

(+ REPRODUCTIVE)

THE EFFECT OF MYOSTATIN GENE MUTATION ON PRODUCTIVE TRAITS IN MARCHIGIANA YOUNG BULLS IN PERFORMANCE TEST

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DI SCIENZE AGRARIE,
ALIMENTARI E AMBIENTALI



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AFNAE Dipartimento di Agronomia,
Alimenti, Risorse naturali,
Animali e Ambiente



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Normal



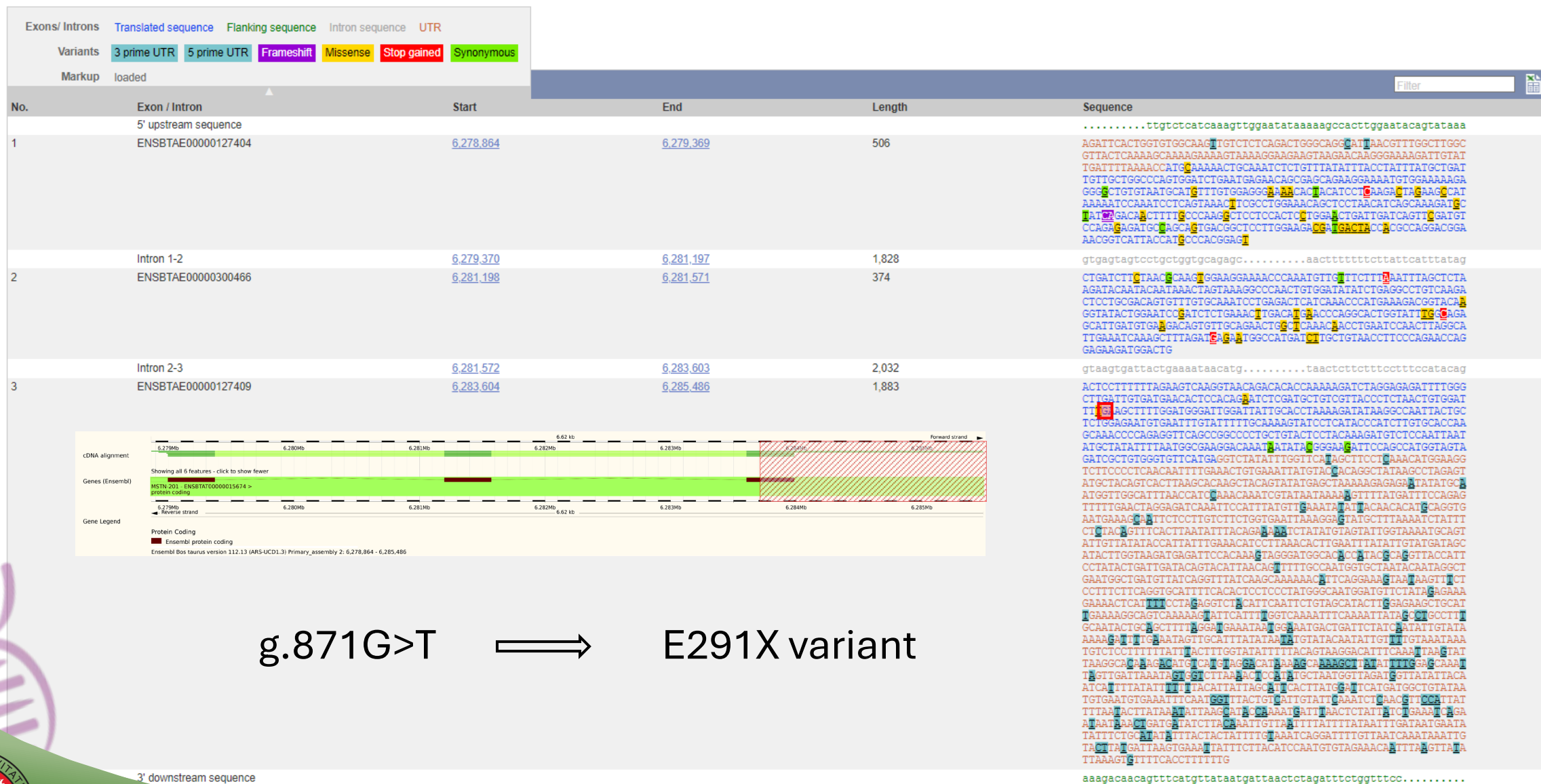
Hypertrophic - heterozygous



Hypertrophic - homozygous



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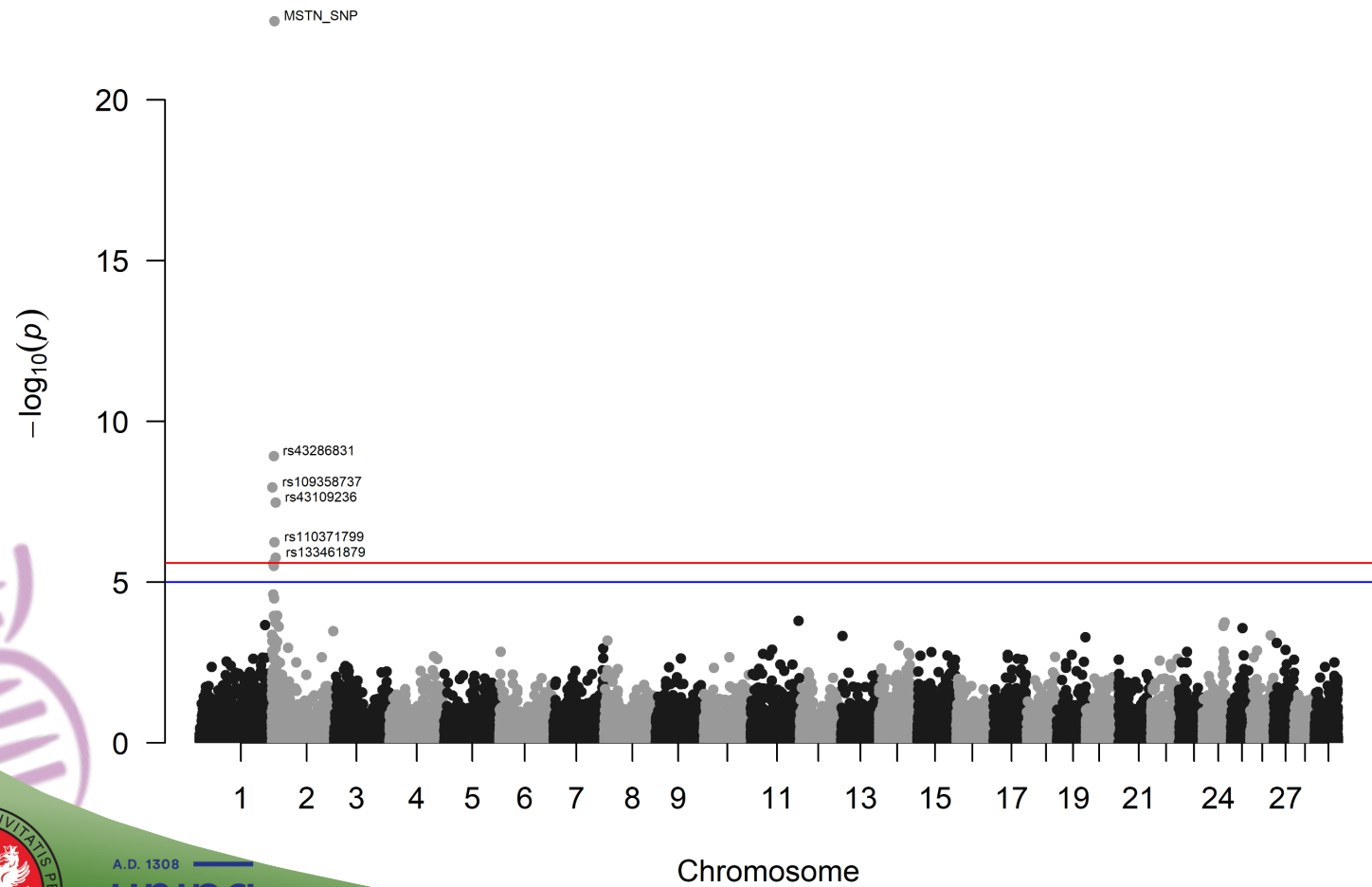
g.871G>T $\Rightarrow$ E291X variant



