

Juvenile-onset Laryngeal Paralysis and Polyneuropathy (also called NVSA or POANV) in Rottweilers.

Dennis O'Brien DVM PhD and Gary Johnson DVM PhD, University of Missouri, College of Veterinary Medicine

JLPP? NVSA? POANV? Why the alphabet soup of names?

The parable of the blind men and the elephants tells of blind men touching an elephant for the first time. Each comes away with a different impression of what an elephant is like (a wall, a tree, a brush) depending on which part they touched. When veterinary researchers first see a new disease, they describe what they observe but may be blind to other aspects of the disease that latter come to light.

In the late 1990s, veterinary neurologists in America and Europe recognized a new hereditary disease in Rottweilers. The affected pups showed coordination problems (spinocerebellar ataxia) and a very unusual form of degeneration in the brain at post-mortem with basically holes (vacuoles) developing within the brain cells (neurons). This syndrome was originally describes as "Neuronal Vacuolation and Spinocerebellar Ataxia" or NVSA for short. As more cases were studied, however, it was recognized that these pups can usually suffer from breathing difficulties due to paralysis of the muscles of the voice box (laryngeal paralysis) as well as weakness in the limbs. These symptoms were due to a degeneration of the nerves supplying these muscles (polyneuropathy). Laryngeal paralysis is common in old dogs, but since these dogs were affected at a young age, this portion of the syndrome was called Juvenile-onset Laryngeal Paralysis and Polyneuropathy (JLPP). Finally it was noticed that many of these pups also had cataracts along with other abnormalities within the eye (ocular abnormalities). Like laryngeal paralysis, cataracts are common in older dogs, but if they are found in a juvenile dog, that suggests a hereditary problem. Once it was clear that all these symptoms were part of a common disease, the broader term Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation (POANV) to encompass all aspects of the disease. For sake of simplicity, the term JLPP is still commonly used.

The typical affected pup begins showing symptoms at around 3 months of age. The initial symptoms can be breathing difficulties, especially with exercise or excitement, or they can begin with weakness and incoordination of the legs. Cataracts or other changes may be visible on careful examination of the eyes. The breathing problems from the laryngeal paralysis can be improved with surgery, but the disease is progressive. The weakness and coordination problems become more severe and the dogs can develop problems swallowing. All affected dogs have been euthanized before a year of age to prevent suffering.

Recently, researchers at the University of Missouri began studying a hereditary disease in Black Russian Terriers that was originally called juvenile-onset laryngeal paralysis and polyneuropathy (JLPP) because the first two dogs identified by Dr. Nicholas Granger in England presented for breathing difficulties and weakness of the limbs. As with the Rottweilers, further study showed that the Black Russian Terriers also had cataracts and the same vacuolation of the brain cells. Dr. Gary Johnson's lab at the University of Missouri identified the mutation responsible for this polyneuropathy with ocular abnormalities and neuronal vacuolation (POANV) in the Black Russian Terriers. Recognizing the similarity between this disease and the one in Rottweilers, they investigated that breed as well and found that the same mutation caused the disease in both breeds. This is not surprising since Rottweilers were used as foundation stock for the creation of the Black Russian Terrier breed. For further information on the disease please see our website <http://www.caninegeneticdiseases.net/RottweilerJLPP/> .

With the discovery of the mutation, a DNA test was developed and is available through the Orthopedic Foundation for Animals. The test can be ordered from the OFA website (www.ofa.org) under “Juvenile Laryngeal Paralysis Polyneurpathy” in Order DNA Tests section. The DNA test can aid your veterinarian in definitively diagnosing the disease if a pup is showing signs of the disease. It is important to make a definitive diagnosis since other, potentially treatable diseases, may show similar symptoms.

The DNA test can also identify carriers of the mutation so that breeders can make wise breeding decisions. While the initial response is often to want to eliminate all carriers of a mutation from the breeding pool, this is not the recommended approach. Eliminating all the carriers will narrow the gene pool and limit the breeding choices available. This reduces desirable genetic diversity in the breed and may inadvertently increase the incidence of other hereditary problems that may be lurking the lines that are free of the JLPP mutation. There are often highly desirable traits in the lines with a mutation that made them popular to begin with. Eliminating those lines can eliminate those desirable traits, throwing out the baby with the bathwater. The best approach is to test breeding stock and ensure that two carriers are never bred together so that no affected pups are produced. A carrier that has desirable traits can be bred to a dog who is tested clear of the mutation since no affected pups can result from such a breeding. Then the carrier status of the offspring should be one factor in determining who will be kept for the next generation of breeding stock. Over time, the prevalence of the mutation in the breed will decline while a genetic bottle-neck is avoided. The key is to never breed two carriers so that no affected pups are produced.