How is neuromuscular diseased diagnosed?

1. *Blood samples - haematology and biochemistry*
2. *Other blood tests*
3. *Electrophysiology*
4. *Muscle and nerve biopsy*
5. *Other imaging*

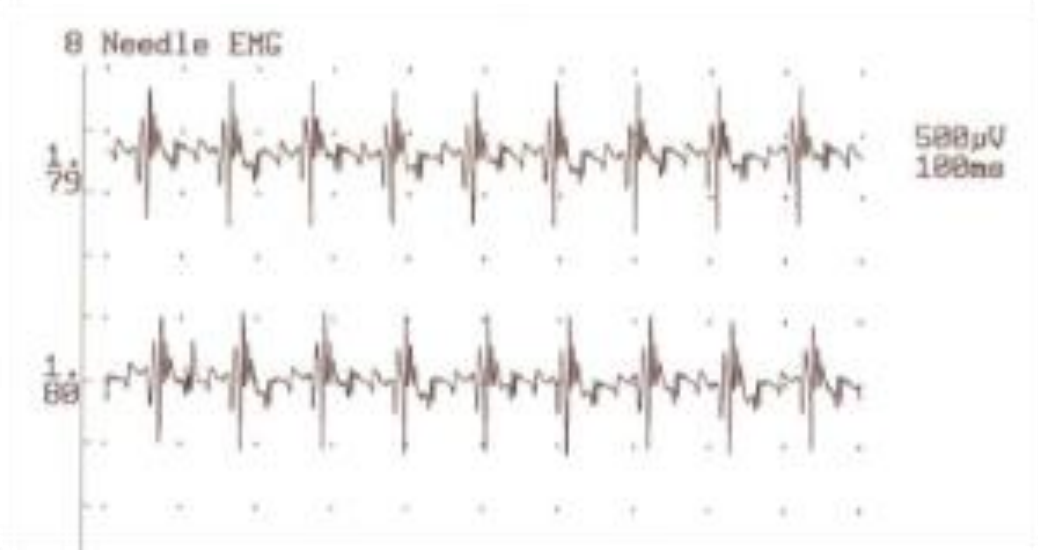
1. Blood samples - haematology and biochemistry

A lot of diseases can be eliminated/suggested by routine tests for example electrolyte disorders (hypokalaemia, hypocalcaemia,) and endocrinopathies (Cushing's syndrome, hypothyroidism, hyperthyroidism, diabetes mellitus). A significantly elevated creatine kinase (>500u/l) indicates muscle damage and is suggestive of a dystrophy, inflammation or metabolic imbalance such as hypokalaemia. However creatine kinase can also increase after prolonged recumbency or because the animal is breaking down muscle for energy. In protozoal diseases such as toxoplasmosis or Neosporosis, liver parameters may also be elevated.

