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Estimation of genetic parameters for visual score traits: a simulation study

Estimativa de parâmetros genéticos para características de escore visual: um estudo de simulação

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Abstract: Visual scores are assigned with no basis on an absolute reference. In this case, evaluation references are different for each group, depending only on the structure of the group. Thus, factors that change the structure of evaluation groups, such as group size, can affect visual score assignment, as well as estimation of genetic parameters and prediction of breeding values, respectively. The objective of this study to determine the consequences of assigning scores based on a relative reference on the estimation of genetic parameters and breeding values considering different evaluation group sizes. A simulation study was conducted considering 6 scenarios formed by the combination of two populations (selected and unselected) and 3 group sizes (10, 40, and 100 animals) for evaluation. Genetic parameters and breeding values were estimated using two threshold models that differed in their correction for environmental effects. Heritability estimates for the two populations were close when the model corrected for environmental effects (0.407 – 0.418). However, heritability levels fell over generations when the model did not consider environmental effects (0.172 - 0.247). The size of the evaluation group mainly affected the accuracy of breeding values prediction, since higher accuracy was related to larger evaluation group sizes (0.576 - 0.715). We concluded that the way in which visual scores are assigned does not affect the estimation of genetic parameters and values. However, evaluation groups of more than 40 animals should be used to obtain greater accuracy and genetic progress.

Keywords: accuracy; environmental effects; heritability.

Resumo: Escores visuais são atribuídos sem considerar uma referência absoluta. Nesse caso, as referências de avaliação são diferentes para cada grupo avaliado, dependendo apenas da estrutura do grupo. Assim, fatores que alteram a estrutura dos grupos de avaliação, como o tamanho do grupo, podem afetar a atribuição do escore visual, bem como a estimação e predição de parâmetros genéticos e valores genéticos aditivos, respectivamente. O objetivo deste estudo é determinar as consequências da atribuição de escores com base em uma referência relativa na estimativa de parâmetros genéticos e valores genéticos aditivos, considerando diferentes tamanhos de grupo de avaliação. Foi realizado um estudo de simulação considerando 6 cenários formados pela combinação de duas populações (selecionadas e não selecionadas) e 3 tamanhos de grupos (10, 40 e 100 animais) para avaliação. Parâmetros e valores genéticos foram estimados usando dois modelos de limiar que diferiram em sua correção para efeitos ambientais. As estimativas de herdabilidade para as duas populações foram próximas quando o modelo corrigiu os efeitos ambientais (0,407 – 0,418). No entanto, os níveis de herdabilidade caíram ao longo das gerações quando o modelo não considerou os efeitos ambientais (0,172 - 0,247). O tamanho do grupo de avaliação afetou principalmente a precisão das predições do valor genético, uma vez que maior precisão estava relacionada a tamanhos de grupo de avaliação maiores (0,576 - 0,715). Concluímos que a forma como os escores visuais são atribuídos não afeta a estimativa de parâmetros e valores genéticos. No entanto, grupos de avaliação de mais de 40 animais devem ser utilizados para obter maior precisão e progresso genético.

Palavras-chave: acurácia; efeitos ambientais; herdabilidade.

1. Introduction

Visual score measurements in beef cattle aid in the selection of more productive and environmentally adapted animals as scores are genetically correlated with economically important traits, especially growth (1-6). These are obtained through visual evaluation by assigning scores to different levels of phenotypic expressions. Scores are usually assigned in relation to a group, using the group's mean phenotype as a reference.

Given the process used to assign scores, two aspects must be considered when estimating the genetic parameters and breeding values. The first is the categorical nature of these phenotypes (split into five or six levels). Many studies consider these phenotypes to be continuous and use linear models for genetic analyses (3, 7-11). However, the use of a linear model can affect the classification of animals for selection, particularly when heritability and the number of categorical scale levels are low (12,13). Faria et al. (14) compared estimates of genetic parameters using linear and threshold models for visual scores but found no significant difference in parameter estimates. However, the threshold model has been recommended because of its statistical properties, which guarantee accurate estimations. The second aspect that differentiates visual scores from other traits is the way they are assigned, with no basis for an absolute reference. In this case, the evaluation references are different for each group, depending only on the structure of the group (animals in the group), which does not allow for phenotypic comparison of animals belonging to different groups (7). Factors that change the structure of the evaluation groups, such as group size (number of animals), can affect the visual score assignment.

