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ORIGINAL ARTICLE

## Y-chromosomal variation of local goat breeds of Turkey close to the domestication centre

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### Keywords

AMELY; goat; SRY; Y chromosome; ZFY.

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### Summary

Genetic variations in chromosome Y are enabling researchers to identify paternal lineages, which are informative for introgressions and migrations. In this study, the male-specific region markers, sex-determining region-Y (*SRY*), amelogenin (*AMELY*) and zinc finger (*ZFY*) were analysed in seven Turkish native goat breeds, Angora, Kilis, Hair, Honamlı, Norduz, Gürcü and Abaza. A SNP in the *ZFY* gene defined a new haplotype Y2C. All domestic haplogroups originate from *Capra aegagrus*, while the finding of Y1A, Y1B, Y2A and Y2C in 32, 4, 126 and 2 Turkish domestic goats, respectively, appears to indicate a predomestic origin of the major haplotypes. The occurrence of four haplotypes in the Hair goat and, in contrast, a frequency of 96% of Y1A in the Kilis breed illustrate that Y-chromosomal variants have a more breed-dependent distribution than mitochondrial or autosomal DNA. This probably reflects male founder effects, but a role in adaptation cannot be excluded.

### Introduction

The goat is one of the first domesticated ruminants (Mannen *et al.* 2001; Zeder 2008; Larson *et al.* 2014). Three wild species, bezoar (*Capra aegagrus*), markhor (*C. falconeri*) and ibex goats (*C. ibex*), have been considered as ancestors of domestic goats (*C. hircus*) (Harris 1962; Takada *et al.* 1997; Wang *et al.* 2008). Archaeological evidence and mitochondrial DNA (mtDNA) studies now indicate the bezoar from south-eastern Turkey, northern Iraq and north-west Iran as the ancestor population (Luikart *et al.* 2001; Naderi *et al.* 2008; Pereira *et al.* 2009; Cinar Kul & Ertugrul 2011; Zeder 2011). Arbuckle *et al.* (2014) hypothesized that genotype and phenotype of early domestic goat populations were subject to interactions between ecogeographic conditions, cultural preferences, domestic animals and wild populations and changed already before the westward expansion from south-eastern Turkey to Europe.

Modern domestic goats are generally bred as small groups in often marginal rural areas for products such as meat, milk, skin, fibre and horns. Goats are especially important in developing countries ('the poor man's cow', MacHugh & Bradley 2001). Selective breeding is not as common as for other farm animals. Furthermore, the lack of pedigree records and uncontrolled breeding among goat breeds has maintained a high level of variation, but also a phylogeographic differentiation that is higher than for other livestock species (Cañón *et al.* 2006; Groeneveld *et al.* 2010).

Mitochondrial DNA and non-recombining parts of the Y-chromosomal DNA give complementary information on the domestication history of livestock species, migrations, introgression events and genetic relationships among breeds (Boissinot & Boursot 1997; Meadows *et al.* 2006; Pereira *et al.* 2008; Groeneveld *et al.* 2010). Although autosomal DNA sequences harbour a wealth of information on phylogenetic and geographic origin, the non-recombining

mitochondrial and Y-chromosomal haplotypes directly trace the maternal and paternal lineages, respectively, to ancestral species, geographic origin and/or introgressing populations (Petit *et al.* 2002; Edwards *et al.* 2011; Hurler & Jobling 2001; Ferenčaković *et al.* 2012). Pidancier *et al.* (2006) examined amelogenin (*AMELY*) and zinc finger Y-chromosomal (*ZFY*) polymorphisms to describe the evolution of the genus *Capra*. Lenstra *et al.* (2005), Pereira *et al.* (2008, 2009) used SNPs in the sex-determining region-Y (*SRY*) and *ZFY* genes for genotyping European, North African, Turkish and Asian goat breeds.

