Lafora disease

Lafora disease is a genetic disease caused, in humans, by mutations in EPM2A and NHLRC1 genes, resulting in accumulation of polyglucosan bodies within neurons. Affected subjects present progressive neurological signs characterised primarily by myoclonic epilepsy. In dogs, Lafora disease has been described mainly in miniature wire-haired Dachshunds, where a dodecamer expansion in NHLRC1 gene has been identified. The same mutation has then been detected in the Basset Hound, Beagle, Chihuahua and Pembroke Welsh Corgi breeds. This is the first case of a Newfoundland dog with myoclonic epilepsy diagnosed with Lafora disease based on confirmed dodecamer expansion in the NHLRC1 gene. Lafora disease is being progressively recognised in different unrelated breeds suggesting a wider distribution in the canine population than previously thought [1)].

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