

Geographical contrasts of Y-chromosomal haplogroups from wild and domestic goats reveal ancient migrations and recent introgressions

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Abstract

By their paternal transmission, Y-chromosomal haplotypes are sensitive markers of population history and male-mediated introgression. Previous studies identified biallelic single-nucleotide variants in the SRY, ZFY, DDX3Y genes, which in domestic goats identified four major Y-chromosomal haplotypes Y1A, Y1B, Y2A and Y2B with a marked geographic partitioning. Here, we analyze whole-genome sequences of 386 domestic goats from 75 modern breeds and 7 wild goat species that were generated by the VarGoats goat genome project. Phylogenetic analyses indicated domestic haplogroups corresponding to Y1B, Y2A and Y2B, respectively, whereas Y1A is split into Y1AA and Y1AB. All five haplogroups were detected in 26 ancient DNA samples from southeast Europe or Asia. Haplotypes from present-day bezoars are not shared with domestic goats and are attached to deep nodes of the trees and networks. Haplogroup distributions for 180 domestic breeds indicate ancient paternal population bottlenecks and expansions during the migrations into northern Europe, eastern and southern Asia and Africa south of the Sahara. In addition, sharing of haplogroups indicates male-mediated introgressions, most notably an early gene flow from Asian goats into Madagascar and the crossbreeding that in the 19th century resulted in the popular Boer and Anglo-Nubian breeds. More recent introgressions are those from European goats into the native Korean goat population and from Boer goat into Uganda, Kenya, Tanzania, Malawi and Zimbabwe. This study illustrates the power of the Y-chromosomal variants for reconstructing the history of domestic species with a wide geographic range.

Geographical contrasts of Y-chromosomal haplogroups from wild and domestic goats reveal ancient migrations and recent introgressions

Running title: Y-chromosomal variation in goats

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Abstract

By their paternal transmission, Y-chromosomal haplotypes are sensitive markers of population history and male-mediated introgression. Previous studies identified biallelic single-nucleotide variants in the *SRY*, *ZFY*, *DDX3Y* genes, which in domestic goats identified four major Y-chromosomal haplotypes Y1A, Y1B, Y2A and Y2B with a marked geographic partitioning. Here, we analyze whole-genome sequences of 386 domestic goats from 75 modern breeds and 7 wild goat species that were generated by the VarGoats goat genome project. Phylogenetic analyses indicated domestic haplogroups corresponding to Y1B, Y2A and Y2B, respectively, whereas Y1A is split into Y1AA and Y1AB. All five haplogroups were detected in 26 ancient DNA samples from southeast Europe or Asia. Haplotypes from present-day bezoars are not shared with domestic goats and are attached to deep nodes of the trees and networks. Haplogroup distributions for 180 domestic breeds indicate ancient paternal population bottlenecks and expansions during the migrations into northern Europe, eastern and southern Asia and Africa south of the Sahara. In addition, sharing of haplogroups indicates male-mediated introgressions, most notably an early gene flow from Asian goats into Madagascar and the crossbreeding that in the 19th century resulted in the popular Boer and Anglo-Nubian breeds. More recent introgressions are those from European goats into the native Korean goat population and from Boer goat into Uganda, Kenya, Tanzania, Malawi and Zimbabwe. This study illustrates the power of the Y-chromosomal variants for reconstructing the history of domestic species with a wide geographic range.

Keywords :

goat, Y-chromosome, phylogeography, haplogroup, domestication, migration, introgression

Introduction

Goat (*Capra hircus*), sheep (*Ovis aries*), cattle (*Bos taurus*) and pigs (*Sus scrofa*) are four major livestock species, which after their domestication in southwest Asia about 10,000 BP (Larson & Fuller, 2014; Stiner et al., 2022) spread to all inhabited continents. Because of their relatively small size, sheep and goats were the earliest domesticates, but have become less important than cattle and pigs as suppliers of food. However,

sheep and goat are suitable for extensive management by smallholders or hobby breeders with goats being favored in conditions of poverty (Peacock, 2005). Whereas the high-quality goat cashmere wool and mohair fibres do not attain the volume of sheep wool, the demand of goat milk and cheese has increased considerably since the 1960s (Dubeuf, Morand-Fehr, & Rubino, 2004; Miller & Lu, 2019). During the last four decades, this has doubled the global number of goats to around 1 billion heads (Utaaker, 2021), which now approaches the numbers for sheep and cattle (Hegde, 2019).

As for other livestock, genetic isolation, adaptation and selection have created numerous local goat populations, whereas a restricted number of high-performing breeds play a major role in the agricultural production. The genetic diversity of goat breeds has been studied extensively (Ajmone-Marsan et al., 2014; Amills et al., 2017; Deniskova et al., 2021; Zheng et al., 2020). This demonstrated for autosomal DNA a geographic partitioning of the diversity (Colli et al., 2018), which is in sharp contrast to similar studies of sheep (Belabdi et al., 2019; Ciani et al., 2020; Kijas et al., 2012), but has been confirmed by similarities of ancient and modern DNA samples from the same regions (Cai et al., 2020; Daly et al., 2018, 2021). However, the goat Y-chromosome as a marker for the paternal lineages has not yet been studied at a worldwide scale.

