

MYOPHOSPHORYLASE DEFICIENCY

The Clinical disease

Myophosphorylase is an enzyme that releases glucose from glycogen stored in muscle. In the absence of myophosphorylase activity, cattle are unable to rapidly mobilise glucose and muscles lose function, resulting in exercise intolerance. Affected calves tire easily and when driven may collapse, but recover after a period of rest. The condition progresses to such an extent that the animal is unable to move and is usually destroyed for humane reasons.

Mode of inheritance

Myophosphorylase deficiency is inherited as an autosomal recessive condition. Affected calves are those that inherit a mutant myophosphorylase gene (m) from each parent. The parents are clinically normal, but they carry one copy of the mutant gene (m) as well as one copy of the normal gene (M). These animals are known as carriers or heterozygotes (Mm). Fifty percent of the progeny of a heterozygote will be heterozygous (Mm). When two heterozygotes are mated, 25% of calves will be affected (mm), 50% will be heterozygotes (Mm) and 25% normal (MM).

Genotype

Scientific name	abbreviation	Common name
Homozygous wildtype	MM	Normal
Heterozygous	Mm	Carrier
Homozygous mutant	mm	Affected

Testing for myophosphorylase deficiency

The mutation responsible for myophosphorylase deficiency in Charolais cattle has been identified. A procedure to genotype cattle for myophosphorylase deficiency has been established at the Elizabeth Macarthur Agricultural Institute, Menangle. The test procedure utilises the awesome power of polymerase chain reaction (PCR) to amplify DNA from hair roots, blood or semen. The preferred sample is hair roots as they are simpler to collect, ship and process than blood.