ANIMAL GENETICS

CENTRONUCLEAR MYOPATHY (CNM)

Centronuclear Myopathy (CNM), previously known as HMLR, or Hereditary Myopathy, is an autosomal recessive mutation that causes insufficient muscle function in the Labrador Retriever breed. This is due to the centralisation of the nuclei in muscle fibers, caused by a missense insertion in the PTPLA gene.

Puppies are born apparently normal; however, it quickly becomes evident that there is a problem. The puppy will often not gain weight adequately due to decreased muscle tone in the esophagus. Within 2 to 5 months, the disease has usually progressed to display the full range of symptoms, including a loss of muscle tone and control, an awkward gait, and extreme exercise intolerance. This condition is exacerbated in cold conditions.

Unfortunately, there is no cure for CNM, as the dog will never develop properly functioning muscle tissue. The dog usually has a normal life span but he will always be plagued with the symptoms of Centronuclear Myopathy.

Testing Tips

Centronuclear myopathy is a recessive disorder, meaning that the dog must have two copies (CNM/CNM) of the defective gene to suffer from the disease. A dog can also be a carrier (CNM/n) of this disease and will not display any symptoms. A carrier dog will pass on the mutation that causes CNM to 50% of it's offspring. If mated with another carrier dog, there is a 25% chance of producing an offspring affected by Centronuclear Myopathy.

Reliable genetic testing is important for determining breeding practices. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers to each other is not recommended. Labrador Retrievers that are not carriers of the mutation have no increased risk of having affected pups.

Source: https://www.animalgenetics.eu/Canine/Genetic_Disease/CNM.asp

The prevalence of this condition in South Africa is unknown but is likely uncommon in purebred, registered Labradors. Some breeders test locally bred dogs for the condition and most breeders do test if the dog is imported. Note that many breeders will include carriers of the disease in their breeding programme, always making sure that the mate is clear. This carefully implemented strategy is done to ensure continued genetic diversity in the breed and poses no risk to the puppies.

BREEDING IMPLICATIONS			MATERNAL CANDIDATE					
			CLEAR		CARRIER		AFFECTED	
			G	G	G	A	A	A
PATERNAL CANDIDATE	CLEAR	G G	GG	GG	GG	GA	GA	GA
			ALL CLEAR		50% CLEAR 50% CARRIER		ALL CARRIER	
	CARRIER	G A	GG	GA	GG	GA	GA	AA
					GA	AA		
			50% CLEAR 50% CARRIER		25% CLEAR 50% CARRIER 25% AFFECTED		50% CARRIER 50% AFFECTED	
	AFFECTED	A A	GA	GA	GA	AA	AA	AA
			ALL CARRIER		50% CARRIER 50% AFFECTED		ALL AFFECTED	

Source: Inqaba Biotec