Microsatellite genotyping and mtDNA variations of five goat breeds in Turkey have recently been reported (Cinar Kul & Ertugrul 2011; Korkmaz Agaoğlu & Ertugrul 2012). However, data on Y-chromosomal variation from the breeds close to the domestication centre are only limited (Lenstra *et al.*, 2005). We have characterized Y chromosome diversity of seven domestic goat breeds by sequencing the Y-chromosomal gene segments investigated by Lenstra *et al.* (2005), Pereira *et al.* (2008) as well as Pidancier *et al.* (2006).

## Materials and methods

### Sample collection and DNA extraction

Whole blood samples were collected from 149 unrelated male goats representing specific phenotypes of seven indigenous Turkish goat breeds: Angora (ANG, *n* = 25); Kilis (KLS, *n* = 25); Hair goat (HG, *n* = 27); Honamlı (HNM, *n* = 24); Norduz (NRD, *n* = 24); Gürcü (GRC, *n* = 12); and Abaza (ABZ, *n* = 12). The samples were handled in accordance with local ethical approvals by the Animal Welfare Act. Ethical Committee of the Ankara University (2009-48-239 and 2011-119-456). DNAs were extracted using the standard phenol–chloroform extraction protocol (Sambrook *et al.* 1989). Additional previous data for the Angora (*n* = 6) and Hair goat (*n* = 9) (Lenstra *et al.* (2005) were combined with our data.

### Polymerase chain reactions

The amplicon sizes, annealing temperatures and primer pairs (Sentromer DNA ltd, Istanbul, Turkey) for amplifying the *AMELY*, *ZFY*, *SRY* 3' untranslated region (*UTR*) and *SRY* promoter (*SRY-ORF*) are listed in the Table S1. PCRs were performed in a 30 µl reaction mixture, containing 200 µM of the four dNTPs, 10 pmol of each primer, 1× PCR buffer, 1.5 IU Taq polymerase (MBI Fermentas, Vilnius, Lithuania) and

100 ng of genomic DNA template. The concentrations of MgCl<sub>2</sub> were shown in Table S1. DMSO (1.5%, v/v) was used as a PCR enhancer to both *SRY* reactions. An initial denaturation step at 94°C for 4 min was followed by 30 cycles of 94°C for 30 s, annealing (Table S1) for 30 s, 72°C for 60 s and a final extension at 72°C for 5 min. Amplicons for the *AMELY* and *AMELX* genes were separated by gel electrophoresis, and the *AMELY* amplicons were purified using the Gene Jet gel extraction kit (MBI Fermentas).

### Sequence analysis

The PCR products were sequenced using a BigDye Terminator v3.1 cycle sequencing kit and an ABI 3730XL automatic sequencer (Applied Biosystems, Foster City, CA, USA). Amplification primers were used for sequencing, and each fragment was sequenced in both directions. *AMELY* exon 5 sequences were aligned to *C. aegagrus* and/or *C. hircus* (Y1, AY082488; Y2, AY082491; C3, AY082492), *C. aegagrus* (C4, AY08248) and *C. falconeri* (AY082491) homologues. *ZFY* sequences were aligned to *C. aegagrus* and/or *C. hircus* (Y1 and C3, AY082496; Y2, AY082500), *C. aegagrus* (C4, AY082495) and *C. falconeri* (AY082499 and AY082498) homologues. *SRY* sequences were aligned to *C. hircus* homologous sequence (D82963).

The variable positions of the genes and haplotypes frequencies within the breeds were calculated by DNASP (Librado & Rozas 2009), MEGA 5.2 (Tamura *et al.* 2011) and ARLEQUIN (Excoffier & Lischer 1997).