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Genome-wide association study for muscularity in Marchigiana breed



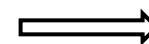
rs	BTA	bp	A1	MAF	β	SE(β)	P-value	Candidate genes
MSTN_SNP	2	6283727	T	0.132	0.9037	0.0887	3.64E-23	MSTN
rs43286831	2	4636218	A	0.296	0.3858	0.0628	1.22E-09	AMMECR1L, SFT2D3, LIMS2, MYO7B, SAP130, UGGT1, HS6ST1
rs109358737	2	1283089	G	0.235	0.4146	0.0720	1.14E-08	TUBGCP5, IMP4, PTPN18, AMER3, ARHGEF4, CYFIP1, NIPA1, NIPA2, HERC2
rs43109236	2	8826383	A	0.227	0.3979	0.0714	3.37E-08	TFPI, CALCRL
rs110371799	2	5909758	G	0.450	0.2904	0.0577	5.73E-07	MFSD6, NAB1, INPP1, NEMP2, HIBCH, C2H2orf88, MSTN
rs133461879	2	8634840	A	0.280	0.3262	0.0678	1.75E-06	TFPI, CALCRL

Colombi et al. 2024. Population structure and identification of genomic regions associated with productive traits in five Italian beef cattle breeds. Scientific Reports. <https://doi.org/10.1038/s41598-024-59269-z>



MSTN mutation in Marchigiana breed

- ✓ Increased muscularity
- ✓ Higher dressing percentage
- ✓ Lower content of fat
- ❖ Homozygous mutated genotype is unwanted
- ❖ The positive or negative effects of the mutation on growth traits are still debated
- ❖ Evidences in mice that *MSTN* deletion leads to impairment of testicular development



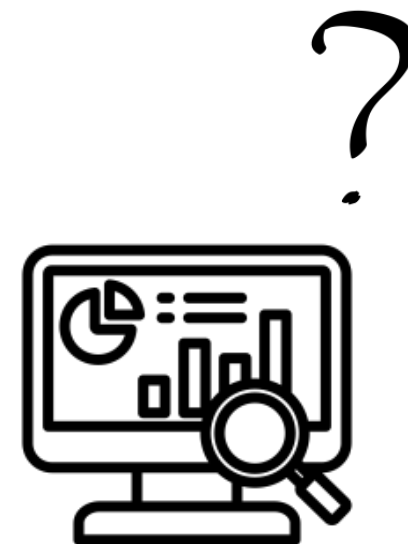
Also heterozygous animals
are often looked upon with
suspicion by breeders



Research questions

- Are Marchigiana bulls carrying this mutation (i.e., heterozygous GT) different from the individuals homozygous for the wild allele in terms of productive or reproductive traits?
- Does it positively or negatively affect animal's performances?

↳ **Is the heterozygous genotype favourable?**



Phenotypic data

Productive traits

1. **Morphological score** (MOR, score)
2. **Bull selection index** (IST)
3. **Average daily gain index** (ADG_IDX)
4. **Muscularity index** (MUS_IDX)
5. **Average daily gain before performance test** (ADG_PRE, kg/d)
6. **Average daily gain during performance test** (ADG_TEST, kg/d)
7. **Muscularity** (MUS, score)
8. **Live weight at one year** (WEI, kg)

Reproductive traits

Testicular morphometric traits

9. **Scrotal circumference** (SC, cm)
10. **Right testicle diameter** (RTD, cm)
11. **Right testicle length** (RTL, cm)

Sperm quality traits

12. **Sperm volume** (SV, ml)
13. **Sperm concentration** (Sconc, mln of spermatozoa/ml)
14. **Abnormal spermatozoa** (ABN, %)
15. **Total motility** (SM, %)

Genotypic data

- RFLP-PCR
- GeneSeek Genomic Profiler Bovine LDv4 33K chip

780 GG
294 GT

458 GG
166 GT (SC)

451 GG
161 GT (RTD, RTL)

223 GG
75 GT



Statistical analysis

- ANOVA

(ADG_PRE, WEI, SC, RTD, RTL)

$$Y_{ijkl} = \mu + \alpha_i + \beta_j + \gamma_k + \delta_l + \varepsilon_{ijkl}$$

