

## VETERINARY TECHNICAL DATASHEET

Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in Brazilian Terrier



Mutation Found In :Terrier Brasileiro

### 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 Brazilian Terrier and is characterized by severe skeletal dysplasia. Affected puppies are usually euthanized because of the severity of the condition.

### Key Signs

- Dysmorphism
- Skeletal lesions
- Death

### Clinical Description

The first clinical signs of mucopolysaccharidosis VII can be seen in 2 to 4 week old puppies. Affected puppies show facial dysmorphism that includes a short broad face with low set ears. Affected dogs also have a broader body than their unaffected littermates. Affected puppies have difficulty standing and moving due to severe changes in the bone structure (spondyloepiphyseal dysplasia) and joint hyperlaxity.

### 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

Hytönen MK, Arumilli M, Lappalainen AK, Kallio H, Snellman M, Sainio K, Lohi H. A Novel GUSB Mutation in Brazilian Terriers with Severe Skeletal Abnormalities Defines the Disease as Mucopolysaccharidosis. PLoS One 7:e40281, 2012.