds with smaller effective population sizes tended to avoid mating with close relatives, indicating a heightened awareness of their conservation status. This concept is illustrated vividly in Figure 3, indicating the power of the visualization originally proposed by Clark et al. (2019).

Genetic Relatedness and Population Structure

Genetic Relatedness Between Goat Breeds. Various methods were used to analyze the relatedness and population structure. Figure 4 shows the relatedness between the Czech and Slovakian goat breeds and the corresponding breeds from Austria and Switzerland through DAPC, VAE, and Neighbor-Net approaches.

Both DAPC and VAE show the relatedness at both individual and breed levels, whereas Neighbor-Net shows only breed-related connections. Remarkably, the first 2 discriminatory functions of DAPC elucidate 42% of the total variation, with the first and second functions particularly separating CHAPP, CHTSG, and CHSAA from other breeds (see Figure 4A). As shown in Figure 4B, a more pronounced relatedness between goat breeds was determined using the VAE approach implemented in the POPVAE software (Battey et al., 2021). It is noteworthy that the POPVAE representation of breed relatedness shows a country-specific separation, with the exception of ATCHA, which was positioned within the Swiss goats. This is logical, as ATCHA is a transboundary Swiss breed (CHCHA) that also occurs in Italy (Camosciata delle Alpi) and Austria (ATCHA). Interestingly, the Czech and Slovakian breeds are located along a relatively narrow central axis that separates the Austrian breeds from their Swiss counterparts. This unexpected arrangement does not correspond to the geographical expectations arising from the habitats of the goat breeds studied but seems to reflect the historical influence of Saanen goats (here CHSAA) on CZWSH and SKWSH. The recent application of VAE in population genomics has emerged as a versatile and powerful tool for extracting meaningful insights from genetic data. This approach facilitates the revelation of hidden nonlinear structures and allows inference of relatedness and genetic diversity within and between populations (Battey et al., 2021). Despite the promising potential of VAE in demonstrated population

