

## 208. Y-chromosomal haplogroups from wild and domestic goats reveal ancient migrations and recent introgressions

J.A. Lenstra<sup>1</sup>, VarGoats Consortium<sup>2</sup>, I.J. Nijman<sup>1,3</sup>, B.D. Rosen<sup>4</sup>, P. Bardou<sup>5</sup>, T. Faraut<sup>5</sup>, T. Cumer<sup>6</sup>, K.G. Daly<sup>7</sup>, Z. Zheng<sup>8</sup>, Y. Cai<sup>8</sup>, H. Asadollahpour<sup>8</sup>, B. Çınar, Kul<sup>9</sup>, W.-Y. Zhang<sup>10</sup>, G. E<sup>10</sup>, A. Ayin<sup>11</sup>, M. Bakhtin<sup>12</sup>, V.A. Bâlțeanu<sup>13</sup>, D. Barfield<sup>14</sup>, H. Baird<sup>15</sup>, B. Berger<sup>16</sup>, T. Blichfeldt<sup>17</sup>, G. Boink<sup>18</sup>, S.R.A. Bugiwati<sup>19</sup>, Z. Cai<sup>20</sup>, S. Carolan<sup>21</sup>, E. Clark<sup>22</sup>, V. Cubric-Curik<sup>23</sup>, M.I.A. Dagong<sup>19</sup>, T. Dorji<sup>24</sup>, L. Drew<sup>25</sup>, J. Guo<sup>26</sup>, J. Hallsson<sup>27</sup>, S. Horvat<sup>28</sup>, J. Kantanen<sup>29</sup>, F. Kawaguchi<sup>11</sup>, P. Kazymbet<sup>12</sup>, N. Khayatzadeh<sup>16</sup>, N. Kim<sup>30</sup>, M. Kumar Shah<sup>31</sup>, Y. Liao<sup>32</sup>, A. Martínez<sup>33</sup>, J.S. Masangkay<sup>34</sup>, M. Masaoka<sup>11</sup>, R. Mazza<sup>35</sup>, J. McEwan<sup>15</sup>, M. Milanese<sup>36</sup>, F. Md. Omar<sup>37</sup>, Y. Nomura<sup>11</sup>, N.-A. Ouchene-Khelifi<sup>38</sup>, F. Pereira<sup>39</sup>, G. Sahana<sup>20</sup>, S. Sasazaki<sup>11</sup>, A. Da Silva<sup>40</sup>, M. Simčič<sup>28</sup>, J. Sölkner<sup>16</sup>, A. Sutherland<sup>41</sup>, J. Tigchelaar<sup>18</sup>, H. Zhang<sup>23</sup>, Econogene Consortium<sup>42</sup>, P. Ajmone-Marsan<sup>43,44</sup>, D.G. Bradley<sup>7</sup>, L. Colli<sup>43,45</sup>, C. Drögemüller<sup>46</sup>, C. Lei<sup>8</sup>, H. Mannen<sup>11</sup>, F. Pompanon<sup>6</sup>, G. Tossier-Klopp<sup>5</sup> and Y. Jiang<sup>8</sup>