Because of the particularities involved in this process, this study aimed to determine the consequences of assigning scores based on a relative reference for the estimation of genetic parameters and breeding values through a simulation study that considered selected and unselected populations and three evaluation group sizes (10, 40, and 100 animals).

2. Materials and methods

A simulation study was conducted using three evaluation group sizes (10, 40, and 100 animals) in the selected and unselected populations. QMSim (15) was used to simulate the populations. The simulation was performed in two steps. First, a historical population was simulated in which the balance between the rates of genetic drift and mutation was stabilized. Subsequently, the desired population was constructed. The simulated historical population consisted of 1000 generations: from the last generation, we randomly selected 2000 females and 50 males as the base population, after which the animals were randomly mated in the subsequent generations. The number of sires with progeny in each contemporary group depended on size of the evaluation group. Therefore, in groups with 10 animals the number varied from 7 to 10, in groups with 40 animals it varied from 23 to 33, and in groups with 100 animals it varied from 38 to 48 sires.

The simulated genome was made up of 29 pairs of autosomal chromosomes simulating the bovine genome, ranging from 40 to 145 cM in length with a total length of 2333 cM (16). uantitative trait loci (QTLs, 500), randomly distributed throughout the genome, were used to control this trait. The QTLs were biallelic and triallelic and their effects were sampled from a normal distribution. All additive genetic variances were assigned to the QTL effects.

Given the categorical nature of the scores, it was assumed that an animal's observable phenotype results from an underlying Eq (1) unobservable trait with a normal distribution N ~ (0, 1), known as a "liability" (17). Therefore, the observable categorical response results from an animal exceeding the threshold of a specific point for the underlying trait.

The model used to simulate the underlying trait was:

Equation (1)

$$y = \mu + gc + a + e$$

where y is the liability; μ is the mean population, which was equal to zero; gc is the fixed effect of a contemporary group, formed by sex and year of birth, which was simulated from a normal distribution with zero mean and variance 0.2, equivalent to 20% of the total liability variance; a is the additive genetic effect of the animal, calculated as the sum of the allelic effects of each animal's QTLs; and e is the residual, simulated from a normal distribution with zero mean and a variance 0.6. The heritability of the underlying traits was 0.4.

As the population's liability followed a normal distribution, the same was expected to occur in the evaluation groups. Thus, the underlying traits were categorized into five levels within each evaluation group, simulating the methodology used by the evaluators (7). Thresholds within the group were defined based on the normal distribution of the liability. To keep the distribution of scores within the group similar to those found in actual data, the points were set as -1.3, -0.5, 0.5, and 1.3 standard deviations (13) (Figure 1).

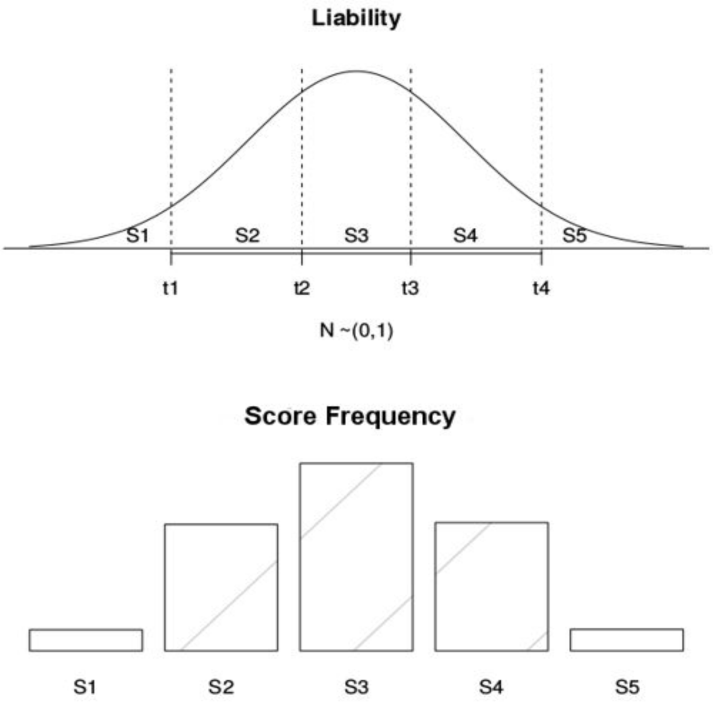

Figure 1. Representation of the visual score and the underlying trait within the group. On top, the liability with the four thresholds that define the five phenotypes. Below, the frequency observed for each phenotypic category.

