Mutations linked with the inherited anomalies in the Russian Aberdeen Angus sires

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Abstract. The purpose of research was to estimate of the gene load caused by the harmful mutations linked with the hereditary abnormalities of Aberdeen Angus cattle breed located in Russia. The object was the sires (n=3960) of five Russian populations which have been genotyped on AM, OS, DD and M1 genetic defects by previously created techniques. The genotyping showed the existence of AMC-, OSC-, DDC- and M1C-individuals among the investigated groups. The AMC-bulls have been revealed in four groups with the frequencies of 1.1-4.9%, the DDC- in all investigated livestock at 1.8-10.4%, the OSC- - in the two groups with 0.1 and 1.0% and the M1C- - in one population with 1.9%. We have also tracked the raising of AMC- and DDC-individuals in the later generations of two sire bull populations. In population No. 1 the AMC- and DDC-individuals frequencies have raised on 1.1 and 8.3%, respectively. In population No. 2 these differences were 2.2 and 2.7%. The monitoring of the genetic anomalies distribution in period 2013-2019 has shown the stable percent of AMC, OSC, and M1-bulls, but for the DD genetic defects the growth tendency has been watched (from the absence in 2013 to 10.4% in 2019).

1. Introduction
Today Aberdeen Angus breed is bred in the many farms of Russian because the animals are good adopted in the different geographic zones and appeared the high genetic potential on meat production, reproductive ability, feed conversion, slaughter features and taste and culinary characteristics of meat [1]. But, the serious drawback of the breed is the manifestation of the hereditary abnormalities caused by the gene mutations [2].

The genetic defects problem has been analyzed abroad and in our country. Earlier investigations showed the presence of the individuals carrying in their genomes of the mutant alleles linked with the diseases in the Russian farms, particularly, of the genetic defects of Arthrogriposis multiplex (AM), Osteopetrosis (OS), Developmental duplication (DD) and Muscular hypertrophy (double muscling, M1) [11]. The clinical traits of these disorders have already been discussed and in the table 1 we summarized the data and give the short characteristics of AM, OS, DD and M1 genetic defects.

At the movement in our disposal there are the tests based on DNA analysis and allowing of the early diagnostics conducting of AM, OS, DD and M1 genetic defects [7-10].
The research purpose was to estimate the level of the AM, OS, DD and M1 carriage and to track the dynamics of their distribution among the Russian Aberdeen Angus sires.

2. Material and methods

2.1. Research material
As a research material were the sires of four populations of Aberdeen Angus cattle breed: No. 1 (n=470), No. 2 (n=150), No. 3 (n=300), No. 4 (n=2980) and No. 5 (n=60). The populations No. 1 contained the animals born in 2013 (n=200) and 2017 (n=270), and have been divided into two subpopulations named 1a and 1b, respectively. The populations No. 2 contained the animals born in 2015 (n=60) and 2018 (n=90) and have been divided into two subpopulations named 2a and 2b, respectively. The population No. 3 has been presented by bulls of 2014 year of birth (n=310), No. 4 – in 2018 (n=2980) and No. 5 – by animals of 2019 year of birth (n=60). A visual representation of the research material is given in the table 2.

Table 2. The description of the research material.

| Population | Year of birth | n   | Population | Year of birth | n   |
|------------|---------------|-----|------------|---------------|-----|
| No. 1a     | 2013          | 200 | No. 3      | 2014          | 310 |
| No. 1b     | 2017          | 270 | No. 4      | 2018          | 2980|
| No. 2a     | 2015          | 60  | No. 5      | 2019          | 60  |
| No. 2b     | 2018          | 90  |            |               |     |

2.2. The methods of the investigation
All the investigated livestock has been genotyped on AM, OS, DD and M1 genetic defects by previously created test systems based on allele-specific polymerase chain reaction (AS-PCR) and PCR with the following restriction fragment length polymorphism analysis (PCR-RFLP) methods [7-10]. On the figure 1 the illustration of the test-system working has been present.

The percent of the individuals-mutant alleles carriers has been counted by formula (1):

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\text{Genotype frequency} = \frac{\text{quantity of animal-carrying}}{\text{total population number}} \times 100
\]
Figure 1. The illustration of the AS-PCR and PCR-RFLP methods for AM, OS and DD genetic defects diagnostics. AMC – Arthrogryposis multiplex carrier, DDC – Developmental duplication carrier, DDF – Developmental duplication free, OSF – Osteopetrosis free, OSC – osteopetrosis carrier, M1F – double muscling free, M1C – double muscling carrier, K - negative PCR control, M – molecular weight marker (500 bp).

To analyze the dynamics of the mutations carriage during the years the populations No. 2b and No. 4 have been united into one group because both of them consisted from the bulls of 2018.

3. Results

The frequencies of the individuals carrying in the genotypes of the AM, OS and DD mutant alleles (named of AMC, OSC and DDC, respectively) presented in the table 3. The AMC-, OSC- and DDC-bulls were absent in groups 1a and 2a.

