PRNP polymorphism in goats from Poland

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Background

Scrapie is one of the fatal, prion diseases and belongs to the group of transmissible spongiform encephalopathies (TSEs). The disease affects sheep and goats. There are two scrapie types: classical and atypical (NOR98). They differ in aetiology and genetics. In Poland, there were three scrapie cases in goats. Scrapie susceptibility is connected with the *PRNP* gene. Codons 146 and 222 in the *PRNP* gene are thought to influence the scrapie resistance in goats. The aim of the study was to investigate the *PRNP* polymorphism in goats from Poland. Here we present the preliminary results of the study.

Carpathian goat

https://odrzechowa.com.pl/zakład/kozy/kozy.html

Material and Methods

- 294 samples were collected in 14 different locations (tab. 1).
- DNA isolation (Sherlock AX; *A&A Biotechnology, Poland*);
- Real-time PCR genotyping (Allelic Discrimination) with TaqMan MGB Probes (*ThermoFisher Scientific, USA*), on StepOnePlus Real-time PCR System; the TaqMan assays were designed with ThermoFisher online tools;
- Sequencing (BigDye® Terminator v3.1 Cycle Sequencing Kit; *Applied Biosystems by ThermoFisher Scientific, USA*) and capillary electrophoresis on 3500xl Genetic Analyzer.

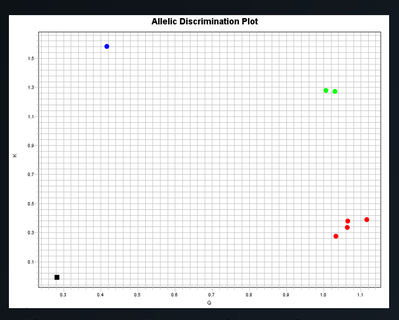


Figure 1. Exemplary plot with results for Q222K TaqMan Genotyping Assay.

Breed	Number of Animals
Carpathian *(native)*	261
Anglo-Nubian	20
Alpine	6
Colorful Polish Noble	3
crossbred	4

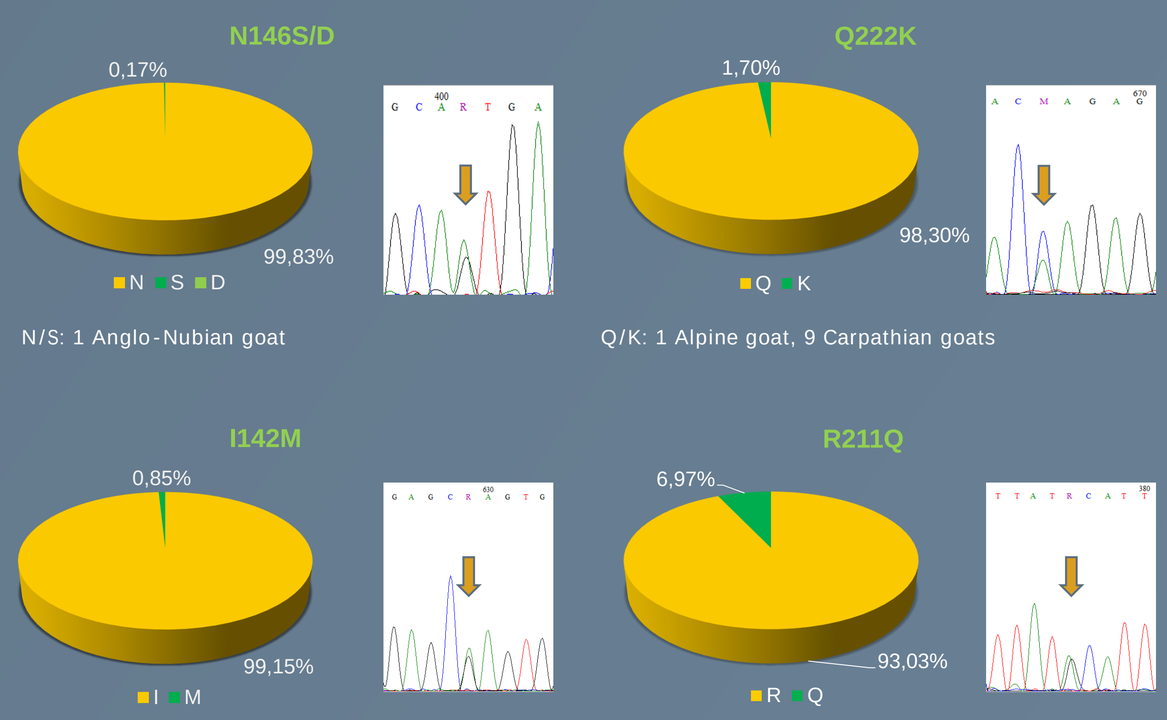
Table 1. Animals used in the study.