<sup>1</sup>Utrecht University, the Netherlands; <sup>2</sup><http://www.goatgenome.org/vargoats.html>; <sup>3</sup>Univ. Medical Center Utrecht, Utrecht University, the Netherlands; <sup>4</sup>Agriculture Research Service, Beltsville, MD, USA; <sup>5</sup>GenPhySE, Univ. Toulouse, INRA, INPT, ENVT, Castanet Tolosan, France; <sup>6</sup>Université Grenoble Alpes, Université Savoie Mont Blanc, CNRS, LECA, Grenoble, France; <sup>7</sup>Trinity College Dublin, Dublin, Ireland; <sup>8</sup>College of Animal Science & Technology, Northwest A&F Univ., Yangling, China; <sup>9</sup>Faculty of Veterinary Medicine, Ankara Univ., Turkey; <sup>10</sup>Southwest University, Chongqing, China; <sup>11</sup>Kobe Univ., Kobe Japan; <sup>12</sup>Astana Medical Univ., Astana, Kazakhstan; <sup>13</sup>Inst. of Life Sciences, Univ. Agricultural Sciences and Veterinary Medicine of Cluj-Napoca, Cluj-Napoca, Romania; <sup>14</sup>Pleystole, Queensland, Australia; <sup>15</sup>AgResearch, Invermay Agricultural Centre, Mosgiel, New Zealand; <sup>16</sup>Univ. Natural Resources and Life Sciences Vienna (BOKU); <sup>17</sup>Norwegian Association of Sheep and Goat Breeders, Aas, Norway; <sup>18</sup>Stichting Zeldzame Huisdierrassen, Wageningen, the Netherlands; <sup>19</sup>Hasanuddin Univ., Makassar, Indonesia; <sup>20</sup>Aarhus Univ., Tjele, Denmark; <sup>21</sup>Old Irish Goat Society, Mulranny, Ireland; <sup>22</sup>Univ. Edinburgh, Edinburgh, United Kingdom; <sup>23</sup>Univ. Zagreb, Zagreb, Croatia; <sup>24</sup>International Centre for Integrated Mountain Development, Kathmandu, Nepal; <sup>25</sup>Finch Hatton, Queensland, Australia; <sup>26</sup>Sichuan Agricultural Univ., Chengdu, China; <sup>27</sup>Agricultural Univ. Iceland, Reykjavik, Iceland; <sup>28</sup>Biotechnical Faculty, Univ. Ljubljana, Ljubljana, Slovenia; <sup>29</sup>Natural Resources Inst. Finland (Luke), Helsinki, Finland; <sup>30</sup>Korea Research Institute of Bioscience and Biotechnology (KRIBB), Daejeon, South Korea; <sup>31</sup>National Animal Nutrition Research Centre, Lalitpur, Nepal; <sup>32</sup>Guangxi Key Laboratory of Livestock Genetic Improvement, Guangxi, China; <sup>33</sup>Universidad de Córdoba, Córdoba, Spain; <sup>34</sup>Univ. of the Philippines, Los Banos, Philippines; <sup>35</sup>Laboratorio Genetica e Servizi, Agrotis srl, Cremona, Italy; <sup>36</sup>Univ. degli studi della Tuscia, Viterbo, Italy; <sup>37</sup>Bangladesh Agricultural Univ., Mymensingh, Bangladesh <sup>38</sup>Science Veterinary Inst., University of Blida, Blida, Algeria; <sup>39</sup>IDENTIFICA Genetic Testing Maia & Centre for Functional Ecology, Porto, Portugal; <sup>40</sup>E2LIM, Univ. Limoges, Limoges, France; <sup>41</sup>New Zealand Arapawa Goat Association, New Zealand; <sup>42</sup><https://cordis.europa.eu/project/id/QLK5-CT-2001-02461>; <sup>43</sup>Univ. Cattolica del S. Cuore di Piacenza and BioDNA Biodiversity and Ancient DNA Res. Centre, Piacenza, Italy; <sup>44</sup>UCSC PRONUTRIGEN Nutrigenomics Res. Centre, Piacenza, Italy; <sup>45</sup>UCSC BioDNA Biodiversity and Ancient DNA Res. Centre, Piacenza, Italy; <sup>46</sup>Univ. Bern, Bern, Switzerland; [j.a.lenstra@uu.nl](mailto:j.a.lenstra@uu.nl)

## Abstract

By its paternal transmission, Y-chromosomal haplotypes are sensitive markers of population history and male-mediated introgression. We used whole-genome sequences (WGSs) of 386 domestic goats from 75 modern breeds and 7 wild goat species generated by the VarGoats goat genome project. Phylogenetic analyses indicated five domestic haplogroups Y1AA, Y1AB, Y1B, Y2A and Y2B. Haplogroup distributions for 180 domestic breeds indicate ancient paternal population bottlenecks during the migration into northern Europe, southern Asia and Africa. Sharing of haplogroups reveals male-mediated introgressions: from Asia into Madagascar and, more recently, into the South-African Boer goat; then from this breed into other southeastern African goats; and from Europe into native Korean and Ugandan goats. This study illustrates the power of the Y-chromosomal variation for the reconstructing the history of domestic species with a wide geographic range.

