

Mucopolysaccharidosis IIIB

Disease Description:

The disease Mucopolysaccharidosis IIB (MPS IIIB), also known as Sanfilippo syndrome type IIIB, is an inherited disease classified as a lysosomal storage disease (LSD). Lysosomes are “bags” within cells of the body and are filled with special enzymes that disassemble molecules in an orderly manner. If one of the enzymes is missing (due to mutations in the gene for that enzyme), the disassembly stops and undegraded molecules accumulate in the lysosomes (hence the term LSD). When this happens the cells become sick or die, which leads to disease. The compound accumulating in MPS IIIB is heparan sulfate and the affected enzyme is N-acetyl-a-D-glucosaminidase (NAGLU).

The inheritance pattern of MPS IIIB is autosomal recessive. The mutant gene may be as far back as eleven generations, and hence may be very broadly distributed in the Schipperke population. The carrier frequency is unknown, but judging from similar diseases in cattle, it may be as high as 15%. Both males and females are equally capable of having the disease, or of being carriers. Carriers are absolutely normal, and will not have signs of the disease. We have a mutation-based test for the NAGLU mutation in the Schipperke breed. Testing offered at the University of Pennsylvania reports a result of “affected,” “carrier,” or “normal.” Results are confidential and are released only to the individual who submitted the sample. Every breeding animal should be DNA tested for this disease, unless the animal is completely descended from animals tested as normals.

Clinical Signs:

Clinical signs in the dogs are related to brain disease, appear between 2-4 years of age, and include tremor and difficulty in balancing, walking, and negotiating obstacles such as stairs. The disease is progressive and there is no effective treatment. Because of this, most owners choose euthanasia 1-2 years after recognizing clinical signs.

Affected Breeds:

Schipperke

Required Samples:

EDTA Blood, 1-2 mL or 2 Buccal Swabs

Cost:

See PennGen Cost Price Sheet for current pricing

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