Animals in the simulated populations were mated 12 times. The female/male ratio was kept constant (2000 females, 50 males). The replacement rates per mating cycle for females and males were 15% and 20%, respectively. The culling criteria were age for unselected populations and age and estimated breeding values (EBVs) for selected populations, with the oldest and worst animals discarded. At each mating cycle, genetic parameters and breeding values were estimated using a threshold model (Eq (2)) that modeled the characteristics underlying the observable phenotype. The general statistical model can be described as follows:

Equation (2)

$$y = X\beta + Zu + e$$

where y is the vector of liability phenotypic (vector of the unobservable underlying scale in the threshold model); β represents the systematics environmental effects; X is the incidence matrix that associates β with y; u is the vector of random direct additive genetic effects; Z is the incidence matrix that associates u with y; and e is the vector of random residual effects. The threshold model considers an underlying scale with continuous and normal distribution.

$$U \vee \theta N(W\theta, I\sigma_e^2)$$

where U is the vector of the underlying scale of order r; θ = (β′, u′) is the vector of location parameters of order s, with β (as systematic effects) and u (as direct additive genetic effects); W is the incidence matrix of order r by s; I is the identity matrix of order r by s; and σ_e^2 is the residual variance. Categorical traits are determined by continuous variables not observable on the underlying scale, where baseline thresholds are set as follows: t1<t2... < tj-1, with t0=-∞ and tj=∞, where j is the number of categories.

Two models were used for each scenario, differing only in that one included the effects of the contemporary group. The first model considered the contemporary group as a fixed effect, while the second model did not include this effect, assuming that the assignment of visual scores within a group corrects for the environmental effects used to group animals for evaluation (18). Table 1 shows the combinations of simulated populations and evaluation models used.

Table 1. Combination between scenarios and models used to estimate genetic parameters and breeding values.

Selected				Unselected			
Name	**GS**	**Model**	**Replications**	**Name**	**GS**	**Model**	**Replications**
Sel10_M1	10	M1	10	Uns10_M1	10	M1	10
Sel10_M2	10	M2	10	Uns10_M2	10	M2	10
Sel40_M1	40	M1	10	Uns40_M1	40	M1	10
Sel40_M2	40	M2	10	Uns40_M2	40	M2	10
Sel100_M1	100	M1	10	Uns100_M1	100	M1	10
Sel100_M2	100	M2	10	Uns100_M2	100	M2	10

GS is the size of the evaluation group, and M1 and M2 are models with and without the contemporary group as fixed effects, respectively.

Variance components and breeding values were estimated by Gibbs sampling using THRGIBBS1F90 (19). A uniform prior distribution was assumed for the systematic effects, and an inverse chi-squared distribution for the variance components. The posterior distribution of the variance and breeding value components was built from a chain of 100,000 samples, of which the first 20,000 were treated as burn-in. The chain size definition was based on the convergence analysis of the Geweke (1992) (20) and Heidelberger e Welch (1983) (21) tests, implemented in the Coda package (22) of the R software (23). The variance components and breeding values were estimated using THRGIBBS1F90, which implements the threshold model under Bayesian estimation. The residual variance was fixed at 1; the thresholds t1 and t2 were fixed at 0 and 1, respectively; and the rest were estimated from the data.

To determine the effects of group size and selection on the estimation of genetic parameters and breeding values for visual score traits, the following criteria were used:

1. Mean Square Error (MSE) (3) was used as an indicator of quality to estimate heritability. The MSE was calculated as described by Casella and Berger (2002) (24):

Equation (3)

$$\frac{1}{n}\sum_{i=1}^{n}\left(\hat{h}_i^2 - h^2\right)^2$$

where n is the number of replications for each simulated scenario, in this case, 10; $\hat{h}_i^2$ is the estimated heritability for each replication; and h^2 is the true heritability value.

2. Pearson's correlations between true breeding values (TBVs) and estimated breeding values (EBVs), as indicators of prediction accuracy (17).

3. Percentage of coincidence (only in the selected population scenarios) between the 10 selected males and 10, 20, and 30 male candidates with the largest TBVs.