Fot. Katarzyna Boguszewska

Conclusions

In the tested goat population, the frequency of *PRNP* alleles considered to be associated with scrapie resistance was very low (S146, K222) and the D146 allele was not found.

Figure 2. The *PRNP* gene allele frequencies [%].



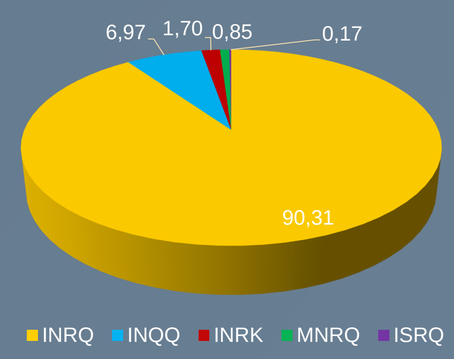
N/S: 1 Anglo-Nubian goat

Q/K: 1 Alpine goat, 9 Carpathian goats

I/M: 1 Alpine goat, 4 Carpathian goats

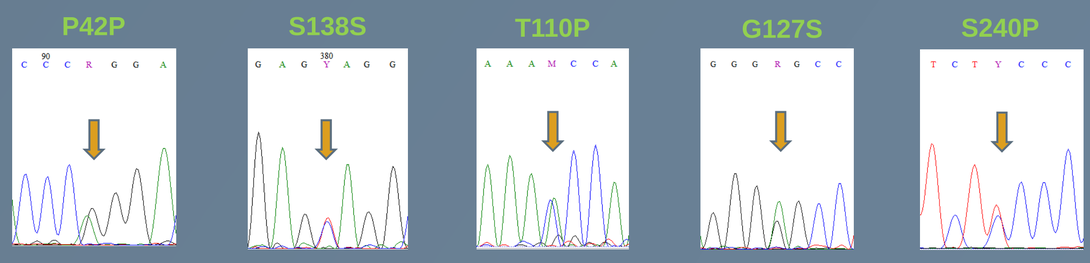
R/Q: 34 Carpathian goats, 1 crossbred
Q/Q: 3 Carpathian goats

Figure 3. The *PRNP* gene haplotypes frequencies [%].



142	146	211	222
I	N	R	Q
I	N	Q	Q
I	N	R	K
M	N	R	Q
I	S	R	Q

Figure 4. Other polymorphic stites found in goat *PRNP* gene.



Results and Discussion

All animals were examined using TaqMan MGB Assays for codons 142, 146, 211 and 222. The TaqMan MGB Assays for codons 146 and 222 were validated with reference samples shared by Barbara Chiappini and used also in two Goat PRNP Genotyping Proficiency tests organized by EURL TSE (Lab passed both tests). Some samples were sequenced (the PRNP coding region) to confirmed the SNP genotyping results for all four codons. The sequencing confirmed results revealed by TaqMan Genotyping.
Several PRNP codon variants (G32stop, G127S, I142M, H143R,N146S/D, R154H, R211Q, and Q222K) were associated with resistance to TSE, however, two codons seems to be the most promising – 146 and 222 [1-4]. In studied population, the allele S146 was present only in one Anglo-Nubian goat and we didn't found any individual with D146 allele. Allele K222 was present in 10 heterozygous samples and Q/K genotype frequency was 3,4%. The frequency of M142 (potentially extend the incubation period) was also low. More frequent was allele Q in codon 211. The frequencies of R/Q (connected to lower susceptibility) and Q/Q genotypes were 11,9% and 1,02%, respectively. The frequencies of S146 and K222 alleles are lower than in Italy [4] and Greece [5] (fig. 2), however we tested smaller group of animals.
The sequencing reveal also other polymorphic sites – silent mutations P42P, S138S and missense mutations T110P, G127S, S240P. However, the sequencing data for the whole group is still being analyzed.

Acknowledgments

We would like to thank Barbara Chiappini from Istituto Superiore di Sanità (Rome, Italy) for sharing the reference samples, which were used to validate our TaqMan MGB Assays.

References

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The study was financed by Ministry of Agriculture and Rural Development - Multiannual Programme no. 03-18-22-09.
20th TSE EURL/NRL Annual Meeting, Rome, Italy, 02-03 October 2023