Spinal Dysraphism in Weimaraner Dogs:

Introduction

Spinal dysraphism (SD) in Weimaraner dogs is a genetic disorder present at birth that results from faulty embryonic development. Affected Weimaraners have a defective spinal canal which leads to neurological abnormalities. Puppies born with SD may have difficulties starting to walk due to weakness of their rear legs. Adults with SD show a typical abnormal gait that includes simultaneous movement of the hind legs or "bunny hopping" in the rear. Additional characteristics include weakness and lack of coordination in the rear, together with normal front end coordination and strength. Rarely a "bunny hopping" gait is observed in the front. The condition is not painful and it does not progress during the life of an affected dog.

SD is an inherited autosomal recessive disease caused by a mutation in the *NKX2-8* gene. Two copies of the mutation are necessary for disease signs to be present with both sexes being affected equally in frequency and severity. Weimaraners that have only one copy of the SD mutation (N/SD) are normal but they are carriers of the disease. When two carriers are bred to each other, 25% of the resulting puppies are expected to be affected and 50% to be carriers. Approximately 1.4% of Weimaraners are estimated to be carriers of SD (N/SD); however, the number of carriers can change with each generation. Weimaraners that are carriers of SD (N/SD) are completely normal, and they can be safely bred to dogs that are non-carriers (N/N) in order to maintain diversity within the breed and to select for other positive attributes in carrier dogs.

	Female		
Male	**N/N**	**N/SD**	**SD/SD**
N/N	100% N/N	50% N/N, 50% N/SD	100% N/SD
N/SD	50% N/N, 50% N/SD	25% N/N, 50% N/SD, 25% SD/SD	50% N/SD, 50% SD/SD
SD/SD	100% N/SD	50% N/SD, 50% SD/SD	100% SD/SD

The VGL offers a DNA test for SD to assist Weimaraner owners and breeders in identifying affected and carrier dogs. The test uses DNA collected from buccal (cheek) swabs, thus avoiding blood sample collection. Breeders can use results from the test as a tool for selection of mating pairs to avoid producing affected dogs.

Results are reported as:

N/N:	no copies of SD mutation; dog is normal
N/SD:	1 copy of SD mutation; dog is normal but is a carrier
SD/SD:	2 copies of SD mutation; dog is affected.

Reference:

Safra N, Bassuk AG, Ferguson PJ, Aguilar M, Coulson RL, Thomas N, Hitchens PL, Dickinson PJ, Vernau KM, Wolf ZT, Bannasch DL. Genome-wide association mapping in dogs enables identification of the homeobox gene, NKX2-8, as a genetic component of neural tube defects in humans. PLoS Genet. 2013 Jul;9(7):e1003646. doi: 10.1371/journal.pgen.1003646. Epub 2013, Jul 18.