μ = mean

α = effect of the genotype i (GG or GT)

β = effect of year of birth j

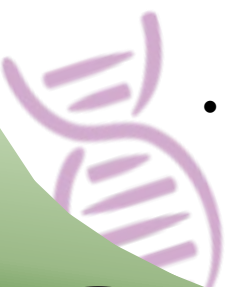
γ = effect of month of birth k

δ = effect of province of birth l (productive traits) or the effect of weeks of life when the testicular morphometry was evaluated (testicular morphometric traits)

ε = random effect.

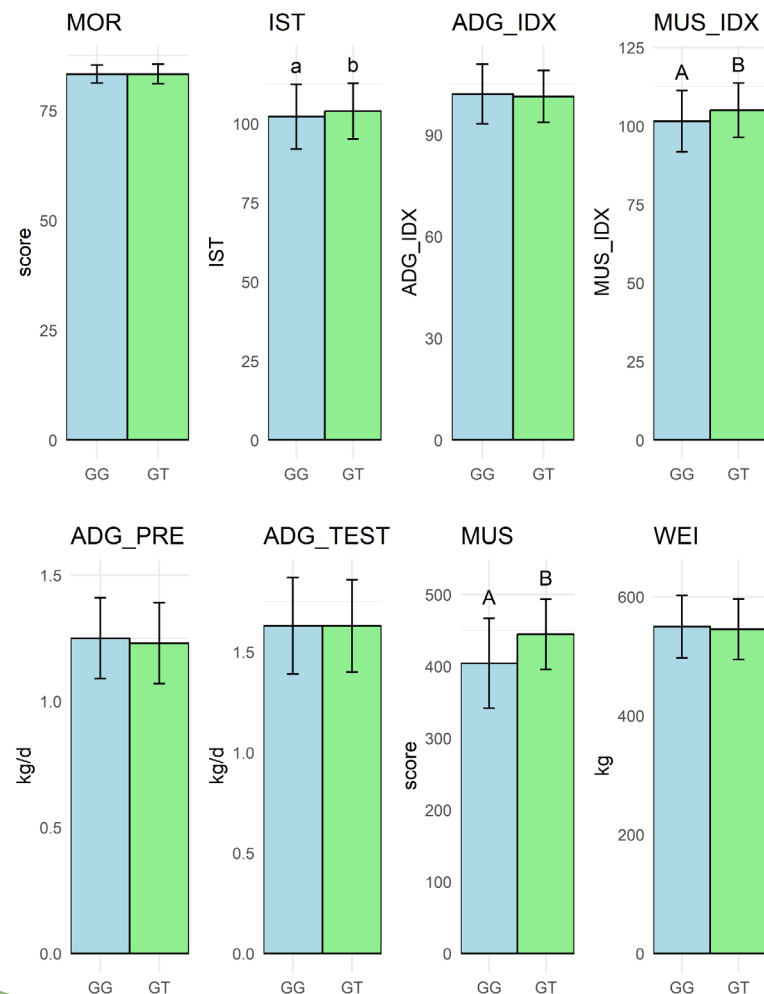
- Mann-Whitney U

- The threshold of significance was set according to the Bonferroni method to 0.0033