## Results and discussion

Combining the sequence information of the *AMELY*, *ZFY* and *SRY* genes sequences (Table 1 and Figure S1) identified the C1 and C2 variants previously found in bezoars and domestic goats (Pidancier *et al.* 2006). These correspond to the Y2 and Y1 haplogroups, respectively, and both split into two haplotypes in our samples (Y1A and Y1B; Y2A and Y2C). In 164 male goats from seven Turkish goat breeds, we found the same haplotypes Y1A, Y1B and Y2 as in European and North African goats (Lenstra *et al.* (2005); Pereira *et al.* 2008), but not the Y1C haplotype observed in one Swiss breed (ECONOGENE consortium, unpublished observations). However, in the non-coding region of *ZFY* gene from one Hair and one Kilis goat, a new mutation (g.453A>C) defined haplotype Y2C (GenBank, KJ767543, Zc3), the original Y2 haplotype now being denoted as Y2A. One SNP in *AMELY* defines another domestic haplotype C3 found in a Romanian

**Table 1** Combined haplotypes for determination of the haplogroups. C3 and C4 are domestic and bezoar haplotypes, respectively and Cf1 and Cf2 are markhor (*C. falconeri*) haplotypes, defined on the basis of ZFY and AMELY sequence (Pidancier *et al.* 2006). The numbering of nucleotide positions is according to the Genbank entries D82963 (SRY, add 108 bp to convert to the numbering of Pereira *et al.* 2008), AY082500 (ZFY) and AY082491 (AMELY), respectively

Haplogroup	AMELY				ZFY						SRY				Found in this study	
	4	6	AGCCCATCCAGCCAC deletion		2	4	5	5	7	TTATATTTTAA	1	2	2	2		
	2	6			3	5	2	6	4	copy number	7	6	8	9		
					6	3	7	2	9		6	0	6	9		
Y1A	C2	C	C	Yes	Zc2	T	A	A	T	T	1	A	T	T	G	+
Y1B		C	C	Yes		T	A	A	T	T	1	A	T	A	G	+
Y1C		C	C	Yes		T	A	A	T	T	1	C	T	A	G	-
Y2A	C1	C	C	No	Zc1	T	A	G	T	T	2	A	A	T	A	+
Y2C		C	C	No	Zc3	T	C	G	T	T	2	A	A	T	A	+
C3		T	C	Yes		T	A	A	T	T	1	?	?	?	?	-
C4		C	A	No		C	A	A	T	T	2	?	?	?	?	-
Cf1		C	C	No		T	A	A	T	C	2	?	?	?	?	-
Cf2		C	C	No		T	A	A	G	C	2	?	?	?	?	-

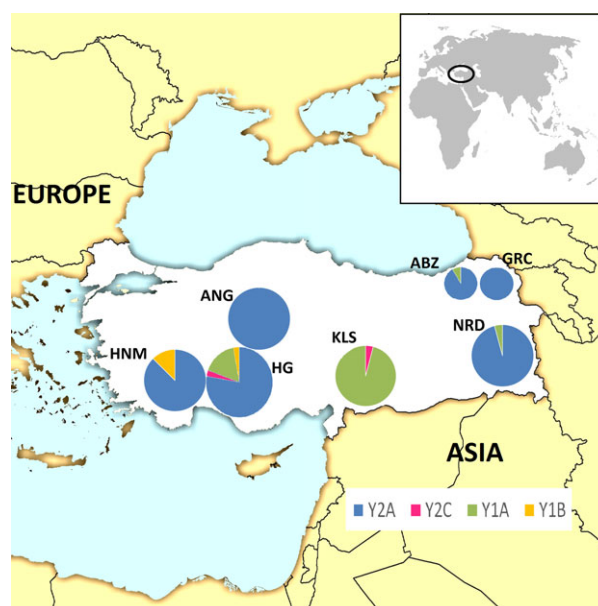
**Table 2** Haplotype distributions among Turkish goat breeds. C1 and C2 are the original haplotype codes (Pidancier *et al.* 2006) and Y1A to Y2C the codes by Pereira *et al.* (2008, 2009) and/or in this paper

