

Episodic Falling (EF)

Affected breeds: Cavalier King Charles Spaniel

Episodic Falling (EF) is an excitement, exercise or stress-induced muscle stiffness of short duration of usually less than 5 minutes. It develops in pups of 3-7 months of age, and affects both males and females. During an episode the limbs appear locked in an extended, rigid position. EF does not shorten life, and many affected dogs lead essentially normal lives between EF episodes; owners can often manage the condition by avoiding environmental triggers. **Not all pups which test as genetically affected will necessarily go on to exhibit EF episodes – it may be that the disorder is influenced by variation in environmental stimuli and/or modifier genes.**



The mutation is recessive, which means that dogs which carry the mutation ("CARRIERS") are normal but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation are at high risk of Episodic Falling

This test is particularly useful for breeders:

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- To conclusively confirm Episodic Falling

This test will be reported as:

CLEAR : no evidence of the Episodic Falling mutation

CARRIER : carries one copy of the defect, which will be passed to 50% of offspring

AFFECTED : carries two copies of the defect, and is at high risk of Episodic Falling

The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:

AFFECTED X AFFECTED	= 100% AFFECTED
AFFECTED X CARRIER	= 50% AFFECTED, 50% CARRIER
AFFECTED X CLEAR	= 100% CARRIER
CARRIER X CARRIER	= 25% AFFECTED, 50% CARRIER, 25% CLEAR
CARRIER X CLEAR	= 50% CARRIER, 50% CLEAR
CLEAR X CLEAR	= 100% CLEAR

References

Gill JL, Tsai KL, Krey C, et al. A canine BCAN microdeletion associated with episodic falling syndrome. *Neurobiol Dis.* 2012;45(1):130-136. doi:10.1016/j.nbd.2011.07.014

Forman OP, Penderis J, Hartley C, Hayward LJ, Ricketts SL, Mellersh CS. Parallel mapping and simultaneous sequencing reveals deletions in BCAN and FAM83H associated with discrete inherited disorders in a domestic dog breed. *PLoS Genet.* 2012;8(1):e1002462.