Spinocerebellar Ataxia with Myokymia and/or Seizures (SCA); g.22141027insC mutation

In brief
Spinocerebellar ataxia is an early-onset neurological disorder characterised by uncoordinated movements and impaired balance. Three different forms of spinocerebellar ataxia have been described in Jack Russel Terriers and Parson Russel Terriers. It has been suspected that there might be other forms of ataxia in these breeds as well. Two different mutations in the KCNJ10-gene have been identified as causes for spinocerebellar ataxia associated with myokymia and/or seizures in these breeds. The disorder is inherited in an autosomal recessive manner.

Clinical overview
The onset of symptoms is usually at the age of 2-6 months. The first observable sign is poor coordination of movements (ataxia). Affected dogs may also suffer from episodic myokymia and/or seizures. Myokymia is characterised by uncontrollable twitching of the muscles that tends to run through a muscle in waves. Frequency of myokymia episodes varies from a few times a week to a few times a month. Myokymia episodes can be precipitated by exercise or excitement. Duration of the episodes can vary remarkably between individuals. Myokymia episodes can last from few minutes to few hours at a time. Myokymia can be generalised or restricted to muscles of one limb for example. The condition is progressive and can lead to neuromyotonia (generalised muscle stiffness). Neuromyotonia episodes are incapacitating attacks during which the dog becomes rigid and falls down. Affected dogs remain aware of their surroundings during myokymia attacks. Neuromyotonia can last from several minutes to several hours causing the body temperature to rise. Affected dogs suffering from neuromyotonia attacks are at risk of overheating. Excessive facial rubbing has also been reported in some cases. Severity of the symptoms leads to poor quality of life of the affected dogs.

Disease severity
Severe

Clinical signs
Ataxia Myokymia Neuromyotonia Seizures

Mode of Inheritance
Autosomal Recessive

Results of the genetic test are reported as follows:
- Clear
- Carrier
- At risk

Mutation(s) found in:
- Fox Terrier - Smooth
- Jack Russell Terrier
- Mixed breed
- Parson Russell Terrier
- Russell Terrier
- Tenterfield Terrier
- Toy Fox Terrier
- Working Jack Russell Terrier
References