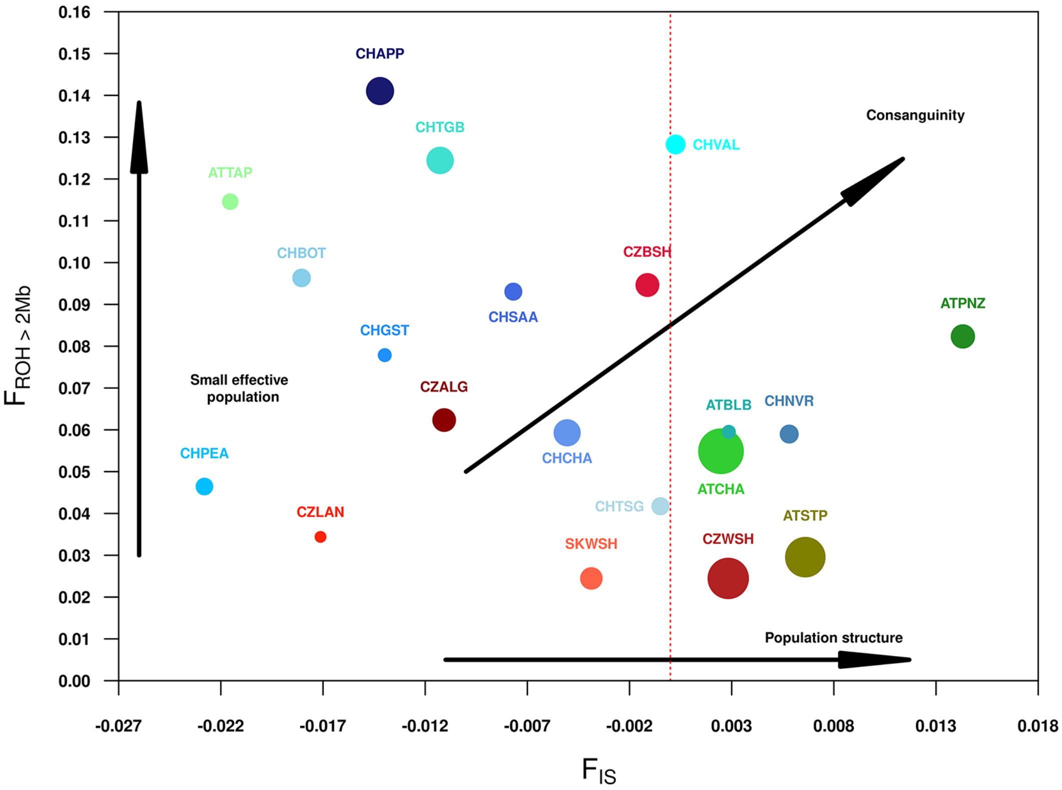

Figure 3. Three-dimensional plot (bubble plot) of the relationship between genomic ROH inbreeding ($F_{ROH >2Mb}$), Wright's inbreeding coefficient (F_{IS}), and gamete/linkage effective population size (Ne_{LD}; proportional to bubble size) in 20 Central European goat breeds. Shades of blue represent the Swiss breeds, shades of red represent the Czech and Slovak breeds, and shades of green represent the Austrian breeds. Austrian breeds (AT): ATBLB (Blobe), ATCHA (Chamois Colored), ATPNZ (Pinzgau), ATSTP (Styrian Pied), ATTAP (Tauern Pied). Czech breeds (CZ): CZWSH (White Shorthair), CZBSH (Czech Brown Shorthair), CZLAN (Czech Landrace), CZALG (Alpine). Slovak breed (SK): SKWSH (Slovak White Shorthair). Swiss breeds (CH): CHAPP (Appenzell), CHGST (Grisons Striped), CHCHA (Chamois Colored), CHNVR (Nera Verzasca), CHPEA (Peacock), CHSAA (Saanen), CHBOT (Booted), CHTSG (Tessin Gray), CHTGB (Toggenburg), CHVAL (Valais).

genomics (Battey et al., 2021), it is noteworthy that, to our knowledge, this study represents the first application of VAE in analyzing relatedness in domestic animals. Consequently, further empirical and theoretical investigations are needed to fully understand the proper use and interpretation of VAE in this context.

The phylogenetic network (Neighbor-Net) derived from Reynolds's pairwise genetic distances, which was formed on a priori defined breeds, complements the results of the DAPC and VAE approaches (Figure 4C). The Swiss breeds were clearly separated from the Austrian breeds, with the exception of ATCHA, which confirmed the VAE results. The other Austrian breeds (ATBLB, ATPNZ, ATSTP, and ATTAP) were between CZBSH and SKWSH on the one hand and between CZBSH, CZLAN, and CZALG on the other. The separation between Austrian and Swiss breeds corresponded to the results of the VAE analysis.

The population differentiation (relatedness) of 20 Central European goat breeds from Austria, the Czech Republic, Slovakia, and Switzerland is shown in Table 3 in the form of pairwise Wright's fixation index (F_{ST}) values together with the mean F_{ST} values of each breed with other 19 breeds analyzed in this study. Pairwise F_{ST} values were calculated between 20 populations ranging from the closely related breeds ATCHA and CHCHA (F_{ST} = 0.007) to the less related breeds CHTGB and ATTAP (F_{ST} = 0.149). The mean F_{ST} value for all pairs was 0.085, which is consistent with Hall's (2022) estimates for SNP arrays (0.080 to 0.160) for goat breeds worldwide. Like ATCHA and CHCHA, CZWSH and SKWSH also showed very close relatedness (F_{ST} = 0.017), with both pairs being transboundary breeds that could be considered subpopulations within a metapopulation. This is consistent with the results of Vostry et al. (2023), who reported small F_{ST} values (0.003 to 0.028) and close relatedness between Holstein subpopulations. According to Wright (1965), F_{ST} values of 0.05 to 0.15 are considered moderately differentiated between breeds (populations). The most distant population was CHAPP with the highest average F_{ST} value of 0.121, whereas the most central breed was CZWSH with the lowest average F_{ST} value of 0.058. In