Because of its absence of recombination, the male-specific part of the mammalian Y-chromosomes is by far the longest haplotype that is stably transmitted across generations (Hughes et al., 2015). In many species, males have a relatively small male effective population size, which makes Y-chromosomal variants highly informative markers for paternal origin that generally show a much stronger phylogeographic differentiation than mitochondrial or autosomal variants. This is now widely exploited in population-genetic studies of humans (Batini & Jobling, 2017; Jobling & Tyler-Smith, 2017; Kivisild, 2017), cattle (Edwards et al., 2011; Ganguly et al., 2020; Xia et al., 2019), horse (Felkel et al., 2019; Wallner et al., 2017; Wutke et al., 2018), water buffalo (Zhang et al., 2016), sheep (Deng et al., 2020; Meadows & Kijas, 2009; Zhang et al., 2014), camel (Felkel et al., 2019), llamas and alpacas ((Marín et al., 2017) pigs (Choi et al., 2020; Guirao-Rico et al., 2018) and dogs (Natanaelsson et al., 2006; Oetjens et al., 2018).

A preliminary analysis of the Y-chromosomal diversity in European and Turkish goats defined the three haplotypes Y1A, Y1B and Y2, which had a strong geographic differentiation (Lenstra & Econogene Consortium, 2005). The same haplotypes were found in goats from Portugal and North-Africa (Pereira et al., 2009), Turkey (Çinar Kul et al., 2015), eastern and southern Asia (Waki et al., 2015; Tabata et al., 2018, 2019) and Switzerland and Spain (Vidal et al., 2017) with the additional haplotypes Y2B in east Asia, Y2C in Turkish Hair and Kilis goats, and Y1B2 as well Y1C mainly in Switzerland (Table S1). However, these haplotypes are based on a low number of SNPs in or near *DDX3Y*, *SRY* and *ZFY* and genotyping in a limited number of domestic goat breeds. Thus, it is not clear if the haplotypes represent major haplogroups or local variants or if other major haplogroups exist. Nor does it inform us on the Y-chromosomal variants that existed in the earlier domestic goats or in their wild ancestor, the bezoar (*Capra aegagrus*) (Amills et al., 2017). Whole-genome sequences (WGSs), however, has confirmed the differentiation of the Y1 and Y2 haplogroups (Xiao et al., 2021; Zheng et al., 2020).

In this study, we used WGS data for a large panel of goat breeds (Denoyelle et al., 2021) to systematically characterize the SNP-level variation in the single-copy male-specific part of the caprine Y-chromosome. In addition, we determined the Y-chromosomal haplogroups in goats originating from several European, Asian or African countries, in ancient goat DNA samples and in the wild bezoar (Alberto et al., 2018; Cai et al., 2020; Daly et al., 2018, 2021; M. Zhang et al., 2014; Zheng et al., 2020). We sought to answer the following questions (1) How are the WGS- based haplogroups related to the previously reported haplotypes? (2) How are the domestic paternal lineages related to those of bezoars from Iran and Anatolia, respectively? (3) How strong is the phylogeographic structure of the caprine male lineages? (4) What does the pattern of diversity tell us about Neolithic and later migrations? (5) Can we also infer other gene flows between or within continents? Answering these questions will contribute to our understanding of the genetic background of the domestic goat, which is relevant for breed management and conservation.

Materials and Methods

WGS data, filtering and phylogenetic analysis

We selected as source of the SNPs four scaffolds that together cover 1,567,760 bp of the male-specific part of the caprine Y-chromosome. These are unplaced in the ARS1 assembly but for a large part closely match a recent Y-chromosomal contig of the Saanen_v1 assembly (Table S2)(Li et al., 2021) and contain the single-copy Y-chromosomal genes *SRY*, *DDX3Y*, *ZFY* with the SNPs that define the major haplotypes Y1A, Y1B, Y2A and Y2B (Çinar Kul et al., 2015; Lenstra & Econogene Consortium, 2005; Waki et al., 2015). The genes *USP9Y*, *UTY*, *DDX3Y* and *ZFY* are proximate near one of the ends of the male-specific Y-chromosomal region and well separated from *SRY* (Li et al., 2021). The selected contigs have a low overall level of apparent heterozygosity, indicating a high frequency of hemizygous markers (Table S2).

In a preliminary study (<https://www.biorxiv.org/content/biorxiv/early/2020/02/17/2020.02.17.952051.full.pdf>), we used WGS data from the Sequence Read Archive for 70 mainly Asian and Moroccan male goats. We extracted the genotypes of 2350 SNPs without female of male-heterozygous scores, <5% missing scores/SNP and a minor allele frequency (MAF) >0.02.