4. The selection efficiency (only in selected population scenarios) was defined as the percentage of true and estimated genetic gains relative to the maximum possible genetic gain. The genetic gain per year was calculated by regressing the true and estimated breeding values over time. It was assumed that the mating cycle was equivalent to one year. The maximum genetic gain (Eq (4)) is obtained when the accuracy is 1. It was calculated as:

Equation (4)

$$\frac{max\Delta G}{year} = \left[\left(i_m + i_f\right) * r_{\hat{a}\mathrm{a}} + \sigma_{\mathrm{a}}\right] / \left(IG_m + IG_f\right)$$

where ΔG/year is the genetic gain per year; i_m is the intensity of male selection; i_f is the intensity of female selection; $r_{\hat{a}\mathrm{a}}$ is the accuracy; σ_{a} is the genetic standard deviation; IG_m is the generation interval of males; and IG_f is the generation interval of females. Thus, the maximum genetic gain (Eq (5)) expected in the populations under selection was:

Equation (5)

$$\frac{max\Delta G}{year} = \frac{[(2.66 + 1.15) * 1 * 0.64]}{4 + 4.5} = 0.286 \text{ unidades da liability}$$

3. Results and discussion

The average heritability over the mating cycles for the simulated scenarios is shown in Figure 2. The inclusion of the contemporary group as a fixed effect in the model improved the heritability estimates in both populations, particularly in the selected population, where the inclusion of the fixed effect corrected for the selection process, as considering groups of animals (contemporary group) with different genetic merits is equivalent to including a linear function that describes the selection process in the model (25). Heritability estimates decreased rapidly because of not correcting for the selection effect, as observed in the selected population models that did not consider the fixed effects of the contemporary group.

In the unselected population scenarios, the differences between the evaluation models were smaller. In the model that did not consider the contemporary group as a fixed effect, the heritability estimates approached the true values as the information used in the estimation increased, particularly when the size of the evaluation group was large. This behavior occurred because when assigning visual scores within a group, they depended on the genetic and residual variance in the group, while the variability caused by environmental effects (contemporary group) was not taken into account as evaluation was performed within a group and the animals within the evaluation group were under the same environmental effects. Therefore, we can infer that the score assigned to each animal was corrected for environmental effects used in the formation of the evaluation groups. From this perspective, animals with the same score belonging to different evaluation groups would be comparable as the score reflects the animals' performance in relation to others in the same environmental conditions.

The size of the evaluation group influenced the heritability estimate, especially in the model that did not consider the effects of the contemporary group (Figure 2). These differences are a consequence of the heterogeneity of the phenotypic variance that exists between contemporary groups. Therefore, when the evaluation group is smaller, the heterogeneity is greater (Figure 3). Another point that contributes to heterogeneity is less genetic representation in small groups, as they have fewer sires with progeny. One of the assumptions of BLUP in genetic evaluation is the homogeneity of variance between the levels of fixed and random effects. When this is not the case, parameters, and to a greater extent, breeding values, are affected (26). Including the contemporary group as a fixed effect reduced the differences in the heritability estimates between the different evaluation group sizes, indicating that the inclusion of this effect corrects the bias in the estimation of variance components when there is variance heterogeneity.

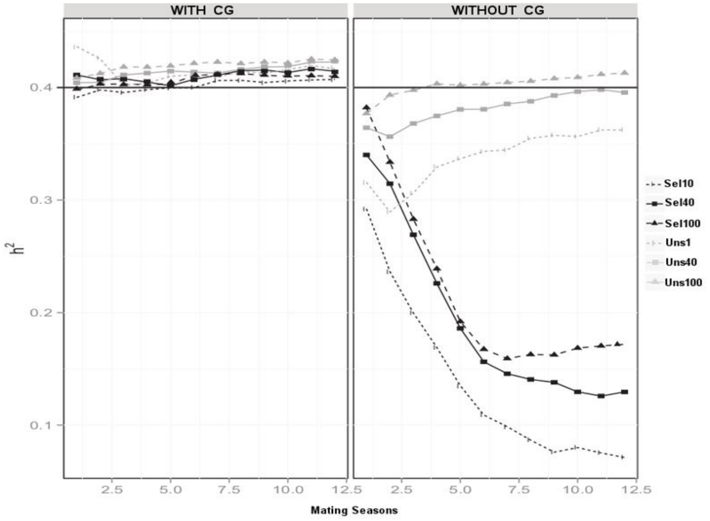

Figure 2. Heritability estimates the simulated scenario throughout mating seasons using two models in the estimation of genetic parameters (including the contemporary group on the left and not including the contemporary group on the right). Sel and Uns indicate selected and unselected populations, respectively; 10, 40, and 100 represent the size of the evaluation groups.

