

VETERINARY TECHNICAL DATASHEET

X-Linked Tremors; mutation originally found in English Springer Spaniel



Mutation Found In :English Springer Spaniel

Disorder Type

- Nervous system

Disease Severity

- Severe

Background

X-linked tremors is a congenital neurological disorder of the central nervous system. Normally, oligodendrocytes produce an insulating myelin sheath that covers the axons of the nerves. Abnormal differentiation of oligodendrocytes causes lack of myelin production, tremors, and premature death. X-linked tremors is encountered in English Springer Spaniels. The condition is inherited in an X-linked recessive manner and only males are severely affected due to this X-linked inheritance. Females may exhibit mild tremors.

Key Signs

- Generalized tremor
- Reduced weight and body size
- Convulsions

Clinical Description

The first signs of X-linked tremors are usually observed by 2 weeks of age. Affected males suffer from severe tremors in the body, head, and limbs. The tremors tend to worsen with activity and decline during rest. Affected puppies suffering from tremors have difficulties standing, moving, and eating. The condition is progressive and leads to seizures. The affected dogs are smaller in size than their unaffected littermates. The life expectancy of affected dogs is not likely to be over 3-4 months.

Mode of Inheritance

- X-linked

Gene Name

- PLP1

Next Steps

Humane euthanasia for affected male dogs is often elected. Female carriers of the disorder may have mild tremors during puppyhood that resolve by the age of 4 to 6 weeks of age.

References

Nadon NL, Duncan ID, Hudson LD. A Point Mutation in the Proteolipid Protein Gene of the Shaking Pup Interrupts Oligodendrocyte Development Development 110:529-537, 1990.

Griffiths IR, Duncan ID, McCulloch M, Harvey MJA. Shaking pups: A disorder of central myelination in the spaniel dog. J Neurol Sci 1981;50:423-433.