For a more comprehensive global coverage, we used the WGS datasets generated by the VarGoats project (Denoyelle et al., 2021, www.goatgenome.org/vargoats.html) or whole-genome sequences that have been published previously or have been submitted to the Short-Read Archive (Table S3). Four Y-chromosomal scaffolds were selected with a low average frequency of heterozygous variants, which cannot belong to a hemizygous Y-chromosomal SNP (Table S2). A VCF file for these scaffolds was imported into Plink and contained 65556 variants, 54032 of which were SNPs. From the 424 male samples, 34 were from wild goats, whereas 354 domestic goats had call rate of >5%. After discarding two domestic goats with unknown breed origin, we kept 386 wild or domestic goats. SNPs were selected in four subsequent steps. (i) In 670 female goats with <1% scores for the Y-chromosomal SNPs (*i.e.* free of male contamination) 36804 SNPs had a call rate of zero and thus were considered male-specific. (ii) 29035 SNPs did not show heterozygous score in the 424 males and were considered as being hemizygous. (iii) 16495 had a call rate in the 388 male goats (354 domestic, 34 wild) of >1%. (iv) 552 SNPs had a MAF in the 354 male domestic goats of >95% and from the remaining 15943, 9977 SNPs had at least one score in wild goats, summing up to a total of 10529 SNPs representing male-specific Y-chromosomal variation in domestic and/or wild goats.

Allele-sharing distances between individuals were calculated using Plink or Mega (Tamura et al., 2011), and visualized in Neighbor-Joining trees by using the programs Splitstree (Huson & Bryant, 2006) and Mega.

For construction of a Median-Joining networks (Bandelt et al., 1999), we selected 286 male goats and 27 bezoars, omitting transboundary breeds outside their region of origin and balancing the breed representation by analyzing [?]18 individuals per breed. With 1734 polymorphic SNPs, the program Popart (Leigh & Bryant, 2015) generated a network of 91 haplotypes (Table S4).

Haplogroups distribution in 180 goat breeds

We differentiated the major Y-chromosomal haplogroups Y1AA, Y1AB, Y1B, Y2A and Y2B for 2223 domestic goats from 180 breeds by combining data from several sources as detailed per breed in Table S5:

1. From the goat panel collected for the Econogene project, DNA samples of 353 male goats from 38 European or southwestern Asian breeds were analyzed by PCR amplification and dideoxy-sequencing of *DDX3Y*, *SRY* and *ZFY* segments (Lenstra & Econogene Consortium, 2005) as described previously for bovine samples (Edwards et al., 2011; Nijman et al., 2008) and using the primers listed in Table S6.
2. We used published data from five Portuguese breeds or from Moroccan goats (Pereira et al., 2009), 12 Asian breeds or national populations (Waki et al., 2015, Tabata 2018, 2019, combined with unpublished data), for eight Turkish breeds (Cinar Kul et al., 2015) and for 26 Spanish and Swiss breeds (Vidal et al., 2017).
3. Genotypes of 85 breeds in the VarGoats dataset were derived from their position in the phylogenetic trees (Fig. S1) and/or the alleles of the diagnostic SNPs (Table S1) without any conflicting assignments.

4. 368 DNA samples from several sources, including the AdaptMap panel (Colli et al., 2018) were genotyped by the KASP assay (Kompetitive allele specific PCR assay), at the Van Haeringen Laboratory (Wageningen, Netherlands) and differentiated Y1 vs Y2 and Y1A vs Y1B.
5. For 31 breeds genotypes were obtained by Blast searching of the Short-Read Archive (SRA) or derived from the original WGS data. SRA data for pools of individuals were only used if this allowed an unambiguous identification of the haplogroup composition.

For samples collected by Vidal *et al.* (2017), a few samples analyzed by Cinar-Kul *et al.* (2015) and samples analyzed by KASP before genomic data became available, Y1AA and Y1AB have been both scored as Y1A haplotype. Several of these samples did not contain Y1A or belonged to breeds for which additional data were available (Table S1). However, for 17 breeds we only have the Y1A frequency. Likewise, Vidal *et al.* (2017) and the KASP assays did not differentiate Y2A and Y2B. Since the VarGoats and Econogene panels with a comprehensive coverage of Europe and Africa did not contain a single Y2B-carrying goat, we assigned Y2 scores in other European and African goats to Y2A.

Haplotypes of ancient DNA samples

Daly *et al.* (2021) assigned ancient DNA samples from southwest Asia and southeast Europe to the domestic Y-chromosomal haplogroups on the basis of their positions in a phylogenetic tree. Cai *et al.* (2020) and Zheng *et al.* (2020) described eight Chinese ancient DNA samples and one medieval sample from the northern Caucasian region. Sample YJL2G (coverage 13.4x) clustered with the Y1AB haplogroup, but the low coverage of the others (from 0.013 to 0.118x) resulted in an insufficient overlap with the 552 SNPs that are variable in domestic goats and did not display female scores or male heterozygote scores (see above). Therefore, we relaxed the latter requirements and allowed SNPs scoring in 1% of the females. After removal of two male samples, each representing a unique haplotype, with a high proportion of heterozygote scores, we excluded SNPs with male heterozygote scores only if this occurred in a panel of 78 individuals representing different haplotypes. This resulted in 5593 SNPs, 1018 of which were also scored in the low-coverage aDNA samples. The combined phylogenetic signals (Table S7) showed for sample GTM6G a high proportion of inconsistent scores, presumably due to contamination, but allowed plausible haplogroup assignment for samples KA1G, SMG11 and YJL2G and a tentative assignment for GB3.