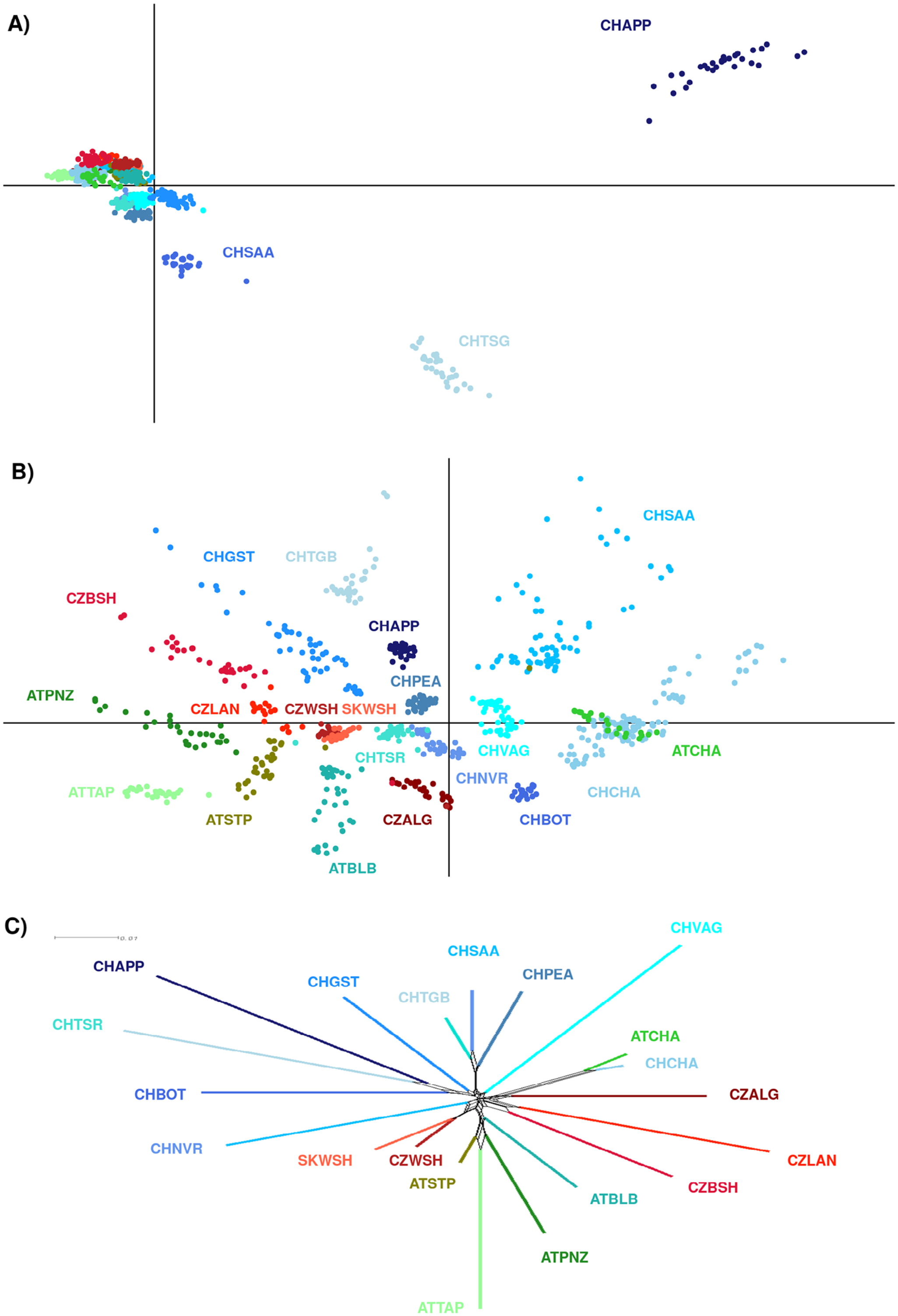

Figure 4. Relatedness between 20 Central European goat breeds. (A) DAPC scatter plot presenting a variation of the first 2 discriminant functions. (B) POPVAE-generated (default settings) clustering based on variational auto encoders (VAE) and presented in a scatter plot to illustrate population genetic variation in the reduced dimensionality of latent variables. (C) Phylogenetic network (Neighbor-Net) inferred from pairwise Reynolds's genetic distances. Shades of blue represent the Swiss breeds, shades of red represent the Czech and Slovak breeds, and shades of green represent the Austrian breeds. Austrian breeds (AT): ATBLB (Blobe), ATCHA (Chamois Colored), ATPNZ (Pinzgau), ATSTP (Styrian Pied), ATTAP (Tauern Pied). Czech breeds (CZ): CZWSH (White Shorthair), CZBSH (Czech Brown Shorthair), CZLAN (Czech Landrace), CZALG (Alpine). Slovak breed (SK): SKWSH (Slovak White Shorthair). Swiss breeds (CH): CHAPP (Appenzell), CHGST (Grisons Striped), CHCHA (Chamois Colored), CHNVR (Nera Verzasca), CHPEA (Peacock), CHSAA (Saanen), CHBOT (Booted), CHTSG (Tessin Gray), CHTGB (Toggenburg), CHVAL (Valais).

general, the average F_{ST} value observed between Czech and Slovak goats (0.085) reflects the relatedness between 2 randomly selected breeds analyzed in this study.

Population Structure. To identify population structure and estimate admixture levels, we employed the STRUCTURE algorithm, known for its ability to uncover "hidden structure" and quantify admixture without requiring a priori membership in individual clusters. We executed STRUCTURE runs from K = 1 to K = 22. The Ln Pr(G|K) value consistently increased until K = 19, at which point the stability between runs of the same K-value started to decrease (see Figure 5A). The highest rate of Ln Pr(G|K) change between successive K-values was observed at K = 19 (see Figure 5B). Following the recommendations of Evanno et al. (2005), Falush et al. (2007), and Pritchard et al. (2000), it is likely that K = 19 is the most suitable value for the analyzed data set. Hence, we have presented the STRUCTURE results for admixture membership at K = 19. In addition, the results at K = 11, K = 13, and K = 17 are shown to illustrate the clustering process conducted in this study (see Figure 5C).