The AMC-bulls have been found in four of five populations in frequencies of 1.1 to 4.9%. Only in population 5 and subpopulations 1a and 2a the same individuals were absent. The DDC-bulls have been found in the all investigated populations in frequencies 1.8-10.4% and the subpopulations 1a and 2a were missing. The OSC-bulls were found only in a little percent in two of five populations: No. 3 (1.0%) and No. 4 (0.1%). The bulls-carriers of nt821del11 mutation (M1C) were revealed in only group No.4 from all investigated livestock at 1.9%.

| Population | 1a | 1b | 2a | 2b | 3 | 4 | 5 |
|------------|----|----|----|----|---|---|---|
| AMC        | 0.0| 1.1| 0.0| 2.2| 4.9| 1.7| 0.0|
| DDC        | 0.0| 8.3| 0.0| 2.7| 1.8| 10.3| 10.4|
| OSC        | 0.0| 0.0| 0.0| 0.0| 1.0| 0.1| 0.0|
| M1C        | 0.0| 0.0| 0.0| 0.0| 0.0| 1.9| 0.0|

The average frequencies of sires-carriers of mutant allele associated with AM, DD, OS and M1 gene anomalies were 1.4, 4.5, 0.2 and 0.3%, respectively.

It is noteworthy that the observed missing of AMC- and DDC bulls in the subpopulations 1a and 2a has changed on the existence of such individuals in 1b and 2b ones. The figure 2 illustrates the raising of AMC- and DDC-frequencies in populations 1 and 2 during four and three years, respectively.
Figure 2. The raising of the AMC- and DDC-bulls frequencies in the later-age Aberdeen Angus herds.

In the later born subpopulations (1b and 2b) we have found the enhancement of AMC- and DDC-bulls. For Arthrogryposis multiplex, the variances in the frequencies of AMC-animals between 1a-1b and 2a-2b consist 1.1 and 2.2%, respectively. For Developmental duplication, these differences were 8.3 and 2.7%, respectively.

The monitoring of the mutations accumulation during the period 2013-2019 has indicated the upward trend of DDC-bulls with the pick in 2019 (10.4% on the figure 3).

The picks of the AMC-sires were in 2014 (4.9%) and in 2018 (3.1%) but the upward trend was not been observed.

The number of OSC-bulls was nonsignificant; such individuals were found only in two populations of 2014 and 2018 in 1.0 and 0.4%, respectively. The growth tendency relating to the defect we haven’t observed.

Figure 3. The dynamics of the distribution of AMC-, DDC, OSC- and M1C-sires distribution among the Russian populations in 2013-2019.
4. Discussion
The obtained data completely confirmed the previous results displayed the existence of the individuals carrying the congenital anomalies among Russian populations of Aberdeen Angus cattle breed [11]. The analysis has shown that the situation relating to the congenital disorders in Russia is not tragically, because in the most bull populations there were only single individuals carrying in their genotypes the mutations linked with AM, OS and M1 genetic defects. However, for the polymelia, the observed upward tendency of the DDC-sires is troubled considering the appearance of the calves with the signs of this anomaly registered in the some herds [confidence personal communication].

It should be noted that nt821del11 mutation linked with M1 genetic defect more typical of the Belgian blue cattle breed and as shown the previous studies was fixed in Belgian Blue cattle resulting of the longtime selection on enlarged muscle mass [12, 13]. In the current investigation, M1C-bulls have been found in only one population (No.4) of 2018 in a little frequency (1.9%).

Among the all investigated livestock, only two subpopulations were free from AM, OS and DD genetic defects (1a and 2a). Notably these subpopulations consisted from the first generation bulls; in the later born individuals of the same farms, the AMC- and DDC-animals have been revealed. These data concord with the previous ones indicating the raising of the frequency of the gene mutations linked with the congenital abnormalities in the populations of Aberdeen Angus and Ayrshire cattle breeds [11].

It’s necessary to understand that the complete elimination of the harmful mutations in part associated with the congenital anomalies is impossible due the features of their inheritance and the fact that crossing of the most productive individuals between each other results of the inbreeding level raising [14]. The sires are the key link in the selection process because the widely using in meat cattle breeding of artificial insemination proposes the use one sire bull for the covering of the several cows that, also, contribute the inbreeding degree raising and buildup of the inherited anomalies in next generations [15].

Due these facts, the maximum that the breeders could be achieved is the maintaining of the stabile low quantity of individuals-carriers of the genetic anomalies to avoid their distribution with the following manifestation of the clinical incidents of the inherited diseases with the respective economic losses [16].

5. Conclusion
It’s widely known that the basic index of meat cattle breeding profitability is the output of calves, optimally consisting 90-100 calves per 100 females. The clinical cases of the genetic anomalies can significantly decrease the figure.

The conducted investigations have shown that the problem of genetic defects should not be forgotten, because the situation can change over time. Seemingly weak percentages of AMC-, OSC-, DDC-and M1C-individuals can become a big challenge for farmers breeding the Aberdeen Angus cattle over time under the lack of the managing.

Therefore, we encourage breeders of both the Aberdeen Angus and other breeds of cattle to use the latest attainments of agricultural science and regularly conduct DNA testing of animals to avoid the appearance and spread of hereditary abnormalities. Finally, such costs will be paid off by the larger output of healthy calves.

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