

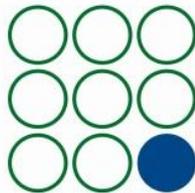


# Genetic Studies of Idiopathic Epilepsy in the Border Collie at the Animal Health Trust

**THE KENNEL CLUB**  
GENETICS CENTRE  

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AT THE ANIMAL HEALTH TRUST





# The Study

- Funded by PetPlan Charitable Trust
- **Overall aim:**

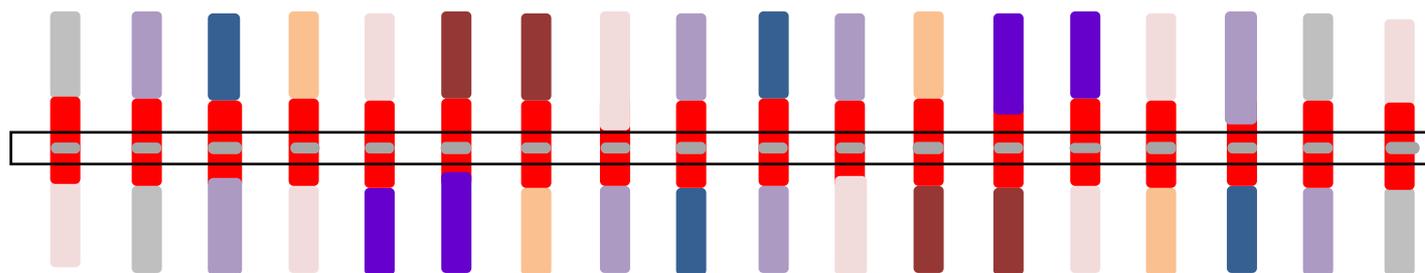
Identify mutations associated with Idiopathic Epilepsy (IE) in the Border Collie, which will help us understand the possible underlying causes of IE in this breed.
- For the study to be successful we require:
  - DNA from Borders Collies with **a confirmed diagnosis of IE - cases**
  - DNA from **older Border Collies who have never had a seizure - controls**



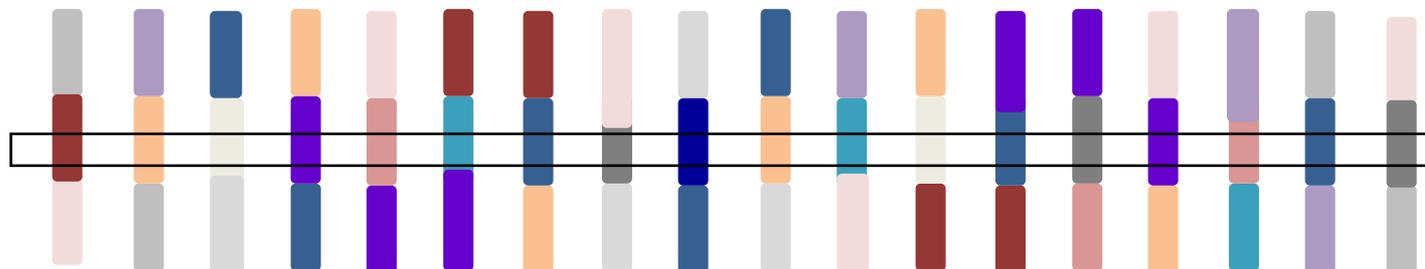
# Methodology

**Genome Wide Association Study (Genome Scan):** compare thousands of DNA markers in cases with those in controls to look for regions in the genome that are consistently shared among cases, but that are different in the controls.

Cases



Controls





# First Genome Scan: 2014

- In the first part of the study we had DNA from:
  - **73 cases** and **89 controls**
- The results suggested a region of the genome that could have potentially been associated with IE in the Border Collie.
- We needed to perform a second scan using DNA from additional dogs to confirm or disprove this finding.





## Second Genome Scan: 2015

- With help from Border Collie breeders and owners we collected an additional **29 cases** and **64 controls** for a second genome scan.
- The data from both genome scans were combined and analysed together to give:
  - **102 cases** and **153 controls**
- Unfortunately these results did not identify any regions of DNA which might be involved in idiopathic epilepsy.
- This means that the region identified in the first set is most likely not associated with idiopathic epilepsy.





# Moving Forward

- We have applied for funding to investigate epilepsy in the Border Collie using a different approach.
- We hope to use a whole-genome sequencing approach, which would involve reading the entire DNA in Border Collies and other breeds affected by idiopathic epilepsy.
- Using this approach we aim to identify common biological pathways that underlie genetic predisposition to idiopathic epilepsy.
- We also aim to identify gene variants that influence severity of disease and which might be involved in determining responsiveness to drugs.

***To progress this research we still need DNA from more dogs affected with idiopathic epilepsy***





# How Owners Can Help

- Owners of epileptic dogs are asked to:
  1. Fill in a short questionnaire, designed by Luisa De Risio, Head of Neurology
  2. Give the AHT permission to contact their vet for more information, if necessary
  3. Provide five cheek swabs from their dog
- All information is kept in complete confidence and is not shared with anyone who does not work at the AHT.





## Please help us...

- By contacting us if you have a Border Collie with epilepsy
- By sharing information about the AHT's epilepsy research with anyone you know who has a Border Collie
- Breed Clubs, Associations, Societies, Health groups etc.
  - please consider sharing information about this research with your members, by email or using social media
- For more information and to request a questionnaire contact:

[chris.jenkins@aht.org.uk](mailto:chris.jenkins@aht.org.uk) or [bryan.mclaughlin@aht.org.uk](mailto:bryan.mclaughlin@aht.org.uk)