The optimal number of identified clusters (K = 19) resulted from the presence of 2 transboundary breeds that were not distinguished by the STRUCTURE algorithm. In contrast, 2 distinct clusters were identified in ATBLB. Neither the DAPC nor the VAE approach were able to distinguish ATCHA from CHCHA, or CZWSH from SKWSH, as can be seen in Figure 4A and 4B, where these breeds either overlapped or were closely branched (see Figure 4C).

Admixture Quantified by Effective Number of Parental Clusters. The STRUCTURE algorithm revealed considerable admixture within the analyzed goat breeds. For example, admixture was observed in ATBLB, ATCHA, ATSTP, CHTGB, and CHCHA, with CZLAN particularly standing out as an extremely admixed breed (see Figure 5C). Building on the STRUCTURE results at K = 19 (admixture coefficients), we further quantified the admixture present in each breed using new metrics termed ENPC, as shown in Table 2. The ENPC values numerically supported the admixture visualized in Figure 5C, indicating that the highest ENPC value (4.2) was observed in CZLAN, followed by ATBLB (ENPC = 2.5), ATCHA (ENPC = 2.1), ATSTP (ENPC = 2.1), CHTGB (ENPC = 2.1), and CHCHA (ENPC = 2.0). The 3 other Czech breeds and 1 Slovakian breed we focused on were relatively homogeneous, with ENPC values between 1.1 and 1.4, which was below the median value of 1.6 observed in 20 goat breeds (see Table 2). The calculation of the ENPC follows the logic of the definition of the effective number of alleles proposed by Kimura and Crow (1964), which is defined here as the effective number of parental clusters that contribute equally to a breed (population). For example, an ENPC value of 4.0 would

Table 3. Population differentiations in 20 Central European goat breeds from Austria, the Czech Republic, Slovakia, and Switzerland[1]

