

Mucopolysaccharidosis VI (MPS VI)

About the disease

A rare mutation (L476P) in the *ARSB* gene causes the severe form of mucopolysaccharidosis VI (MPS VI Severe). This is a lysosomal storage disease and affected cats typically show signs of wide faces, shortened noses, small ears and retarded growth. As the cat ages other problems develop, such as severe hind-limb mobility or paralysis. The mutation is inherited in an autosomal-recessive fashion, hence Affected cats have two copies of the mutation.



A second variant (D520N) in the *ARSB* gene has been reported to cause 'mild' MPS VI. However, it is now considered that cats with this variant (MPS VI Mild) are unaffected by disease UNLESS they also carry the severe mutation mentioned above (i.e. MPS VI Severe/MPS VI Mild). It has also been shown that the D520N variant is VERY common in MANY cat breeds, and negative selection for this variant could be very detrimental to the genetic diversity of many breeds, whilst having little value for the prediction of MPS VI disease*.

Since results of D520N screening SHOULD NOT be used to inform breeding decisions we have decided to ONLY offer this test when breeders know that their cat is a Carrier of MPS VI Severe (i.e. to detect MPS VI Severe/MPS VI Mild cats), or to test a potential mate for their cat that carries the MPS VI Severe mutation (i.e. to avoid producing MPS VI Severe/MPS VI Mild kittens).

*Lyons et al. BMC Veterinary Research (2016) 12:136

About the test

At the Molecular Diagnostic Unit, we have developed a PCR-based pyrosequencing assay to quickly and accurately identify the MPS VI Severe mutation that is known to cause MPS VI. For cats shown to carry the MPS VI Severe mutation we can also screen them, or potential mates, for the MPS VI Mild variant.

This assay will allow owners and breeders to identify affected and carrier cats and will enable them to inform breeding programs to reduce MPS VI in their kittens.

What breeds are at risk?

Siamese and related breeds.

Reception Hours

Mon-Fri 9am - 5pm

Contact Us

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Interpretation of results

A **Normal** result on the MPS VI Severe genetic test means that the cat does not have the MPS VI Severe (L476P) mutation. It may have one or two copies of the MPS VI Mild variant (D520N), but there is no evidence that this variant is associated with disease.

A **Carrier** result on the MPS VI Severe genetic test means that the cat carries one copy of the MPS VI Severe (L476P) mutation. This cat is a Carrier of MPS VI Severe and should also be tested for the MPS VI Mild (D520N) variant.

An **Affected** result on the MPS VI Severe genetic test means that the cat has two copies of the MPS VI Severe (L476P) mutation and will be affected by MPS VI.

Each certificate we issue will specify whether the cat is Normal, Carrier or Affected for the MPS VI Severe mutation.

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