COVID-19 is an emerging, rapidly evolving situation.
Get the latest public health information from CDC: https://www.coronavirus.gov.
Get the latest research from NIH: https://www.nih.gov/coronavirus.

FULL TEXT LINKS

Neuromuscul Disord. 2020 Apr 16;S0960-8966(20)30081-X. doi: 10.1016/j.nmd.2020.03.005.
Online ahead of print.

Pathogenic Variants in COL6A3 Cause Ullrich-like Congenital Muscular Dystrophy in Young Labrador Retriever Dogs

Véronique Bolduc 1, Katie M Minor 2, Ying Hu 1, Rupleen Kaur 1, Steven G Friedenberg 3, Samantha Van Buren 2, Ling T Guo 4, Joseph C Glennon 5, Katia Marioni-Henry 6, James R Mickelson 2, Carsten G Bönemann 7, G Diane Shelton 8

Affiliations
PMID: 32439203 DOI: 10.1016/j.nmd.2020.03.005

Abstract
The collagen VI-related muscular dystrophies in people include a broad spectrum of diseases ranging from the severe Ullrich congenital muscular dystrophy to the mild Bethlem myopathy. Clinical features are attributable to both muscle and connective tissue and include progressive muscle weakness and respiratory failure, hyperlaxity of distal joints, and progressive contracture of large joints. Here we describe two different COL6A3 pathogenic variants in Labrador Retriever dogs that result in autosomal recessive or autosomal dominant congenital myopathies with hyperlaxity of distal joints and joint contracture, similar to the condition in people.

Keywords: Canine; Collagen VI; Muscle; Myopathy.

Copyright © 2020 Elsevier B.V. All rights reserved.

LinkOut - more resources
Full Text Sources
ClinicalKey
Elsevier Science
Miscellaneous
NCI CPTAC Assay Portal