Breed	ATBLB	ATCHA	ATPNZ	ATSTP	ATTAP	CZALG	CZBSH	CZLAN	CZWSH	SKWSH	CHAPP	CHCHA	CHGST	CHNVR	CHPEA	CHSAA	CHBOT	CHTSG	CHTGB	MF_{ST}
ATBLB																				0.075
ATCHA	0.059																			0.067
ATPNZ	0.065	0.070																		0.085
ATSTP	0.040	0.041	0.046																	0.059
ATTAP	0.095	0.099	0.095	0.064																0.110
CZALG	0.070	0.059	0.081	0.054	0.108															0.082
CZBSH	0.077	0.065	0.086	0.058	0.112	0.092														0.087
CZLAN	0.089	0.079	0.101	0.073	0.128	0.086	0.084													0.097
CZWSH	0.045	0.038	0.054	0.028	0.082	0.057	0.062	0.063												0.058
SKWSH	0.049	0.043	0.059	0.031	0.087	0.062	0.066	0.069	0.017											0.062
CHAPP	0.114	0.111	0.126	0.097	0.154	0.120	0.126	0.139	0.095	0.100										0.121
CHCHA	0.064	0.007	0.075	0.048	0.101	0.060	0.068	0.080	0.043	0.047	0.109									0.069
CHGST	0.077	0.070	0.088	0.060	0.115	0.083	0.087	0.099	0.060	0.063	0.122	0.073								0.086
CHNVR	0.070	0.062	0.082	0.055	0.108	0.075	0.082	0.093	0.053	0.056	0.118	0.066	0.077							0.078
CHPEA	0.070	0.063	0.082	0.055	0.110	0.076	0.083	0.094	0.053	0.057	0.118	0.066	0.078	0.053						0.079
CHSAA	0.089	0.077	0.099	0.069	0.125	0.090	0.096	0.108	0.065	0.068	0.132	0.077	0.096	0.090	0.089					0.095
CHBOT	0.088	0.071	0.102	0.072	0.129	0.092	0.100	0.112	0.070	0.075	0.131	0.074	0.097	0.088	0.090	0.107				0.096
CHTSG	0.059	0.051	0.071	0.043	0.098	0.065	0.071	0.084	0.042	0.046	0.107	0.055	0.067	0.034	0.047	0.081	0.076			0.068
CHTGB	0.110	0.106	0.122	0.092	0.149	0.116	0.121	0.134	0.091	0.098	0.137	0.104	0.113	0.113	0.110	0.130	0.120	0.102		0.116
CHVAL	0.103	0.095	0.118	0.089	0.144	0.110	0.115	0.128	0.086	0.091	0.152	0.095	0.115	0.106	0.105	0.120	0.123	0.095	0.147	0.112

[1] Austrian breeds (AT): ATBLB = Blobe, ATCHA = Chamois Colored, ATPNZ = Pinzgau, ATSTP = Styrian Pied, ATTAP = Tauern Pied. Czech breeds (CZ): CZWSH = White Shorthair, CZBSH = Czech Brown Shorthair, CZLAN = Czech Landrace, CZALG = Alpine. Slovak breed (SK): SKWSH = Slovak White Shorthair. Swiss breeds (CH): CHAPP = Appenzell, CHGST = Grisons Striped, CHCHA = Chamois Colored, CHNVR = Nera Verzasca, CHPEA = Peacock, CHSAA = Saanen, CHBOT = Booted, CHTSG = Tessin Gray, CHTGB = Toggenburg, CHVAL = Valais.

