Expression, determinants and genetic architecture of neck conformation traits in Pura Raza Española horse

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Resumen

Neck conformation plays a key role in equine biomechanics, balance, and locomotor performance, and is particularly relevant in Pura Raza Española (PRE) horses because of its functional and morphological importance in breeding programmes. Despite its relevance, several specific cervical conformation defects have not been systematically characterized as independent traits, which may limit their biological interpretation and genetic evaluation.

The objectives of this study were to estimate the prevalence of eight neck conformation defects in PRE horses, analyse the main non-genetic factors associated with their expression, identify phenotypic profiles using a multivariate clustering approach, and estimate genetic parameters.

A total of 66,515 PRE horses (21,241 males and 45,274 females), evaluated between 2012 and 2025 within the official studbook evaluation system, were included. Eight cervical defects were analysed: poorly and very marked head–neck junction, low and high neck–trunk junction, closed and open intermandibular space, and thin and thick upper neck border. Traits were scored on a three-class ordinal scale (0 = absence, 1 = moderate expression, 2 = severe expression). Statistical analyses included tests of proportions, contingency-table association analyses, k-means clustering, and genetic parameter estimation using Bayesian threshold animal models implemented in the BLUPF90 family of programs (GIBBSF90+).

Results showed that the proportion of unaffected animals was consistently higher across all traits, although prevalence varied among defects. The most frequent defect was poorly marked head–neck junction (37.63%), followed by closed intermandibular space (33.54%) and thick upper neck border (30.89%). Heritability estimates were generally moderate, with the highest values observed for low neck–trunk junction (0.286–0.330), closed intermandibular space (0.292–0.299), and open intermandibular space (0.286–0.289). Relevant genetic correlations were identified, including both positive and antagonistic relationships among traits.

Cluster analysis revealed three distinct phenotypic profiles of cervical conformation, reflecting structured combinations of defects rather than isolated traits. In addition, sex, age, birth period, proportionality index (height at the withers * 100 divided by the scapular-ischial length), and neck length to withers height ratio were significantly associated with defect expression and cluster distribution. The moderate heritability values indicate that these defects have a relevant additive genetic component and may respond to selective breeding. The identification of integrated phenotypic profiles highlights the importance of considering neck conformation as a complex trait, supporting the inclusion of defect-specific information in breeding programmes to improve both functionality and morphological quality in the PRE breed.

Keywords: heritability, morphological defect, equine, genetic selection

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