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Cause of neurological disorder in Belgian Shepherds discovered

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Fawn and Black Belgian Malinois Puppy on Green Grass. Via Pexels (https://www.pexels.com/photo/fawn-and-black-belgian-malinois-puppy-on-green-grass-209115/).

Malinois dogs are working animals known for being used by the Secret Service to guard the White House. These dogs, a subtype of the Belgian Shepherd breed, are robust, with an average life expectancy of 10–12 years. But some puppies are afflicted by a genetic condition called spongy degeneration with cerebellar ataxia (SDCA). A puppy diagnosed with SDCA quickly loses all coordination and needs to be put down. In the August issue of *G3*, Mauri *et al.* (http://www.g3journal.org/content/7/8/2729) report the genetic cause of one type of SDCA, a finding that will allow breeders to eliminate the problem from their lines.

The group previously showed (http://www.g3journal.org/content/7/2/663) that there is more than one type of SDCA in Belgian Shepherds. They identified causal mutations in one gene, *KCNJ10*, but that gene didn't account for all cases of SDCA. In their current study, they examined the genomes of several other affected puppies and found mutations in a gene called *ATP1B2*. Since this was a different gene than identified previously, they propose to call the new form of the disorder affecting these puppies SDCA2.

ATP1B2 encodes a subunit of the enzyme Na^+/K^+-ATPase, a protein complex essential for maintaining ion gradients across the cell membrane. In guinea pigs, inhibiting the enzyme causes seizures and makes the brain take on a spongy appearance, similar to that found in dogs with SDCA. Mice without their version of *ATP1B2* also

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have spongy brains and rapidly progressing motor disturbances. Although no *ATP1B2* variants have been found in humans, mutations in genes encoding the other subunits of Na^+/K^+-ATPase cause neurological problems, such as certain types of migraines and a fast-onset form of Parkinsonism.

The function of *ATP1B2* is also similar to that of the gene that causes SDCA1. Both are involved in maintaining potassium homeostasis—especially in the cerebellum—further substantiating the conclusion that mutations in *ATP1B2* are the cause of SDCA2. The evidence presented by Mauri *et al.* will enable development of genetic tests for both types of SDCA. Now that they've found that mutations in *ATP1B2* cause SDCA2 in dogs, checking people with family histories of cerebellar disorders without known causes for problems with *ATP1B2* might provide insight into previously inexplicable conditions.

CITATION:

Mauri, N.; Kleiter, M.; Dietschi, E.; Leschnik, M.; Högler, S.; Wiedmer, M.; Dietrich, J.; Henke, D.; Steffen, F.; Schuller, S.; Gurtner, C.; Stokar-Regenscheit, N.; O'Toole, D.; Bilzer, T.; Herden, C.; Oevermann, A.; Jagannathan, B.; Leeb, T.

A SINE Insertion in *ATP1B2* in Belgian Shepherd Dogs Affected by Spongy Degeneration with Cerebellar Ataxia (SDCA2).
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