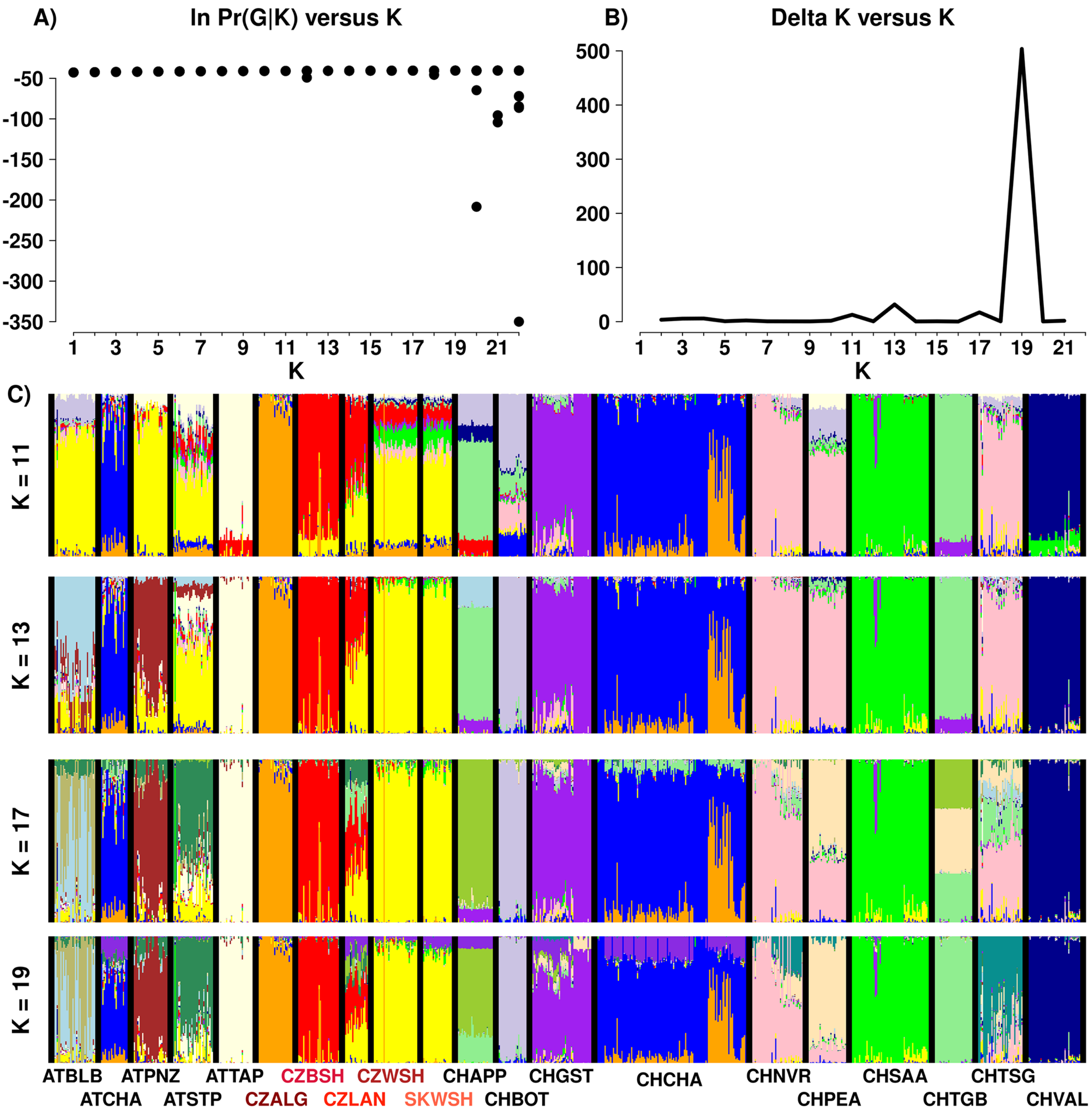

Figure 5. Population structure and admixture of 20 Central European goat breeds (defined a priori) from Austria, the Czech Republic, Slovakia, and Switzerland. Selection of representative number of clusters (K) based on plotting Ln Pr(G|K) values as a function of the number of clusters, 10 runs (A), plotting ΔK as a function of the number of clusters (B), and graphical representation of Bayesian unsupervised clustering (K = 4, K= 5, and K = 6) of 740 individual goats, performed with the algorithm STRUCTURE and visualized with the software CLUMPAK (C). Each goat is represented by a single vertical line, divided into K colored segments whose length is proportional to the estimated membership of the inferred cluster to illustrate the presence of admixture. The breeds indicated by the red text on the x-axis are the target breeds in this study. Austrian breeds (AT): ATBLB (Blobe), ATCHA (Chamois Colored), ATPNZ (Pinzgau), ATSTP (Styrian Pied), ATTAP (Tauern Pied). Czech breeds (CZ): CZWSH (White Shorthair), CZBSH (Czech Brown Shorthair), CZLAN (Czech Landrace), CZALG (Alpine). Slovak breed (SK): SKWSH (Slovak White Shorthair). Swiss breeds (CH): CHAPP (Appenzell), CHGST (Grisons Striped), CHCHA (Chamois Colored), CHNVR (Nera Verzasca), CHPEA (Peacock), CHSAA (Saanen), CHBOT (Booted), CHTSG (Tessin Gray), CHTGB (Toggenburg), CHVAL (Valais).

correspond to an admixture of 4 breeds (populations), each contributing 25% of the genes, whereas an ENPC value of 2.0 would correspond to a breed (population) in which 2 breeds each contribute 50% of the genes. In this study, we found that ENPC is an appropriate metric that quantifies admixture with a single number.