Results

Phylogeny of Y-chromosomal haplogroups

A phylogenetic tree of the wild and domestic goats (Fig. 1A) shows an intermingling of bezoars and the domestic goat. From the other wild goat species, the markhor is the closest relative of the bezoar and the domestic goat.

We found 107 different haplotypes in our panel of 352 domestic goats and 27 bezoars with markhor as outgroup. A phylogenetic tree (Fig. 1B) shows haplogroups that correspond to the haplotypes Y1B, Y2A (Lenstra & Econogene Consortium, 2005) and Y2B (Waki et al., 2015) whereas the Y1A haplotypes are split in haplogroups Y1AA and Y1AB. This is confirmed in a dataset of mainly Asian and Moroccan goats (Fig. S1). All domestic haplotypes differ from the Iranian or Anatolian bezoar haplotypes, which also differ from each other. The bezoar haplotypes are associated with the domestic Y1AA or Y2B clusters (CaY1AA and CaY2B, respectively), are linked to the Y1 or Y2 roots (CaY1 and CaY2) or are outside the domestic cluster (Y0).

Fig. S2 shows subtrees containing all 352 domestic goats. This figure also indicates goats with the previously proposed local haplotypes (Vidal et al., 2017; this study, Table S2): Y1AB2 (this study), Y1B2 and Y1C. Y1B2 is represented by Swiss, French and Dutch goats. Our goat panel does not contain goats with the Y2C diagnostic allele (Cinar Kul et al., 2015).

The phylogenetic relationships of domestic and bezoar haplotypes are confirmed by a Median-Joining Network (MJN, Fig. 2). Both the NJ tree and the MJN network allow a few interesting observations to be made: (i) a close relationship of Y1B sequences from Switzerland and Korean Native goats, suggesting a

recent crossbreeding; (ii) likewise, a close relationship of Y1AA sequences from Central as well as South Asia, from South-African Boer goats and from other Y1AA goats in southern and eastern Africa and iii) a clear divergence of the Malagasy Y2A haplotypes from the African continental haplotypes.

Geographic distribution of haplogroups

Figs. 2 and 3 show the haplogroup distributions in 80 breeds from Europe, Asia and Africa. Remarkably, there is clear difference between the ancient and modern DNA samples. All five haplogroups were detected in ancient goat samples from Southeast Europe and southwest Asia (Fig. 3A). In contrast, we observe a strong geographic differentiation of domestic goats:

1. Haplogroup Y1B is predominant in central and northern Europe, but outside Europe and North Africa it has only been found in one Ugandan Karamonja goat, in the Korean native breed and in exported Saanen populations.
2. In northern and central Europe Y2A is only found in a single French des Fosses sample and together with Y1AA in the crossbred Anglo-Nubian. It is the predominant haplogroup in Spain, Anatolia and Africa south of the Sahara, but it is not found in China and Southeast Asia except in about 20% of the goats in the Philippines and Sulawesi. Remarkably, most Y2A haplotypes on Madagascar are more related to Asian than to continental-African Y2A haplotypes.
3. Y2B is absent in Europe, continental Africa and west Asia, but is a major haplotype in east and southeast Asia. It is also observed in one Malagasy Diana sheep.
4. Y1AA is in Europe only represented by three haplotypes in the local Ciocara breed and the island Montecristo population (Somenzi et al., 2022). The Italian haplotypes are outside the cluster of closely related south-Asian and southeast-African Y1AA haplotypes (Figs. 2, S2).
5. The available data suggest a contrast of Y1AB dominating in northern China and Y1AA with Y2B in the south.

Discussion

One of the benefits of the current availability of WGS datasets is the access to an abundance of sequence variants, which allow a comparison of individuals or populations for several purposes. This is especially useful for the analysis of Y-chromosomal diversity, which used to be restricted by the availability of Y-chromosomal markers. The male-specific part of the Y-chromosome constitutes the longest haplotype in the mammalian genome and may serve as marker for mammalian paternal lineages. Here, we combined the dataset of the VarGoats project with published data and genotyped diagnostic SNPs in male goat samples from several sources.

The Y-chromosomal phylogeny of wild and domestic goats is in agreement with the Y-chromosomal tree on the basis of *AMELY* and *ZFY* gene fragments (Pidancier *et al.* 2006) and with a phylogeny of WGS sequences (Grossen et al., 2020; Cai et al., unpublished). MtDNA trees confirm the close relationship of markhor (*Capra falconeri*) with bezoar and domestic goat, but do not show the separation of these species and the other wild goats. In addition, mitochondrial DNA (mtDNA) sequences of some, but not all East-Caucasian turs (*Capra cylindricornis*) cluster with the mtDNAs of markhor, bezoar and domestic goat, illustrating a separate history of maternal and paternal lineages in cross-fertile species (Chen et al., 2018; Marin et al., 2017; O'Connell et al., 2014; Zhang et al., 2020; Zhang et al., 2016).

