Polymyositis in Vizsla

Muscle wastage, weakness, difficulty eating / drinking, drooling

Please note: Dr Rusbridge now at Fitzpatrick Referrals -http://www.fitzpatrickreferrals.co.uk/

DNA Collection Pack:
1. Instruction Sheet
2. Consent Form Sheet 2
3. Phenotype From Sheet 3 (3 pages)
4. Phenotypic inclusion/exclusion criteria (Sheet 4)
5 UK DNA Archive for Companion Animals Information Sheet. (Sheet 5)

Instructions for DNA collection

Instruction Sheet (Sheet 1)

Thank you very much for agreeing to participate in this project aiming to collect DNA from Hungarian Vizslas and any affected/unaffected relatives (sire, dam, siblings, offpring).

- **Blood - Remaining blood from a diagnostic test, such as a serum creatine kinase is put in an EDTA tube (purple/pink top).**
- **Saliva - Collection kits available from di.addicott@gmail.com. Full instructions provided.**

Owner should read and understand the UK DNA Archive for Companion Animals Information (sheet 5) or Leaflet provided .

For each dog sampled:.

1) Sign the **DNA Archive Consent Form** (Sheet 2)

2) Complete the **Phenotype Form** (sheet 3) for each dog sampled. It is vital that we have the dog's correct pedigree name and registration number if available.

3) Send completed **Consent and Phenotypes Forms with blood or saliva sample** (Sheets 2 -3)

Please also include a copy of the dog's pedigree. Send to

CIGMR Medical School Stopford Building
The University of Manchester Oxford Road
Manchester M13 1BJ

4) Post or fax a duplicate of phenotype Form 3 together with a copy of the pedigree

to **Dr Clare Rusbridge, Stone Lion Veterinary Hospital ,**
41 High Street, Wimbledon, SW19 5AU.
Confidential Fax line 44 (0) 208 786 0525

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Informed consent (sheet 2)

Name _____ ID#_____

Reg No _____

1. I have read and understood the accompanying information leaflet explaining the UK DNA Archive for Companion Animals.

2. I appreciate that in order to advance our understanding of veterinary diseases there is a need to determine how a particular condition relates to the genetic profile of the animal.

3. I understand that any genetic tests relating to my animal will not provide specific information about its condition but will contribute to the general body of knowledge about the disease in the species. I realise that no specific information regarding genetic tests on my animal will be reported back to me.

4. I agree to DNA being extracted from a **blood or saliva sample** taken from my animal and that this will be used entirely for research purposes. I give consent for the material to be stored and made available to *bona fide* scientists in the field of animal disease and genetics.

5. I understand that all information I give will be held in strict confidence and the source of the archived DNA will not be divulged

6. I understand that this research will not benefit my animal directly, but in the future may be of benefit to other animals.

7. I understand that the custodianship of the DNA resides with the University of Liverpool but I retain the right to remove my animal's sample from the archive in the future if so wished.

Signed…… Date…….

DNA collection coordinator Clare Rusbridge, ~~Stone Lion Veterinary Hospital , 41 High Street, Wimbledon, SW19 5AU~~
~~Tel: 44 (0) 20 8946 4228~~ ~~www~~.veterinary-neurologist.co.uk Confidential fax 44 (0)20 87860525

DNA Archive for Companion Animals

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Phenotype Form

Form /Sheet 3

DATE -

Breed

Pedigree Name :

Kennel Registration number Date of birth:d/m/y

Colour Gender M MN F FN Weight (kg)

Call name Owners name .

Owner or Veterinary addresses and contact no. / email

Possible signs of Polymyositis - past or present.

No clinical signs of polymyositis (no weakness, muscle atrophy or eating difficulty ☐

Signs	Yes/No/Don't know	Date of onset	Comments
Eating difficulty			
Swallowing difficulty			
Drooling saliva			
Difficulty opening mouth			
Pain on opening mouth			
Regurgitation[1]			

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UK DNA Archive for Companion Animals

Masticatory muscle atrophy [2]			
Other muscle atrophy			Specify which muscles
Weakness			
Poor exercise tolerance			Describe
Lameness			Specify which limbs
Diagnostic tests	**Yes/No/Don't know**	**Date**	**Result(s)**
Megaoesophagus [3] (x-rays +/- Barium studies)			
Fluoroscopy			
Creatine kinase[4] (initial)			
Creatine kinase (subsequent)			
Type 2M muscle fibre autoantibodies [5]			
Acetylcholine receptor antibody titre [6]			
Electromyography			
Biopsy			Please attach copy report

1. Bringing up undigested food
2. Loss of head muscle especially on the top of the head. Skull bones become very prominent
3. Enlarged, dilated, flaccid oesophagus
4. Also known as "CK" or "muscle enzymes"
5. Test for Masticatory Myositis
6. Test for Myasthenia Gravis

Other comments or clinical signs -

(*please include computer medical history if likely to be helpful)*

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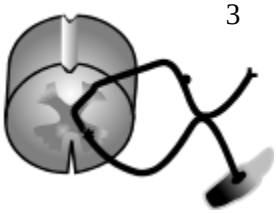

Phenotype Form

Form /Sheet 3

TREATMENT

Corticosteroids

Drug name -
Initial Dose -
Date started -
Length of time on this dose -
Subsequent changes