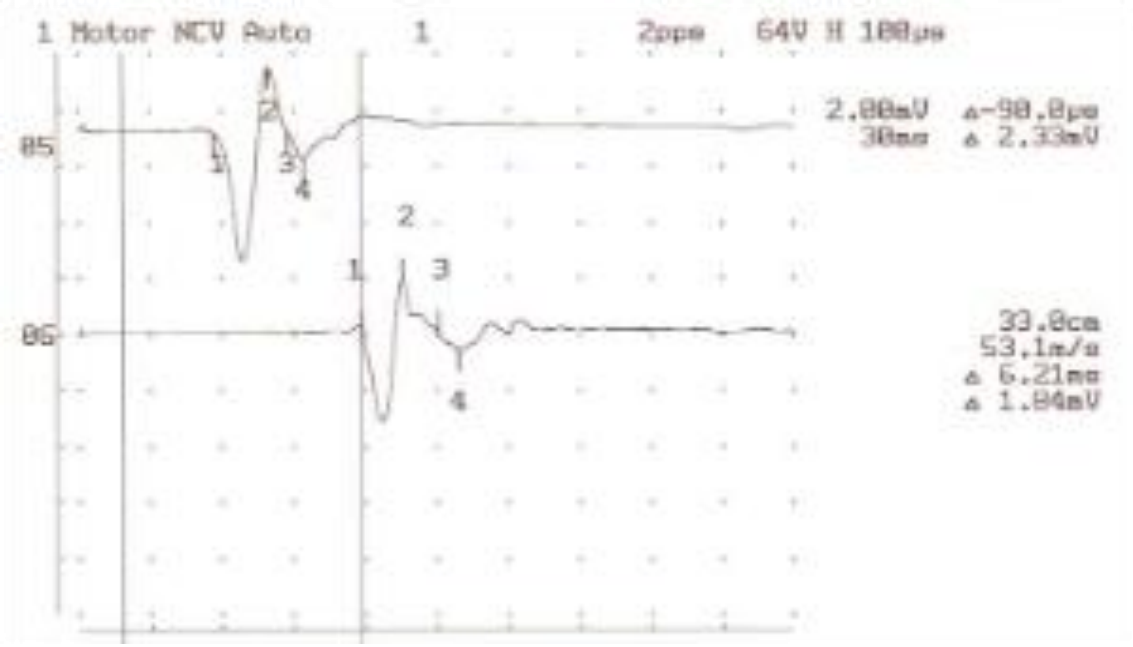
2. Other blood tests

If serum biochemistry is suggestive then specific tests for endocrine disorders may be recommended. Blood samples may also be recommended to test for myasthenia gravis (acetylcholine receptor antibody); http://vetneuromuscular.ucsd.edu/ protozoal problems (*Toxoplasma gondii and Neospora caninium*) and / or mitochondrial myopathies(blood pH and abnormal levels of organic acids). It pet's problem suggests a specific inherited problem then a specific genetic test may be recommend e.g. Centronuclear Myopathy (Labrador retriever myopathy) or exercise induced collapse in Labrador Retrievers.

3. Electrophysiology At Stone Lion Veterinary Centre electrophysiology (**electromyography** (EMG) with **nerve function tests**) is used for diagnosing neuromuscular disease and specifically localising the problem to the nerve, neuromuscular junction or a neuropathy. It can also provide information as to the aetiology of the problem; for example it is possible to ascertain if the disease is more likely to involve the myelin or the axon.



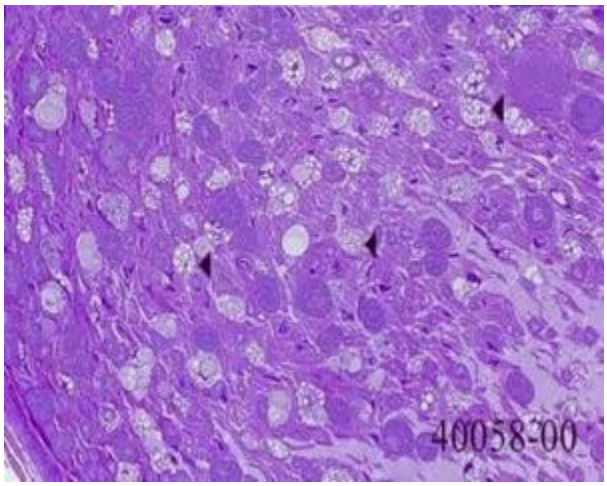
An EMG recording from a muscle. Abnormal spontaneous activity can indicate a neuropathy or myopathy



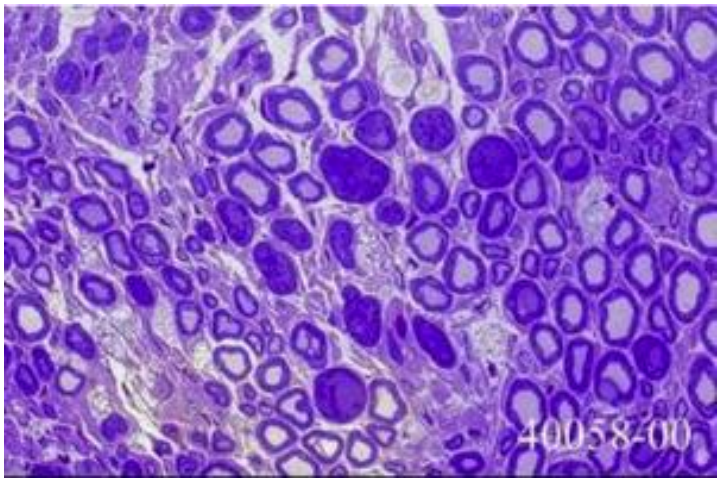
Trace of a sciatic motor nerve conduction velocity. The amplitude of the compound muscle action potential (numbered wave form) is reduced and so is the conduction velocity (53.1m/s). This dog had a neuropathy.