On the basis of WGS data, Zheng *et al.* (2020) and Xiao *et al.* (2021) reproduced the divergence of the domestic Y1 and Y2 haplogroup previously found on the basis of SNPs within or near Y-chromosomal genes (Lenstra et al., 2005). Here we report a further differentiation of haplogroups, resulting in a phylogeny supported by a largely independent preliminary dataset (Fig. S1) and two phylogenetic algorithms. We found that the major haplogroups corresponds with haplotypes defined by SNPs (Cinar Kul et al., 2015; Lenstra & Econogene Consortium, 2005; Pereira et al., 2009; Tabata et al., 2018, 2019; Vidal et al., 2017; Waki et al., 2015), but Y1A haplotypes belong to either haplogroup Y1AA or haplogroups Y1AB.

The phylogeny also indicates that these haplogroups diverged after the split of the markhor and the cluster of

the wild bezoar and the domestic goats. The domestic goats and the two bezoar populations from Anatolia and Iran do not share haplotypes, whereas the bezoar haplotypes are attached to deep nodes in the tree of domestic haplotypes. This suggests an absence of male gene flow between the bezoar populations and between the bezoar and domestic goats from the same region. Thus, domestic goats, which possibly were derived from bezoar populations not sampled in this study, maintained their paternal lineages during the migration from the Fertile Crescent via Anatolia to Europe, this in spite of indications of management of wild goats in central Anatolia (Stiner et al., 2022).

Geographic plots of domestic haplogroup frequencies show a considerable spatial differentiation, which resonates with the strong phylogeography displayed by autosomal SNPs (Colli et al., 2018), but is in clear contrast with the weak phylogenetic structure displayed by the major mtDNA haplogroups (Luikart et al., 2001; Naderi et al., 2007, 2008; Zhao et al., 2014b, 2014a, Colli et al., 2015). Remarkably, Y-chromosomal haplotypes from all five haplogroups have been found in ancient DNA samples from Southwest Asia or Southeast Europe. The locations of ancient Y1AA, Y1B and Y2B samples are well outside the range of the corresponding domestic haplogroups (Fig. 3). This indicates that during the Neolithic and later worldwide migrations a series of bottlenecks and expansions in the domestic male lineage created a strong geographic differentiation of the haplogroup distribution (Fig. 4):

1. The dominance of haplogroup Y1B in central and northern Europa may very well reflect population bottlenecks during the Neolithic introduction of agriculture via the Danube route (Cymbron et al., 2005; Rivollat et al., 2015; Tresset & Vigne, 2007)
2. Y2A and Y1AA are almost the only haplogroups in Africa south of the Sahara. The two African Y1AA haplotypes are related to those of Asian, indicating that only Y2A expanded during the first introduction of domesticated goats in central and southern Africa
3. Y2B has been found in two Neolithic Iranian samples whereas related CaYB2 haplotypes are present in Iranian and Anatolian bezoars. However, by population bottlenecks during the global spreading of domestic goats Y2b now only occurs in Asia east of the Indus River and in one goat from Madagascar (see below).
4. Y1AA was found in Neolithic samples in southeast Europe, but now has a low frequency in Europe. In Asia it expanded together with Y2B and later came to South Africa when Asian goats were used to breed the Boer goat (see below).
5. Y1AB is the most frequent haplogroup in north China. The distributions of Y1AB and Y1AA/ Y2B in East Asia correspond to ranges of the north-Chinese cashmere goats and the small Southeast Asian 'katjang' type, respectively (Porter et al., 2016). This obviously reflects the large difference of climate between northern and southern China, which determined a similar distribution of taurine and indicine cattle. These two types of cattle are supposed to have entered China via a northern and southern migration route, respectively (Chen et al., 2018; K. Zhang et al., 2020), which support the separate eastern expansions of the Y1AB and Y1AA/Y2B goats, respectively.

Exceptions to these geographic patterns follow from close relationships between haplotype from different continents, which are most likely explained by later major introgressions. Interestingly, in the phylogenetic trees and networks (Figs. 2 and S2) the Y2A haplotypes on Madagascar are closely related to Asian haplotypes and one Diana goat from northern Madagascar even has an eastern-Asian Y2B haplotype. However, autosomal DNA shows that the Malagasy goats are more related to the southern and eastern African continental goats (Colli et al., 2018; Denoyelle et al., 2021). This parallels a recent finding that Malagasy cattle combines Indian and admixed African zebu ancestry (Magnier et al., 2022). The Malagasy language has an Austronesian origin, which testifies the colonization of Madagascar by immigrants from southeastern Asia about 500 CE. Thus, it is likely that these immigrants brought Austronesian goats, cattle and possibly also other livestock from their region of origin to Madagascar.

Other introgressions are more recent. The exceptional Y1AA and Y2A in the English Anglo-Nubian is explained by the documented import during the 19th century of Indian and African goats to England. These served on the ships as source of milk and meat, but surviving males were crossed with English goats, which

resulted in the emergence of a popular transboundary breed.