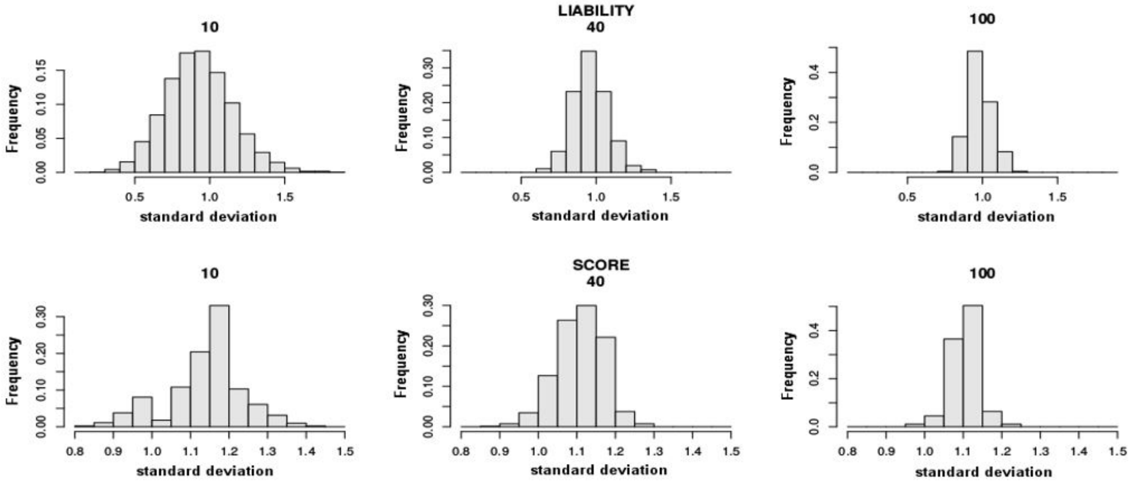

Figure 3. Distribution of the phenotypic standard deviations of contemporary groups for the different evaluation group sizes (10, 40, and 100 animals) in the observable and underlying phenotype. Data were taken from a replication.

Table 2 shows the mean heritability and MSE values for each scenario. It was important to include the contemporary group as a fixed effect in the model in populations under selection and/or heterogeneity of variances as the heritability estimates are closer to the true value and the variability of the estimates decreases (less MSE). In the absence of selection and heterogeneity (scenarios SS100_M1 and SS100_M2, Table 2), the differences between the models were minimal.

Table 2. Mean of the ten replications of each scenario for the true heritability (h^2), estimated heritability ($\hat{h}^2$), and mean square error (MSE).

Selected				Unselected			
Scenario *	**h^2**	**$\hat{h}^2$**	**MSE**	**Scenario ***	**h^2**	**$\hat{h}^2$**	**MSE**
Sel10_M1	0.397	0.172	0.0075	Uns10_M1	0.408	0.359	± 0.0005
Sel10_M2	0.390	0.407	0.0004	Uns10_M2	0.402	0.418	0.0008
Sel40_M1	0.387	0.224	0.0071	Uns40_M1	0.404	0.400	+0.0002
Sel40_M2	0.389	0.418	0.0003	Uns40_M2	0.397	0.418	0.0003
Sel100_M1	0.379	0.247	0.0029	Uns100_M1	0.399	0.418	0.0003
Sel100_M2	0.383	0.416	0.0004	Uns100_M2	0.399	0.421	0.0002

** Sel and Uns are the selected and unselected populations, respectively; 10, 40, and 100 are the evaluation group sizes; and M1 and M2 are models without and with the contemporary group as a fixed effect, respectively.*

Table 3 presents the correlations between EBVs and TBVs for each simulated scenario. Including the contemporary group effect in the model increased the accuracy of the breeding values in both selected and unselected populations. The SS100 scenario (with unselected populations and a group size of 100) presented the smallest difference between the scenarios, in agreement with the results obtained at the estimation of the variance components. Group size influenced accuracy because, as previously indicated, small groups have a greater heterogeneity of phenotypic variances, which leads to a loss of accuracy and increased bias in the prediction of breeding values.

Table 3. Mean of the tem replications of each scenario for the correlations between the true and estimated breeding values ($r_{\hat{a}a}$), and the standard error (SE).