Success (or not) -
Current dose -

Azathioprine

Initial Dose -
Date started -
Length of time on this dose -
Subsequent changes

Success (or not) -
Current dose

Other

Drug name -
Initial Dose -
Date started -
Length of time on this dose -
Subsequent changes

Success (or not) -
Current dose -

If additional drugs used then please duplicate this sheet (*include computer medical history if helpful)*

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Phenotype – inclusion and exclusion criteria

Form/ Sheet 4

Disease description A breed-specific polymyositis (generalized inflammatory myopathy) has been recognized in Hungarian Vizsla dog (Foale *et al*, BSAVA 2008 and Haley *et al*,ACVIM 2009). The most common presenting signs are eating and swallowing difficulty (pharyngeal dysphagia) with loss of the muscles on the head (masticatory muscle atrophy). Other common signs include regurgitation, drooling saliva and difficulty and/or pain on opening the jaw. The creatine kinase is elevated to values greater than 1000 U/L - at least in the early stages of the disease. Thoracic radiographs may reveal a megaoesophagus (dilated, usually air filled, oesophagus). Fluoroscopy may detect oesophageal motility disorders. Electromyography can be normal or may reveal spontaneous activity suggestive of polymyositis (prolonged insertional activity and spontaneous activity including pseudomyotonia). The tongue and pharynx are the most useful muscles for electrophysiological evaluation. MRI may reveal hyperintensity within muscle groups. Histopathologic examination of muscle biopsy may reveal multifocal areas of lymphohistiocytic and plasmacytic myositis with fibrosis however active inflammation may not be appreciated if end stage muscle or limited areas are biopsied.

Inclusion criteria

- **Any breed with a histopathological diagnosis of polymyositis**
- **Vizsla with at least *one* of the following *in addition* to dysphagia (eating, drinking and swallowing difficulty with or without excessive salivation)**
    - **Creatine kinase > 1000U/L**
    - **Exercise intolerance**
    - **Megaoesophagus identified on thoracic radiographs**
    - **MRI changes consistent with polymyositis**
    - **Electrophysiological changes consistent with muscle disease**
- **Vizslas with siblings diagnosed with polymyositis**
- **Vizslas with offspring diagnosed with polymyositis**

Ideally all cases would have histopathological diagnosis however because this option is not available for all owners and, because some samples may not have active inflammation, the inclusion criteria is widened to include dogs with a consistent phenotype and biochemical or other evidence of polymyositis. In anticipation of future genetic studies DNA from unaffected relatives is also being collected (e.g. if remaining EDTA blood from routine screening) in addition to “control” cases from other breeds with polymyositis.

Exclusion criteria

- **Cases which do not fulfil the above criteria**
- **Dogs with normal creatine kinase that do not fulfil Inclusion Criteria**
- **Vizslas with positive antibody titres against type 2M muscle fibres and acetylcholine receptors are *not excluded* if other inclusion criteria are fulfilled**

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Information DNA Archive

Sheet/Form 5

UK DNA ARCHIVE FOR COMPANION ANIMALS

Great advances in veterinary medicine have been made recently and many of theses have centred around new developments in body imaging, new treatments and surgical procedures, and the identification of genes, which cause disease. Major developments in molecular biology have taken place in the last few years, making it possible to quickly analyse the DNA of both human and animals.

This is helping scientists work out what the underlying causes are for diseases and why some individuals become ill, whereas others remain well. Many of the diseases seen in companion animals, including dogs, cats and horses, are caused by a combination of genes from their parents (this is often referred to as “nature”) and the external or environmental factors they have experienced during their lives (this is called “nurture”). Most scientists now accept that for the majority of features about ourselves and our animals, they are the result of a mixture of nature and nurture. For example, body weight and height are in part caused by which genes are inherited and in part caused by our nutritional intake. In the same way, diseases such as diabetes in dogs, sarcoid in horses and renal failure in cats are likely to be caused by a combination of both nature and nurture. The analogy often given to explain why such diseases develop is that of requiring both the seed (nature) and the soil (nurture) before a plant can grow.

If researchers can identify which genes and environmental factors (such as vaccination, infections, nutrition, drugs) are important and interact together to cause diseases, we may be able to use this information to improve animal welfare. For example it may be possible to advise owners which foods or vaccinations their pets should avoid (or alternatively have) to reduce the risks of certain diseases developing.

Researchers from the UK Veterinary Schools and referral practices are now beginning to investigate the genetic and environmental factors underlying a wide range of diseases in companion animals. To do this it is important to collect large numbers of DNA samples from animals where the clinical features of diseases are clearly defined. Rather than have many small or duplicated collections across the UK, the Vet Schools have agreed to work together in assembling one National UK DNA Archive.

The information collected will be kept strictly confidential. The samples and clinical data will be made available through application to a review committee to *bona fide* research groups working on these conditions and where the projects have been deemed to be ethically sound. It is possible that samples will also be made available to research groups working in collaboration with non-academic and industrial partners.

The DNA sample being submitted to the Archive will usually be derived from blood leftover from the routine pathology tests being performed. Samples will only be included if the owners give their written consent. The sample will be anonymous once it is entered into the Archive.

The owner will also retain the right to remove the sample from the Archive in the future if so wished. No information regarding tests performed on the DNA sample will be given back to the owner. This is because it will only be possible to find out which genes and environmental factors are important by identifying patterns in large numbers of affected and unaffected animals.

Should you require further clarification of any issues raised please contact vetdna@liverpool.ac.uk

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