We consider that ENPC is useful to explain the variations of recent inbreeding ($F_{ROH >8Mb}$), as the negative linear regression coefficient of $F_{ROH >8Mb}$ on ENPC (over 20 breeds) was significant ($P = 0.048$), with a remarkable coefficient of determination ($R^2 = 0.201$).

Gene Flow Between Breeds. The assessment of contemporary gene flow between the 20 goat breeds analyzed (Figure 6) sheds light on the admixture patterns observed with the STRUCTURE algorithm. We plotted 10 migration rates, and only those with Bayesian 95% credible intervals (**95%CrI**) that did not overlap 0.01. It is noteworthy that the observed low migration rates show predominantly unidirectional trends. The highest migration rate was observed from ATSTP to ATBLB (m = 0.066 with 95%CrI of 0.043 to 0.094). Only bidirectional migrations were identified between ATCHA and CHCHA, with a higher migration rate from CHCHA to ATCHA (m = 0.060 with 95%CrI of 0.024 to 0.106) and a slightly lower migration rate from ATCHA to CHCHA (m = 0.018 with 95%CrI of 0.012 to 0.024).

However, no bidirectional pattern was evident between CZWSH and SKWSH, which could be considered a transboundary breed. Thus, the contemporary migration rate from CZWSH to SKWSH was estimated (m = 0.063 with 95%CrI of 0.037 to 0.090), but not vice versa. In particular, BayesAss reaches its limits when estimating long-term migration rates, which prevents the exploration of historical interactions. Although the exchange of goats within the same country is expected, some cross-border migrations raise interesting questions, such as the migration rate from CZALG to CHCHA (m = 0.043 with 95%CrI of 0.032 to 0.056). Overall, the contemporary migration rates inferred among goat breeds provide insights into the observed admixture and complement the STRUCTURE approach. However, caution should be exercised when interpreting these estimates, especially when modeling assumptions are compromised. The BayesAss estimate of gene flow proves to be particularly sensitive when genetic differentiation between breeds is low ($F_{ST} <0.02$). For example, the F_{ST} between ATCHA and CHCHA was 0.007, and the F_{ST} between CZWSH and SKWSH was 0.017. The low F_{ST} values even prompt consideration of whether these breeds should be delineated as separate entities (breeds) or as a single metapopulation subdivided into subpopulations. Conversely, even under violated modeling assumptions, there are accurate estimates of migration rates when F_{ST} estimates exceed 0.100, which is evident in several pairwise estimates in this study (see Table 3 and Supplemental Table S3). Rigorous and comprehensive analyses, including model selection from 3 optimal replicates of 10, mitigated the convergence and other issues associated with BayesAss estimation in this study (Faubet et al., 2007; Meirmans, 2014).