## Introduction

The genetic diversity of goats has been reported to show a weak phylogeography of the mtDNA (Luikart *et al.*, 2001), but a strong geographic partitioning of the autosomes (A Mills *et al.*, 2017; Colli *et al.*, 2018; Zheng *et al.*, 2020). This is in sharp contrast to similar studies of sheep (Kijas *et al.*, 2012; Ciani *et al.*, 2020), but has been confirmed by studies of ancient DNA (Daly *et al.*, 2022). However, the goat Y-chromosome as a marker for the paternal lineages has not yet been studied at a worldwide scale. For many species, Y-chromosomal variants have proved to be highly informative markers for paternal origin, which generally show a much stronger phylogeographic differentiation than mitochondrial or autosomal variants (Jobling and Tyler-Smith, 2017).

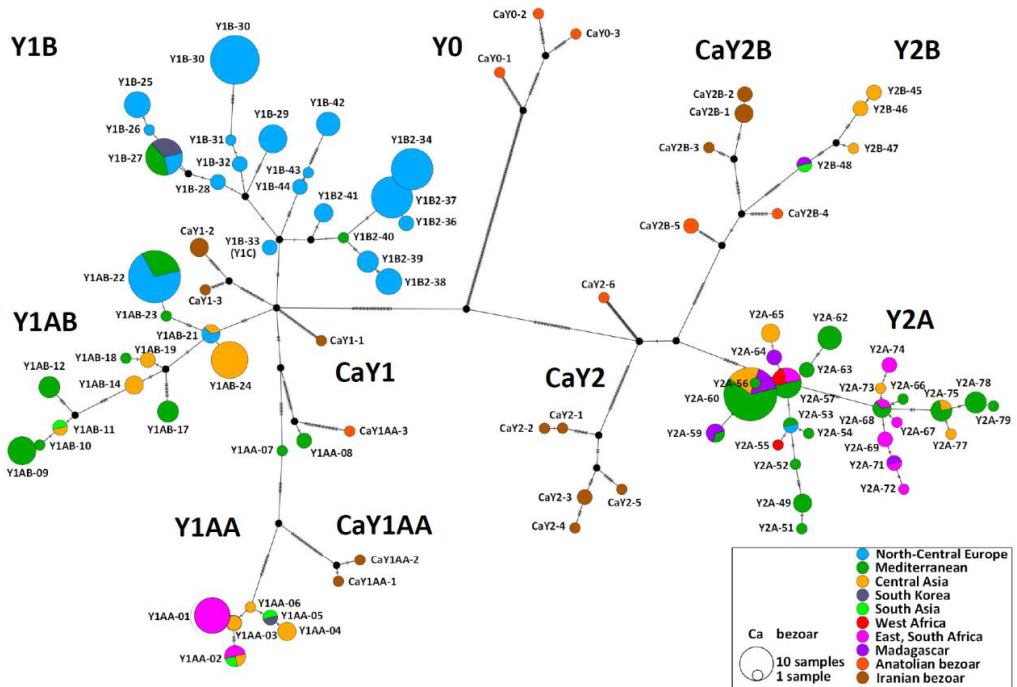
Previous analyses of Y-chromosomal diversity in European and Asian goats on the basis of low number of SNPs defined four haplotypes Y1A, Y1B, Y2A and Y2B, which have a strong geographic differentiation (Lenstra and the Econogene Consortium, 2005; Tabata *et al.*, 2018). Here, we use WGS data from the VarGoats project for a broad sample of European, Asian and African goats in order to define the major haplogroups, their phylogeny and the relationships with haplotypes from Iranian and Anatolian bezoar. After combining with data from other sources, we characterize the phylogeographic patterns across the three continents. We present evidence for population bottlenecks during the ancient migration of domestic goat and for more recent introgressions within and between continents.

## Materials & methods

We extracted SNPs from four contigs in the male-specific part of the Y-chromosome, which after quality control were used for constructing Neighbour-Joining (NJ) trees and a Median-Joining Network of bezoars and domestic goats. We identified Y-chromosomal haplogroups for 2,214 goats on the basis of: (1) the VarGoats WGS data; (2) dideoxy sequencing of DNA segments covering SNPs that are diagnostic for the haplogroups; (3) published data for French, Swiss, Turkish and South-Asian goat breeds; (4) KASP genotyping of diagnostic SNPs; and (5) SNP alleles extracted from WGS data.