Selected			Unselected		
Scenario *	**$r_{\hat{a}a}$**	**SE**	**Scenario ***	**$r_{\hat{a}a}$**	**SE**
Sel10_M1	0.576	0.0014	Uns10_M1	0.612	0.0007
Sel10_M2	0.652	0.0018	Uns10_M2	0.656	0.0012
Sel40_M1	0.631	0.0011	Uns40_M1	0.676	0.0008
Sel40_M2	0.688	0.0014	Uns40_M2	0.694	0.0010
Sel100_M1	0.651	0.0011	Uns100_M1	0.702	0.0009
Sel100_M2	0.700	0.0011	Uns100_M2	0.715	0.0008

** Sel and Uns are the selected and unselected populations, respectively; 10, 40, and 100 are the evaluation group sizes; and M1 and M2 are models without and with the contemporary group as a fixed effect, respectively.*

Two criteria were used to assess selection efficiency in the scenarios under selection: the percentage of coincidence between the best male candidates for selection and those selected, and the percentage of genetic gain obtained in relation to the maximum possible gain (Table 4). For the first criterion, the coincidence percentage is generally low. As expected, this increased when evaluated with a larger number of better males. When the size of the evaluation group increased, the coincidence increased, indicating that the breeding values were estimated with greater accuracy, as shown in Table 3. Again, there were differences between the models, and the model that considered the fixed effects of the contemporary group had higher percentages of coincidence as it corrected for the effect of selection.

Table 4. Means of the ten replications of the selected population scenarios for the percentage of coincidence between the top male candidates for selection with those selected in each mating cycle, and for the efficiency of the selection measured in the true and estimated genetic gain expressed as a percentage of the maximum genetic gain.

Scenario	Coincidence % - Top Males			% Genetic gain	
	10	20	30	True ΔG	Estimated ΔG
Sel10_M1	10.73	18.00	23.27	51.39	3.50
Sel10_M2	16.64	27.82	34.36	61.99	63.59
Sel40_M1	14.18	21.64	29.91	58.16	5.38
Sel40_M2	21.64	32.00	39.82	66.34	71.38
Sel100_M1	17.73	28.55	36.55	65.08	6.76
Sel100_M2	21.45	33.27	43.27	70.33	78.08

Although the percentage of coincidence of the selected males was low, the percentage of true genetic gain (Table 4) was high, indicating that the chosen males had a high breeding value, even though they were not the best. The estimated genetic gain was close to the true gain when the model considered the contemporary group as a fixed effect, whereas the model without the fixed effect underestimated the genetic gain in relation to the true gain because of the lower estimated genetic variance that occurs if the effect of the selection is not corrected (Figure 2). The evaluation group size influenced the response to selection, which increased with the use of larger evaluation group sizes. This was expected because these scenarios had a higher accuracy.

The results showed that the assignment of visual scores with reference to the mean profile of the assessed animals corrected for environmental effects. However, the fixed effects of the contemporary group should be retained in the models, as this enables them to correct for the effects of selection, leading to more accurate estimations of breeding values and, consequently, greater genetic progress. In real data, the contemporary group effect also corrects for the evaluator effect as the groups are not evaluated by the same evaluator. The size of the evaluation group mainly influenced the accuracy of the breeding value estimates, demonstrating the importance of having groups of 40 or more animals to obtain more accurate breeding values.

4. Conclusion

Parameter estimates and breeding values were not affected by the assignment of scores with an evaluation group as a reference. However, care must be taken when using evaluation groups with fewer than 40 animals as this decreases the accuracy of breeding value predictions.

Conflicts of interest statement

The authors declare that there is no conflict of interest.

Data availability statement

The data will be provided upon request.

Author contributions

Conceptualization: L. O. Diutama, A. T. H. Utsunomiya, M. M. Farah, L. Cavani, and R. Fonseca. Data curation: L. O. Diutama, R. K. Ono, and R. Fonseca. Formal Analysis: L. O. Diutama, D. P. Silva, É. S. Harada, and R. Fonseca. Investigation: L. O. Diutama, L. Cavani, and R. Fonseca. Methodology: L. O. Diutama, A. T. H. Utsunomiya, M. M. Farah, and R. Fonseca. Project management: L. O. Diutama, R. K. Ono, L. Cavani, and R. Fonseca. Supervision: L. O. Diutama and R. Fonseca. Writing (original draft): L. O. Diutama, D. P. Silva, S. Harada, and R. Fonseca. Writing (review and editing): L. O. Diutama, D. P. Silva, É. S. Harada, and R. Fonseca.

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