An introduction to the importance of ACAN testing for miniature Horses:-

Chondrodysplasia-like dwarfism in Miniature horses and Shetland ponies is a disproportionate type of dwarfism which involves malformations of the cartilage and subsequent maturation to bone during foetal development and subsequent growth of the individual during sexual maturation. This type of dwarfism is not considered a desirable trait due to its association with serious health issues including breathing problems, malformed mouth that causes eating difficulties, and abnormal bone growth leading to chronic health issues.

Disproportionate dwarfism involves the shortening and malformation of all the long bones of the body and causes abnormal growth of the other bones within the body such as the skill.

A recent research by John Eberth and Ernie Bailey identified four mutations in the ACAN gene which are associated with Chondrodysplasia-like dwarfism. The four mutations are termed D1, D2, D3 and D4.

Trait of Inheritance

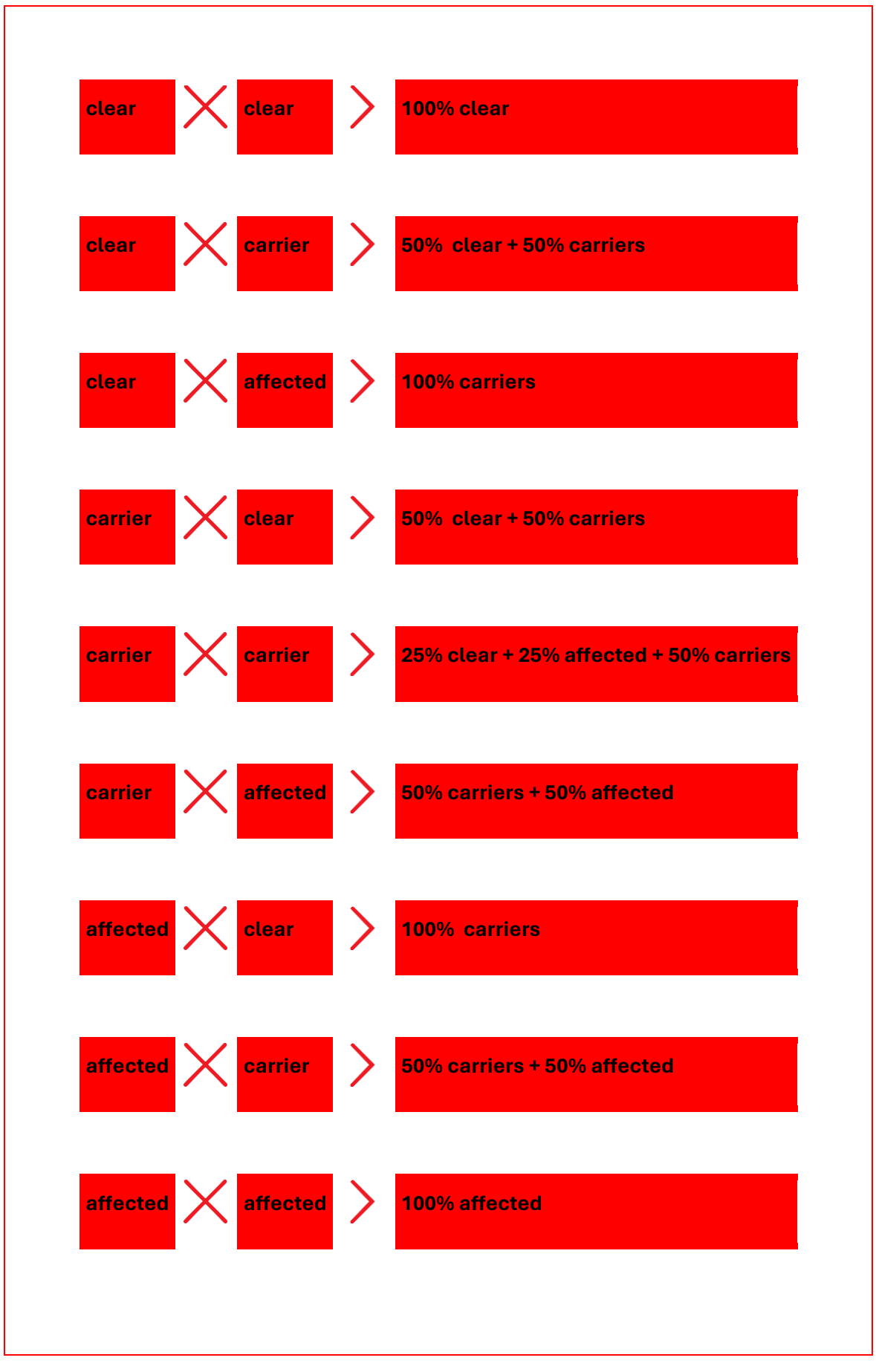
The disease follows an autosomal recessive trait of inheritance and therefore a horse with one copy of the mutation : N/D1, N/D2, N/D3 or N/D4 are clinically normal and have a normal appearance but they are genetically carriers, a carrier will pass the mutation to its offspring with a 50% probability.

The D1 mutation is considered lethal if combined with any other mutation: D1/D1, D1/D2, D1/D3 and D1/D4. These horses will die in the womb and will consequently be aborted or reabsorbed.

Dwarf horses have two copies of any mutation except D1, and therefore will have one of the following genotypes: D2/D2, D2/D3, D2/D4, D3/D4, and all have similar symptoms of dwarfism, which vary from mild to severe.

It is worth noting that the genotypes D3/D3 and D4/D4 have unknown effects, as no samples with these genotypes have been found so far.

Sire	Dam	Offspring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Clear

Genotype: N / N [Homozygous normal]

The horse is noncarrier of the mutant gene.

It is very unlikely that the horse will develop Dwarfism (Chondrodysplasia) ACAN. The horse will never pass the mutation to its offspring, and therefore it can be bred to any other horse.

Carrier

Genotype: N / d [Heterozygous]

The horse carries one copy of the mutant gene and one copy of the normal gene.

The horse is clinically normal and has a normal appearance but it is genetically a carrier, a carrier will pass the mutation to its offspring with a 50% probability.

Affected

Genotype: d / d [Homozygous mutant]

The horse carries two copies of the mutant gene and therefore it will pass the mutant gene to its entire offspring.

The horse will be dwarf (Chondrodysplasia) and will pass the mutant gene to its entire offspring

Further reading

Eberth, John E., 'Chondrodysplasia-Like Dwarfism in the Miniature Horse