## Results

Neighbour-Joining phylogenetic trees (not shown) and a Median-Joining Network (Figure 1) on the basis of the VarGoats data gave completely consistent patterns of clustering and show five domestic haplogroups that correspond to the previously found haplotypes: Y1AA, Y1AB (both representing a distinct cluster of Y1A haplotypes), Y1B, Y2A and Y2B. 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 (CaY1AA) or Y2B (CaY2B) clusters, are linked to the Y1 or Y2 roots (CaY1 and CaY2) or are between the Y1 and Y2 haplogroups (Y0, in the NJ tree close to the root).



**Figure 1.** Median-Joining Network of 286 male domestic goats and 27 bezoars.

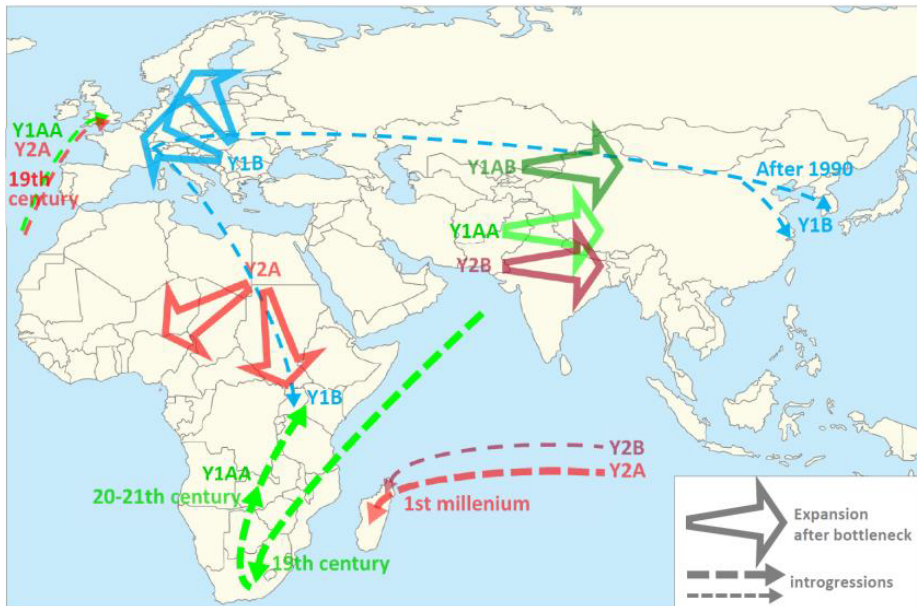
The network further shows a remarkable geographic differentiation:

1. Haplogroup Y1B is predominant in central and northern Europe, but is also found in South Korea and Uganda.
2. Y2A is almost absent in central and northern Europe, but is the predominant haplogroup in Spain, Anatolia and Africa. The British Anglo-Nubian (not shown) carries Y2A and Y1AA because of its Asian and African ancestry. Remarkably, most Y2A haplotypes on Madagascar are more related to Asian than to continental-African Y2A haplotypes.
3. Y2B does not occur in Europe, continental Africa or west Asia, but is a major haplotype in east and southeast Asia. It was also found in a single goat from Madagascar.
4. Y1AA is sporadic in Europe, but is a major lineage in Asia. Two Y1AA haplotypes in East and South Africa are closely related to Asian haplotypes. This reflects the Asian ancestry of the popular Boer breed, which has been crossed into other southeastern African goats.

## Discussion

We propose that the phylogeographic pattern is the result of ancient population bottlenecks and recent introgressions (Figure 2). The Y-chromosomal link between Madagascar and Asia is in line with the genomic composition of the Malagasy zebu (Magnier *et al.*, 2022) and the Austronesian origin of the Malagasy language.

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.



**Figure 2.** Bottlenecks, expansions and migrations inferred from the distribution of Y-chromosomal haplotypes.

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