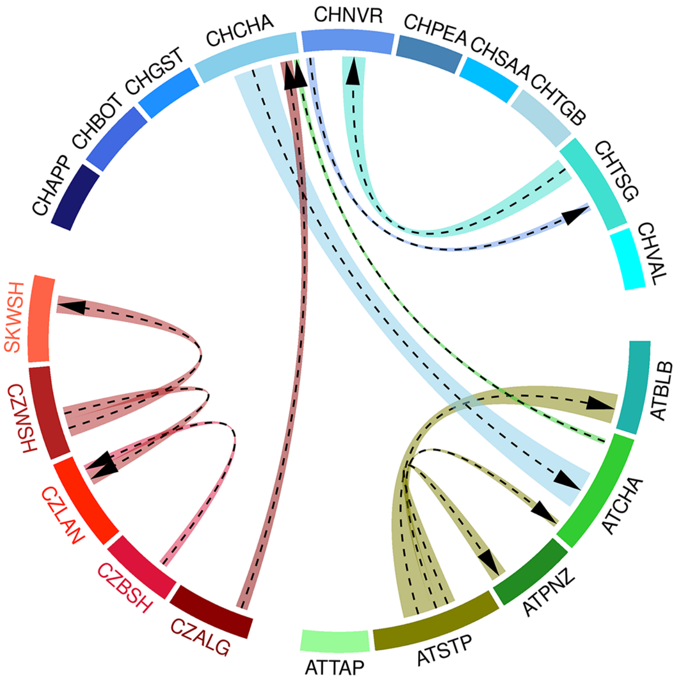

Figure 6. Estimated relative migration pattern between 20 Central European goat breeds from Austria, the Czech Republic, Slovakia, and Switzerland. The most intense migrations (>1%) are indicated with gene flow directions by black dashed arrows. Shades of blue represent the Swiss breeds, shades of red represent the Czech and Slovak breeds, and shades of green represent the Austrian breeds. Austrian breeds (AT): ATBLB (Blobe), ATCHA (Chamois Colored), ATPNZ (Pinzgau), ATSTP (Styrian Pied), ATTAP (Tauern Pied). Czech breeds (CZ): CZWSH (White Shorthair), CZBSH (Czech Brown Shorthair), CZLAN (Czech Landrace), CZALG (Alpine). Slovak breed (SK): SKWSH (Slovak White Shorthair). Swiss breeds (CH): CHAPP (Appenzell), CHGST (Grisons Striped), CHCHA (Chamois Colored), CHNVR (Nera Verzasca), CHPEA (Peacock), CHSAA (Saanen), CHBOT (Booted), CHTSG (Tessin Gray), CHTGB (Toggenburg), CHVAL (Valais).

CONCLUSIONS

In 4 Czech goat breeds and 1 Slovakian goat breed, the parameters for conservation status—such as gene diversity, HR, $F_{ROH >2Mb}$, and Ne_{LD}—were above the median values of all 20 breeds analyzed, with the exception of the CZBSH. Notably, the Ne_{LD} estimated by GONE (95%CI from 115 to 220) was not considered critical even for CZBSH. However, for all 5 breeds, an examination of historical effective population size indicated a substantial decline approximately 8 to 22 generations ago. In addition, our study revealed that the Czech and Slovakian breeds are not fully consolidated; for instance, CZWSH and SKWSH were not clearly distinguishable. Considerable admixture, especially in CZLAN (ENPC = 4.2) was observed, together with estimated low contemporary migrations mainly within Austrian, Czech, and Swiss breeds. These results provide valuable insights for future breeding programs and management initiatives aimed at preserving the genetic diversity of local Czech and Slovak goat breeds.

NOTES

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Nonstandard abbreviations used: 95%CrI = Bayesian 95% credible interval; ATBLB = Austrian Blobe; ATCHA = Austrian Chamois Colored; ATPNZ = Austrian Pinzgau; ATSTP = Austrian Styrian Pied; ATTAP = Austrian Tauern Pied; BSH = Brown Shorthair; CHAPP = Swiss Appenzell; CHBOT = Swiss Booted; CHCHA = Swiss Chamois Colored; CHGST = Swiss Grisons Striped; CHNVR = Swiss Nera Verzasca; CHPEA = Swiss Peacock; CHSAA = Swiss Saanen; CHTGB = Swiss Toggenburg; CHTSG = Swiss Tessin Gray; CHVAL = Swiss Valais; CZALG = Czechian Alpine; CZBSH = Czech Brown Shorthair; CZLAN = Czech Landrace; CZWSH = Czech White Shorthair; DAPC = discriminant analysis of principal components; ENPC = effective number of parental clusters; F_{IS} = individual inbreeding coefficient; F_{ROH} = runs of homozygosity genomic inbreeding coefficient; F_{ST} = Wright's fixation index; H_E = expected heterozygosity; H_O = observed heterozygosity; HR = haplotype richness; HRR = haplotype richness by rarefaction; K-value = number of (sub)populations or clusters assumed to exist in the analyzed dataset; m = migration rate; Ne_{LD} = contemporary effective population size; ROH = runs of homozygosity; SKWSH = Slovak White Shorthair; VAE = variational auto encoders; WSH = White Shorthair.

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