Results

Productive traits

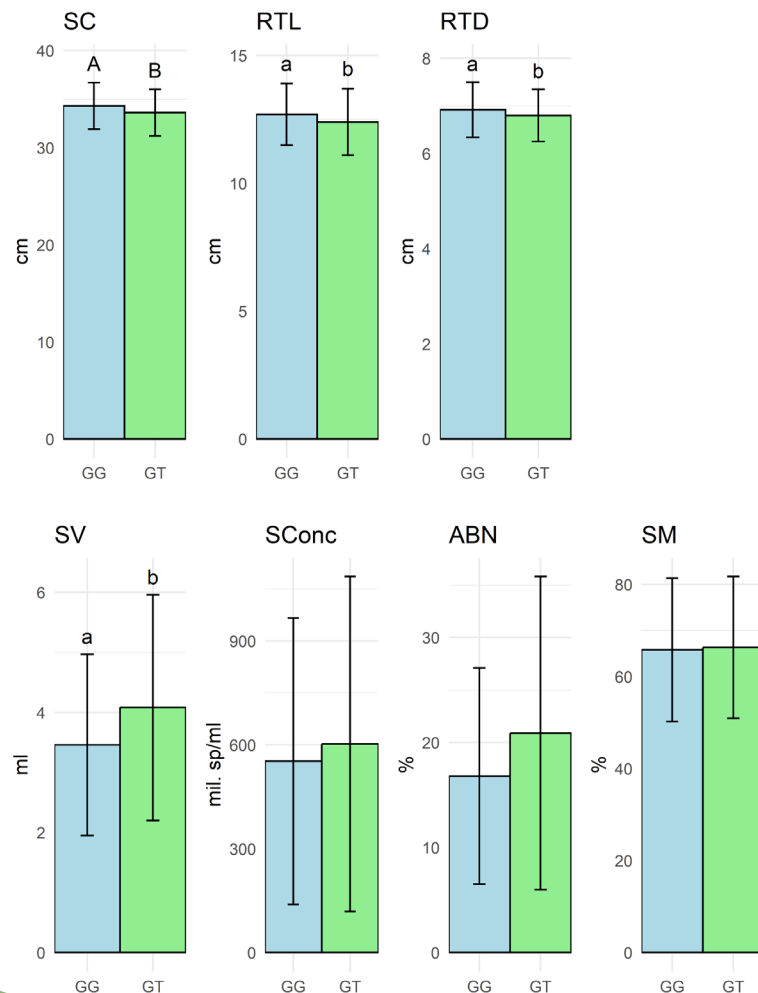


Trait	μ (σ) GG	μ (σ) GT	Nominal P-value
Morphological score (score)	83.31 (2.06)	83.31 (2.25)	0.98
<i>Bull selection index</i>	102.23 (10.23)	103.98 (8.84)	0.0034
Average daily gain index	101.97 (8.80)	101.25 (7.65)	0.42
Muscularity index	101.55 (9.72)	105.05 (8.66)	1.95e-07
Average daily gain before performance test (kg/d)	1.25 (0.16)	1.23 (0.16)	0.19
Average daily gain during performance test (kg/d)	1.63 (0.24)	1.63 (0.23)	0.51
Muscularity (score)	404.35 (62.31)	444.56 (49.04)	5.42e-24
Weight at one year old (kg)	549.41 (52.59)	545.25 (50.87)	0.27



Results

Reproductive traits



Trait	μ (σ) GG	μ (σ) GT	Nominal P-value
<i>Testicular morphometry traits</i>			
Scrotal circumference (cm)	34.3 (2.4)	33.6 (2.4)	9.12e-05
<i>Right testicle length (cm)</i>	12.7 (1.2)	12.4 (1.2)	0.0093
<i>Right testicle diameter (cm)</i>	6.92 (0.58)	6.80 (0.55)	0.0136
<i>Sperm quality traits</i>			
<i>Sperm volume (ml)</i>	3.46 (1.51)	4.08 (1.88)	0.01
<i>Sperm concentration (mil. sp/ml)</i>	552.5 (413.4)	602.2 (483.2)	0.76
<i>Abnormal spermatozoa (%)</i>	16.8 (10.3)	20.9 (14.9)	0.83
<i>Sperm motility (%)</i>	65.8 (15.6)	66.3 (15.4)	0.07



Conclusions

Muscularity related traits are strongly increased by *MSTN* mutation in heterozygous Marchigiana bulls.

Other productive traits and reproductive features were not negatively affected by such genotype in a relevant manner.

Heterozygous Marchigiana bulls can be a valuable resource for genetic selection and improvement with regards to muscularity.



Thank you for your attention



Acknowledgements

This work was supported by the projects: “Italian Biodiversity Environment Efficiency Fitness”—I-BEEF 1 and 2—2014–2020 and 2020–2023. PSRN: Support for the conservation, use and sustainable development of genetic resources in agriculture. Sub-measure 10.2.