The worldwide popular Boer goat also is of mixed origin (Porter et al., 2016; Vidal et al., 2017) and carries exclusively Y1AA haplotypes. This breed is supposed to be a crossbred of local African and Indian goats, possibly mediated by incrossing of Anglo-Nubian males (Porter et al., 2016). The crossbred origin is consistent with the results of Colliet *al.* (2018): a separate phylogenetic position of the Boer relative the other African and Asian goats and the K=3 pattern of model-based clustering showing African and west-Asian ancestry. The Indian ancestry is entirely in agreement with a close clustering of the Boer and Pakistani Y1AA haplotypes (Figs. 1B and 2).

Subsequently, the Boer became itself a source of introgression. The same Y1AA haplotypes are closely related to Y1AA haplotypes in local breeds in Uganda, Malawi, Mozambique and Zimbabwe. In these countries crossbreeding with Boer goats from Africa is popular because of its excellent meat production (Banda et al., 1993; Garrine, 2007; Lu, 2011; Onzima et al., 2018). Therefore, it is most likely that the Y1AA in eastern and southern African goats originates from the Boer.

There were three out-of-range findings of Y1B, in the Ugandan Karamonja, in the Korean native goat and in the indigenous goats kept on the Chongmin Island in Shanghai. Because of the popularity of Swiss dairy goats in both Uganda (NAADS, 2005) and Korea (Kim et al., 2019), crossbreeding again is the most likely explanation. There are no data on European admixture in the Chongmin goats (Gao et al., 2020). Thus, exotic occurrence of Y chromosomal variants are direct and sensitive indicators of admixture events, but are to be complemented with quantitative admixture tests, such as model-based clustering, the f_3 and f_4 test or, ideally, identification of introgressed segments across the genome.

The latter approach may also lead to clues on the phenotypic consequences of the introgression via the identification of the admixed genes (Chen et al., 2018; Lv et al., 2014; Wang et al., 2015; Zheng et al., 2020). A more direct link with the Y-chromosomal variation would be provided if this can be linked to male phenotypic traits, but even in human genetics this has scarcely been investigated (Matsunaga et al., 2021; Yang et al., 2018; X. Zhang et al., 2021). Breeds in which different Y-chromosomal haplogroups occur may allow to study an association of haplogroups with typically male traits such as male fertility and dominance behavior. It would be interesting if Y-chromosomal variants can be related to the climate or other environmental features, because this would imply that the geographic differentiation of the Y-chromosomal variation is driven by regional adaptation.

Most introgressions described in this study contribute to the expansion of popular breeds at the expense of the original local breeds. On the one hand, depending on the extent of the gene flow this may decrease the diversity of the genetic resources; on the other hand, it does not necessarily disrupt the environmental adaptation, arguably one of the most consequential components of the phenotypic repertoire. If properly managed, admixture of productive breeds may also contribute to the sustainable conservation of local populations and illustrates that genetic diversity has never been a static phenomenon.

We conclude that the Y-chromosomal variation of goats reveals bottlenecks, expansions and introgressions, illustrating the power of Y-chromosomal markers for inferring the genetic origin of mammalian populations.

Ethics Approval

Blood samples were collected according to the regulations of veterinarian practice in the country where the sampling was carried out.

Conflict Of Interest

The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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Data Availability

The novel whole-genome sequences from European, Pakistan and African goats analyzed in this study were obtained from the VarGoats project (www.goatgenome.org/vargocats.html) (Denoyelle et al., 2021).

Authors' Contributions

IJN, PAM and JAL designed the study; GT-K and LC coordinated the VarGoats project; IJN and BÇK carried out the ABI sequencing; BDR, PB, TF, ZZ, YJ, YC, ZC, MMi and GS analyzed the WGS data; TC, FPo and CD supplied the bezoar genotypes; KGD, DGB, YC and YJ supplied the aDNA data; HM, FK, SS, MMa, YN, AA, JSM, IAD, SRAB, FMD, TD, MKS, MB and PK provided genotypes for south Asian goats; BDR supplied most of the African samples for KASP genotyping; VAB, DB, BB, TB, SC, VC-C, LD, JG, JH, JK, NKh, NK, AM, RM, JMc, NAOC, FPe, AdS, MS, JS, AS, JT and HZ collected material and/or data for other breeds; JAL and IJN performed the downstream analysis; JAL wrote the first draft; and KGD, AM, FPe, BÇK, JMc, MM, MS, PAM, LC, CD and GTK contributed to the text.