Haplotypes	Angora	Norduz	Honamli	Hair	Kilis	Abaza	Gurcu	Total
C1								
Y2A	31 (100%)	23 (95.8%)	21 (87.5%)	28 (77.7%)	-	11 (91.7%)	12 (100%)	126 (76.8%)
Y2C	-	-	-	1 (2.7%)	1 (4.0%)	-	-	2 (1.2%)
C2								
Y1A	-	1 (4.2%)	-	6 (16.7%)	24 (96.00%)	1 (8.32%)	-	32 (19.5%)
Y1B	-	-	3 (12.5%)	1 (2.8%)	-	-	-	4 (2.44%)
Total	31	24	24	36	25	12	12	164

breed (Pidancier *et al.* 2006), which on the basis of its ZFY and AMELY sequences can be assigned to the Y1 haplogroup (Table 1). As all haplotypes found so far in domestic goat are subvariants of the Y1 or Y2 haplogroups, it is plausible that these correspond to paternal lineage originating from the bezoar.

The C4 haplotype previously identified in Anatolian bezoars deviates from both Y1 and Y2 and has so far not been found in domestic goats, indicating that not all paternal lineages of the bezoar have been domesticated. This is consistent with the incomplete domestication of the bezoar maternal lineages (Naderi *et al.* 2008; Cinar Kul & Ertugrul 2011).

The haplotypes Y1A, Y1B, Y2A and Y2C were observed in 32 (19.5%), 4 (2.4%), 126 (76.8%) and 2 (1.2%), goats, respectively (Table 2). This is in agreement with the frequencies of 2/14 and 12/14 for Y1 and Y2, respectively, that are obtained from Turkey by Pidancier *et al.* (2006). This suggests a south-west Asian origin of the major goat Y-chromosomal haplotypes, which presumably traces back to the ancestral bezoar population, but *C. falconeri* ancestry



**Figure 1** The haplogroups distribution of the Turkish goat breeds. Circle sizes are proportional to the sample size.

and/or male-mediated admixtures (from *C. falconeri* and *C. aegagrus*) cannot be excluded. Otherwise haplotype C4 could also be domesticated but lost again due to drift.

The distribution of the haplotypes in Turkish goat breeds is shown in the Figure 1 and Table 2. The most striking observation is the dominance of Y1A without any Y2A in the Kilis breed, contrasting to the dominance of Y2A in other Turkish breeds. However, mtDNA and autosomal markers did not indicate such strong genetic differentiation of Kilis from the other breeds (Cinar Kul & Ertugrul 2011; Korkmaz Agaoglu & Ertugrul 2012).

An obvious explanation of the contrasting Y-chromosomal haplotype distribution is the occurrence of founder effects accompanying small male population sizes. An alternative explanation may be based on a correlation between heat stress and testicular development, which is mainly regulated by Y chromosome genes (Hansen 2009) and might lead to selection of a different Y-chromosomal haplotype in the relatively hot climate in the region of the Kilis breed. However, the uneven distribution in Europe of the Y1 and Y2 haplogroups, with Y2 dominant in the warmer southern regions and Y1 central and northern regions as well as in Italy and in a minority of the Iberian goats (Lenstra *et al.* 2005; Pereira *et al.* 2009), cannot be explained readily by selection of Y1-patrilines for heat resistance. Additional studies will be needed to verify any effect of selection the distribution of the Y-chromosomal haplotypes.

We presented comprehensive data provided on Y-chromosomal polymorphisms close of the domestication centre. Further insight into the origin of goat paternal lineages would require more Y-chromosomal sequence data from domestic, wild and ancient south-west Asian goat samples.

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### Authors contributions

Conceived and designed the experiments: BCK, BA, OE. Performed the experiments: BCK, NB. Analysed

the data: BCK, NB. Contributed reagents/materials/analysis tools: BCK, NB, OKA, BA, OE. Contributed to the writing of the manuscript: BCK, JAL, BA, NB, OKA.

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## Supporting Information

Additional Supporting Information may be found in the online version of this article:

**Figure S1** Alignment of the variable positions of each haplotype, defined on the basis of *ZFY* (A) and *AMELY* (B) sequences.

**Table S1** The primer sequences, amplicon sizes and annealing temperatures used for the amplification of the genes.