Cerebellar Degeneration, Purkinje Cell Abiotrophy – Italian Spinone

Contributors:
Linda Shell, DVM DACVIM Neurology

Synonyms:
Cerebellar cortical degeneration
Cerebellar abiotrophy
Cerebellar Purkinje cell abiotrophy
Cerebellar atrophy

Disease description:
Cerebellar degeneration (abiotrophy) is the loss of differentiated cerebellar tissue, in this case the Purkinje cells. Purkinje cell degeneration is more commonly described than granule cell degeneration. Clinically, both types of degeneration produce signs of cerebellar dysfunction and the only way to distinguish between them is histopathological evaluation of cerebellar tissue. Neuropathologically, Purkinje degeneration is characterized by progressive loss of Purkinje cells and gliosis.

CLINICAL SIGNS AND DIAGNOSIS
Signs of cerebellar degeneration are the same signs as found with any cerebellar dysfunction: head and body tremors, hypermetria, a broad base stance, truncal swaying, bunny hopping, ataxia, decreased menace response and nystagmus. In general, signs are progressive over time but speed of progression varies with breed and type of degeneration.

A tentative diagnosis is based on the signalment, the absence of other systemic diseases, the presence of progressive cerebellar signs, and MRI findings of a smaller than normal cerebellum. Cerebrospinal fluid evaluation is of value if an inflammatory disorder is one of the differential diagnoses. A definitive diagnosis requires histopathological evaluation of cerebellar tissue.

Disease description in this species:
Cerebellar Purkinje cell degeneration or abiotrophy is the most common type of cerebellar cortical abiotrophy and the best described type of cerebellar degeneration in dogs. Both purebred and mixed breed dogs have been affected.

PATHOPHYSIOLOGY

The most common histopathological feature is degeneration and loss of Purkinje cells of varying degrees, depending on the chronicity of the degeneration. As the disease progresses, there is thinning of the molecular and granular cell layers.

CLINICAL SIGNS AND DIAGNOSTICS
Affected animals are normal at birth and develop signs of cerebellar dysfunction weeks after birth. Clinical signs reflect the loss of function of inhibitory cerebellar cortical neurons: head and body tremors, hypermetria, a broad base stance, truncal swaying, bunny hopping, ataxia, nystagmus, and decreased menace response.

A tentative diagnosis is based on signalment, progressive cerebellar signs, absence of systemic signs, and MRI findings of a smaller-than-normal cerebellum. Cerebrospinal fluid evaluation is of value if an inflammatory disorder is one of the differential diagnoses. Definitive diagnosis requires histopathological evaluation of cerebellar tissue. There is no treatment.

In the Italian Spinone, signs are first seen at 5 months of age. Signs include progressive tremor, hypermetria and ataxia; initially in hind limbs. Proceeds to death or euthanasia by 10-11 months.

**Etiology:**
Autosomal recessive defect with a tightly linked marker. The defective gene and mutation have not been identified.

Genetic, hereditary

**Breed predilection:**
Italian Spinone

**Age predilection:**
Juvenile

**Clinical findings:**
AFEBRILE
ANOREXIA, HYPOREXIA
ATAXIA, INCOORDINATION
Cachexia, weight loss
CENTRAL NERVOUS SYSTEM (CNS) SIGNS
Cerebellar signs
Disoriented
Dysmetria
Falling
GAIT ABNORMAL
Gait bunny hopping
Head tremors
Hypermetria
Intention tremors
Malaise
Menace response absent or decreased
Muscle fasciculations
NYSTAGMUS, EYE MOVEMENT RAPID
Signs progressive
Stance wide based
TREMORS
Walking difficulty
ZZZ INDEX ZZZ
Diagnostic procedures:  
Brain CT or MRI scan  
Biopsy and histopathology of the brain

Diagnostic results:  
Cerebellar size decreased  
Axonal degeneration  
Degeneration of caudate nuclei  
Degeneration of cerebellar granular layer  
Degeneration of cerebellar molecular layer  
Degeneration of cerebellar Purkinje layer  
Degeneration of substantia nigra

Linked-marker based genetic test

Treatment/Management/Prevention:  
SPECIFIC  
1) None  
SUPPORTIVE  
1) Keep affected animal on non-slick surfaces.  
2) Avoid stairwalking, swimming, and other high risk activities

Preventive Measures:  
1) Test relatives of affected dogs, or dogs at risk, and avoid breeding carrier dogs to each other.  
2) Replace carrier parents with normal testing offspring.

Differential Diagnosis:  
Cerebellar hypoplasia  
Storage diseases  
Canine distemper encephalitis  
Encephalitis  
Toxoplasmosis  
Neuroaxonal dystrophy  
Neoplasia  
Cystic structures

Mode of Inheritance  
Autosomal Recessive

Human Disease Homolog  
Autosomal recessive, juvenile-onset cerebellar ataxia – none linked

Available Tests/Testing Facilities  

OMIA: 000175-9615  OMIM: 213200

References:  