Fig. 1A

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migrations-and-recent-introgressions

Fig. 1B

Fig. 1. Neighbor-joining tree of allele-sharing distances calculated on the basis of 10529 hemizygous male-specific SNPs extracted from the VarGoats WGSs. **A.** Fourteen wild goats and five domestic goats with indicated haplogroup. **B.** 101 different haplotypes in 352 male domestic goats and 27 male bezoars and (not shown) markhor as outgroup. Individual codes consist of **(i)** Country codes, first two letters: AZ, Azerbaijan; BD, Bangladesh; CH, Switzerland; CN, China; DK, Denmark; ET, Ethiopia; FI, Finland; FR, France; IE, Ireland; IL, Israel; IR, Iran; IS, Iceland; IT, Italy; KE, Kenya; KO, Korea; MG, Madagascar; ML, Mali; MO, Morocco; MZ, Mozambique; NL, Netherlands; NZ, New Zealand; PK, Pakistan; RU, Russia; SD, Sudan; TR, Turkey; TJ, Tadjikistan; TZ, Tanzania; UZ, Uzbekistan; ZA, South Africa; ZW, Zimbabwe); **(ii)** Species code, 3rd-4th letter: CA (*Capra aegagrus*), bezoar; CC (*Capra caucasica*, *Capra cylindricornis*), West- and East-Caucasian tur, respectively; CF (*Capra falconeri*), markhor; CH (*Capra hircus*), domestic goat; CI (*Capra ibex*) Alpine ibex; CN (*Capra nubiana*), Nubian ibex; CS (*Capra sibirica*), Siberian ibex. **(iii)** Codes for domestic breeds (three letters), or bezoar populations (four letters): see Figure 3. **(iv)** Sample number. Numbers near the nodes indicate bootstrapping values (%) after carrying out 100 bootstrap replicates. Y0, CaY1, CaY1AA, CaY2 and CaY2B indicate bezoar haplotypes.

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Fig. 2. Median-Joining Network of 286 male domestic goats and 27 bezoars, omitting transboundaries breeds outside their region of origin and balancing the breed representation by reducing the sample size of ALP and SAA to 18 individuals. This panel has 1734 SNPs with MAF > 0.01, and 91 different haplotypes (Table S3)

Fig. 3a

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Fig. 3b

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Fig. 3c

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Fig. 3d

Fig. 3. Haplogroup distributions for 171 breeds (2210 goats). (a) Ancient DNA samples and bezoars; (b) European breeds; (c) Asian breeds; (d) African breeds; Breed or bezoar population codes: ABA, Abaza; ABR, Abergelle; ALG, Algarve; ALP, Alpine; ANB, Anglo-Nubian; AND, Androy; ANK, Angora; APP, Appenzell; ARA, Arabia; ARG, Argentata dell'Etna; ARR, Arran; ARW, Arapawa; BAG, Bagot; BAL, Balearic; BBL, Black Bengal; BCM, British Composite Dairy; BEE, Beetal Black Pakistan; BEY, Bermeya; Bhutan; BIO, Bionda dell'Amadello; BLA, Blanca Andaluza; BLB, Bilbery; BLO, Blobe; BOE, Boer; BRI, Barbari (Bari); BRV, Bravia; BUK, Polish Fawn Colored; BZA1, Algorz bezoar; BAAz, Azerbaijan bezoar; BZFa, Fars bezoar; BZMa, Markazi bezoar; BZTr, Anatolian bezoar; BZZa, Zagros bezoar; CAG, Capra Grigia; CAM, Cambodjan; CAP, Capore; CCG, Ciociara; CDB, Chengdu Brown; CDM, Chaidamu Qinhai; CHQ, Charnequeira; CHV, Cheviot; COR, Corsican; CPC, Changtangi Pashmira Cashmere; CRO, Croatian Spotted; CRP, Carpathian; DAM, Damani; DIA, Diana; DJA, Djallonke; DKL, Danish Landrace; DPG, Dutch Pied Goat; DRZ, Dreznica; DSA, Dutch Saanen; DUA, Du'an; DUK, Dukati; DUL, Dutch Nordic Goat; ESF, Esfahan; ETH, Ethiopian; FIN, Finnish; FLR, Florida; FSS, de Fossés; FUE, Fuenteventura; GAL, Galla; GAR, Garganica; GDR, Guadarrama; GGT, Girgentata; GMO, Grigia Molisana; GRG, Greek; GRS, Grisons Striped; GUA, Guadeloupe Creole; GUE, Guéra; GUR, Gurcu; HAI, Hair (Anatolian Black); HAS, Hasi; HNM, Honamli; IND, Indian; IRA, Iran; IRQ, Iraq; IST, Istrian; JBL, Jintang Black; JPA, Shjiba; JSA, Saanen; KCC, Kacchan; KHA, Khazakhstani; KHU, Khurasani; KIG, Kigezi; KLS, Kilis; KMO, Karamonja; KMR, Kamorr; KNG, Korean Native; KSA, Korean Saanen; LAO, Laos; LBA, Lori-Bakhtiari; ICL, Icelandic; LGL, Longlin; LIQ, Liqenasi; LOR, Lorraine; MAK, Makatia; MAT, Mati; MAU, Maure; MEN, Menabe; MGL, Mongolian; MLG, Malagueña; MLI, Mali; MLT, Maltese; MLW, Malawi; MLY, Malya; MNT, Montecristo; MOR, Moroccan; MSH, Mashona; MTB, Matebele; MUB, Mubende; MUG, Murciano Granadina; MUL, Mulranny; MUZ, Muzhake; MYA, Myanmar; MZ, Mozambique; MZA, M'Zabite; NAC, Nacki; NCG, Norwegian Coastal; NDG, Norwegian Dairy; NDZ, Norduz; NEP, Nepalese; NGD, Nganda; NKA, Naine de Kabylie; ORO, Orobica; PCG, Peacock; PAT, Pateri; PER, Percy Island; PEU, Peulh; PHI, Pilippine; PIZ, Pinzgauer; PRW, Pare-White; PTV, Poitevin; PVC, Provencale; PYR, Pyrenean; PYY, Payoya; QIN, Qin; QHI, Qinhai; RAN, Rangeland; RAO, Henan Raoshan White; RAS, Rasquera; ROV, Rove; RSK, Nigerian Maradi (Red Sokoto); SAA, Saanen; SAR, Sarda; SAV, Savoye; SCA, Shaanbei Cashmere; SDN, Sudan; SEA, Small East African; SEB, Sebei; SER, Serrana; SGB, St Gallen Booted; SHL, Shahel; SHN, Nigerian Sahel; SKO, Skopelos; SNJ, Sanja; SOF, Sofia; SOU, Southwest; SRP, Serpentina; SSG, Steirische Schecken; SSU, South Sulawesi; TAS, Tauernschecken; TED, Teddi; TER, Teramo; THA, Thari; TIB, Tibetan; TNF, Tinerfena; TOG, Toggenburg; TWZ, Thuringian Forest; TZ, Tanzania; VAG, Valais Blackneck; VAG, Valais Blackneck; VAL, Valdostana; VIE, Vietnamese; VRT, Verata; VZC, Verzasca; WAD, West African Dwarf; XJI, Xinjiang; YBL, Youzhou Black Skin; YIM, Shandong Yimeng White; ZAR, Zaraiba.

