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Identification of three distinguishable phenotypes in golden retriever muscular dystrophy

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ABSTRACT. Duchenne muscular dystrophy (DMD) is a human disease characterized by progressive and irreversible skeletal muscle degeneration caused by mutations in genes coding for important muscle proteins. Unfortunately, there is no efficient treatment for this disease; it causes progressive loss of motor and muscular ability until death. The canine model (golden retriever muscular dystrophy) is similar to DMD, showing similar clinical signs. Fifteen dogs were followed from birth and closely observed for clinical signs. Dogs had their disease status confirmed by polymerase chain reaction analysis and genotyping. Clinical observations of musculoskeletal, morphological, gastrointestinal, respiratory, cardiovascular, and

renal features allowed us to identify three distinguishable phenotypes in dystrophic dogs: mild (grade I), moderate (grade II) and severe (grade III). These three groups showed no difference in dystrophic alterations of muscle morphology and creatine kinase levels. This information will be useful for therapeutic trials, because DMD also shows significant, inter- and intra-familiar clinical variability. Additionally, being aware of phenotypic differences in this animal model is essential for correct interpretation and understanding of results obtained in pre-clinical trials.

Key words: Duchenne muscular dystrophy; Animal disease; Model; Phenotype

