VETERINARY TECHNICAL DATASHEET

Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in German Shepherd Dog

Mutation Found In :German Shepherd Dog

Disorder Type

- Metabolic

Disease Severity

- Severe

Background

Mucopolysaccharidoses (MPS) are a group of rare inherited lysosomal storage disorders resulting from a deficiency in the enzymes required for degradation of glycosaminoglycans. Enzyme deficiency causes accumulation of metabolic by-products in cells disrupting their normal function. Several different forms of mucopolysaccharidoses with different clinical signs have been recognized in dogs. Mucopolysaccharidosis Type VII (MPS VII) is seen in the German Shepherd and is characterized by severe skeletal dysplasia, corneal clouding, and difficulty standing and moving due to the severe changes in bone structure and joint laxity. Affected puppies are usually euthanized because of the severity of the condition.

Key Signs

- Dysmorphia
- Skeletal lesions
- Death

Clinical Description

The first clinical signs of mucopolysaccharidosis VII can be seen in dogs 2 to 5 months of age and typically involves hind limb weakness that progresses to ataxia of all four limbs. The affected dogs also exhibit growth retardation, facial and other skeletal dysmorphisms, and corneal clouding. These dogs may show severe epiphyseal dysplasia when radiographed and exhibit extreme laxity of joints that can result in subluxation with minimal effort. Neutrophils and lymphocytes in the blood or CSF may have cytoplasmic granules. Several other organs may be involved showing hepatomegaly, tracheal dysplasia, and cardiac abnormalities. Affected puppies are usually euthanized because of the severity of the condition.

Mode of Inheritance

- autosomal recessive

Gene Name

- GUSB

Next Steps

Treatment is supportive care, however, affected puppies are usually euthanized on welfare grounds because of the severity of the condition. Experimental gene therapy may prove curative in the future.

References

Ray J, Bouvet A, Desanto C, Fyfe JC, Xu DB, Wolfe JH, Aguirre GD, Patterson DF, Haskins ME, Henthorn PS. Cloning of the canine beta-glucuronidase cDNA, mutation identification in canine MPS VII, and retroviral vector-mediated correction of MPS VII cells. Genomics 48:248-253, 1998.