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Fig. 4. Bottlenecks, migrations and introgressions of domestic goat. Fill block arrows indicate the approximated place where population-bottlenecks that have altered regional haplogroup distributions. Open block arrows indicate subsequent migrations. Dashed arrows indicate later introgressions. This figure does not indicate export of transboundary breeds.

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Supplemental Figures

Fig. S1. Neighbor-joining tree of allele-sharing distances calculated on the basis of 2350 hemizygous male-specific SNPs extracted from WGS of 70 male goats (Table S1) and the markhor sequence as an outgroup.

BDCH-BEE, Beetal Black; CNCH-QHI, Qinghai; CNCH-RAO, Henan Raoshan white; CNCH-YIM, Shandong Yimeng Black; CNCH-TIB, Tibetan; CNCH-XJI, Xinjiang; CNCH-YIM, Chinese Shandong Yimeng White FRCH-ALP, French Alpine; FRCH-PTV, Poitevine; IRCH-IRA, Iran; KRCH-KNG, Korea native; KRCH-KSA, Korean Saanen; MOCH-MOR, Morocco; NLCH-LNR, Dutch Landrace; NLCH-SAAA, Dutch Saanen; PKCH-BEE, Pakistan Beetal Black. SDCH-SDN, Sudan; SNPs with missing values were omitted. Figure near the nodes indicate bootstrapping values (%).

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Fig. S2. Neighbor-joining tree of allele-sharing distances for haplogroups Y1AA and Y1AB (left), Y1B (middle) and Y2A and Y2B (right).

Supplemental Tables (see separate file)

Table S1. Diagnostic SNPs used in this and previous studies. Mutations are relative to the goat reference sequence, which has haplotype Y1AB. Haplotype assignments are consistent with their position on the phylogenetic trees.

Table S2. Selection of Y-chromosomal scaffolds in the ARS1 assembly with the lowest frequency of heterozygote SNP scores in male goats. Scaffolds used as source of SNPs are printed in bold.

Table S3. Individual codes (see Figs. 1 and 3) and SRA accession numbers of WGS data used retrieved from the Sequence Read Archive and used for phylogenetic analysis. Other SRA accession codes are in Table S5.

Table S4. Y-chromosomal haplotypes in domestic goats (A) and bezoars (B) used for Figs. 1b and 2. See Fig. 1 for the individual codes. Horizontal lines separate animals with the same haplotype. Haplotypes for the Neighbor-joining tree in Fig. 1b are based on allele-sharing distances. For the Median-Joining Network in Figure 2 trees, transboundary goats outside their countries have been omitted and [?]¹⁸ goats per breed were considered. Eight haplotypes for Fig. 1b were fused to other haplotypes by the default settings of the program Popart used for the construction of the Median-Joining Network.

Table S5. Haplogroup distributions in breeds or populations, in ancient DNA and in the bezoar. Kasp, (Kompetitive allele-specific PCR assay); ddSeq, dideoxy sequencing. Codes starting with ERX or SRX indicate Short Reads Archive accessions.

Table S6. Primers for amplification and dideoxy sequencing of *DDX3Y*, *SRY* and *ZFY* fragments and genomic coordinates of sequence covered by the sequence reads excluding the most upstream and most downstream primer sequences.

Table S7. Identification of haplogroups for ancient DNA samples. Minor allele frequencies are based on a panel of 354 male goats with call rates >5%.