Cerebellar Abiotrophy

Affected breeds: Arabian, Bashkir Curly, Danish Sport, Trakehner, Welsh Pony.

Equine cerebellar abiotrophy (CA) is an inherited neurological condition and is characterized by the degeneration of a specific cell type in the brain called Purkinje cells; these cells play a fundamental role in controlling movement. CA foals are apparently normal at birth, but between the ages of 6 – 16 weeks develop signs of CA which include head tremor and a lack of balance. Consequently, affected foals may show a splayed stance in an attempt to balance themselves, and may fall and be unable to rise easily. The severity of the signs displayed by affected foals varies widely and symptoms can be confused with other conditions. Affected foals are often euthanized as they are unsafe to ride.

Research carried out at the Veterinary Genetics Laboratory in Davis, California has identified a mutation that is associated with CA. CA is found mainly in Arabian horses, but is also seen at a lower level in several other breeds including the Bashkir Curly Horse, Danish Sport Horse, Trakehners and Welsh ponies. The appearance of the defect in these breeds is due to Arabian ancestry; the CA test is therefore recommended for horses that have Arabian horses in their pedigree.

CA is inherited in a recessive manner. This means that carrier horses which have one copy of the defective gene appear healthy, but can pass this on to their offspring. The breeding of two carriers will produce CA-affected foals 25% of the time. The breeding of a carrier with a clear horse will not result in affected foals, though 50% of offspring will be carriers themselves. The CA test provided by Animal DNA Diagnostics Ltd can also be used to definitively diagnose foals suspected of having CA.

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 foals.
- To conclusively diagnose CA in sick foals.

This test will be reported as:

CLEAR : no evidence of the CA mutation
CARRIER : carries one copy of the mutation, which will be passed to 50% of offspring
AFFECTED : carries two copies of the mutation and will develop CA

Typical Breeding Outcomes:

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

References

Brault LS, Cooper CA, Famula TR, Murray JD, Penedo MC Mapping of equine cerebellar abiotrophy to ECA2 and identification of a potential causative mutation affecting expression of MUTYH. Genomics 97(2): 121-9.
Brault LS, Penedo MC The frequency of the equine cerebellar abiotrophy mutation in non-Arabian horse breeds. Equine Vet J. doi: 10.1111/j.2042-3306.2010.00349.x.