

Mucopolysaccharidosis VI

Description:

MPS VI disease is due to an arylsulfatase B deficiency and has been reported in Siamese and domestic shorthair cats, as well as Miniature Pinschers, Miniature Schnauzers, Welsh Corgis, and Chesapeake Bay Retrievers. A DNA based test is available to screen Siamese cats, in other cats an enzyme activity test is required (metabolic screening). Similarly, a DNA test has recently been developed for Miniature Pinschers with MPS VI.

In mucopolysaccharidosis, certain large sugars (polysaccharides) of the body are not properly broken down. In MPS VI, the deficient enzyme is arylsulfatase B, which is responsible for degrading dermatin sulfate. MPS VI is autosomal recessively inherited and has been seen in humans, cats and, more recently, dogs.

Clinical Signs:

The ensuing cellular accumulation results in skeletal deformities, including defects in the sternum, vertebrae and particularly the hip joints. To varying degrees they may also experience corneal cloudiness and facial dysmorphism.

The discovery of a disease-causing mutation in the arylsulfatase gene in Miniature Pinschers enabled the development of a DNA test which allows the identification of MPS affected, carrier, and normal (clear) animals. Affected Miniature Pinschers have not only been found in various states in the US, but also abroad. Thus, it is believed to be more wide spread and under-diagnosed than previously thought.

Affected Breeds:

Siamese, Miniature Pinscher, Miniature Schnauzer, Welsh Corgi, Chesapeake Bay Retriever and others.

DNA tests are available for Miniature Pinschers, Siamese and DSH.

Required Samples:

EDTA Blood, 1-2 mL or 2 Buccal Swabs (DNA Tests)

Urine, 1-2 mL (urine-screening tests for breeds without DNA tests)

Cost:

See PennGen Cost Price Sheet for current pricing

*To submit for an MPS VI DNA test please use our **DNA Submission Form***