4. *Muscle and nerve biopsy*

Although relatively easy to perform it is important that biopsies are handled correctly and submitted to an appropriate laboratory. A dedicated neuromuscular laboratory should be able to provide the full range of histologic and histochemical stains and electron microscopy e.g. Neuromuscular Laboratory, Edinburgh University and Comparative Neuromuscular Laboratory, Davis University, California



Nerve biopsy from a 1 year old female domestic shorthair diagnosed with a neuropathy. The male sibling was more severely affected. The nerve biopsy revealed lipid droplets within macrophages endoneurium & subperineurally (arrows) suggestive of a lysosomal storage disease. Histopathology sections were prepared, analysed and photographed by Dr Caroline Hahn Neuromuscular Laboratory, University of Edinburgh.



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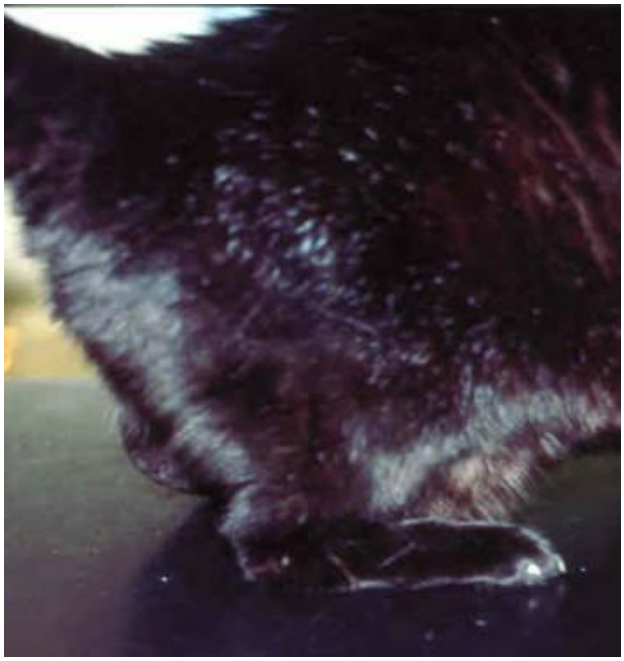
Other imaging

Other tests e.g. radiographs (x-rays) or ultrasound may be recommended to look for associated or contributing diseases e.g. myasthenia gravis can be induced by a tumour in the chest called thymoma

How is neuromuscular diseased treated?

Therapy obviously depends as to the precise nature of the problem. Some neuromuscular diseases have a specific therapy for example endocrine disorders (hyperthyroidism, diabetes mellitus); Myasthenia gravis (Pyridostigmine and immunosuppressive therapy); and antiprotozoal agents for Toxoplasmosis and Neosporosis. For many degenerative neuropathies there is no treatment however some improve when given dietary supplements such as the mitochondrial co-factor L-Carnitine 50mg/kg twice daily and anti-oxidants such as gamma linoleic acid and omgea-3-fatty acids. Many severely tetraparetic animals will require considerable supportive care as well. For example, a pet which is unable to swallow may require feeding through a (PEG) tube into the stomach.



This 16 year old cat has diabetes mellitus and associated neuropathy particularly of the sciatic nerve. The cat is weak and walking on her hocks (plantigrade stance). She improved after the diabetes mellitus was stabilised and her diet was supplemented with L-Carnitine and omega 3 and 6 fatty acids